

**EXPLORING GENETIC DISCRIMINATION EXPERIENCED
BY PARENTS OF CHILDREN WITH RARE DISEASES
IN MALAYSIA: A QUALITATIVE STUDY**

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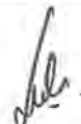
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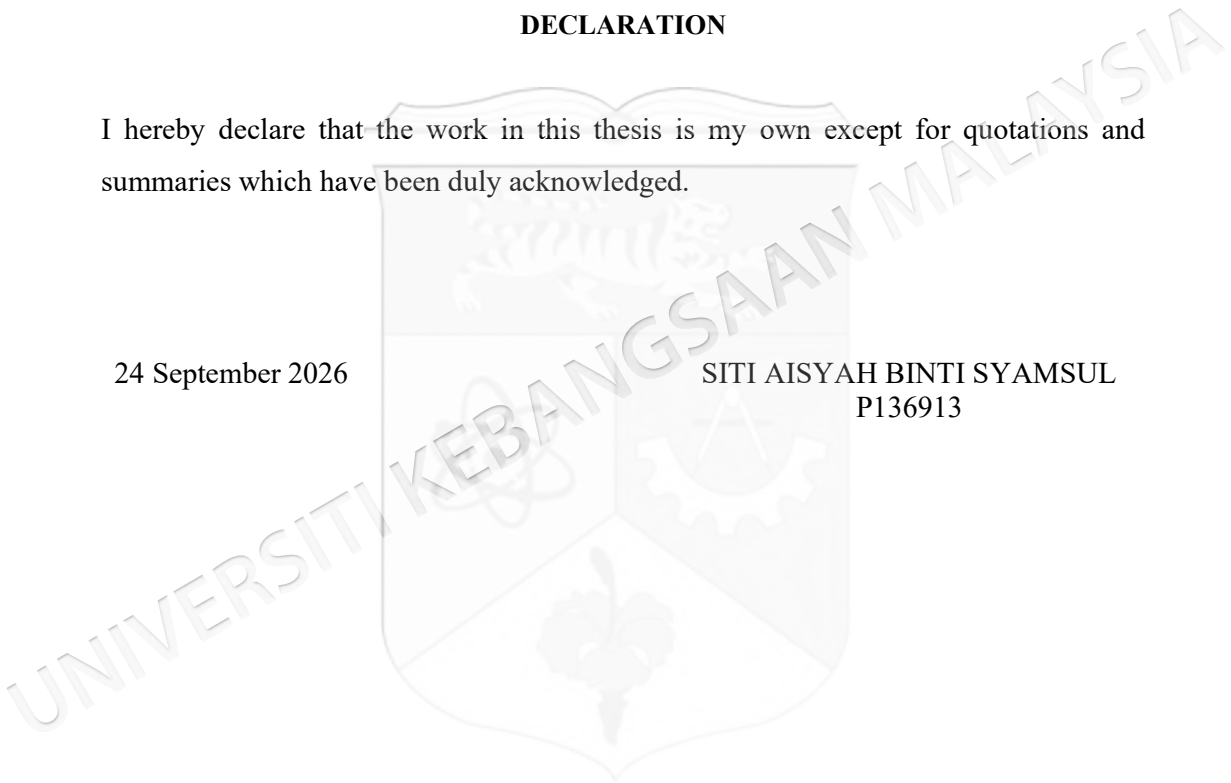
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DECLARATION

I hereby declare that the work in this thesis is my own except for quotations and summaries which have been duly acknowledged.

24 September 2026

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ABSTRAK

Latar Belakang

Kemajuan dalam ujian genetik telah meningkatkan keupayaan diagnosis dan pengurusan penyakit genetik jarang jumpa. Namun, kebimbangan mengenai diskriminasi genetik dan implikasi sosial yang berkaitan masih wujud. Kebanyakan kajian terdahulu memfokuskan kepada sindrom kanser keturunan dan ujian genetik prediktif, namun masih kurang pengetahuan tentang pengalaman keluarga lokal yang terjejas disebabkan penyakit genetik jarang jumpa. Kajian ini meneroka pengalaman diskriminasi genetik dalam kalangan ibu bapa kepada kanak-kanak dengan penyakit genetik jarang jumpa di Malaysia, termasuk konteks berlakunya diskriminasi, kesannya terhadap keluarga, strategi daya tindak, serta cadangan untuk meningkatkan sokongan.

Kaedah Kajian

Kajian ini menggunakan reka bentuk kualitatif. Temu bual separa berstruktur dijalankan dengan 15 orang ibu bapa kepada kanak-kanak yang didiagnosis dengan penyakit genetik jarang jumpa, yang kebanyakannya direkrut melalui Malaysia Rare Disease Society (MRDS). Data dianalisis menggunakan pendekatan analisis tematik refleksif oleh Braun dan Clarke. Lima tema utama telah dikenal pasti iaitu: (1) pengalaman diagnosis dan ujian genetik, (2) diskriminasi genetik, (3) impak emosi dan sosial, (4) strategi daya tindak, dan (5) cadangan serta keperluan yang tidak dipenuhi.

Hasil Kajian

Peserta menggambarkan proses mendapatkan diagnosis sebagai satu perjalanan yang mencabar dan sering melibatkan tempoh yang panjang tanpa jawapan yang jelas, manakala ujian genetik memainkan peranan penting dalam mengesahkan diagnosis. Peserta melaporkan pengalaman diskriminasi dan ketidakadilan dalam konteks penjagaan kesihatan, pekerjaan, insurans, pendidikan dan sosial. Halangan berkaitan insurans merupakan bentuk diskriminasi langsung yang paling jelas, manakala pengalaman lain termasuk kekurangan persekitaran pendidikan yang sesuai, kebimbangan tidak diberi perhatian sewajarnya dalam sistem penjagaan kesihatan, serta salah faham masyarakat terhadap penyakit genetik jarang jumpa. Begitu juga stigma berkaitan ketidakupayaan, ketidakadilan berdasarkan fenotip dan halangan struktur. Pengalaman tersebut menyumbang kepada tekanan emosi dan beban penjagaan, maka ibu bapa menggunakan pelbagai strategi daya tindak seperti advokasi, pendedahan maklumat secara terpilih dan sokongan sosial. Peserta turut menekankan keperluan kepada penjagaan bersepadu, peningkatan kesedaran dalam kalangan profesional dan masyarakat, serta perlindungan kewangan yang lebih kukuh untuk keluarga yang terjejas.

Kesimpulan

Pengalaman diskriminasi dalam kalangan keluarga yang terjejas oleh penyakit genetik jarang jumpa berkait rapat dengan realiti harian penjagaan anak dan cabaran berhadapan dengan sistem yang kurang bersedia untuk menyokong. Sebagai antara kajian kualitatif terawal mengenai diskriminasi genetik dalam kalangan keluarga penyakit genetik jarang jumpa di Malaysia, kajian ini menekankan keperluan untuk meningkatkan kesedaran, memastikan akses yang adil kepada perkhidmatan, serta memperkukuh perlindungan dasar, di samping menyokong pelaksanaan Dasar Penyakit Jarang Jumpa Kebangsaan Malaysia (2025).

ABSTRACT

Background

Advances in genetic testing have transformed the landscape of rare disease diagnosis and clinical management. Yet, alongside these benefits, concerns remain regarding genetic discrimination and the broader social consequences of living with a genetic condition. While existing research has focused largely on hereditary cancer syndromes and predictive genetic testing, little is known about how local families affected by rare genetic conditions experience discrimination in everyday life. This study explored the experiences of genetic discrimination among parents of children with rare genetic conditions in Malaysia, including the settings in which discrimination occurred, its impact, coping strategies and recommendations for improving support.

Methods

A qualitative study design was employed. Semi-structured interviews were conducted with 15 parents of children diagnosed with rare genetic conditions, primarily recruited through the Malaysia Rare Disease Society (MRDS). Data were analysed using Braun and Clarke's reflexive thematic analysis approach. Five major themes: 1) diagnosis and genetic testing experience, 2) genetic discrimination, 3) emotional and social impact, 4) coping strategies, 5) suggestions and unmet needs were identified.

Results

Participants frequently characterised diagnostic journey as challenging, often involving prolonged periods without clear answers, while genetic testing played a crucial role in establishing diagnosis. Participants reported experiences of discrimination and disadvantage across healthcare, employment, insurance, education and social settings. Insurance-related barriers represented the clearest form of direct discrimination, while experiences in other settings included limited educational accommodations, perceived dismissal of concerns within healthcare settings, and social misunderstanding of rare genetic conditions. Some experiences also reflecting disability-related stigma, phenotype-based disadvantage, and structural barriers. These experiences contributed to emotional distress and caregiver burden, while parents responded through advocacy, selective disclosure, and social support. Participants highlighted the need for integrated care, greater professional and public awareness, and stronger financial protection for families affected by rare genetic conditions.

Conclusion

The findings suggest that experiences of discrimination among rare disease families are closely intertwined with the everyday realities of caregiving and navigating systems that are inadequately prepared to support rare conditions. As one of the first qualitative studies to explore genetic discrimination among rare disease families in Malaysia, the findings highlight the need for greater awareness, equitable access to services, and stronger policy protections, while supporting the implementation of Malaysia's National Policy for Rare Diseases (2025).

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LIST OF ABBREVIATIONS

EL	Emergency Leave
GD	Genetic Discrimination
GINA	Genetic Information Nondiscrimination Act
MRDS	Malaysian Rare Disease Society
OKU	Orang Kurang Upaya
PDK	Program Pemulihan Dalam Komuniti
PPKI	Program Pendidikan Khas Integrasi
SMA	Spinal Muscular Atrophy
TABTAR	Tunku Ampuan Besar Tuanku Aishah Rohani Hospital
UNESCO	the United Nations Educational, Scientific and Cultural Organization
UKM	Universiti Kebangsaan Malaysia
WHO	World Health Organization



CHAPTER I

INTRODUCTION

1.1 RESEARCH BACKGROUND

Rare diseases are increasingly being acknowledged as an important public health concern worldwide despite their individual low prevalence. It is estimated that more than 300 million people to be living with one of over 7,000 identified rare diseases globally, with approximately 70–80% having a genetic origin (Nguengang Wakap et al. 2020). Advances in genomic medicine and the increasing availability of genetic testing technologies have substantially improved the diagnosis of these conditions by enabling earlier detection and more accurate disease characterization to better inform clinical management (Posey 2019; Marshall et al. 2020; Hong et al. 2024). As described by the World Health Organization, genomic technologies have the potential to transform healthcare to be more personalised approaches for disease prevention, diagnosis as well as treatment¹. The integration of genomics into routine healthcare has therefore expanded the attention beyond their clinical benefits where their ethical, legal and social implications are included (Stark et al. 2019).

One important concern is genetic discrimination generally refers to the differential treatment of individuals based on either their actual or perceived genetic characteristics rather than their current health status or abilities (Otlowski et al. 2012; Kim et al. 2021). Genetic discrimination may occur in many different settings, including insurance, employment, healthcare, education, and social interactions. In the context of

¹ World Health Organization. Human genomics in global health

rare genetic conditions, however, experiences of discrimination may be complex and may intersect with other forms of stigma, disadvantage, or differential treatment, particularly where the condition has physical, developmental, or functional manifestations (von der Lippe et al. 2017; Fournier et al. 2023). In addition to actual discriminatory experiences, there are also concerns regarding potential discrimination which may influence decisions about genetic testing in terms of disclosure of genetic information and engagement with healthcare services (Bombard et al. 2009; Tiller et al. 2024). The United Nations Educational, Scientific and Cultural Organization (UNESCO) recognized these concerns in the Universal Declaration on the Human Genome and Human Rights by stating that "no one shall be subjected to discrimination based on genetic characteristics"². The UNESCO International Declaration on Human Genetic Data has also similarly emphasised that there is need to protect individuals from inappropriate use of genetic information (UNESCO 2003).

Although several countries have been exploring genetic discrimination, existing research has largely focused on hereditary cancer syndromes, Huntington disease, and predictive genetic testing (Bombard et al. 2012; Gopalakrishnan et al. 2024). Parents of children with rare genetic diseases are an important group to study because they frequently interact with healthcare, school, insurance, and community systems on behalf of their children. They may therefore encounter different forms of discrimination and disadvantage associated with their child's condition. Comparatively little attention has been given to families affected by rare genetic diseases, particularly within Asian and middle-income country contexts. In Malaysia, awareness of rare diseases and access to genetic services have increased in recent years (Shafie et al. 2020), yet little is known about how families experience and interpret discrimination related to a genetic diagnosis.

Given that there is limited Malaysian evidence in this area, the need has become apparent to better understand how genetic discrimination is experienced by families affected by rare genetic diseases. In order to provide valuable insights into the broader social implications of genomic medicine and help inform genetic counselling practice,

² UNESCO. Universal Declaration on the Human Genome and Human Rights (1997)

healthcare service development, exploring these experiences may be aimed for future policy discussions to support affected individuals and their families. Therefore, this study seeks to explore the experiences of genetic discrimination among parents of children with rare genetic diseases in Malaysia through an in-depth qualitative approach.

1.2 PROBLEM STATEMENT

Genetic discrimination has emerged as an important ethical, legal, and social concern with the increasing use of genetic testing and genomic medicine (Otlowski et al. 2012; Kim et al. 2021). There were reports in previous studies on experiences of discrimination primarily in relation to insurance, employment, healthcare, and social interactions among individuals affected by hereditary conditions, particularly (Bombard et al. 2009; Bombard et al. 2012). However, relatively little attention given to families affected by rare genetic diseases as much of the existing evidence has focused on hereditary cancer syndromes and Huntington disease (Gopalakrishnan et al. 2024).

Existing research on rare disease has primarily focused on caregiver burden, emotional distress, healthcare challenges, and quality of life although its psychosocial impact has been increasingly recognized. Consequently, the understanding of whether parents of children with rare genetic diseases experience genetic discrimination and how such experiences may affect their lives, is still limited. This gap is particularly evident in Malaysia, where research examining genetic discrimination remains scarce despite growing access to genetic testing and increasing awareness of rare diseases. As primary caregivers and advocates, they may encounter various forms of discrimination, stigma, exclusion, or differential treatment associated with their child's diagnosis. While Malaysian studies have highlighted the challenges faced by rare disease families, including caregiving burden and difficulties navigating healthcare systems alongside psychosocial challenges (Ch'ng et al. 2022; Supian et al. 2024; Shukor et al. 2026), the specific issue of genetic discrimination remains largely unexplored. As a result, the nature, contexts, and impact of these experiences within the Malaysian setting remain poorly understood.

Therefore, the need to explore the experiences of genetic discrimination among parents of children with rare genetic diseases in Malaysia has remained. To address an important gap in the Malaysian literature, it is important to understand the forms and contexts in which these experiences occur, their emotional and social consequences before identifying the support needed by affected families. The findings of this study are expected to contribute to the growing knowledge on the ethical, legal, and social implications of genomic medicine to provide valuable insights for genetic counselling practice and healthcare services while supporting future initiatives to improve awareness, advocacy, and support for families living with rare genetic diseases in Malaysia.

1.3 RESEARCH QUESTIONS AND HYPOTHESES

1.3.1 Research Questions

1. What are the types of genetic discrimination experienced by the caregivers and patients with rare diseases in Malaysia?
2. In what settings does this discrimination most commonly occur?
3. What is the emotional and social impact on the affected individuals and families?
4. What protections or support do parents of rare disease children perceive as necessary?

1.3.2 Research Hypothesis

1. Families of children with rare diseases in Malaysia experience both enacted and perceived genetic discrimination.
2. Genetic discrimination among these families occurs most frequently in healthcare, education, employment, insurance and social settings.
3. Experiences of genetic discrimination led to emotional strain, social stigma and isolation for both caregivers and affected children.

4. Affected individuals and families perceive need for multifaceted protections and support, including legal protection, equitable access to healthcare and insurance, psychosocial support, public awareness, and inclusive service.

1.4 OBJECTIVE OF RESEARCH AND SCOPE OF WORKS

This study seeks to address this knowledge gap by exploring the experiences of parents of children with rare genetic diseases in Malaysia through an in-depth qualitative approach. The aim of this study is to understand the forms and contexts of genetic discrimination, its emotional and social consequences followed by identifying the support needed by affected families. The scope of the study encompasses participants' experiences across multiple settings, including healthcare services, educational institutions, workplaces, insurance matters as well social environments. In addition, the study explores the coping strategies adopted by families and their recommendations for improving support systems and services.

1.4.1 General objective

To explore the lived experiences of genetic discrimination among the parents of children with rare genetic conditions in Malaysia.

1.4.2 Specific objective

The specific objectives of this study are:

1. To identify the settings where genetic discrimination commonly occurs, including employment of the parents, schooling of the children, insurance coverage, healthcare and social settings.
2. To understand how individuals interpret the act of discrimination, affected by, respond to and cope with such experiences.
3. To gather suggestions from parents of affected children on how discrimination can be reduced through support and policy.

1.5 STUDY JUSTIFICATION

As genomic medicine becomes increasingly integrated into healthcare, greater attention is needed to ensure that individuals and families benefit from genetic advances without experiencing unintended social disadvantages. The challenges may extend beyond medical management to include barriers encountered within healthcare, education, insurance, employment, and community settings for families affected by rare genetic conditions. As these experiences is important because they may influence wellbeing, access to services and participation in society, it is crucial to make effort on understanding them. By exploring the perspectives of parents of children with rare genetic diseases, this study provides insights into an underexplored aspect of rare disease care in Malaysia. Through this study, evidence supporting more inclusive healthcare practices and improved support services may be generated to inform better policy development for affected families.

CHAPTER II

LITERATURE REVIEW

2.1 RARE DISEASES AND GENOMIC MEDICINE

Rare diseases encompass a heterogeneous group of conditions that each affect only a small proportion of the population, yet together they contribute substantially to the global burden of disease. Although definitions vary across countries, rare diseases are generally characterized by their low prevalence and are frequently chronic, progressive, debilitating, or life-threatening in nature (Nguengang Wakap et al. 2020). It is estimated that more than 300 million people worldwide live with over 6,000 recognised rare diseases, underscoring their considerable cumulative impact on health systems and society (Nguengang Wakap et al. 2020). Importantly, approximately 70–80% of rare diseases have a genetic basis, making advances in genetics and genomics particularly relevant to their diagnosis and management (Posey 2019). In Malaysia, rare diseases are increasingly acknowledged as a public health issue because of their long-term care needs, complex management, and effects on affected individuals and families (Shafie et al. 2020).

A major challenge associated with rare diseases is the difficulty of obtaining an accurate diagnosis. Many rare conditions present with non-specific clinical features, marked phenotypic variability, or symptoms that overlap with more common disorders, which makes diagnostic identification particularly difficult. Consequently, patients and families often experience a prolonged diagnostic journey involving multiple healthcare consultations, referrals, investigations, and inconclusive results before a definitive diagnosis is established (Zurynski et al. 2017). Delayed diagnosis can limit access to appropriate treatment, supportive services, and reliable information about prognosis and disease management. The prolonged uncertainty associated with this process has

become widely recognized as a hallmark experience of many rare disease families (Han et al. 2017).

Recent developments in genomic medicine have changed the way rare genetic conditions are diagnosed. Next-generation sequencing technologies, including whole-exome sequencing and whole-genome sequencing, have greatly improved the identification of disease-causing variants and made molecular diagnoses possible in cases where this was previously not achievable (Marshall et al. 2020; Vinkšiel et al. 2021). These technologies have also supported the wider growth of precision medicine, in which prevention, diagnosis, and treatment are adapted to an individual's genetic profile (Stark et al. 2019; Hong et al. 2024). For people suspected of having rare genetic conditions, genomic testing has become an increasingly valuable clinical tool because it can support earlier diagnosis, provide more accurate prognostic information, and improve healthcare decision-making (Marwaha, Knowles & Ashley 2022).

Nevertheless, genetic diagnoses do not always remove uncertainty. Testing may identify novel variants, variants of uncertain significance, or conditions for which little is known about natural history and treatment outcomes. Han et al. (2017) proposed that uncertainty within genomic medicine extends beyond scientific uncertainty regarding genetic findings to include personal uncertainty about future health outcomes and practical uncertainty relating to healthcare and life decisions. Nevertheless, the increasing integration of genomic technologies into healthcare has fundamentally changed how rare diseases are diagnosed and managed, resulting in greater reliance on genetic testing and specialist genetic services. For this reason, understanding genetic counselling and genetic testing is essential to understanding the experiences of individuals and families affected by rare genetic conditions.

2.2 GENETIC COUNSELLING AND GENETIC TESTING

The rapid development of genomic medicine has transformed the role of genetics within healthcare and increased the importance of genetic counselling and genetic testing in clinical practice. Genetic counselling is a communication process that aims to help individuals and families understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease (Resta et al. 2006).

Contemporary genetic counselling has encompassed a broader range of activities, including facilitating informed decision-making, providing psychosocial support, promoting adaptation to genetic conditions as well as supporting communication within families while traditionally associated with risk assessment and the provision of genetic information (McCarthy Veach, LeRoy & Callanan 2018). As genomic technologies become increasingly integrated into routine healthcare, genetic counsellors play a critical role in helping patients navigate complex genetic information and its implications.

Advances in genomic technologies, especially next-generation sequencing, have made it possible to identify disease-causing variants with much greater speed and accuracy (Marshall et al. 2020; Vinkšiel et al. 2021). Genetic testing has become a crucial part of diagnosing and managing rare diseases. For people suspected of having rare genetic conditions, genetic testing may confirm a diagnosis, improve access to appropriate management and guide reproductive decisions while reducing uncertainty about the cause of the disease (Marwaha, Knowles & Ashley 2022). The wider availability of whole-exome sequencing and whole-genome sequencing has also broadened diagnostic possibilities, particularly for those who remained undiagnosed despite extensive clinical workups (Posey 2019; Hong et al. 2024). As a result, genetic testing is now widely viewed as a key part of precision medicine, where healthcare decisions are tailored to a person's genetic profile (Stark et al. 2019).

Despite its clinical benefits, genetic testing may also generate significant psychological and emotional challenges for individuals and families. The process of undergoing testing and receiving results can evoke a wide range of responses, including relief, anxiety, uncertainty, fear, and concerns regarding future health outcomes (McCarthy Veach, LeRoy & Callanan 2018). Where genetic findings may be difficult to interpret, such challenges may be particularly pronounced in rare disease settings due to poorly understood disease progression and treatment options may be limited. Han et al. (2017) argued that uncertainty in genomic medicine extends beyond scientific uncertainty regarding genetic findings and includes personal uncertainty relating to future outcomes as well as practical uncertainty surrounding healthcare decisions and everyday life. These uncertainties often persist even after a diagnosis has been

established, highlighting the importance of ongoing support throughout the genetic testing process.

In response to these challenges, genetic counsellors play an important role in supporting individuals and families before, during, and after genetic testing. The Accreditation Council for Genetic Counselling emphasises competencies related to communication, psychosocial assessment, ethical practice, patient advocacy, and facilitation of informed decision-making (ACGC 2023). In rare disease settings, genetic counsellors may assist families in understanding test results, coping with uncertainty, adapting to a diagnosis, and accessing relevant healthcare and community resources. Effective communication between healthcare professionals and families has also been identified as an important factor influencing trust, satisfaction, and adaptation following diagnosis (Gómez-Zúñiga et al. 2019). As such, genetic counselling represents an important component of patient-centred care within genomic medicine.

Unlike many other forms of medical information, genetic information may reveal future health risks, family relationships, reproductive risks, and inherited conditions. Hence, the growing use of genetic testing has also raised broader ethical, legal, and social questions about how genetic information is used and interpreted. This has led to concerns about privacy, confidentiality, informed consent, and the possible misuse of genetic information by institutions and society (Ascencio-Carbajal et al. 2021). These concerns have helped increase interest in the idea of genetic discrimination, especially as genetic information becomes more deeply integrated into healthcare systems and everyday life. Understanding these issues is important for appreciating the wider implications of genetic testing beyond its clinical value and provides the basis for the next discussion on genetic discrimination.

2.3 GENETIC DISCRIMINATION AS A CONCEPT

The increasing use of genetic testing has generated growing interest in the concept of genetic discrimination and its potential implications for individuals and families. As advances in genomic medicine enable the identification of genetic variants associated with disease susceptibility, carrier status, and inherited conditions, concerns have emerged regarding how such information may be used by employers, insurers,

healthcare providers, educational institutions, and society more broadly (Otlowski, Taylor & Bombard 2012). Unlike many other forms of health information, genetic information may reveal not only an individual's current health status but also future health risks and information relevant to biological relatives. Consequently, the possibility that individuals could experience disadvantage or unequal treatment because of their genetic characteristics has become an important ethical, legal, and social concern within genomic medicine (Kim et al. 2021).

Even though the topic has been widely discussed in academic and policy literature, there is still no universal definition of genetic discrimination. In general terms, genetic discrimination refers to unfair or unequal treatment based on a person's actual or perceived genetic characteristics (Otlowski, Taylor & Bombard 2012). Early discussions were largely driven by concerns that genetic information could be used to deny employment opportunities or insurance coverage to people identified as being at increased risk of future disease (Joly, Braker & Huynh 2010). For that reason, early literature mainly framed genetic discrimination as misuse of predictive genetic information. More recent scholarship, however, has recognised that genetic discrimination may occur in a wider range of settings and may involve subtler forms of disadvantage than initially expected (Bombard et al. 2012).

Several conceptual distinctions have been introduced to better explain the complexity of genetic discrimination. One commonly used distinction is between enacted discrimination and perceived discrimination. Enacted discrimination refers to actual experiences of unfair treatment that can be linked to genetic information, while perceived discrimination refers to a person's belief or concern that they have been treated differently because of a genetic condition or genetic risk (Bombard et al. 2012). Researchers have also distinguished between direct discrimination, where genetic information is explicitly used in decision-making, and indirect discrimination, where policies, procedures, or institutional practices disproportionately disadvantage people with genetic conditions even without clear discriminatory intent (Kim et al. 2021). These distinctions matter because disadvantage can occur even when discrimination is subtle, unintentional, or built into wider systems and structures.

The emergence of genetic discrimination as a policy concern has led to the development of various ethical and human rights frameworks intended to protect individuals from misuse of genetic information. The UNESCO Universal Declaration on the Human Genome and Human Rights emphasises that no individual should be subjected to discrimination based on genetic characteristics and highlights the importance of protecting human dignity and fundamental rights (UNESCO 1997). Similarly, the UNESCO International Declaration on Human Genetic Data underscores the need to ensure appropriate safeguards regarding the collection, storage, and use of genetic information (UNESCO 2003). These principles reflect broader concerns regarding privacy, confidentiality, autonomy, and fairness in the application of genetic technologies (Ascencio-Carbajal et al. 2021). More recently, scholars have argued that protection against genetic discrimination should be considered within a broader human rights framework that promotes equity and social inclusion for individuals affected by genetic conditions (Rud et al. 2026).

While concerns regarding genetic discrimination traditionally centred on asymptomatic individuals undergoing predictive genetic testing, recent literature suggests that the concept may need to be interpreted more broadly. Advances in genomic medicine have increased the visibility of genetic conditions and expanded the contexts in which genetic information is used, raising questions regarding how disadvantage may be shaped not only by the misuse of genetic information but also by clinical manifestations, functional limitations, stigma, social attitudes, and structural barriers associated with genetic conditions (Kim et al. 2021). Such perspectives have contributed to ongoing debates regarding the boundaries of genetic discrimination and whether existing definitions adequately capture the experiences of individuals and families affected by genetic conditions. Understanding these evolving perspectives is important for interpreting contemporary research on genetic discrimination and provides the foundation for examining the evidence reported across different populations and settings.

2.4 INTERNATIONAL EVIDENCE ON GENETIC DISCRIMINATION

Research on genetic discrimination has grown considerably over the past two decades, largely because of increasing concern about the wider availability of genetic testing and genomic information. Much of the early work focused on individuals at risk of hereditary conditions who underwent predictive testing, especially Huntington disease. Huntington disease became a common focus because predictive testing allows asymptomatic individuals to learn whether they carry a pathogenic variant associated with future disease onset. This raised concern that people might face adverse treatment despite not yet being clinically affected. In one of the largest studies of individuals at risk of Huntington disease, Bombard et al. (2009) found that participants perceived genetic discrimination in several areas, including insurance, family relationships, social interactions, and healthcare. Later studies showed that discrimination extended beyond formal institutions and often involved interpersonal and social consequences, highlighting the broader effect of genetic information on everyday life (Bombard et al. 2012).

As genetic testing became increasingly available for other hereditary conditions, research attention expanded to populations affected by hereditary cancer syndromes such as hereditary breast and ovarian cancer syndrome and Lynch syndrome. Similar concerns emerged regarding the potential use of genetic information by employers, insurers, and other organizations. Recent studies have shown that individuals with hereditary cancer syndromes continue to report concerns regarding disclosure of genetic information and fears of discrimination despite the existence of legal protections in some jurisdictions (Gopalakrishnan et al. 2024). Participants in these studies described uncertainty regarding whether to disclose genetic information to employers, insurers, and even healthcare providers due to concerns about potential negative consequences. These findings suggest that the perceived risk of discrimination may influence behaviour even when actual discriminatory events are not always evident.

Insurance has consistently emerged as one of the most frequently reported areas of concern within the genetic discrimination literature. Concerns regarding access to life, disability, and health insurance were among the primary factors motivating

discussions about genetic discrimination internationally (Joly, Braker & Huynh 2010). A systematic review conducted by Joly, Ngueng Feze and Simard (2013) found that insurance-related discrimination represented one of the most commonly reported forms of genetic discrimination across multiple countries. More recently, studies from Australia demonstrated that concerns regarding the use of genetic information in life insurance continue to persist despite the introduction of industry-led regulatory measures intended to limit such practices (Tiller et al. 2021; Tiller et al. 2024). These findings indicate that insurance remains a particularly sensitive area in discussions regarding the fair use of genetic information.

Employment discrimination among affected individuals has also received substantial attention, although the evidence on its prevalence has been less consistent than for insurance-related concerns. People undergoing predictive genetic testing have expressed concern that genetic information could affect hiring, promotion, workplace accommodations, and perceptions of future productivity (Bombard et al. 2009; Otlowski, Taylor & Bombard 2012). However, several studies have found that fears of employment discrimination are often more common than documented cases of actual discrimination (Wauters & Van Hoyweghen 2016). This distinction shows the importance of considering both actual experiences and perceived risks when evaluating the effects of genetic discrimination. Whether discrimination actually occurs or not, concern about it may influence people's willingness to undergo testing, disclose results, or take part in research. On the other hand, less attention has been given to the employment experiences of family members and parents of individuals with genetic or rare diseases. Existing research indicates that caregiving responsibilities can substantially affect parents' employment, with parents reporting reduced working hours, withdrawal from paid employment, and other changes to their working lives (Pelentsov et al. 2016; Baumbusch, Mayer & Sloan-Yip 2018). A scoping review similarly found that caregivers of children with rare diseases may reduce working hours, leave employment, decline career opportunities, take leave, or change jobs, with some reporting insufficient employer support for balancing work and caregiving responsibilities (Buckle et al. 2024). Importantly, research on genetic disease has also raised concerns that being known as a caregiver of an affected family member may itself

create vulnerability to workplace discrimination, including through employment gaps resulting from caregiving responsibilities.

While insurance and employment have traditionally dominated discussions of genetic discrimination, recent scholarship suggests that discriminatory experiences may occur in a wider range of contexts. Kim et al. (2021) argued that existing conceptualizations of genetic discrimination have been largely shaped by Western legal and healthcare frameworks and may not fully capture experiences in other cultural contexts. Furthermore, emerging evidence indicates that individuals with genetic conditions may encounter disadvantage through healthcare interactions, educational systems, social relationships, and broader institutional practices (Gopalakrishnan et al. 2024). These findings have contributed to a growing recognition that genetic discrimination may extend beyond the misuse of genetic information alone and may involve broader forms of stigma, exclusion, and structural disadvantage. Despite these developments, comparatively little research has explored such experiences among individuals and families affected by rare genetic conditions, particularly within Asian settings. As a result, important gaps remain in understanding how genetic discrimination is experienced outside the populations and contexts that have traditionally dominated the literature.

2.5 THEORETICAL CONCEPTS ON DISCRIMINATION AND STIGMA

Understanding genetic discrimination requires consideration of broader theories relating to stigma, social exclusion, and responses to perceived disadvantage. While genetic discrimination is often discussed in relation to the misuse of genetic information, researchers have increasingly recognized that experiences of disadvantage may also be influenced by social attitudes, cultural beliefs, and societal perceptions of difference (Pachankis et al. 2018; Baynam, Gomez & Jain 2024). Consequently, concepts such as stigma and social exclusion have become important theoretical frameworks for understanding the experiences of individuals and families affected by genetic conditions.

Stigma refers to the process by which certain characteristics become socially discredited, leading to negative stereotypes, prejudice, or unequal treatment of affected

individuals (Pachankis et al. 2018). Shakespeare (2017) explains that stigma may arise when people are viewed as different from social norms and may then experience exclusion or disadvantage. In the context of genetic conditions, stigma may occur because these conditions are visible, poorly understood, inherited, or linked to assumptions about disability, dependency, or reduced capability (Baynam, Gomez & Jain 2024). Although stigma and discrimination are different concepts, stigma is often seen as an important step towards discriminatory behaviour because negative perceptions may shape how people are treated in healthcare, education, work, and social settings.

While genetic discrimination and genetic stigma are conceptually distinct, the two phenomena frequently overlap in practice. Individuals affected by genetic conditions may experience disadvantage not only through the direct use of genetic information but also through negative stereotypes, social judgement, exclusion, and structural barriers associated with their condition. Consequently, several scholars have argued that understanding the lived experiences of affected individuals requires consideration of both discrimination and stigma-related processes (Kim et al. 2021; Baynam, Gomez & Jain 2024; Rud et al. 2026). This perspective is particularly relevant for rare genetic conditions, where experiences of disadvantage may arise from the condition itself, its visible manifestations, or societal responses to rarity rather than solely from the disclosure or use of genetic information.

Closely related to these ideas is social exclusion, which refers to the processes that prevent individuals or groups from participating fully in social, economic, educational, or community life (Shakespeare 2017). For people living with rare diseases, social exclusion may result from physical limitations, inaccessible environments, limited accommodations, or misunderstanding of the condition. Research has shown that people affected by rare diseases often report isolation, marginalisation, and fewer opportunities for social participation than their peers (Zhu, Smith & Parrott 2017). These experiences may happen even when there is no deliberate discrimination and may instead reflect broader structural barriers in society.

Another important concept within the literature is concealment and disclosure. Because genetic information may be invisible or not immediately apparent to others, individuals and families often face decisions regarding whether, when, and to whom genetic information should be disclosed. Disclosure may facilitate access to support, accommodations, and healthcare services, but may also increase vulnerability to stigma and discrimination (Bombard et al. 2009; Gopalakrishnan et al. 2024). Conversely, concealment may protect individuals from negative treatment but may also create psychological stress and limit opportunities to access support. These considerations are particularly relevant in genetic conditions where family members may be concerned about how disclosure could affect social relationships, educational opportunities, employment prospects, or insurance access.

Social support has consistently been identified as an important protective factor for individuals and families affected by rare diseases. Peer support groups, patient organisations, and rare disease communities provide opportunities for information sharing, emotional support, and collective advocacy (Joo et al. 2022). Participation in these support networks has been associated with reduced isolation and better adaptation among people living with rare conditions (Fendrich et al. 2025). As a result, current understandings of discrimination and stigma recognise not only the factors that create disadvantage, but also the resources and coping strategies that help individuals and families manage these experiences. Together, these theoretical ideas provide a useful framework for understanding how genetic discrimination may be experienced, interpreted, and responded to by families affected by rare genetic conditions.

2.6 IMPACT OF RARE DISEASES ON FAMILIES

Rare diseases often affect not only the individual diagnosed but also the wider family unit. Due to the chronic, complex, and frequently lifelong nature of many rare genetic conditions, parents and family members commonly assume substantial caregiving responsibilities that extend beyond routine parenting roles. These responsibilities may include coordinating healthcare appointments, managing treatments, monitoring symptoms, advocating for services, and providing ongoing emotional and practical support. Consequently, the impact of rare diseases is frequently experienced at the

family level, influencing emotional wellbeing, family relationships, social participation, employment, and financial stability (von der Lippe, Diesen & Feragen 2017; von der Lippe, Neteland & Feragen 2022).

A substantial amount of research has shown the psychological burden experienced by parents of children with rare diseases. Caregivers often report high levels of stress, anxiety, uncertainty, and emotional exhaustion linked to long-term caregiving responsibilities (Pelentsov, Laws & Esterman 2015; Boettcher et al. 2021). These emotional difficulties may begin during the diagnostic process and continue throughout the child's life as families adapt to changing healthcare needs and uncertain future outcomes. Parents frequently express concerns about disease progression, treatment effectiveness, future independence, educational opportunities, and long-term quality of life (Davidson et al. 2024). Systematic reviews have also shown that caregivers of children with rare diseases often have poorer psychological wellbeing and lower quality of life than the general population (Boettcher et al. 2021; von der Lippe, Neteland & Feragen 2022).

The impact of rare diseases also extends to practical and socioeconomic aspects of family life. Families frequently encounter challenges navigating fragmented healthcare systems, accessing specialist services, and coordinating multidisciplinary care (Baumbusch, Mayer & Sloan-Yip 2018). Many caregivers report reducing work hours, changing employment arrangements, or leaving the workforce altogether to accommodate caregiving responsibilities, resulting in financial strain and reduced household income (Currie & Szabo 2019). Additional expenses related to healthcare, therapies, transportation, specialized equipment, and educational support may further increase the economic burden experienced by affected families (Domaradzki, Jabkowski & Walkowiak 2025). Such challenges demonstrate that the consequences of rare diseases often extend beyond medical management and affect multiple dimensions of daily life.

Importantly, researchers increasingly recognize that the burden experienced by rare disease families may not come solely from the medical condition itself. Social stigma, misunderstanding, limited awareness, inadequate accommodations, and

difficulty accessing appropriate support services may further contribute to family distress and lower quality of life (Zhu, Smith & Parrott 2017; Baynam, Gomez & Jain 2024). Families may feel isolated, excluded, or marginalised when their needs are not properly understood or accommodated in healthcare, education, work, or community settings. For this reason, the impact of rare diseases is increasingly understood as the result of interactions between disease-related challenges and broader social and structural factors shaping the experiences of affected people and their families (Shakespeare 2017).

Within Malaysia, similar challenges have been reported among patients and caregivers affected by rare diseases. Ch'ng et al. (2022) found that spinal muscular atrophy affected multiple aspects of life, including emotional wellbeing, education, employment, social participation, and financial stability. Likewise, Supian et al. (2024) reported that Malaysian patients and caregivers frequently experienced difficulties related to healthcare access, service coordination, and obtaining adequate support. Collectively, these findings suggest that the burden of rare diseases extends far beyond the clinical manifestations of the condition and reflects a complex interaction between medical, psychosocial, financial, and societal factors. Understanding these broader impacts is important for appreciating the lived experiences of rare disease families and the contexts in which experiences of discrimination and disadvantage may occur.

2.7 MALAYSIAN CONTEXT AND RESEARCH GAP

Rare diseases have received increasing attention in Malaysia in recent years, reflecting growing recognition of the challenges faced by affected individuals and their families. Despite advances in genomic medicine and expanding access to genetic services, rare disease care in Malaysia continues to face several obstacles, including delayed diagnosis, limited specialist expertise, fragmented service delivery, and financial burden associated with long-term management (Shafie et al. 2020). To address these challenges, efforts have been made to strengthen rare disease services through improved policy development, healthcare planning, and patient advocacy initiatives. Nevertheless, access to specialized services and support remains uneven, particularly

for families requiring multidisciplinary care and long-term follow-up (Supian et al. 2024).

Existing Malaysian research has mainly focused on the psychosocial and practical challenges experienced by individuals and families affected by rare diseases. In a mixed-method study involving patients with spinal muscular atrophy and their caregivers, Ch'ng et al. (2022) reported substantial effects on emotional wellbeing, education, employment, financial stability, and social participation. Similarly, Supian et al. (2024) found that patients and caregivers often had difficulties navigating healthcare services, obtaining suitable support, and coordinating care across multiple providers. More recently, Shukor, Norhayati and Ahmad (2026) reported that Malaysian parents caring for children with rare diseases commonly experienced emotional distress, caregiver burden, uncertainty about the future, and reliance on different coping strategies to manage these challenges. Taken together, these studies show the considerable effect of rare diseases on Malaysian families and the need for holistic support that goes beyond medical management alone.

Alongside healthcare challenges, Malaysia has also made efforts to improve educational inclusion for children with disabilities and special educational needs through initiatives such as the Zero Reject Policy introduced by the Ministry of Education. This policy aims to ensure that children with disabilities can access formal education regardless of their functional limitations or health conditions (Ensimau, Hamzah & Yassin 2022). However, evidence suggests that children with complex health needs and rare conditions may still face barriers related to educational accommodation, accessibility, and coordination between healthcare and educational services (UNICEF 2014; Verger et al. 2021). These observations underline the importance of examining how wider social systems respond to the needs of people affected by rare diseases.

Despite growing research on rare disease experiences in Malaysia, an important gap remains within the existing literature. Comparatively little attention has been given to experiences of genetic discrimination, particularly among parents of children with rare genetic conditions. This is because previous Malaysian studies have predominantly

focused on caregiver burden, healthcare experiences, psychosocial wellbeing, quality of life, and service needs among patients and families affected by rare diseases (Ch'ng et al. 2022; Supian et al. 2024; Shukor, Norhayati & Ahmad 2026). Furthermore, much of the international literature on genetic discrimination has focused on hereditary cancer syndromes, Huntington disease, and predictive genetic testing populations (Bombard et al. 2009; Bombard et al. 2012; Gopalakrishnan et al. 2024), with limited exploration of how discrimination may be experienced within the context of rare diseases.

This gap is particularly important because the experiences of families affected by rare genetic conditions may differ substantially from those reported in traditional genetic discrimination research. Families may encounter disadvantage through healthcare interactions, educational systems, employment challenges, insurance access, social stigma, and structural barriers associated with rarity itself, rather than concerns arising solely from predictive genetic information. Moreover, little is known about how such experiences are interpreted and navigated within the Malaysian sociocultural and healthcare context. Therefore, this study seeks to address this gap by exploring the experiences of genetic discrimination among parents of children with rare genetic conditions in Malaysia, with the aim of generating evidence that may inform genetic counselling practice, healthcare service development, and future policy initiatives.

CHAPTER III

METHODOLOGY

3.1 INTRODUCTION

This chapter describes the methodology employed to explore the experiences of genetic discrimination among parents of children with rare genetic diseases in Malaysia. It outlines the research design, study setting, participant selection criteria, sample size determination, development of research instruments, interview procedures, data analysis and ethical considerations. The methodological approach was carefully selected to facilitate an in-depth understanding of parents' lived experiences, perceptions, and support needs related to actual or perceived genetic discrimination within the Malaysian context. Given the limited evidence on genetic discrimination among families affected by rare genetic diseases in Malaysia, a qualitative approach was considered appropriate to capture the complexity and contextual nature of participants' experiences.

3.2 RESEARCH DESIGN

This study employed an exploratory qualitative research design using semi-structured individual interviews. Semi-structured interviews were selected as the primary method of data collection due to their flexibility in exploring sensitive topics while ensuring consistency across interviews.

Participants were recruited through purposive sampling using two recruitment pathways: collaboration with the Malaysia Rare Disease Society (MRDS) and referrals from the Genetics Clinic at Tunku Ampuan Besar Tuanku Aishah Rohani Hospital (TABTAR), UKM Children's Specialist Hospital. Eligible participants were parents of children diagnosed with rare genetic conditions who met the study inclusion criteria.

An interview guide was developed based on the literature review and study objectives to facilitate discussion while allowing participants the flexibility to elaborate on issues they considered important. The interview questions were drafted in Malay and organised thematically according to the anticipated areas of inquiry relevant to the study objectives. The interview guide was subsequently pilot tested with five parents who met the study inclusion criteria to assess the clarity, comprehensibility, and flow of the questions. Feedback obtained during the pilot interviews, together with the researcher's reflections on the interview process, was used to refine the wording, sequencing, and probing questions within the guide. Data collected during the pilot study were not included in the final analysis.

Following refinement of the interview guide, semi-structured interviews were conducted either face-to-face at TABTAR Hospital or via an online platform, depending on participants' preferences and availability. All interviews were conducted in Malay and lasted approximately 45 to 90 minutes. Open-ended questions and appropriate probing techniques were employed to encourage participants to provide detailed accounts of their experiences and perspectives. With participants' informed consent, all interviews were audio-recorded.

The audio recordings were transcribed verbatim using online transcription software called AudiotoText. The resulting transcripts were subsequently verified for accuracy by comparing them against the original audio recordings. All transcripts were pseudonymised to protect participant confidentiality, and the audio recordings were permanently deleted following verification. Data analysis was conducted using ATLAS.ti version 24 following Braun and Clarke's (2006) six-phase thematic analysis approach. Initial codes were generated systematically from the transcripts, followed by the development, review, and refinement of themes. To enhance the trustworthiness of the findings, peer debriefing was conducted with research team to confirm the accuracy of interpretations and emerging themes. Finally, the findings were reported using illustrative participant quotations to support and contextualise each theme identified.

Figure 3.1 shows an overview of the research design.

Figure 3.1 Overview of the research design

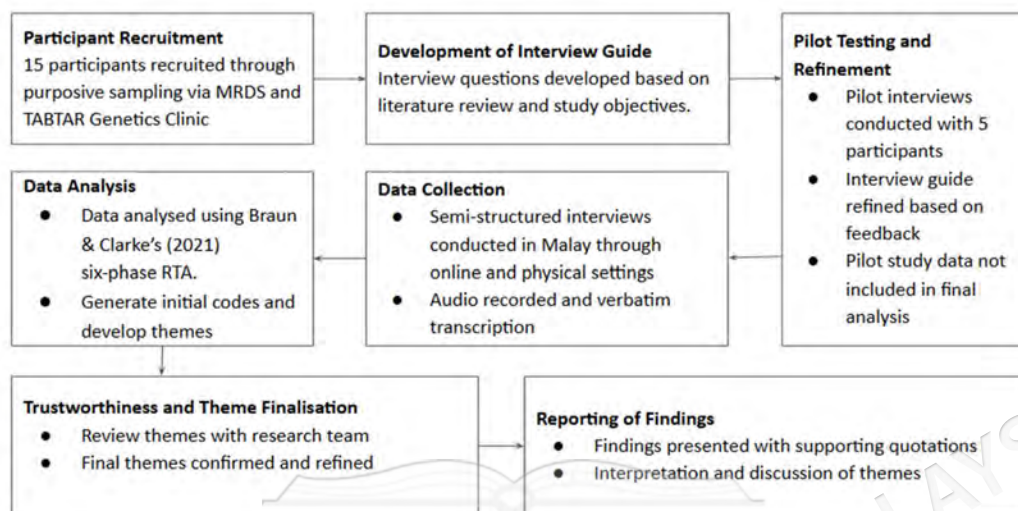


Figure 3.1 Flowchart illustrating the research designs, from participant recruitment to reporting of findings.

3.3 STUDY SETTING

This study was conducted in Malaysia and involved parents of children diagnosed with rare genetic diseases. Participants were recruited through the Malaysia Rare Disease Society (MRDS) and during regular clinic visits at the Genetics Clinic, Tunku Ampuan Besar Tuanku Aishah Rohani Hospital (TABTAR), UKM Children's Specialist Hospital. These recruitment pathways enabled access to parents and caregivers of children with a range of rare genetic conditions residing in different regions of Malaysia.

Data collection was conducted through semi-structured interviews using either face-to-face or online modalities based on participants' preferences and availability. Face-to-face interviews were conducted at TABTAR, UKM Children's Specialist Hospital, while online interviews were conducted via Google Meet. The use of both interview formats facilitated participation from families across different geographical locations while accommodating participants' caregiving responsibilities and logistical constraints.

3.4 INCLUSION AND EXCLUSION CRITERIA

3.4.1 Inclusion Criteria

Participants were eligible for inclusion in this study if they met the following criteria:

1. Parents of a child diagnosed with a rare genetic disease.
2. Aged 18 years and above.
3. Malaysian citizens or permanent residents residing in Malaysia.
4. Willing and able to provide informed consent for participation.
5. Willing to participate in an audio-recorded interview.

3.4.2 Exclusion Criteria

Individuals were excluded from the study if they met any of the following criteria:

1. Parents of children without a confirmed or clinically suspected rare genetic disease.
2. Individuals who were unable to communicate effectively in either Malay.
3. Participants involved in the pilot study, as their interview data were used solely for refinement of the interview guide and were not included in the final analysis.

3.5 RECRUITMENT AND SAMPLE SIZE

Participants were recruited using purposive sampling through two recruitment pathways. The primary recruitment route was through the Malaysia Rare Disease Society (MRDS), where a study invitation poster and registration link were disseminated through the society's communication channels. A total of 16 individuals registered their interest by completing an online Google Form. Following screening against the inclusion and exclusion criteria, 13 individuals were found to be eligible and agreed to participate in the study. The remaining registrants were excluded as they did not meet the study criteria, withdrew from participation, or were unable to arrange a suitable time for the interview. Interviews with the eligible participants were subsequently scheduled according to their preferred dates, times, and mode of interview.

To supplement recruitment, the researcher conducted regular visits to the Genetics Clinic at Tunku Ampuan Besar Tuanku Aishah Rohani Hospital (TABTAR), UKM Children's Specialist Hospital. During these visits, follow-up cases attending the clinic were reviewed and screened for eligibility based on the study inclusion and exclusion criteria. Two eligible parents were identified, approached, and agreed to participate in the study. These participants were subsequently interviewed and included in the final dataset. Together with the 13 participants recruited through MRDS, this resulted in a total sample of 15 participants. A flowchart illustrating the participant recruitment process is presented in Figure 3.2.

In qualitative research, sample size is determined by the depth, relevance and adequacy of the data rather than statistical considerations. This study was informed by the concept of information power (Malterud, Siersma and Guassora, 2016), which proposes that studies with a focused aim, a specific participant group, and rich qualitative data may require fewer participants. As the present study explored the experiences of a well-defined group of parents of children with rare genetic conditions through in-depth semi-structured interviews, a relatively small sample was considered appropriate. Data collection and preliminary analysis were undertaken concurrently to monitor the emergence of new concepts and themes. Participant recruitment continued until thematic saturation was achieved, defined as the point at which no new codes or themes were identified in subsequent interviews. Consequently, recruitment ceased after 15 participants had been interviewed.

Figure 3.2 Participant recruitment process

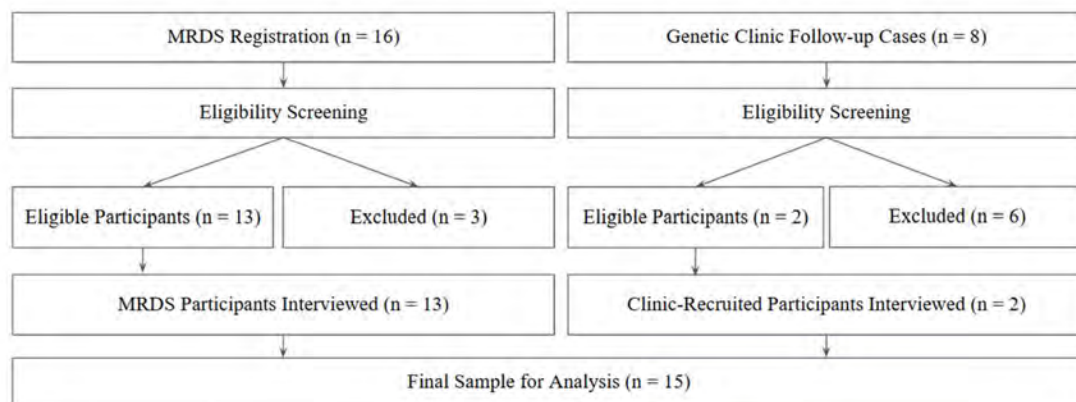


Figure 3.2 Flowchart illustrating the participant recruitment process. Sixteen parents registered their interest through the Malaysia Rare Disease Society (MRDS) recruitment platform, of whom 13 met the eligibility criteria and participated in the study. Three registrants were excluded due to not meeting the study criteria, withdrawal from participation, or inability to arrange an interview. An additional two eligible participants were recruited through screening of follow-up cases attending the Genetics Clinic at Tunku Ampuan Besar Tuanku Aishah Rohani Hospital (TABTAR), UKM Children's Specialist Hospital. A total of 15 participants were included in the final analysis.

3.6 DEVELOPMENT OF RESEARCH INSTRUMENT

A semi-structured interview guide was developed based on the study objectives and relevant literature on rare diseases, genetic discrimination, psychosocial adaptation and genetic counselling. The guide was reviewed by experts in genetic counselling and qualitative research to assess the relevance, clarity and comprehensiveness of the questions. Revisions were made based on their feedback prior to data collection. The interview guide consisted of open-ended questions and probing prompts to facilitate in-depth exploration of participants' experiences. It was developed in the Malay language and covered six key areas: (1) Journey to Diagnosis, (2) Genetic Testing and Genetic Counselling Experiences, (3) Genetic Discrimination, (4) Emotional and Social Impact, (5) Coping Strategies, and (6) Suggestions and Unmet Needs (Appendix A).

Prior to the main study, the interview guide was pilot tested with five eligible parents recruited from the Genetics Clinic at Tunku Ampuan Besar Tuanku Aishah Rohani Hospital (TABTAR), UKM Children's Specialist Hospital. Pilot participants were not included in the final analysis. The pilot study was conducted to assess the clarity, appropriateness and flow of the interview questions and procedures. Based on participant feedback and researcher reflections, minor revisions were made to improve the wording, sequencing and clarity of the questions before the interview guide was finalized for the main study.

3.7 DATA COLLECTION AND INTERVIEW PROCEDURES

Data were collected through individual semi-structured interviews conducted by the primary researcher. Following recruitment, eligible participants were contacted to arrange an interview at a date and time convenient to them. Thirteen interviews were

conducted online via Google Meet, while two interviews were conducted face-to-face at the Genetics Clinic, Tunku Ampuan Besar Tuanku Aishah Rohani Hospital (TABTAR), UKM Children's Specialist Hospital. Prior to the interview, participants were provided with a Participant Information Sheet explaining the study and were given the opportunity to ask questions before providing written informed consent.

The interviews were guided by a semi-structured interview guide (Appendix A) and began with a brief introduction and rapport-building session. Open-ended questions and probing techniques were used throughout the interviews to facilitate in-depth discussion and encourage participants to elaborate on their experiences and perspectives. With participants' consent, all interviews were audio-recorded to ensure accurate capture of the discussions. Interview duration ranged from approximately 45 to 90 minutes, depending on the complexity of participants' experiences and the depth of information shared. Data collection continued until thematic saturation was achieved, whereby no new codes or themes emerged from subsequent interviews. Following data collection up until thematic saturation achieved, all audio recordings were transcribed verbatim. The transcripts were reviewed repeatedly while listening to the original audio recordings to verify their accuracy and completeness. Identifying information was removed during transcription and replaced with participant identification codes to maintain confidentiality prior to data analysis.

3.8 REFLEXIVE THEMATIC ANALYSIS

Data generated from the semi-structured interviews were analysed using ATLAS.ti version 24 following Reflexive Thematic Analysis (RTA) as described by Braun and Clarke (2006, 2021). This approach was selected because it is well suited to exploratory qualitative research and enables an in-depth examination of participants' experiences, perspectives, and meaning-making processes while recognising the active role of the researcher in knowledge generation.

The analysis followed the six phases of Reflexive Thematic Analysis. The first phase involved familiarisation with the data through repeated reading of the interview transcripts and listening to the audio recordings. During the second phase, 286 initial codes were generated by systematically identifying meaningful segments of data

relevant to the research questions. Coding was primarily inductive, allowing patterns and concepts to emerge from participants' accounts rather than being restricted to predetermined categories. In the third phase, related codes were grouped and organised into six preliminary themes: (1) Journey to Diagnosis, (2) Genetic Testing and Genetic Counselling Experiences, (3) Genetic Discrimination, (4) Emotional and Social Impact, (5) Coping Strategies, and (6) Suggestions and Unmet Needs.

The fourth phase involved reviewing and refining the preliminary themes to ensure internal coherence and clear distinction between themes. During this process, the themes “Journey to Diagnosis” and “Genetic Testing and Genetic Counselling Experiences” were merged as they primarily provided contextual understanding of participants' experiences rather than directly addressing the study objectives. In the fifth phase, the themes were further refined, defined, and named to accurately represent the underlying patterns within the dataset. Finally, in the sixth phase, the themes were interpreted and integrated into a coherent narrative in relation to the study objectives, existing literature, and the Malaysian context. The final analysis generated five themes: (1) Journey to Diagnosis and Genetic Testing Experiences, (2) Genetic Discrimination, (3) Emotional and Social Impact, (4) Coping Strategies, and (5) Suggestions and Unmet Needs. Representative quotations were selected to illustrate each theme and preserve the authenticity of participants' voices. All quotations were anonymised to maintain participant confidentiality.

3.9 LANGUAGE MANAGEMENT AND TRANSLATION

All interviews were conducted in Malay, which was one of the study inclusion criteria and served as a common language understood by participants from different ethnic backgrounds in Malaysia. Conducting interviews in Malay enabled participants to communicate their experiences in a familiar language and facilitated richer descriptions of sensitive experiences related to discrimination, stigma and caregiving. Audio recordings were transcribed verbatim in Malay, after which coding and theme development were conducted in English to facilitate analysis, reporting, and thesis writing. Throughout the analytic process, the researcher frequently referred back to the original Malay transcripts to ensure that codes and themes accurately reflected

participants' intended meanings and preserved important cultural and contextual nuances. As participants' quotations are presented in their original Malay language in the findings chapter, direct translation of quotations into English was not undertaken. Instead, English explanations and interpretations were provided within the surrounding text to contextualize the quotations while preserving the authenticity and meaning of participants' original narratives.

3.10 TRUSTWORTHINESS OF THE STUDY

Several measures were undertaken to enhance the trustworthiness of the study. Credibility was supported through repeated engagement with the data, iterative coding, and regular discussions with research supervisors to review interpretations and refine theme development. Transferability was facilitated by providing detailed descriptions of the study context, participant characteristics, and research procedures. Dependability was enhanced by documenting the analytical process, including the development and refinement of codes and themes throughout the study, allowing the progression from raw data to final findings to be clearly traced. Confirmability was supported through researcher reflexivity and ongoing reference to the original transcripts to ensure that interpretations remained grounded in participants' accounts.

Researcher reflexivity was maintained throughout data collection and analysis. As a Master's student in Genetic Counselling with academic and clinical exposure to genetic conditions and genetic healthcare, the researcher recognised the potential for prior knowledge and preconceptions to influence the interpretation of participants' experiences. During interviews, open-ended and non-leading questions were used to allow participants to describe and interpret their experiences in their own words, without assuming that an experience constituted genetic discrimination. During analysis, the researcher repeatedly returned to the original transcripts and discussed emerging interpretations with research supervisors to examine alternative perspectives and challenge potential assumptions. These processes helped to ensure that the findings remained grounded in participants' accounts while acknowledging the researcher's role in the interpretative process.

Consistent with Braun and Clarke's (2021) reflexive thematic analysis approach, member checking was not undertaken because themes were viewed as the researcher's interpretative understanding of the data rather than findings requiring participant validation.

3.11 ETHICAL CONSIDERATIONS

Ethical approval for this study was obtained from the Universiti Kebangsaan Malaysia Research Ethics Committee prior to participant recruitment and data collection (Appendix D). Participation was entirely voluntary and all participants were provided with a Participant Information Sheet (Appendix B) outlining the study objectives, procedures, potential risks and benefits, confidentiality measures and participants' rights. Written informed consent (Appendix C) was obtained before participation. Participants were informed that they could decline to answer any question or withdraw from the study at any time without penalty. Given the potentially sensitive nature of discussions surrounding genetic discrimination, participants were reminded that they could pause or terminate the interview at any stage. A designated genetic counsellor was identified as a support person for participants who experienced emotional distress during or after the interview, and contact information was provided in the Participant Information Sheet.

All interviews were audio-recorded with participants' consent and transcribed verbatim. To maintain confidentiality, identifying information was removed from transcripts and replaced with participant identification codes. Audio recordings, transcripts, consent forms, and related research documents were stored separately and securely, with electronic files maintained in password-protected storage systems and hard-copy documents kept in locked storage facilities accessible only to the research team.

CHAPTER IV

RESULTS AND FINDINGS

4.1 INTRODUCTION

This chapter presents the findings of the study exploring the lived experiences of genetic discrimination among parents of children with rare genetic conditions in Malaysia. The findings were derived from in-depth semi-structured interviews conducted with 15 parents and analysed using Reflexive Thematic Analysis. Five themes were identified: (1) Journey to Diagnosis and Genetic Testing Experiences, (2) Genetic Discrimination, (3) Emotional and Social Impact, (4) Coping Strategies, and (5) Suggestions and Unmet Needs. Prior to presenting the thematic findings, the demographic characteristics of the participants and their children are described to provide context for the study sample. The findings are subsequently presented according to themes and subthemes, supported by representative verbatim quotations from participants. To maintain confidentiality, all participants were assigned identification codes (P1–P15), and only anonymised quotations are presented throughout this chapter.

4.2 PARTICIPANT CHARACTERISTICS

A total of 15 parents participated in this study, all of whom were mothers of children diagnosed with rare genetic conditions. Participants ranged in age from 25 to 58 years. The majority were Malay (80.0%), while Chinese (13.3%) and Indian (6.7%) participants were also represented. Most participants were married (86.7%), with two participants identifying as single mothers (13.3%). Participants demonstrated diverse educational and socioeconomic backgrounds. Three participants (20.0%) had completed secondary education (SPM/Sijil), while the majority (80.0%) had attained tertiary education (Diploma and above). Most participants were employed (73.3%), while four participants (26.7%) were not employed or identified as homemakers. Based

on the Malaysian household income classification, five participants (33.3%) belonged to the B40 group ($\leq$ RM5,249), seven participants (46.7%) were classified as M40 (RM5,250–RM11,819), and three participants (20.0%) belonged to the T20 group ($\geq$ RM11,820), indicating substantial socioeconomic diversity within the sample. Participants were recruited from six states across Peninsular Malaysia. Nearly half resided in Selangor and Kuala Lumpur (46.7%, $n = 7$), followed by Johor (26.7%, $n = 4$). One participant each was from Kedah (6.7%), Penang (6.7%), Perak (6.7%), and Melaka (6.7%). The majority of participants resided in urban areas (80.0%), while three participants (20.0%) lived in rural or semi-rural settings. Table 4.1 presents the sociodemographic characteristics of the participants.

Table 4.1 Sociodemographic Characteristics of Participants (N = 15)

Characteristic	Frequency (n)	Percentage (%)
Gender		
Female	15	100.0
Male	0	0.0
Ethnicity		
Malay	12	80.0
Chinese	2	13.3
Indian	1	6.7
Marital Status		
Married	13	86.7
Single mother	2	13.3
Highest Educational Attainment		
Secondary education (SPM/Sijil)	3	20.0
Tertiary education (Diploma and above)	12	80.0
Employment Status		
Employed	11	73.3
Not employed / homemaker	4	26.7
Monthly Household Income		
$\leq$ RM5,249	5	33.3

RM5,250–RM11,819	7	46.7
≥ RM11,820	3	20.0
Geographic Distribution		
Selangor and Kuala Lumpur	7	46.7
Johor	4	26.7
Kedah	1	6.7
Penang	1	6.7
Perak	1	6.7
Melaka	1	6.7
Residential Setting		
Urban	12	80.0
Rural / semi-rural	3	20.0

Table 4.1 Summary of the demographic characteristics of study participants, including gender, ethnicity, marital status, educational attainment, employment status, household income, geographic distribution, and residential setting (N = 15).

The geographical distribution of participants across different regions of the country, together with the diversity in ethnicity, educational attainment, employment status, income classification, and residential setting, enabled the study to capture a broad range of experiences and perspectives regarding parenting a child with a rare genetic condition within the Malaysian context. Participants represented diverse socioeconomic, educational and geographical backgrounds, with families residing across multiple regions of Peninsular Malaysia except none were from East Coast and East Malaysia.

The children represented a diverse range of rare genetic conditions affecting multiple body systems, including neurological, developmental, neuromuscular, craniofacial, skeletal, and haematological disorders. Their ages ranged from 10 months to 20 years, reflecting experiences across infancy, childhood, adolescence, and early adulthood. This diversity enabled the study to capture a broad spectrum of parental experiences associated with caring for children at different developmental stages and with varying healthcare and support needs. Table 4.2 summarises the characteristics of the children with rare genetic conditions whose parents participated in this study.

Table 4.2 Characteristics of Children with Rare Genetic Conditions (N = 15)

Participant	Child Age	Sex	Diagnosis
P1	8 years	Male	Neurofibromatosis Type 1
P2	2 years	Female	Treacher Collins Syndrome
P3	2 years 6 months	Male	Duchenne Muscular Dystrophy
P4	6 years	Female	PIGG-related neurodevelopmental disorder
P5	20 years	Female	Fibrodysplasia Ossificans Progressiva (FOP)
P6	1 year 8 months	Female	Spinal Muscular Atrophy (SMA) Type 1
P7	10 months	Male	Cardio-Facio-Cutaneous (CFC) Syndrome
P8	17 years	Female	Wolf-Hirschhorn Syndrome
P9	1 year 3 months	Male	Schinzel-Giedion Syndrome
P10 ¹	17 years, 14 years and 11 years	Male, Male and Female	Haemophilia A (two children) and Microdeletion Syndrome (one child)
P11	5 years	Male	Xq25 Duplication Syndrome
P12	14 years	Female	Angelman Syndrome
P13	17 years	Female	15q Duplication Syndrome
P14	8 years	Female	Spinal Muscular Atrophy (SMA) Type 1C
P15	1 year 7 months	Male	Achondroplasia

Table 4.3 The table summarizes the age, sex, and genetic diagnoses of the children represented in the study. The children ranged in age from 10 months to 20 years and were diagnosed with a diverse range of rare genetic conditions affecting multiple body systems, including neurological, developmental, neuromuscular, craniofacial, skeletal, and haematological disorders.

This study includes 15 families affected by distinct rare genetic conditions affecting multiple body systems. The conditions also varied in their clinical manifestations, with some associated with more readily recognisable physical or developmental features, while others may be less apparent without knowledge of the underlying diagnosis. This diagnostic and phenotypic diversity provided a broad range

¹ Participant P10 had three children diagnosed with rare genetic conditions

of contexts for exploring parental experiences across different diagnoses and stages of life.

4.3 OVERVIEW OF THEMES

Thematic analysis of the interview data resulted in the identification of five major themes. These themes reflect participants' experiences from the period of seeking a diagnosis for their child through to their encounters with genetic discrimination, emotional responses, coping mechanisms, and recommendations for improving support for families affected by rare genetic conditions.

Theme 1, Journey to Diagnosis and Genetic Testing Experiences, describes participants' pathways towards obtaining a diagnosis and their experiences with genetic testing and genetic services. Theme 2, Genetic Discrimination, represents the core findings of this study and encompasses experiences occurring across institutional including healthcare, employment and insurance followed by exclusions within education and social settings. Theme 3, Emotional and Social Impact, captures the psychological and emotional consequences associated with these experiences. Theme 4, Coping Strategies, describes the various approaches used by participants to manage challenges associated with caregiving and discrimination. Finally, Theme 5, Suggestions and Unmet Needs, presents participants' recommendations for improving support systems, public awareness, and policies affecting families living with rare genetic conditions. Table 4.3 presents a summary of the themes and subthemes identified through the analysis.

Table 4.3 Summary of Themes and Subthemes

Theme	Subtheme
Theme 1: Journey to Diagnosis and Genetic Testing Experiences	1a. The Diagnostic Pathway and Confirmation
	1b. Post-Diagnostic Psychological Adjustment
Theme 2: Experiences of Genetic Discrimination	2a. Institutional and Structural Discrimination
	2b. Educational Exclusion and Differential Treatment
	2c. Social Stigma and Community Alienation
Theme 3: Emotional and Social Impact	3a. Psychological Distress and Future-Focused Anxiety
	3b. Socio-Emotional Impacts on Child Identity and

Wellbeing	
Theme 4: Coping Strategies	4a. Internal Coping: Acceptance and Positive Reframing
	4b. External Coping: Strategic Disclosure and Structural Adaptation
Theme 5: Suggestions and Unmet Needs	5a. Systemic Reforms: Integrated Care, Policy and Financial Protection
	5b. Community Reforms: Inclusive Education and Public Awareness

Table 4.3 Thematic framework generated from the analysis of participants' narratives.

The themes and subthemes illustrate the progression of participants' experiences from diagnosis and genetic testing, through experiences of genetic discrimination with its social and emotional impact, to coping strategies and suggestions for improving support, services and protections for families affected by rare genetic conditions.

The following sections present each theme in detail, supported by representative quotations from participants.

4.4 THEME 1: JOURNEY TO DIAGNOSIS AND GENETIC TESTING EXPERIENCES

The journey towards obtaining a diagnosis was a significant aspect of participants' experiences. Prior to receiving a confirmed diagnosis, many parents described a period of uncertainty characterised by concerns regarding their child's development, repeated healthcare consultations, and difficulties obtaining clear explanations for their child's symptoms. For several participants, the diagnostic process involved multiple referrals, investigations, and interactions with various healthcare professionals before a definitive diagnosis was established. The eventual diagnosis and genetic testing results often represented an important turning point, providing both answers and new challenges for families. Two subthemes emerged within this theme: 1a) The Diagnostic Pathway and Confirmation and 1b) Post-Diagnostic Psychological Adjustment.

4.4.1 Theme 1a) The Diagnostic Pathway and Confirmation

Many participants described a prolonged and complex pathway to diagnosis. Parents often noticed developmental differences, unusual physical features, or health concerns long before receiving a confirmed diagnosis. During this period, uncertainty regarding

the cause of their child's condition contributed to considerable anxiety and frustration. Despite early recognition of potential abnormalities, obtaining a definitive diagnosis was often a lengthy and complex process. Several participants described being referred between multiple healthcare providers and specialists before receiving a confirmed diagnosis. One participant likened the experience to being repeatedly passed between services without obtaining clear answers.

“Kita macam kena hantar banyak specialist... dekat hospital swasta tu dia macam tak selalu jumpa condition macam ni. Jadi kita rasa macam kita ditolak sini, tolak sana... dalam dua tahun... tak dapat jawapan...macam ‘merry go round’” (P11)

This participant's account illustrates the fragmented healthcare pathway experienced by some families affected by rare genetic conditions. Even after seeking help from multiple specialists, the family remained without a definitive diagnosis for approximately two years, contributing to considerable uncertainty and frustration.

Most participants generally perceived genetic testing as an important step towards obtaining clarity regarding their child's condition to identify appropriate interventions and support. For several participants, the decision to proceed with genetic testing was made following discussions with healthcare professionals. One participant described receiving pre-test genetic counselling, which helped her understand the purpose of testing and informed her decision-making.

“Dia cakap dulu dia bagitahu dulu bila kita even masa nak ambil darah anak untuk genetic test itu pun dia orang memang kaunseling dulu... it's good to know what's wrong with her so that we can take extra precaution... so akhirnya saya sign jugalah untuk genetic test untuk anak saya dan untuk kami suami isteri.” (P8)

This participant valued the opportunity to receive information and counselling before testing, which enabled her to make an informed decision regarding genetic investigations for both her child and family.

Participants also described seeking genetic testing because they wanted answers and guidance regarding future management. For some, obtaining a diagnosis was viewed as a means of ensuring that no important interventions were overlooked.

“Saya macam mana... yang nak jawapan... at least ada petunjuk...saya buat betul atau ada apa intervention yang saya terlepas pandang... saya rasa salah satu petunjuk untuk tolong dalam plan intervention.” (P11)

The desire for diagnostic certainty reflected participants' efforts to better understand their child's condition and optimise available care and support.

However, some participants described anxiety and uncertainty while awaiting results despite recognising the potential benefits of genetic testing. The possibility of receiving confirmation of a genetic condition was often emotionally challenging.

“Masa nak buat tu memang takutlah... mana ada parent yang nak result tu positif... yang mula-mula tu asyik-asyik macam deny sendirilah mungkin dia... ciri-ciri dia sama dengan datuk, sama dengan nenek.” (P7)

This participant described a period of denial and hope that her child's features might be attributable to normal familial traits rather than an underlying genetic condition.

Overall, participants' accounts highlight the challenges associated with diagnosing rare genetic conditions, particularly when symptoms are non-specific or unfamiliar to healthcare providers. The participants also expressed opinion of genetic testing as a valuable diagnostic tool, although the decision-making process was often accompanied by fear and uncertainty regarding the implications of the results.

4.4.2 Theme 1b) Post-Diagnostic Psychological Adjustment

Receiving a diagnosis represented a significant turning point for many families. Although participants initially experienced a range of emotional reactions, including worry and sadness, a confirmed diagnosis often provided a sense of clarity and direction. One participant explained that the diagnosis largely confirmed what

healthcare professionals had already suspected, enabling the family to gradually accept the situation and focus on supporting their child.

“So lepas dapat result tu dia just confirm kan apa yang doktor suspect je macam doktor dah suspect there is something wrong... kita redha, kita pasrah dan kita memang cuba buat yang terbaiklah untuk dia.” (P8)

For this participant, diagnostic confirmation facilitated acceptance and allowed the family to move forward with their caregiving responsibilities.

Following diagnosis, several participants actively sought information and support from other families facing similar experiences. In the absence of local support networks, some turned to international online communities for guidance and reassurance.

“So apa yang saya buat adalah saya join Facebook group yang dia international group... at least nampak macam ada budak-budak lain yang sama masalah... support dekat sini memang sangat sikit.” (P7)

Connecting with other families helped participants gain information, share experiences, and feel less isolated in managing a rare genetic condition.

In summary, participants' journeys towards diagnosis were frequently characterised by uncertainty, repeated healthcare consultations and prolonged periods without clear answers. Genetic testing played a crucial role in establishing diagnostic certainty and informing future management. Although receiving a diagnosis was often emotionally challenging, it also facilitated acceptance, access to information, and engagement with support networks that helped families adapt to their caregiving roles.

4.5 THEME 2: EXPERIENCES OF GENETIC DISCRIMINATION

Experiences of genetic discrimination emerged as a prominent aspect of participants' narratives. Parents described various forms of discrimination, exclusion, and disadvantage encountered while navigating healthcare, employment, insurance,

educational, and social environments on behalf of their children. While some experiences involved direct barriers to services and opportunities, others reflected more subtle forms of disadvantage associated with limited awareness, inadequate accommodations, negative assumptions, and the rarity of the conditions themselves. Collectively, these experiences illustrate how families may encounter challenges across multiple domains of daily life following a rare genetic diagnosis. Three subthemes emerged within this theme: 2a) Institutional and Structural Discrimination, 2b) Educational Exclusion and Differential Treatment, and 2c) Social Stigma and Community Alienation.

4.5.1 Theme 2a) Institutional and Structural Discrimination

Healthcare setting

Experiences within healthcare settings were frequently discussed by participants and represented one of the earliest contexts in which perceived discrimination occurred. Although many participants expressed appreciation for the healthcare professionals involved in their child's care, several described encounters characterized by dismissal of parental concerns, difficulties accessing specialist services, limited awareness of rare genetic conditions and pressure to comply with recommended medical decisions. These experiences contributed to feelings of frustration, helplessness, and a perception that their knowledge and concerns as primary caregivers were not always recognized.

One common concern raised by participants was the perceived dismissal of parental observations regarding their child's symptoms. As primary caregivers, participants often felt they were the first to recognize changes in their child's condition. However, some described situations in which these concerns were not taken seriously by healthcare staff. One participant recalled seeking assistance after noticing abnormal breathing patterns in her child shortly after birth. Despite expressing her concerns, she felt that her observations were overlooked.

“Saya cakap Nurse, saya punya anak saya bernafas macam ni normal ke? Nurse tu macam, takpe nanti kejam lagi saya tengok... lepas saya cakap dengan dia tu, dia tak tengok pun.” (P2)

The participant perceived that her concerns were not given sufficient attention despite her repeated attempts to seek reassurance regarding her child's condition. Such experiences contributed to uncertainty and anxiety, particularly during the early stages of her child's healthcare journey.

Another participant described repeated difficulties obtaining referral to a specialist despite ongoing concerns regarding her child's hearing and long-term health outcomes. She explained that several healthcare providers had advised that specialist assessment was unnecessary, prompting her to actively advocate for further review.

“So pergi sana, jumpa bahagian pergigian...pergi bahagian speech therapy...apa yang berlaku kat dia ni semua dengan saya berusaha....saya bersyukur sebab ada doktor yang tolong jumpakan dengan timbalan pengarah hospital...timbalan pengarah tolong refer kan pergi doktor plastik...Doktor Plastik terus book untuk operation...” (P13)

Reflecting on this experience, the participant expressed concern that other families may not receive the same opportunities to access specialist care.

“Tak mustahil saya fikir ada insan yang terlepas dalam bab ni... saya kebetulan kerja bahagian kesihatan jadi saya cuba juga sebab saya nak dengar sendiri penjelasan daripada pihak yang sepatutnya.” (P13)

The participant perceived that her healthcare background enabled her to navigate the healthcare system and advocate for specialist referral. However, she worried that families with less knowledge of the healthcare system might not receive appropriate referrals or access to specialist services. This highlights how access to care may depend not only on clinical need, but also on a family's ability to navigate and advocate within the healthcare system.

Beyond communication and access challenges, some participants described experiences involving perceived institutional pressure when their healthcare decisions differed from medical recommendations. One participant recounted refusing hospital

admission for her child and subsequently feeling that her parental judgement was questioned by healthcare authorities.

“Lepas ambil darah... beberapa botol tu anak saya sawan kat situ... dia orang kata, okay kena warded. Saya kata saya tak nak warded... dia orang dah report polis...akta kanak kanak, so dia orang panggil counsellor hospital tu dia orang panggil 3-4 orang counsellor datang kaunseling kan saya.” (P9)

The participant perceived the response as excessive and felt that her decision-making capacity as a parent was not respected. Although healthcare professionals may have acted out of concern for the child's welfare, the participant experienced the situation as coercive and disempowering.

Collectively, these findings suggest that experiences interpreted as discrimination within healthcare settings were often linked to dismissal of caregiver concerns, difficulties accessing appropriate specialist services, limited recognition of parental expertise, and perceived imbalances in decision-making power between healthcare professionals and families. Conversely, participants reported more positive experiences when healthcare providers demonstrated awareness of rare genetic conditions, respected parental knowledge, and engaged in collaborative care. These findings highlight the importance of patient-centred and family-centred approaches in supporting families affected by rare genetic conditions and reducing experiences of marginalization within healthcare settings.

Employment setting

Parent's employment emerged as another setting in which participants experienced challenges associated with caring for a child with a rare genetic condition. Although participants did not commonly report overt workplace discrimination, many described difficulties balancing employment responsibilities with the extensive caregiving demands associated with their child's healthcare needs. Frequent medical appointments, hospital admissions, therapy sessions, and caregiving responsibilities often affected work performance, career progression, and employment decisions.

Several participants described workplace environments that were not always accommodating of the caregiving responsibilities associated with raising a child with a rare genetic condition. Frequent medical appointments, hospital admissions, and unexpected health emergencies often required parents to take emergency leave or adjust their work schedules. In some cases, participants perceived employers and supervisors lacked understanding regarding these responsibilities, resulting in strained workplace relationships and feelings of disadvantage. One participant described how repeated emergency leave taken to care for her child eventually resulted in pressure from her department head to resign from her position.

“Ketua jabatan masa tu tak memahami... sampai dia suruh saya berhenti kerja sebab saya kerap sangat EL (emergency leave).” (P10)

The participant perceived that her caregiving responsibilities were viewed negatively within the workplace. Rather than receiving accommodation or support, she felt pressured to choose between maintaining employment and fulfilling her child's healthcare needs. This experience reflected a perceived lack of workplace flexibility and understanding towards parents caring for children with complex medical conditions.

Similarly, another participant anticipated that employers and colleagues would expect her to perform at the same level as employees without significant caregiving responsibilities.

“Kalau saya dah masuk kerja saya rasa memang semua orang akan expect saya kerja macam saya tak ada masalah lain... dekat situ... untuk orang memahami benda-benda yang you go through personally itu memang secondary.” (P7)

This participant perceived that personal caregiving challenges were often regarded as secondary to workplace expectations. Although she did not report direct discriminatory treatment, she anticipated limited understanding and accommodation for the additional responsibilities associated with caring for a child with a rare genetic condition.

Other than perceived lack of workplace understanding and accommodation, participants also described the ongoing challenge of balancing employment responsibilities with the complex healthcare needs of their children. Frequent medical appointments, hospital admissions, and caregiving demands often competed with professional obligations, creating tension between their roles as employees and caregivers. One participant explained that despite her commitment to her profession, her child's healthcare needs inevitably affected the amount of time and attention she could devote to work.

“Saya cuba sehabis baik... jadi pekerja yang committed... tapi bila...kena appointment, sawan, kena masuk hospital... benda-benda itu akan menarik masa untuk saya beri kepada tugas saya sebagai guru... kalau kena berhenti kerja pun saya akan berhenti.” (P8)

This participant's account illustrates the difficult choices faced by many parents of children with rare genetic conditions. Although she remained committed to her career, her child's healthcare needs frequently required her to prioritize caregiving responsibilities over work-related commitments. For this participant, maintaining her child's wellbeing ultimately took precedence over employment, reflecting the substantial caregiving burden experienced by many families.

Overall, participants' experiences within employment settings were characterized by limited workplace accommodation, competing work and caregiving responsibilities, and concerns regarding employers' understanding of the challenges associated with raising a child with a rare genetic condition. While some participants experienced direct pressure or negative responses from supervisors, others anticipated limited flexibility and support within their workplaces. These experiences often influenced career decisions, work participation, and long-term employment opportunities. Collectively, the findings suggest that employment-related discrimination may manifest not only through explicit workplace actions, but also through structural barriers and workplace expectations that fail to accommodate the realities of caregiving for a child with complex healthcare needs.

Insurance setting

Experiences relating to insurance and financial assistance emerged as one of the most commonly reported forms of perceived genetic discrimination. Two of the fifteen participants reported that they had never held insurance coverage. Of the four participants who had existing insurance policies, most described exclusions related to genetic conditions or difficulties maintaining coverage following their child's diagnosis. The remaining participants reported either being discouraged from applying after disclosing their child's condition or having their insurance applications rejected because of the genetic nature of the condition. Many felt that families affected by rare genetic conditions were disadvantaged despite having greater healthcare needs and higher long-term medical expenses.

On closer inspection, several participants reported that obtaining insurance became significantly more difficult once a genetic diagnosis had been established. One participant expressed frustration that children with rare genetic conditions appeared to be excluded from insurance coverage despite being among those who would benefit most from financial protection.

“Lepas kita dapat diagnosis...saya rasa kalau dah tahu then nak ambil insurans memang tak dapat lah... itu saya rasa sangat unfair lah sebab budak-budak yang ada masalah macam ni yang memerlukan insurans lebih daripada budak-budak yang sihat.” (P7)

The participant perceived the exclusion as particularly unjust because children with complex healthcare needs were often the very individuals who required insurance protection the most. This experience contributed to feelings of unfairness and financial vulnerability.

Other participants described practical difficulties obtaining insurance coverage even when they actively sought protection for their children. One participant explained that insurance arrangements became problematic following her child's diagnosis, eventually leading the family to discontinue coverage.

“Tapi insurans yang selepas lahir tu yang banyak masalah, kita terus terminate...biasanya insurer kalau budak tak cecah umur dua tahun dia susah nak cover.” (P4)

This participant perceived that insurance providers imposed significant restrictions and limitations that reduced the usefulness of coverage for families caring for children with complex medical needs.

Some participants reported direct rejection of insurance applications because of their child's medical condition. One participant recalled being informed that her child would not be eligible for insurance until significant improvements in physical functioning had occurred which is almost impossible for a Spinal Muscular Atrophy (SMA) Type 1 patient.

“Apa-apa insurans kena reject... saya pernah kena dia kata tunggu anak boleh jalan dulu baru boleh apply.” (P14)

The participant interpreted this requirement as exclusionary because eligibility was linked to the child's functional abilities rather than individual insurance needs. Such experiences reinforced perceptions that children with rare genetic conditions faced systematic barriers to obtaining financial protection.

Participants also reported situations where insurance policies ceased to provide meaningful coverage after a genetic condition had been identified. One participant explained that although insurance had been purchased when her child was younger, claims were subsequently denied because the condition was classified as genetic.

““Dulu masa dia kecil kami dah ambil insurans tapi lepas tu bila dia masuk hospital...tak cover sebab...ini masalah genetik... kami pun berhentilah sebab no point lah kalau tak cover...” (P5)

For this participant, the exclusion of genetic conditions from coverage rendered the insurance policy ineffective and contributed to additional financial burden during periods of hospitalization and treatment.

Overall, participants consistently described insurance systems as a source of financial disadvantage for families affected by rare genetic conditions. Experiences of rejection, exclusions and restricted coverage contributed to perceptions that insurance policies did not adequately accommodate the needs of individuals with genetic disorders. These findings suggest that insurance-related discrimination may occur either through eligibility restrictions, coverage exclusions or policy practices that disproportionately disadvantage families already facing substantial healthcare and caregiving costs.

4.5.2 Theme 2b) Educational Exclusion and Differential Treatment

Participants described a range of experiences relating to education and childcare settings. While some reported receiving valuable support from early intervention programs and specialized educational services, others described experiences of exclusion, inadequate accommodation, and negative assumptions regarding their children's abilities. These experiences occurred at various stages of the educational journey, from school enrolment and placement decisions to the implementation of accommodations for children with specific healthcare needs.

One of the most significant concerns raised by participants was the failure of educational institutions to adequately accommodate their children's medical conditions despite receiving relevant information and documentation. One participant described providing the school with information regarding her child's hemophilia diagnosis, however she felt that these precautions were not appropriately considered by the school.

“Saya dah bagi surat... pamphlet hemofilia... tapi sama juga... anak saya balik kaki dia tak boleh jalan dah sebab cikgu tak beritahu ada ujian fizikal...” (P10)

Despite proactively disclosing her child's condition and providing supporting documentation, the participant reported that important information regarding physical

activities was not communicated to the family. As a result, the child participated in activities that led to physical complications. This experience illustrates how inadequate accommodation and communication within educational settings may place children with rare genetic conditions at increased risk of harm.

Participants also described experiences of educational exclusion and placement decisions that they perceived as discriminatory. One participant recounted being informed that her child was unsuitable for placement within a special education program.

“Guru besar dekat Program Pendidikan Khas Integrasi (PPKI) dekat sekolah menengah suruh dia keluar daripada sekolah tu... anak saya tak sesuai masuk PPKI... tak boleh menulis nama sendiri... dia hanya boleh meniru... motor dia slow...” (P13)

The participant perceived that her child's abilities were assessed primarily through deficits in academic and motor performance rather than individual strengths or support needs. The decision to remove the child from the program was experienced as exclusionary and contributed to feelings of disappointment and frustration.

Beyond direct experiences of exclusion, some participants described anticipating discrimination before their children entered educational settings. Concerns regarding how schools might respond to disability status influenced decisions about disclosure during the enrolment process. One participant explained that although her child was eventually accepted into school, the family initially chose not to disclose the child's disability card because of concerns that this information might result in rejection.

“Kami dah jumpa penolong kanan hal ehwal murid... kita orang tak disclose...ada kad Orang Kurang Upaya (OKU)... kita takut dia reject terus bila dia dengar OKU...” (P4)

This account reflects anticipatory stigma, whereby families modified disclosure practices because of fears that disability status might negatively influence access to

educational opportunities. Although rejection did not occur, the concern itself demonstrates the perceived vulnerability experienced by parents when navigating school enrolment processes.

Participants also described encountering negative assumptions regarding their children's abilities and future potential. One participant explained that she often felt that others viewed her child through a deficit-based lens.

“Memang betul dia pandang rendah lah... macam dia layan, ‘oh anak awak macam tak bagus lah... anak awak susah lah... anak awak bodoh lah’... macam dia bagi saya rasa macam itulah.” (P11)

Although these perceptions were not always expressed directly, the participant interpreted attitudes and behaviors from others as reflecting low expectations of her child's abilities. Such experiences contributed to feelings of marginalization and reinforced concerns regarding societal acceptance of children with rare genetic conditions.

Despite these challenges, participants also highlighted positive experiences with educational and developmental support services. Several parents described the important role played by community-based rehabilitation program and specialized intervention centres in supporting their children's development. One participant spoke positively about both community-based rehabilitation services and private therapy programs.

“Memang sangat bagus Program Pemulihan Dalam Komuniti (PDK)... very helpful... ada private center... kita bayar... dia orang kena buat elok-elok kan.” (P8)

The participant's account demonstrates that supportive educational and developmental services were highly valued and could positively influence both the child's progress and the family's overall experience. Access to such services was perceived as particularly important for children with complex developmental needs.

Overall, participants' experiences within education and childcare settings reflected both supportive and exclusionary practices. Experiences interpreted as discriminatory included inadequate accommodation of medical needs, exclusion from educational placements, and negative assumptions regarding children's abilities. At the same time, participants recognized the value of specialized educational and developmental services when these were responsive to their children's needs. These findings suggest that discrimination within educational settings may occur not only through direct exclusion but also through institutional practices and attitudes that fail to adequately support children with rare genetic conditions.

4.5.3 Theme 2c) Social Stigma and Community Alienation

Participants described diverse experiences within social and community settings. While some reported acceptance and support from family members, friends, and support networks, others encountered misunderstanding, negative assumptions, and subtle forms of stigma related to their child's rare genetic condition. Social discrimination was often less overt than discrimination experienced in insurance or educational settings, but nevertheless influenced how participants interacted with their communities and perceived societal attitudes towards disability and rare diseases.

A common experience described by participants was the perception that others viewed their child differently because of their condition. Several participants reported receiving negative looks, judgmental reactions or subtle indications that their child was perceived as different from other children. One participant described experiencing negative reactions from members of the public who appeared uncomfortable with her child's condition.

“Ada setengah tu macam pandang rendah... macam tengok dengan cara dia... buat muka...” (P13)

Although these behaviours were often indirect, the participant interpreted them as expressions of stigma and negative judgement towards her child. Such experiences reinforced feelings of social difference and highlighted the challenges of navigating public spaces with a child who appeared visibly different from societal expectations.

Similarly, another participant described experiencing subtle social exclusion in her interactions with others. Although she did not perceive overt rejection or discrimination, she felt that some people were less willing to engage with her affected child compared to her other children who did not have a rare genetic condition.

"Macam disisihkan ni tak adalah, tapi macam kadang-kadang tu nampaklah macam dorang tak layan sangat... dari segi layanan tu lah." (P1)

This account illustrates how stigma may be experienced through subtle differences in social interactions rather than explicit exclusion. The participant's comparison between her child with a rare genetic condition and her other children suggests a perceived difference in the way others responded to and engaged with the affected child. Although such behaviours may appear minor or ambiguous, repeated experiences of reduced social engagement may contribute to feelings of social difference and reinforce perceptions of stigma among affected families.

Participants also described situations in which family members and community members misunderstood the genetic nature of their child's condition. In some cases, this lack of understanding resulted in blame being directed towards parents. One participant explained that her in-laws attributed her child's condition to careless parenting rather than recognizing it as a medical condition.

"Kadang-kadang anak I dia jalan tak stable...dia akan jatuh. Bapa mertua I akan kata...youlah tak jaga dia... bagi dia jatuh. Tapi bukan... bukan sebab kita...penyakit itu buat dia macam lemah..." (P3)

Similarly, another participant described being questioned about her behavior during pregnancy and experiencing suggestions that her child's condition may have resulted from inadequate maternal care.

"dia cakap tak jaga pemakanan ke...waktu mengandung ke... saya cakap jaga ...saya terima sajalah." (P6)

These accounts show how limited understanding of genetic conditions may contribute to courtesy stigma, where parents experience stigma or negative judgement because of their association with an affected child. They also show a gendered form of parental blame, particularly when the child's condition was attributed to the mother's behaviour during pregnancy. Rather than recognizing the genetic basis of the condition, relatives attributed the child's difficulties to parental care or maternal practices. Such assumptions may contribute to guilt, emotional burden, and social judgement among parents. These experiences show that the social impact of genetic conditions can extend beyond the affected individual to family members, particularly mothers.

Experiences of stigma also influenced how some participants engaged with social activities and community events. One participant explained that concerns regarding judgement, misunderstanding, and managing her child's behaviour in public contributed to social avoidance.

“Jadi nak mengelakkan benda tu saya tak ada lah bawa anak saya ke keraian ke keramaian ke... kita jumpa bila perlu sahaja.” (P14)

The participant's account illustrates how anticipated stigma may lead families to withdraw from social situations. Although this strategy reduced potentially uncomfortable interactions, it also limited opportunities for social participation and community engagement.

Despite these challenges, many participants also reported positive social experiences characterized by acceptance, empathy and support. Friends, family members, and peer support groups were often described as important sources of encouragement and understanding. One participant spoke positively about the support she received from friends and disability support communities.

“Alhamdulillah kawan-kawan semua support dan kita pun ada join support group... walaupun tak ada diagnosis sama...kita dah join group cerebral palsy... kami buat aktiviti, join run... kawan-kawan semua bila nampak perubahan... diorang betul-betul impressed lah.” (P4)

The participant's account demonstrates the value of supportive social networks in promoting inclusion and reducing feelings of isolation. Participation in support groups also provided opportunities to connect with families facing similar challenges and fostered a sense of belonging.

In summary, participants' experiences within social and community settings reflected both stigma and acceptance. Negative experiences were primarily associated with public misunderstanding, parental blame, and judgmental attitudes towards children with rare genetic conditions. However, many participants also reported receiving meaningful support from friends, family members and disability communities. These findings suggest that social discrimination is often rooted in limited awareness and understanding of rare genetic conditions, while positive social environments can promote acceptance, inclusion, and resilience among affected families.

4.6 THEME 3: EMOTIONAL AND SOCIAL IMPACTS

Participants described a range of emotional and social consequences arising from experiences of discrimination, stigma, exclusion, and unequal treatment encountered across healthcare, educational, insurance, workplace, family, and community settings. While these experiences occurred within the broader context of raising a child with a rare genetic condition, participants frequently linked discriminatory encounters to feelings of sadness, frustration, anxiety, stress, and social isolation. In some cases, the impact extended beyond the parents themselves and affected the emotional wellbeing, social participation, and self-perception of their children. Two subthemes were identified: 3a) Psychological Distress and Future-Focused Anxiety and 3b) Socio-emotional Impacts on Child Identity and Wellbeing.

4.6.1 Theme 3a) Psychological Distress and Future-Focused Anxiety

Participants described feelings of sadness, hurt and emotional distress when they or their children experienced exclusion, rejection, or stigmatizing attitudes from others. Such experiences could be particularly distressing when they occurred within settings where families expected support and acceptance, such as schools or among extended

family members. One participant described experiencing stigma within her own family, where relatives frequently made indirect remarks regarding her child's condition. Although these comments were not openly confrontational, the participant acknowledged that they were emotionally hurtful and chose to remain silent rather than respond.

“Family *sendiri... perlu-perlu kan... kita pun terasa lah... tapi kita diam je.*”
(P13)

The participant's account illustrates that genetic discrimination may occur not only in institutional settings but also within family relationships, where subtle forms of stigma and judgement can contribute to emotional distress. The participant's decision to remain silent further reflects how individuals may internalize these experiences rather than openly challenge them.

Similarly, another participant described the emotional impact of repeatedly seeking preschool placement for her child, only to perceive some kindergartens were unwilling to accept the child because of the diagnosis. The experience left the participant feeling rejected, pressured, and discouraged.

“*Memang dia layan macam, oh sekolah kita tak boleh ambil... jadi saya rasa sedih lah sebab masa itu dah banyak sekolah saya cari... so saya macam pressure...*” (P11)

For this participant, the difficulty in securing an educational placement extended beyond a practical challenge and became an emotionally distressing experience. The perceived rejection reinforced concerns about whether her child would be accepted within mainstream environments and highlighted the emotional burden that may arise when families encounter barriers to opportunities that are readily available to other children. Collectively, these findings demonstrate how experiences of exclusion and stigma can negatively affect the emotional wellbeing of parents and contribute to feelings of sadness and frustration.

Participants also expressed anxiety regarding their children's future opportunities and long-term financial security. These concerns were often linked to experiences of discrimination or perceived barriers that limited access to services, support, and opportunities available to others. Such experiences contributed to uncertainty about whether their children would be able to access appropriate care, education, and social inclusion in the future.

One participant described concerns regarding the financial burden associated with raising a child with a rare genetic condition, particularly because therapies and interventions were largely funded through private healthcare. The participant linked these concerns to difficulties obtaining adequate insurance coverage, resulting in uncertainty about future healthcare costs and access to ongoing treatment.

“Setakat ni memang rasa bimbang masa depan dia sebab terapi semua saya buat private kan... risau juga...” (P7)

This finding illustrates how experiences of insurance-related discrimination may extend beyond immediate financial consequences and contribute to ongoing anxiety regarding the future wellbeing of affected children.

In addition, some participants expressed concerns about their children's future social acceptance. Although these concerns were often anticipatory rather than based on direct experiences, they reflected worries that the child might face rejection or exclusion because of their condition. One participant described frequently thinking about whether her child would be accepted by peers and be able to form meaningful social relationships.

“Risau...risau jugalah... sebab takut tiada orang nak kawan dengan dia... Masalah sosial tu je saya memang dari sekarang pun saya dah terfikir-fikir.” (P2)

This concern highlights how the anticipation of future discrimination may itself become a source of emotional burden for parents. Even in the absence of direct exclusion,

worries about future acceptance in friendship and social participation can contribute to persistent anxiety regarding a child's long-term quality of life. Together, these findings demonstrate that experiences and perceptions of discrimination may influence not only present circumstances but also parents' expectations and concerns regarding their children's future opportunities and security.

4.6.2 Theme 3b) Socio-Emotional Impacts on Child Identity and Wellbeing

Participants also described how experiences of stigma, exclusion and differential treatment affected their children's social wellbeing and sense of identity. These impacts were particularly evident within school environments, where children were exposed to peer reactions, social comparison, and varying levels of acceptance from others. Parents observed that repeated experiences of being perceived as different could influence children's social interactions, emotional wellbeing, as well as self-perception.

One participant described how her child was subjected to teasing at school because of the child's disability status. According to the participant, the child frequently returned home upset and questioned why they were different from other children.

“Anak saya kena ejek... anak OKU... dia balik dia akan mengadu, Umi kenapa saya tak sama macam orang lain?” (P10)

This experience illustrates how discriminatory attitudes and peer stigma may affect a child's developing sense of identity. Being labelled and treated differently by peers appeared to contribute to feelings of self-consciousness and prompted the child to question their own differences. The participant's account suggests that the impact of discrimination may extend beyond immediate emotional distress and influence how children perceive themselves and their place within their social environment.

Similarly, another participant described how her child's social interactions at school were limited because peers appeared uncomfortable with the child's differences in behaviour and communication. The participant expressed concern that others might not be willing to accept her child, resulting in restricted opportunities for social engagement.

“Pergaulan dia dekat sekolah baru ni agak terbatas lah sebab semua orang agak pelik lah dengan dia ni... takut orang tak boleh nak terima...” (P12)

These finding highlights how social exclusion may occur even in the absence of overt bullying or rejection. Perceived discomfort and lack of acceptance from peers can create barriers to meaningful social participation, contributing to feelings of isolation and reduced opportunities to develop friendships.

To summarize, participants reported a range of emotional and social consequences associated with experiences of discrimination, stigma or exclusion encountered across family, educational, insurance and community settings. These experiences contributed to feelings of sadness, frustration, anxiety, and uncertainty, particularly when parents perceived that their children were being treated differently or denied opportunities because of their condition. Participants also expressed concerns regarding their children's future social acceptance and financial security, while some observed negative effects on their children's self-esteem and sense of belonging. These findings suggest that discriminatory experiences may affect not only parents but also children, influencing emotional wellbeing, social participation along with perceptions of future opportunities.

4.7 THEME 4: COPING STRATEGIES

Participants employed a range of coping strategies to manage experiences of stigma, exclusion, discrimination, and the challenges associated with navigating systems that were often not designed to accommodate the needs of children with rare genetic conditions. Instead of responding passively to these experiences, many parents demonstrated resilience through acceptance, adaptation, strategic decision-making, or active problem-solving. Some participants coped by reframing negative experiences and focusing on acceptance of their child's condition, while others developed practical strategies to manage social interactions or overcome barriers encountered in employment, healthcare, and insurance settings. Two subthemes emerged from the data: 4a) Internal Coping: Acceptance and Positive Reframing and 4b) External Coping: Strategic Disclosure and Structural Adaptation.

4.7.1 Theme 4a) Internal Coping: Acceptance and Positive Reframing

Several participants described acceptance as an important coping strategy that helped them manage negative attitudes and judgement from others. Rather than allowing external perceptions to influence their wellbeing, these parents focused on accepting their children as they were and reframing societal reactions as a reflection of others' lack of understanding rather than a deficiency within the child or family. One participant explained that negative views from others no longer affected her because she and her family had accepted her child unconditionally.

“Setakat ni...saya rasa apa pun pandangan orang...bagi saya dia tak affect saya lah, tak affect family saya sebab kami dah boleh terima dia seadanya.”

(P8)

Similarly, another participant described gradually becoming less concerned about societal judgement and choosing to focus on her family's wellbeing rather than attempting to gain acceptance from others.

“Kita macam kita bahagia cara kita...kita dah tak fikir apa orang luar nak cakap tu... kalau kita nak jelaskan pun macam diorang tak faham pun.” (P12)

These accounts demonstrate how acceptance and positive reframing served as important psychological coping strategies, enabling participants to reduce the emotional impact of stigma and maintain a positive outlook despite challenging social experiences.

4.7.2 Theme 4b) External Coping: Strategic Disclosure and Structural Adaptation

Some participants managed anticipated judgement and misunderstanding through selective disclosure of their children's condition. Rather than concealing the diagnosis entirely, they strategically shared information when they felt it would reduce questioning, facilitate understanding or prevent negative assumptions.

One participant described initially feeling different from others because she had three children with special needs. However, she found that openly disclosing this

information often reduced personal discomfort and allowed her to explain her situation more confidently.

“Saya rasa saya berbeza dengan orang... bila saya cakap saya ada 3 anak keperluan khas, saya rasa lega sebab dah beritahu orang.” (P10)

Similarly, another participant explained that informing others that the child had a rare disease often discouraged further questioning and judgement.

“Bila cakap rare case mulut semua orang tutup...” (P9)

These findings suggest that disclosure was not merely a means of sharing information but also functioned as a coping strategy that helped participants manage social interactions and reduce misunderstandings to navigate situations where they anticipated stigma or negative judgement.

Participants also demonstrated practical adaptation when faced with barriers in employment, healthcare, insurance, and service access. Many sought alternative solutions that enabled them to continue supporting their children's needs rather than accepting these barriers passively. One participant described leaving a workplace that offered little flexibility and seeking alternative employment that better accommodated caregiving responsibilities.

“Awal-awal tu memang saya kerja dekat tempat yang tak ada work from home... memang susah lah dia tak faham tapi lepas tu saya cari tempat kerja yang... lebih fleksibel.” (P11)

Meanwhile, in response to limitations in insurance coverage, some participants adopted practical strategies to minimize the financial impact of their child's condition. One participant explained that she had purchased insurance before her child's diagnosis was confirmed, allowing coverage for non-genetic related medical conditions to remain active. At the same time, she relied on government healthcare services and disability-related benefits for healthcare needs associated with the genetic condition.

"... dia punya insurance awal-awal sudah beli masa saya mengandung... confirm dia punya achondroplasia, all the side effects itu tak boleh cover... Kalau dia influenza saya boleh pergi untuk insurance punya... Kalau yang mahal-mahal tu, langsung boleh datang government hospital... dia ada kad OKU... semua boleh waived lah..." (P15)

Rather than relying solely on private insurance, she strategically utilized both existing insurance coverage and government-funded healthcare services to meet her child's healthcare needs. This adaptation helped reduce the financial burden associated with services and complications that were excluded from insurance coverage due to the genetic nature of the condition.

All in all, participants demonstrated various coping strategies to manage experiences of stigma, discrimination and barriers encountered while caring for a child with a rare genetic condition. Acceptance and positive reframing helped parents reduce the emotional impact of negative attitudes and social judgement, while strategic disclosure enabled them to navigate social interactions and minimize misunderstanding. Participants also adopted practical solutions to overcome structural barriers, including seeking more flexible employment, adjusting financial planning and utilizing alternative resources when access to services or insurance coverage was limited. This is to ensure that their children's needs continued to be met despite limitations in existing support systems. Collectively, these findings highlight the resilience and adaptability of parents in responding to challenges associated with discrimination and exclusion across different aspects of daily life.

4.8 THEME 5: SUGGESTIONS AND UNMET NEEDS

Participants identified numerous areas where improvements could better support families affected by rare genetic conditions and reduce experiences of discrimination. Recommendations focused on strengthening healthcare coordination, improving professional awareness, expanding psychosocial and practical support services, and addressing financial and societal barriers. These suggestions were informed by participants' lived experiences navigating healthcare systems, educational settings, employment challenges, and community environments. Two subthemes emerged within

this theme: 5a) Systemic Reforms: Integrated Care, Policy and Financial Protection and 5b) Community Reforms: Inclusive Education and Public Awareness.

4.8.1 Theme 5a) Systemic Reforms: Integrated Care, Policy and Financial Protection

Participants highlighted the need for more coordinated and integrated services for families affected by rare genetic conditions. Many described navigating fragmented healthcare systems involving multiple specialists, services, and institutions that often operated independently from one another. One participant suggested the need for coordinated medical guidance and better communication between healthcare providers.

“Kalau ada macam support dari medical knowledge... macam mentor, coach... sebab kadang-kadang kita dalam loop... tak ada solution... especially swasta semua specialist tak bercakap dengan each other.” (P11)

The participant perceived that families would benefit from a more coordinated care model in which healthcare professionals communicate effectively and provide clearer guidance throughout the care journey.

Similarly, another participant envisioned a more integrated support system involving healthcare, educational, and home-based services delivered by professionals with specialised rare disease knowledge.

“Kalau boleh kan, buat perubahan dari segi perubatan, pendidikan, bantuan... macam yang saya nampak kat luar negara... buat rawatan datang ke rumah... cikgu ke pakar ke adalah yang dalam bidang khusus yang lebih tahu pasal rare disease ni.” (P12)

These suggestions reflect participants' desire for comprehensive support systems that address the diverse and interconnected challenges faced by families affected by rare genetic conditions.

Many participants have also consistently identified a lack of support directed towards caregivers themselves. While healthcare services often focused on the child,

participants felt that parents' emotional wellbeing and caregiving needs were frequently overlooked. One participant highlighted the absence of dedicated programmes aimed at supporting parents.

“Saya pernah usulkan... satu program untuk parents... kita lack of moral support untuk parents... kita banyak focus on the SMA fighters... tapi untuk parents tak ada.” (P14)

Similarly, another participant felt that healthcare professionals should pay greater attention to caregivers' emotional wellbeing.

“Doktor kena lah tanya kita, kita okay ke tak... sebab kadang-kadang doktor tu just tanya baby okay ke tak tapi tak tanya emosi mak... kita pun nak orang tanya kita okay ke tak.” (P2)

Beyond emotional support, participants also emphasized the need for practical caregiving assistance. Several described relying heavily on grandparents and extended family members to help manage childcare responsibilities. One participant suggested the development of specialised childcare centre or respite services for children with complex healthcare needs.

“Saya rasa memang kena lebih ada macam tempat-tempat lah pusat penjagaan ke bantuan penjagaan untuk budak-budak inilah... kalau you tak ada datuk ke nenek ke... mak ayah tu memang kena berhenti bekerja... satu income sahaja is very difficult...” (P7)

This participant highlighted how practical support services could reduce caregiver burden and help parents remain employed to reduce overall family financial burden. In similar way, many other participants believed that existing financial support mechanisms were insufficient to address the ongoing costs associated with genetic testing, therapy, healthcare and specialised equipment. One participant emphasised the importance of financial assistance for both diagnosis and long-term therapy.

“Saya rasa first dari segi diagnosis kan... mungkin boleh beri bantuan kewangan untuk detect lah, untuk test tu dulu... lepas tu kalau dah dapat tu saya rasa perlu adanya macam bantuan kewangan... kalau dia dapat bagi berkala tu bolehlah dengan ibu bapa ni nak hantar anak-anak pergi terapi macam OT, fisio semua tu kan mahal.” (P4)

Participants also advocated for more inclusive insurance policies that provide at least basic protection for children with rare genetic conditions.

“I really wish lah macam ada insurance company yang... at least untuk warded lah bayaran untuk wad saja pun boleh... buka lah apa salahnya kan.” (P8)

Collectively, participants emphasised the need for healthcare systems, support services, and policies that are better coordinated, more responsive to caregiver needs, and capable of reducing the long-term financial burden associated with rare genetic conditions.

4.8.2 Theme 5b) Community Reforms: Inclusive Education and Public Awareness

Participants frequently emphasised the importance of improving awareness and understanding of rare genetic conditions among educators, healthcare professionals as well as other service providers. They believed that greater knowledge would reduce exclusionary practices and improve the quality of support received by affected children and families. One participant called for greater empathy, responsibility and preparedness among educators supporting children with disabilities.

“Sepatutnya macam cikgu... buatlah budak tu macam anak dia sendiri... jangan terus, dia rasa dia tak nak ambil kerja lebih... sepatutnya dia tahu macam mana uruskan budak-budak OKU.” (P13)

Most participants believed that educators should receive sufficient training to understand the needs of children with disabilities and provide inclusive educational environments. Similarly, another participant highlighted the importance of proactive learning among professionals who encounter individuals with rare diseases.

“Kalau boleh... setiap individu yang berprofesi sebagai doktor ke, cikgu ke, assistant ke... kena ada take the initiative untuk belajar... explore benda itu, check, tanya kat mana-mana... so at least boleh dapat membantulah.” (P12)

Several participants also suggested that rare disease education should be incorporated into professional training programs.

“Kalau doctor-doktor tu... masukkan ilmu ataupun bab rare disease ni... syllabus untuk doktor-doktor ni pun perlu lebih inklusif termasuk dengan rare disease ni.” (P8)

In addition, participants believed that increasing public awareness would reduce stigma and improve acceptance of children with rare diseases within schools and communities.

“Awareness tu kurang... kalau sekolah tu kan ada orang dengar macam rare disease tu dia orang macam oh budak ni dia rare disease, I tak mahu ambil dia lah. So sebab ada banyak orang I rasa dia orang tak faham kot.” (P3)

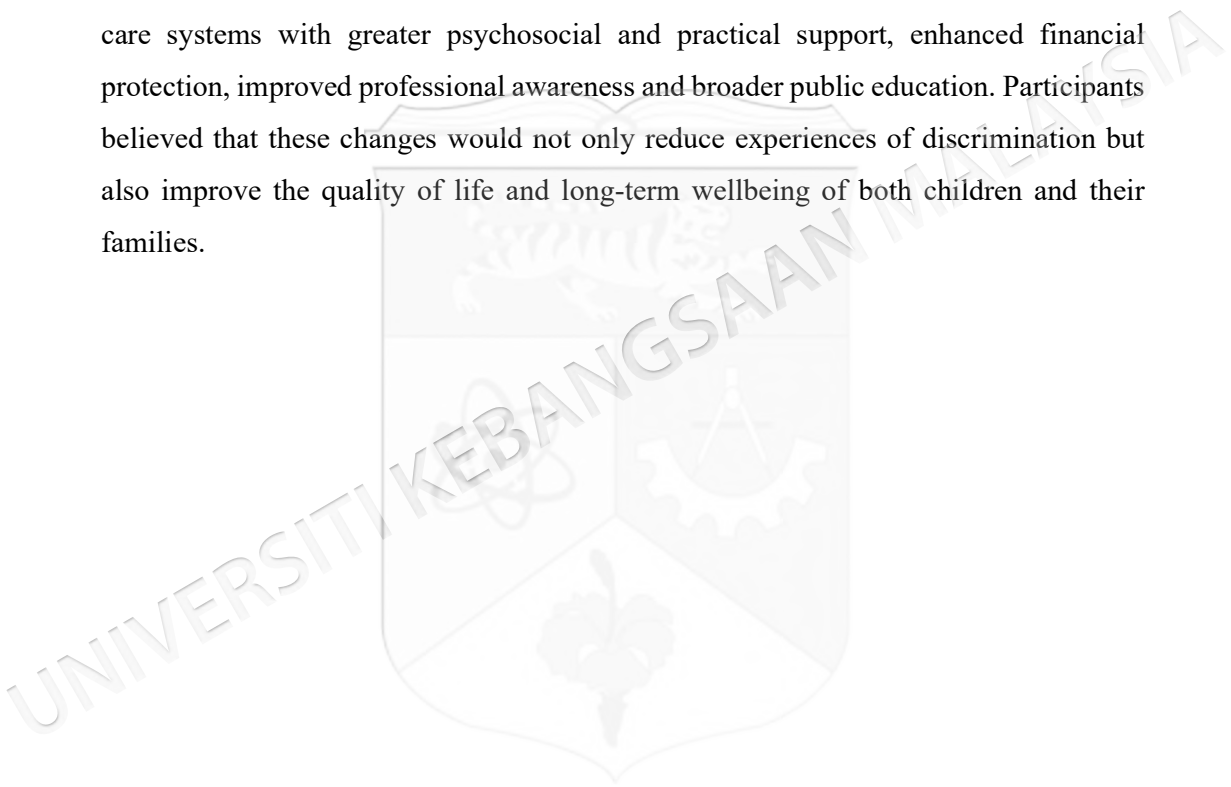
This participant perceived that exclusion and reluctance to accept children with rare diseases were often rooted in a lack of understanding rather than intentional discrimination. Greater awareness was therefore viewed as an important step towards reducing misconceptions and promoting inclusion.

Another participant emphasised the importance of involving families, educators, authorities, and other stakeholders in rare disease awareness programmes to improve understanding of the diverse experiences and needs of affected individuals.

"Untuk keluarga, pihak sekolah, authorities... saya rasa dia orang kena banyak join our program... baru boleh faham dengan lebih luas sebab SMA ni type dia banyak dan sangat berbagai." (P14)

The participant highlighted stakeholder engagement as a practical strategy for increasing awareness and fostering greater understanding of rare diseases. Instead of relying solely on information campaigns, she suggested that direct participation in awareness activities and interactions with affected families could help educators, policymakers, and community members develop a more informed and empathetic understanding of rare conditions.

All in all, participants recommended a more coordinated and inclusive approach to supporting families affected by rare genetic conditions. Priorities included integrated care systems with greater psychosocial and practical support, enhanced financial protection, improved professional awareness and broader public education. Participants believed that these changes would not only reduce experiences of discrimination but also improve the quality of life and long-term wellbeing of both children and their families.



CHAPTER V

DISCUSSIONS

5.1 INTRODUCTION

This chapter discusses the findings presented in Chapter 4 in relation to the study objectives and relevant literature. The discussion aims to provide a deeper interpretation of participants' experiences and examine how the findings contribute to the current understanding of genetic discrimination among families affected by rare genetic conditions. Drawing on existing research, the chapter explores the settings in which participants experienced discrimination, the impact of these experiences on individuals and families, and the strategies used to cope with the challenges encountered throughout their caregiving journey. The discussion also considers participants' recommendations for improving support and reducing discrimination, with particular attention given to the implications for healthcare services and policy development within the Malaysian context.

5.2 TYPES AND SETTINGS WHERE GENETIC DISCRIMINATION EXPERIENCED BY PARENTS WITH RARE DISEASE CHILDREN

The first objective of this study was to identify the types and settings in which genetic discrimination commonly occurs among parents of children with rare genetic conditions in Malaysia. The findings revealed that participants encountered experiences perceived as discriminatory, exclusionary, or disadvantageous across multiple aspects of daily life. These experiences occurred within healthcare settings, educational institutions, employment, insurance access, and social environments, although the nature and severity of these experiences varied between participants. While some experiences involved direct exclusion, others were more subtle and were linked to limited

awareness, inadequate accommodation, communication challenges, and structural barriers within existing systems.

These findings are consistent with previous research demonstrating that families affected by rare diseases frequently encounter challenges that extend beyond the medical consequences of the condition itself. Previous studies have reported experiences of stigma, social exclusion, healthcare barriers, educational difficulties, and financial challenges among individuals living with rare diseases and their families (von der Lippe et al., 2022; Supian et al., 2024). The findings of the present study further suggest that experiences of discrimination and disadvantage among families affected by rare genetic conditions may be embedded within everyday interactions and institutional practices, influencing not only access to services but also the overall wellbeing of affected families. The following sections discuss these experiences according to the settings in which they occurred.

5.2.1 Experience of Genetic Discrimination in Healthcare Settings

The findings suggest that experiences perceived as discriminatory in healthcare settings were rarely overt or directly linked to the use of genetic information. Instead, participants' accounts point towards a form of structural disadvantage in which the rarity of a condition contributed to delayed recognition, difficulties accessing specialist care, and increased reliance on parental self-advocacy. This distinction is important because it highlights that the experiences of families affected by rare genetic conditions may extend beyond conventional understandings of genetic discrimination, with disadvantage arising through healthcare systems that are not adequately equipped to respond to uncommon conditions.

A recurring issue was the perceived dismissal of parental concerns. Similar experiences have been reported in rare disease literature, where parents frequently describe needing to repeatedly advocate for their children before receiving appropriate investigations or referrals (von der Lippe et al. 2022). Rather than reflecting discriminatory intent, these experiences may indicate limitations in professional familiarity with rare conditions. Nevertheless, these experiences had consequences that may resemble those associated with discrimination, including frustration, delayed

access to care, and feelings of being unheard. They illustrate how structural features of healthcare systems can contribute to unequal experiences and outcomes even in the absence of deliberate prejudice (Bombard et al. 2012).

Participants also described difficulties navigating referral pathways and accessing specialist services. One participant questioned whether children from families with less confidence or healthcare knowledge might be overlooked if they were unable to advocate effectively for their needs. This observation is particularly significant because it highlights an issue of equity within healthcare systems. If access to appropriate care depends heavily on parental advocacy, then families may experience unequal outcomes despite having similar healthcare needs. Similar concerns have been reported among rare disease families internationally, where access to specialist care is often influenced by the ability of caregivers to navigate complex healthcare systems (Pelentsov, Laws & Esterman 2015; von der Lippe, Neteland & Feragen 2022).

An interesting finding was that several participants were themselves healthcare professionals, including a paediatrician, dentist, nurses, and a dental nurse. Despite their familiarity with healthcare systems and medical terminology, these participants still reported difficulties obtaining referrals and accessing appropriate services. This suggests that the challenges described cannot be attributed solely to differences in health literacy or parental knowledge. Instead, the findings lend further support to the interpretation that rarity itself may function as a source of structural disadvantage within healthcare systems.

At the same time, positive healthcare experiences were also reported. Participants valued healthcare professionals who were prepared, communicated openly, and recognised parental expertise regarding their child's condition. Similar findings have been reported by Gómez-Zúñiga et al. (2019), who found that trust was strengthened when healthcare professionals demonstrated empathy, transparency, and respect for parental knowledge. These findings suggest that although structural barriers may exist, collaborative and family-centred approaches can mitigate some of their negative effects.

Taken together, the findings suggest that healthcare-related experiences among families affected by rare genetic conditions may differ from traditional conceptualisations of genetic discrimination that focus on the misuse of genetic information. Participants rarely described overt prejudice or explicit differential treatment based on genetic test results. Instead, disadvantage was more commonly associated with delayed recognition, fragmented referral pathways, and limited familiarity with rare conditions. Viewed through this lens, the findings align more closely with the concept of structural disadvantage, whereby systemic factors inadvertently place certain groups at a disadvantage despite the absence of intentional discriminatory practices (Bombard et al. 2012; Otlowski et al. 2012). This interpretation highlights the complexity of discrimination and disadvantage experienced by families affected by rare genetic conditions and suggests that improving equity in healthcare requires not only greater awareness of rare diseases but also systems that reduce reliance on parental self-advocacy and facilitate timely access to appropriate care.

5.2.2 Experience of Genetic Discrimination in Parent's Employment Settings

In this setting, the findings suggest that employment-related disadvantage among parents of children with rare genetic conditions was experienced primarily through anticipated discrimination and structural disadvantage rather than direct workplace discrimination based on genetic information. Unlike the traditional genetic discrimination literature, which often focuses on adverse employment decisions resulting from disclosure of genetic risk, participants in this study more commonly described challenges arising from the realities of caregiving and the difficulty of reconciling these responsibilities with workplace expectations.

Several participants described modifying career decisions because they anticipated future difficulties balancing employment with their child's healthcare and support needs. One participant expressed hesitation about accepting a job opportunity because of concerns regarding medical appointments and educational commitments, while another acknowledged the possibility of leaving employment if caregiving demands increased. These accounts are consistent with the concept of anticipated discrimination described by Bombard et al. (2012), whereby individuals alter behaviour

in response to expected barriers or negative consequences rather than actual discriminatory events. Similar observations have been reported by Wauters and Van Hoyweghen (2016), who noted that fears of discrimination are often more common than documented experiences of discrimination itself. This distinction is important because it suggests that employment-related disadvantage may occur even in the absence of overt discriminatory behaviour.

At the same time, participants described workplace environments that appeared insufficiently equipped to accommodate intensive caregiving responsibilities. Experiences such as difficulties obtaining flexibility, pressure associated with repeated emergency leave, and expectations of uninterrupted productivity suggest the presence of structural disadvantage. Rather than reflecting deliberate prejudice, these experiences arose from workplace systems that assumed employees could maintain consistent attendance and performance regardless of caregiving demands. Similar challenges have been reported in studies of parents caring for children with rare and chronic conditions, where employment participation is frequently constrained by ongoing caregiving responsibilities and limited workplace accommodation (Davidson et al. 2024; von der Lippe, Neteland & Feragen 2022).

An important distinction emerged when comparing these findings with existing genetic discrimination literature. Studies involving hereditary cancer syndromes and predictive genetic testing often focus on concerns regarding direct employment discrimination based on an individual's genetic status or future disease risk for example, when genetic information being used in decisions about hiring, promotion, job assignment, or continued employment (Gopalakrishnan et al. 2024). In contrast, participants in the present study rarely reported enacted discrimination directly attributable to genetic information. Instead, disadvantage was experienced through the social and occupational consequences of caring for a child with a rare genetic condition. These findings suggest that employment-related disadvantage in paediatric rare disease contexts may not always conform to traditional conceptualisations of genetic discrimination, as experiences of disadvantage may be closely intertwined with the broader realities of caregiving. Rather than arising solely from the misuse of genetic

information, disadvantage may arise through systems that inadequately accommodate the long-term demands associated with managing a rare condition.

The findings also highlight the close relationship between employment, financial wellbeing, and caregiving burden. Participants described making career decisions based on their children's needs, often at the expense of career progression, income stability, and future financial security. Similar patterns have been reported among families affected by rare diseases, where caregiving responsibilities contribute to reduced workforce participation and increased financial vulnerability (Supian et al. 2024). Importantly, these consequences were experienced not only as economic challenges but also as sources of emotional stress and uncertainty regarding the future.

As a conclusion, the findings suggest that employment-related discrimination in this study was rarely enacted through direct workplace prejudice. Instead, disadvantage was more commonly experienced through employment difficulties, limited workplace accommodation, and structural barriers that made balancing employment and caregiving responsibilities particularly challenging. These findings highlight the importance of flexible working arrangements, caregiver-friendly workplace policies, and greater recognition of the realities faced by employees caring for children with complex and lifelong health conditions.

5.2.3 Experience of Genetic Discrimination in Insurance Access

Among all settings explored in this study, insurance represented the clearest example of enacted genetic discrimination. Unlike experiences in healthcare, education, and employment, where disadvantage was often linked to structural barriers or anticipated discrimination, participants described direct exclusion from insurance coverage following the diagnosis of a rare genetic condition. Insurance applications were rejected, coverage for genetic conditions was excluded, and in some cases existing policies were terminated. These experiences align closely with conventional definitions of genetic discrimination, whereby genetic information is used to determine access to opportunities, services, or resources (Otlowski et al. 2012; Kim et al. 2021).

The findings are consistent with longstanding international concerns regarding the use of genetic information in insurance underwriting (Bombard et al. 2012; Joly, Ngueng Feze & Simard 2013). Similar debates have led to legislative protections and regulatory reforms in several countries, including the United States, Canada, and Australia (Tiller et al. 2021). In contrast, Malaysia currently has no specific legislation addressing genetic discrimination in insurance, highlighting a potential policy gap as genetic testing becomes increasingly integrated into routine healthcare. From the perspective of participants, insurance exclusion was not viewed simply as a financial or actuarial matter but as an issue of fairness, particularly because those most likely to require healthcare support were also those facing the greatest barriers to obtaining financial protection.

At the same time, the findings also revealed evidence of anticipated discrimination. Several participants described obtaining insurance before a diagnosis was confirmed, maintaining policies secured prior to genetic testing, or modifying insurance-related decisions because they expected future difficulties obtaining coverage. Consistent with Bombard et al. (2012), these accounts suggest that the impact of genetic discrimination extends beyond actual discriminatory events and may influence behaviour through the anticipation of future exclusion. Similar patterns have been reported internationally, where concerns regarding insurance discrimination affect decisions related to genetic testing, disclosure, and financial planning (Wauters & Van Hoyweghen 2016; Tiller et al. 2024). The findings therefore demonstrate that insurance discrimination may operate through both enacted and anticipated pathways.

An additional observation was that insurance-related concerns were reported across participants from diverse educational, occupational, and socioeconomic backgrounds. Healthcare professionals, educators, and participants with postgraduate qualifications described experiences similar to those reported by participants with lower educational attainment. This suggests that concerns regarding insurability are not confined to particular demographic groups and may arise regardless of professional knowledge or socioeconomic position once a genetic diagnosis becomes known. Compared with other forms of disadvantage that may be influenced by income or educational resources, insurance exclusion appeared to be more directly associated with

the child's diagnosed genetic condition and the disclosure or recognition of that condition.

An important distinction also emerged when comparing the present findings with much of the existing genetic discrimination literature. Previous studies have largely focused on predictive genetic information among otherwise healthy individuals, particularly those at risk of hereditary cancer syndromes (Gopalakrishnan et al. 2024; Tiller et al. 2024). In contrast, participants in this study described discrimination associated with an existing diagnosed condition affecting their children. Consequently, insurance discrimination was experienced not as a hypothetical future concern but as an immediate and tangible challenge affecting access to financial protection. This distinction further supports the argument that genetic discrimination in rare disease contexts may differ from traditional conceptualisations centred on predictive genetic risk.

Overall, the findings raise broader questions regarding equity for individuals living with rare genetic conditions. Rare diseases often require lifelong monitoring, multidisciplinary care, repeated healthcare utilisation, and substantial financial resources (Nguengang Wakap et al. 2020). Yet participants described a situation in which those most likely to benefit from financial protection were often the least able to obtain it. Viewed through this lens, insurance discrimination represents not only an individual barrier but also a form of structural disadvantage that may amplify the long-term burden experienced by affected families. As genomic medicine continues to expand in Malaysia, consideration may need to be given to how the benefits of genetic testing can be realised without creating unintended barriers to insurance access and financial security.

5.2.4 Experience of Genetic Discrimination in Educational Settings

The findings suggest that educational disadvantage was experienced through a combination of anticipated discrimination, enacted discrimination, and structural barriers to inclusion. Educational challenges were commonly described in relation to concerns regarding acceptance, accommodation, participation, and assumptions about children's capabilities. These findings indicate that discrimination within educational

settings may not arise solely from the child's diagnosis itself but also from how disability and difference are understood and managed within schools.

One particularly noteworthy finding was the presence of anticipated discrimination. A parent reported deliberately withholding disclosure of her child's OKU status during school enrolment because she feared that disclosure might result in rejection. Although no discriminatory act ultimately occurred, the expectation of negative treatment influenced parental decision-making. This finding aligns with Bombard et al.'s (2012) concept of anticipated discrimination, whereby individuals alter their behaviour because they expect future discrimination, regardless of whether discrimination actually takes place. Similar observations have been reported among individuals with disabilities and genetic conditions, where concerns regarding stigma and exclusion often lead to selective disclosure (Wauters & Van Hoyweghen 2016). Malaysia's Zero Reject Policy was introduced in 2019 to support the right of children with disabilities to access education by preventing their rejection from government schools on the basis of disability and promoting their inclusion within the education system. However, despite this policy, the present finding indicates that parental fears of rejection may persist, suggesting a possible gap between formal policy protection and families' perceptions of how effectively such protections are implemented (Ensimau, Hamzah & Yassin 2022). This may discourage parents from disclosing their child's condition, potentially limiting access to appropriate educational support and accommodations.

The findings also suggest that access to education does not necessarily translate into meaningful inclusion. Participants described situations in which schools were informed of a child's condition but failed to provide appropriate accommodations or communicate effectively regarding the child's needs. Rather than reflecting deliberate exclusion, these experiences may be better understood as structural or disability-related barriers to inclusion, in which they may also have discriminatory consequences for children's participation and educational opportunities, where systems are insufficiently prepared to support children with uncommon and complex conditions. Similar concerns have been raised within inclusive education literature, which argues that genuine

inclusion requires not only access but also appropriate support, participation, and safety within the educational environment (UNICEF 2014; Verger et al. 2021).

Another important finding relates to assumptions regarding children's abilities and educational potential. Participants described educational decisions that appeared to be influenced by developmental limitations rather than individual strengths and support needs. Such experiences may reflect enacted discrimination, where assumptions about developmental limitations directly influenced educational opportunities or placement decisions. These findings are consistent with the social model of disability, which argues that disadvantage arises not solely from impairment but from environments and systems that fail to accommodate diversity (Shakespeare 2017). Rather than asking how educational environments can be adapted to support children with rare genetic conditions, responsibility for inclusion appeared to be placed on the child to fit existing educational expectations.

The findings further suggest that educational discrimination may extend beyond institutional practices and manifest through everyday social interactions. Participants described experiences of bullying, peer rejection, and feelings of being different from other children. Such experiences are consistent with stigma processes described by Pachankis et al. (2018), whereby visible differences become associated with negative stereotyping, social distancing, and exclusion. Similar experiences have been reported among children living with rare diseases, who often encounter difficulties forming peer relationships and achieving social inclusion within mainstream educational settings (Adama et al. 2023). Importantly, these experiences affected not only educational participation but also children's social wellbeing and developing sense of identity.

Altogether, the findings suggest that educational discrimination in the context of rare genetic conditions extends beyond denial of school access. Instead, disadvantage was experienced through anticipated discrimination, inadequate accommodation, stigma, and assumptions regarding children's capabilities. These findings support the argument that genetic discrimination within rare disease contexts is often intertwined with broader disability-related stigma and structural barriers to inclusion. Consequently, the challenge is not simply ensuring that children with rare genetic conditions are

admitted to school, but ensuring that they are able to participate meaningfully, safely, and equitably within educational environments. Greater disability awareness, educator training, and stronger implementation of inclusive education principles may therefore be required to reduce educational disadvantage experienced by this population.

5.2.5 Social Stigma and Community Discrimination

The findings suggest that social experiences of families affected by rare genetic conditions existed along a continuum ranging from acceptance and inclusion to stigma and social exclusion. Unlike insurance discrimination, which was frequently experienced as enacted discrimination through direct exclusion from services, social disadvantage was more commonly mediated through stigma processes, misunderstanding, and negative assumptions about disability and genetic conditions. This highlights the complex nature of social discrimination, where disadvantage may arise even in the absence of overt hostility or intentional prejudice.

A prominent finding was that many discriminatory experiences appeared to stem from limited public understanding of rare genetic conditions. Participants described situations in which family members, neighbours, or members of the wider community attributed their child's condition to poor parenting, inadequate nutrition, or other behavioural factors. These experiences are consistent with stigma processes described by Pachankis et al. (2018), whereby visible differences become associated with negative stereotypes and socially undesirable attributes. Similar findings have been reported among families affected by rare diseases, where limited public awareness contributes to misconceptions, judgement, and social marginalisation (Supian et al. 2024). From this perspective, social discrimination may be understood not solely as intentional prejudice but also as a consequence of inadequate knowledge and understanding of rare genetic conditions.

A significant consequence of these misconceptions was parental blame. Several participants reported being questioned about decisions made during pregnancy, parenting practices, or lifestyle factors, implying responsibility for outcomes largely beyond their control. Such experiences reflect a form of enacted stigma, whereby parents become targets of judgement because others seek behavioural or moral

explanations for a child's condition. Similar experiences have been documented among caregivers of children with rare diseases and disabilities, where feelings of guilt, blame, and social scrutiny are common despite the biological basis of the condition (Shukor, Norhayati & Ahmad 2026). This finding is particularly significant because it demonstrates how genetic conditions may be socially interpreted through moral narratives rather than biomedical understanding.

The findings also suggest the presence of anticipated discrimination. Some participants described proactively explaining their child's condition to others in order to prevent misunderstanding, reduce negative assumptions, or avoid potential judgement. Consistent with Bombard et al. (2012), these behaviours can represent as responses to anticipated discrimination, whereby individuals manage information disclosure based on expectations of future stigma or negative treatment. Similar patterns have been reported among families affected by rare diseases, who often assume the role of educators and advocates to navigate social interactions and challenge misconceptions (Zhu, Smith & Parrott 2017). While such strategies may reduce misunderstanding, they also place an additional emotional burden on families who must repeatedly explain and justify their child's condition.

At the same time, participants' accounts also provided evidence that social experiences were not uniformly negative. Several participants described supportive family members, understanding friends, and positive interactions within disability and rare disease communities. Others perceived that public acceptance of disability had improved over time, reducing experiences of overt stigma. These findings provide an important contrast to literature that focuses primarily on discrimination and suggest that increased awareness, exposure to disability, and participation in support networks may promote more inclusive social environments. The coexistence of acceptance and discrimination within participants' narratives highlights the dynamic and evolving nature of societal responses towards rare genetic conditions.

Taken together, the findings suggest that social discrimination in the context of rare genetic conditions is often experienced through stigma processes, anticipated discrimination, and misunderstanding rather than direct hostility. Importantly, the

findings also demonstrate that discrimination and acceptance can coexist within the same social environment. This observation broadens conventional understandings of genetic discrimination by showing that social disadvantage may arise not only from the misuse of genetic information but also from societal interpretations of difference, disability, and rarity. Consequently, efforts to reduce discrimination may require not only legal or policy protections but also broader public education initiatives aimed at improving understanding of rare genetic conditions and reducing stigma within the community.

5.3 INTERPRETATION OF GENETIC DISCRIMINATION, IMPACT AND COPING STRATEGIES

The second objective of this study was to understand how parents of children with rare genetic conditions interpreted experiences of genetic discrimination, how these experiences affected them, and how they responded to the challenges encountered. While Objective 1 identified the settings in which discrimination occurred, the present findings suggest that discrimination was rarely experienced as a discrete or isolated event. Instead, participants described discrimination as being closely intertwined with diagnostic uncertainty, caregiving responsibilities, financial pressures, social stigma, and concerns regarding their children's future. Consequently, the impact of discrimination was often cumulative, influencing multiple aspects of family life simultaneously.

Importantly, the findings suggest that the difficulty in separating discrimination from the broader experience of living with a rare genetic condition may itself represent a meaningful finding rather than merely a methodological limitation. Much of the existing genetic discrimination literature focuses on asymptomatic individuals at risk of future disease, particularly within hereditary cancer syndromes and predictive genetic testing contexts (Bombard et al. 2009; Gopalakrishnan et al. 2024). In contrast, participants in the present study were caring for children with established and often complex conditions requiring ongoing medical, educational, and social support. As a result, discriminatory experiences were frequently embedded within everyday caregiving challenges. This suggests that genetic discrimination in rare disease contexts

may differ qualitatively from traditional conceptualisations because it is often inseparable from the broader realities of caregiving and living with a rare condition.

This interpretation is consistent with research demonstrating that families affected by rare diseases frequently experience multiple and interacting burdens, including emotional distress, financial strain, social isolation, service fragmentation, and uncertainty regarding the future (Pelentsov, Laws & Esterman 2015; von der Lippe, Neteland & Feragen 2022). Rather than operating independently, these stressors often accumulate and reinforce one another over time (Currie & Szabo 2019). The findings therefore suggest that the impact of genetic discrimination may be best understood within a broader ecosystem of disadvantage, where discriminatory experiences interact with existing caregiving and psychosocial challenges to shape family wellbeing.

5.3.1 How Parents Interpreted Experiences of Genetic Discrimination

An important finding of this study was that participants did not consistently interpret their experiences of disadvantage as discrimination, such as deliberate prejudice or intentional unfair treatment. While some experiences, particularly those involving insurance exclusion, were clearly perceived as genetic discrimination, many participants attributed negative experiences to limited awareness, inadequate knowledge, or insufficient understanding of rare genetic conditions. This was especially evident within healthcare, educational, and community settings. Such interpretations align with contemporary understandings of discrimination that recognise disadvantage may arise through structural and institutional processes rather than overt discriminatory acts alone (Otlowski, Taylor & Bombard 2012).

Participants frequently distinguished between the actions of individuals and the limitations of broader systems. Difficulties obtaining specialist referrals, accessing educational accommodations, or securing appropriate support were often interpreted as consequences of systems that were insufficiently prepared to respond to rare conditions. This interpretation is consistent with the concept of structural disadvantage, whereby individuals experience unequal outcomes not because of intentional prejudice but because existing systems are organised around the needs of more common conditions (Zurynski et al. 2017). Viewed through this lens, rarity itself may function as a source

of disadvantage, as uncommon conditions often receive less recognition, fewer resources, and lower institutional preparedness.

The findings also demonstrated considerable variation in how participants defined discrimination. Some participants explicitly labelled experiences as discriminatory, particularly when they involved direct exclusion from insurance coverage, educational opportunities, or services. Others were reluctant to use the term discrimination and instead described their experiences as misunderstanding, ignorance or lack of awareness. This variation reflects ongoing debates within the genetic discrimination literature regarding whether discriminatory intent is necessary for an experience to be considered discrimination (Rud et al. 2026). The present findings support the view that the consequences of discrimination may occur regardless of intent. Whether actions were perceived as deliberate or unintentional, participants frequently described similar outcomes, including exclusion, frustration, emotional distress, and reduced access to support.

Perhaps the most significant finding was that many experiences described by participants may be more accurately understood as perceived discrimination, phenotype-based discrimination, or disability-related stigma rather than discrimination arising directly from genetic information itself. Unlike populations commonly studied in genetic discrimination research, most participants did not routinely disclose their children's genetic test results, specific genetic variants, or molecular diagnoses outside specialist healthcare settings. Instead, disadvantage was often triggered by visible manifestations of the condition, including physical differences, developmental delays, disability status, medical complexity, or the caregiving demands associated with the condition. Consequently, families were often responding not to reactions towards genetic information, but to reactions towards the observable consequences of a genetic condition.

The diversity of diagnoses represented in this study further highlights the importance of considering phenotypic visibility when interpreting these experiences. The children had conditions with varying clinical manifestations, with some associated with more readily recognisable physical or developmental features, while others may

be less apparent without knowledge of the underlying diagnosis. This variation may influence the pathway through which disadvantage is experienced. Where a condition is phenotypically apparent, negative treatment may occur in response to visible physical, developmental, or functional differences without others being aware of the child's specific genetic diagnosis. Conversely, for conditions with less readily apparent manifestations, experiences specifically related to the genetic condition may depend more heavily on disclosure or recognition of genetic information. However, these pathways are not mutually exclusive, as a visible phenotype may lead to diagnostic disclosure and subsequent reactions to genetic information.

This distinction is theoretically important because it suggests that genetic discrimination in rare disease populations may operate differently from traditional models centred on predictive genetic risk. Rather than being driven solely by disclosure of genetic information, experiences occurring in the context of rare genetic conditions may also intersect with social responses to disability, difference, and rarity. The findings therefore suggest that genetic discrimination in rare disease contexts may overlap with, but should not be equated with, phenotype-based disadvantage, disability-related stigma, and structural barriers. Such an interpretation provides a more nuanced understanding of how genetic discrimination is experienced by families affected by rare diseases and represents an important contribution of the present study.

5.3.2 How Parents Are Affected by Experiences of Genetic Discrimination

The findings suggest that the impact of genetic discrimination extended beyond isolated discriminatory events and was experienced through a broader accumulation of emotional, psychological, and social burdens. Participants described distress arising not only from direct experiences of exclusion but also from ongoing encounters with healthcare barriers, educational challenges, financial uncertainty, social stigma, and concerns regarding their children's future. Importantly, the findings reinforce the argument that discrimination in the context of rare genetic conditions is often inseparable from the wider realities of caregiving. Rather than operating as a distinct source of distress, discriminatory experiences frequently interacted with existing caregiving challenges, amplifying their overall impact on family wellbeing.

A key finding was that participants were affected by both enacted discrimination and anticipated discrimination. Experiences such as insurance exclusion, educational barriers, and social stigma represented direct forms of disadvantage that contributed to feelings of frustration, sadness, and helplessness. However, participants were also affected by concerns about future discrimination, particularly regarding their children's education, employment opportunities, financial security, independence, and social acceptance. Consistent with Bombard et al. (2012), these findings suggest that the consequences of discrimination may extend beyond actual events and operate through anticipation of future disadvantage. Similar observations have been reported in genetic discrimination literature, where fear of discrimination can have psychological effects even when direct discriminatory experiences are absent (Wauters & Van Hoyweghen 2016).

The findings also highlight the important role of stigma processes in shaping parents' emotional experiences. Participants described distress associated with blame, misunderstanding, judgement, and negative assumptions regarding their children's conditions. Such experiences are consistent with stigma frameworks which propose that social reactions to difference can contribute to emotional burden, social isolation, and reduced wellbeing (Pachankis et al. 2018). Notably, some participants linked their distress not to the condition itself, but to how others responded to the condition. This suggests that social attitudes and community responses may significantly influence how families experience and adapt to rare genetic conditions.

A significant observation was that participants often struggled with uncertainty regarding the future. Concerns about long-term care, educational opportunities, employment prospects, financial stability, and future caregiving arrangements were common across interviews. Consistent with Han et al. (2017), these findings reflect the broader uncertainty associated with rare genetic conditions, encompassing uncertainty about prognosis, future outcomes, and decision-making. While uncertainty is widely recognised within rare disease literature, the present findings suggest that concerns regarding future exclusion and disadvantage may further intensify this burden. In this way, the psychological impact of discrimination may extend beyond present experiences and shape expectations about future opportunities and quality of life.

At the same time, the findings demonstrated that emotional responses were dynamic rather than static. Several participants described gradual adaptation following diagnosis, particularly after receiving diagnostic confirmation, obtaining information about the condition, or developing confidence in managing their child's needs. This observation is consistent with previous studies showing that parents often move from initial shock and uncertainty towards greater acceptance and adjustment over time (von der Lippe et al. 2022). The finding suggests that although discrimination, stigma, and caregiving challenges may contribute to distress, access to information, support, and appropriate services can strengthen resilience and facilitate adaptation.

Therefore, the findings suggest that the impact of genetic discrimination among families affected by rare genetic conditions extends beyond direct experiences of exclusion. Parents were affected by a combination of enacted discrimination, anticipated discrimination, stigma, uncertainty, and structural disadvantage, all of which interacted with the existing demands of caregiving. This reinforces the notion that genetic discrimination in rare disease contexts may differ from traditional conceptualisations focused on isolated discriminatory events, as its consequences are often embedded within the broader experience of living with and caring for a child with a rare genetic condition.

5.3.3 How Parents Respond to and Cope with Discrimination and Rare Disease Caregiving

The findings suggest that parents employed a range of strategies to manage both actual experiences of discrimination and the anticipation of future stigma or exclusion. While many coping strategies were developed in response to the broader challenges of raising a child with a rare genetic condition, several appeared specifically directed towards managing social judgement, misunderstanding, and potential discrimination. These responses included selective disclosure, advocacy and education, social withdrawal, and seeking support from individuals with shared experiences.

One notable finding was the use of selective disclosure as a strategy to manage anticipated discrimination. For example, one participant chose not to disclose her child's disability status during school enrolment because she feared that disclosure might

reduce the child's chances of acceptance. Consistent with Bombard et al. (2012), this behaviour can be interpreted as a response to anticipated discrimination, whereby individuals modify their actions based on expectations of future negative treatment. Similar patterns have been reported among individuals living with genetic conditions who carefully consider the potential risks and benefits of disclosure before sharing health-related information (Bombard et al. 2009). In the present study, selective disclosure functioned as a protective strategy aimed at reducing the likelihood of exclusion and preserving access to opportunities.

The findings also highlight the important role of advocacy and education as active responses to perceived discrimination and misunderstanding. Rather than avoiding disclosure altogether, many participants chose to proactively explain their children's conditions to teachers, healthcare professionals, family members, and community members. Such efforts were intended to improve understanding, facilitate accommodations, and challenge inaccurate assumptions. This finding suggests that parents frequently assumed the role of advocates, not only for their children but also for rare disease awareness more broadly. Similar observations have been reported in rare disease literature, where parents often become key sources of information and advocates for their children's needs within healthcare, educational, and social settings (Davidson et al. 2024; Gómez-Zúñiga et al. 2019). In this context, advocacy served both as a practical coping strategy and as a means of addressing the underlying causes of discrimination.

At the same time, some responses reflected efforts to minimise exposure to stigma. Participants whose children had visible physical differences, disabilities, or behavioural challenges described avoiding certain social situations, reducing participation in gatherings, or limiting interactions with individuals perceived as judgemental. These behaviours are consistent with stigma management strategies described in the broader stigma literature, where individuals may withdraw from potentially threatening social environments to protect themselves from negative experiences (Pachankis et al. 2018). While such strategies may reduce immediate emotional distress, they may also inadvertently contribute to social isolation and reduced community participation over time (Baumbusch, Mayer & Sloan-Yip 2018).

Interestingly, many participants balanced these protective strategies by actively seeking support from others with similar lived experiences. Peer support groups, rare disease organisations, and online communities were frequently described as sources of emotional support, practical advice, validation, and understanding. These findings are consistent with previous studies demonstrating that peer support networks can strengthen resilience, reduce feelings of isolation, and facilitate adaptation among families affected by rare diseases (Fendrich et al. 2025; Joo et al. 2022). Such communities may also provide an environment where families feel less vulnerable to stigma because their experiences are understood and normalised by others facing similar challenges.

Taken together, the findings suggest that parents responded to discrimination through a combination of active and protective coping strategies. Some sought to challenge discrimination through advocacy, education, and information sharing, whereas others attempted to minimise exposure to stigma through selective disclosure or social withdrawal. Importantly, these responses highlight how families are often required to actively manage environments that may not fully understand or accommodate rare genetic conditions. The findings therefore suggest that reducing discrimination should not rely solely on families' coping efforts but also require broader improvements in awareness, inclusivity, and support across healthcare, educational, workplace as well as community settings.

5.4 SUGGESTIONS FOR REDUCING GENETIC DISCRIMINATION AND IMPROVING SUPPORT FOR FAMILIES AFFECTED BY RARE GENETIC CONDITIONS

The third objective of this study was to explore participants' perspectives on how experiences of discrimination and disadvantage could be reduced for families affected by rare genetic conditions. Importantly, participants rarely focused on discrimination as an isolated problem. Instead, their recommendations centred on improving awareness, strengthening service coordination, increasing psychosocial support, enhancing financial protection, and creating more inclusive environments. This finding is consistent with the broader argument emerging from the study that genetic discrimination in rare disease contexts often operates through structural barriers, stigma,

and limited institutional preparedness rather than through overt discriminatory acts alone.

A recurring recommendation was the need to improve awareness and understanding of rare genetic conditions among healthcare professionals, educators and the wider community. Participants frequently attributed negative experiences to limited knowledge, misunderstanding, and negative assumptions rather than deliberate prejudice or overt hostility. Similar concerns have been reported internationally, where inadequate awareness of rare diseases contributes to delayed diagnosis, fragmented care, stigma, and exclusion from appropriate support (Verger et al. 2021). Increasing professional and public awareness may therefore help reduce indirect forms of discrimination by improving responsiveness, accommodation, and inclusion across different settings. Consistent with Baynam, Gomez and Jain (2024), improving public understanding may also reduce stigma and promote social inclusion. Participants themselves frequently engaged in awareness activities, advocacy efforts, and peer support initiatives, suggesting that affected families can play an important role in challenging misconceptions and promoting greater societal understanding of rare genetic conditions.

Participants also highlighted the importance of more integrated and coordinated services. Difficulties navigating healthcare systems, accessing specialist care, and obtaining educational or therapeutic support were commonly described as sources of frustration and disadvantage. Similar findings have been reported among rare disease families internationally, who often face fragmented care pathways and substantial responsibility for coordinating services themselves (Walton et al. 2023). The introduction of Malaysia's National Policy for Rare Diseases (2025) represents an important step towards addressing these challenges through a more coordinated national approach. However, participants' experiences suggest that further efforts are needed to ensure that services remain accessible, connected, and responsive to families' long-term needs. Established programs in Australia provide useful examples of how these services could be strengthened. The NSW Rare Disease Research-Enabled Model of Care incorporates care coordination, multidisciplinary services and telehealth, while Australia's National Disability Insurance Scheme (NDIS) provides eligible individuals

with disability-related supports such as personal care, therapy and home modifications (Rare Diseases NSW 2026; National Disability Insurance Agency 2026). Drawing from these approaches, Malaysia could further develop community-based and home-based support services that bring care closer to families and reduce the burden of travelling to specialist centres and coordinating multiple services. Such services could include home-based therapy, community care, care coordination and links between healthcare, education and social support, particularly for children requiring long-term multidisciplinary care.

Another important recommendation concerned psychosocial support for caregivers. Participants frequently described emotional exhaustion, uncertainty, and social isolation, yet many perceived that healthcare services focused primarily on the affected child rather than the wellbeing of the family as a whole. Similar observations have been reported in previous studies, where caregivers of children with rare diseases often experience substantial psychological burden while receiving limited formal support (Pelentsov, Laws & Esterman 2015; Shukor, Norhayati & Ahmad 2026). Strengthening access to counselling services, peer support programs, and caregiver-focused interventions may therefore help mitigate some of the emotional consequences associated with discrimination, stigma, uncertainty, and long-term caregiving responsibilities.

Financial protection and insurance access emerged as particularly important areas for improvement. Participants frequently viewed insurance exclusion as unfair because those most likely to require healthcare support were often the least able to obtain financial protection. Internationally, concerns regarding genetic discrimination have contributed to legislative responses such as the Genetic Information Nondiscrimination Act (GINA) in the United States and the Genetic Non-Discrimination Act in Canada. However, an important limitation of GINA is that its protections do not generally extend to information about a disease that has already manifested, meaning that individuals with an existing genetic condition may not receive the same protection as those whose genetic information indicates only a future disease risk. The present findings suggest that legislation targeting misuse of genetic information alone may not fully address the forms of disadvantage experienced by

families affected by rare genetic conditions. While insurance exclusion represented a clear example of enacted discrimination, many other experiences identified in this study were associated with structural barriers, inadequate accommodations, limited awareness, and stigma rather than explicit misuse of genetic information. Consequently, legal protections against genetic discrimination, although important, may be insufficient if not accompanied by broader efforts to improve service accessibility, disability inclusion, rare disease awareness, and social support. This distinction highlights a potential gap between existing legislative models and the lived realities of families affected by rare genetic conditions.

Finally, the findings suggest that reducing genetic discrimination requires more than addressing individual discriminatory acts or restricting misuse of genetic information. Rather, meaningful change may depend on addressing the broader structural and social conditions that give rise to disadvantage, including limited awareness, fragmented services, inadequate accommodations, financial barriers, and stigma. These findings support a more holistic approach to genetic discrimination that extends beyond legal protections alone and recognizes the importance of healthcare, educational, social, and policy interventions in creating more equitable environments for individuals and families affected by rare genetic conditions.

CHAPTER VI

IMPLICATIONS, FUTURE WORKS AND CONCLUSION

6.1 IMPLICATIONS FOR GENETIC COUNSELLING PRACTICE

The findings of this study underscore the important role of genetic counsellors in supporting families beyond the simple delivery of genetic information. Many participants described difficulties related to disclosure, social misunderstanding, access to education, barriers in obtaining insurance, and uncertainty about how others might react to their child's diagnosis. These findings suggest that genetic counselling should include open discussion of possible experiences of stigma and discrimination, including both the potential advantages and risks of revealing a genetic diagnosis in different settings. This may be incorporated as anticipatory guidance, whereby genetic counsellors prepare families for potential psychosocial, social, and practical challenges that may arise following diagnosis or disclosure. Genetic counsellors may also have an important role in correcting misconceptions about genetic conditions, especially when parents encounter blame, judgement, or misunderstanding from relatives, teachers, or the wider community.

The findings also highlight the need for psychosocial assessment within genetic counselling practice. Genetic counsellors receive training in psychosocial assessment and counselling as part of their professional preparation, enabling them to explore patients' and families' emotional responses, coping, family relationships, support systems, and adjustment to genetic conditions (Accreditation Council for Genetic Counseling 2023). Several participants reported considerable emotional distress, anxiety, caregiver burden, and concern about their child's future, highlighting the importance of identifying such concerns early. While genetic counsellors do not replace psychologists or other mental health professionals, they can identify psychosocial needs, provide appropriate counselling and support within their scope of practice, and refer families for further psychological or social support when required. Genetic

counsellors can also connect families with rare disease organizations and peer support networks and advocate for those facing barriers in healthcare, education, or social settings. As genetic services continue to develop in Malaysia, incorporating psychosocial assessment, family advocacy, and discussion of possible stigma and discrimination into routine genetic counselling may help families cope more effectively with the long-term challenges associated with rare genetic conditions.

6.2 IMPLICATIONS FOR HEALTHCARE SERVICES AND POLICY

This study suggests that reducing genetic discrimination requires more than simply preventing obvious acts of unfair treatment. Many participants described disadvantage that arose from limited awareness of rare genetic conditions, fragmented services, inadequate accommodations, and difficulties in accessing appropriate support. These experiences suggest that, in the context of rare diseases, genetic discrimination often appears through structural and institutional processes rather than through clear or deliberate prejudice. For this reason, healthcare services should do more than focus on diagnosis and treatment; they should also be designed to respond to the specific needs of individuals and families affected by rare genetic conditions. The need for integrated and family-centred support identified in this study is also consistent with findings among Malaysian families affected by SMA, where caregivers highlighted the substantial psychosocial and practical burden of managing a rare genetic condition (Ch'ng et al. 2022).

The findings also support the continued implementation of Malaysia's National Policy for Rare Diseases (2025), especially its focus on raising awareness, improving service coordination, and expanding access to supportive care. However, the findings suggest that Malaysia's rare disease support infrastructure remains relatively limited beyond medical care, particularly in the areas of psychosocial, educational, social, and community-based support. Participants described difficulties accessing educational and therapeutic services, limited awareness among the wider community, caregiver burden, and a lack of coordinated support, indicating that families may continue to manage many of these needs on their own. Better awareness among healthcare professionals, educators, insurers, and the wider community may help reduce misunderstanding and

exclusion experienced by affected families. At the policy level, greater attention should therefore be given to strengthening psychosocial support, educational inclusion, community and peer support, and care coordination, alongside reducing insurance-related barriers and ensuring fair access to healthcare, therapies, and support services. The findings of this study provide local evidence of these gaps and may inform the further development and implementation of the National Policy for Rare Diseases by highlighting the psychosocial and social needs that may not be fully addressed through medical care alone.

Several countries have also introduced laws specifically aimed at preventing genetic discrimination, such as the Genetic Information Nondiscrimination Act (GINA) in the United States and the Genetic Non-Discrimination Act in Canada. Although these laws mainly focus on areas such as employment and insurance, they also reflect broader efforts to prevent discrimination based on genetic information before it occurs. As genetic testing becomes more widely available, policymakers in Malaysia may need to consider whether current legal and regulatory frameworks offer enough protection against discrimination in less visible areas such as healthcare, education, and social participation. The findings of this study further suggest that policy responses should not focus solely on preventing the misuse of genetic information, but should also address the broader social and structural disadvantages experienced by families affected by rare genetic conditions.

6.3 STRENGTHS AND LIMITATIONS OF THE STUDY

This study contributes to an underexplored area of research by examining experiences of genetic discrimination among parents of children with rare genetic conditions in Malaysia. To the best of the researcher's knowledge, this is among the first local qualitative studies to specifically explore genetic discrimination within the rare disease context. While previous Malaysian studies have examined caregiver burden, healthcare experiences, and psychosocial challenges, the issue of genetic discrimination has received limited attention. The study therefore addresses an important knowledge gap within the Malaysian literature. In addition, the findings contribute a broader understanding of genetic discrimination by demonstrating that disadvantage among rare

disease families may extend beyond the traditional domains of insurance and employment to include structural barriers, inadequate accommodations, limited awareness, and exclusion across healthcare, educational, and social settings.

Several limitations should nevertheless be considered when interpreting the findings. First, participants were primarily recruited through the Malaysia Rare Disease Society (MRDS), which may have resulted in the inclusion of families who were more connected to support networks, advocacy activities, and available resources than families who are not affiliated with patient organizations. Their experiences may therefore differ from those of less connected or harder-to-reach families. Second, all participants in this study were mothers. Although mothers are often the primary caregivers and were therefore well positioned to discuss their children's experiences, the absence of fathers' perspectives limits the extent to which the findings can be generalized to parents more broadly. Fathers may experience and interpret issues related to caregiving, employment, financial planning, and social interactions differently, and these perspectives were not captured within the present study. Third, although participants were recruited from several states across Peninsular Malaysia, no participants were from the East Coast region or East Malaysia. Given potential differences in healthcare infrastructure, access to genetic services, geographical barriers, cultural contexts, and awareness of rare diseases, the experiences reported in this study may not fully reflect those of families living in other regions of the country.

An additional limitation relates to the complex nature of genetic discrimination within the context of rare diseases. During the interviews, participants frequently discussed experiences of discrimination alongside broader challenges associated with raising a child with a rare genetic condition, including caregiving responsibilities, emotional distress, financial burden, healthcare navigation, and uncertainty regarding the future. Consequently, it was sometimes difficult to distinguish the specific impact of discriminatory experiences from the wider psychosocial impact of living with and caring for a child with a rare disease. However, this overlap may itself reflect an important characteristic of genetic discrimination in rare disease contexts, where experiences of disadvantage are often intertwined with the everyday realities of caregiving rather than occurring as isolated events. The findings should therefore be

interpreted within the broader context of family adaptation and long-term rare disease caregiving.

6.4 RECOMMENDATIONS FOR FUTURE RESEARCH

Future research should build upon the findings of this study by exploring experiences of genetic discrimination from the perspectives of individuals living with rare genetic conditions themselves, particularly adolescents and adults who may face different challenges related to education, employment, relationships, healthcare access, and independent living. Given that the present study primarily captured parental perspectives, including affected individuals would provide a more comprehensive understanding of how genetic discrimination is experienced across different stages of life. Future studies may also explore the perspectives of healthcare professionals, educators, insurers, employers, and policymakers to better understand the systemic factors that contribute to discriminatory experiences and barriers encountered by rare disease families.

In addition, further research is needed to examine the extent and nature of genetic discrimination within the Malaysian context. While the present study provides in-depth qualitative insights, future mixed-methods research combining qualitative approaches with larger-scale survey studies could help establish the prevalence and distribution of discriminatory experiences across different rare disease populations. Such studies would provide a stronger evidence base for understanding the scope of genetic discrimination and identifying groups that may be particularly vulnerable to disadvantage.

The findings of this study further suggest the need for research examining the legal, regulatory, and policy landscape surrounding genetic information in Malaysia. Future studies could evaluate existing protections related to genetic information, insurance practices, privacy, and discrimination, while identifying potential legislative and regulatory gaps as genomic medicine becomes increasingly integrated into healthcare. Such work would provide valuable evidence to inform future policy development and discussions regarding the protection of individuals and families affected by genetic conditions.

Finally, comparative research involving other Asian countries may provide valuable insights into how cultural, healthcare, and policy contexts influence experiences of genetic discrimination. Cross-country studies could help determine whether the patterns identified in Malaysia are unique to the local context or reflect broader regional challenges associated with rare diseases and genomic medicine. Such comparisons may also facilitate the identification of best practices that could inform future service development, public awareness initiatives, and policy responses within Malaysia.

6.5 CONCLUSION

At the beginning of this study, genetic discrimination was understood mainly as unequal treatment based on a person's genetic information. However, the accounts shared by parents showed that the reality is often more complicated. For many families living with rare genetic diseases, disadvantage did not always come through direct discrimination or through the misuse of genetic information itself. More often, it was woven into everyday interactions and systems that were not well equipped to recognise, accommodate, or support rare conditions. Because of this, parents often faced barriers that restricted access to opportunities, services, and support that were available to others, which in turn created feelings of exclusion and disadvantage. In many cases, these experiences seemed to be linked more to the visible effects of a rare genetic condition, such as disability, developmental differences, or complex care needs, than to knowledge of the genetic diagnosis alone.

Another important finding from this study is that discrimination was rarely experienced on its own. Parents did not separate discriminatory experiences from the wider realities of caring for a child with a rare genetic condition. Challenges in healthcare, education, insurance, employment, and social settings were often tied to caregiving demands, financial strain, and uncertainty about the future. This suggests that, in the context of rare disease, genetic discrimination may be better understood as part of a broader pattern of structural disadvantage, where rarity itself can create vulnerability. Rather than appearing as isolated incidents, discrimination was often built

into the everyday experience of moving through systems that were not designed for rare and complex conditions.

Taken together, this study shows the need to look beyond a narrow focus on the misuse of genetic information when thinking about genetic discrimination. Supporting families affected by rare genetic diseases requires more than protection against unfair treatment; it also requires greater awareness, inclusion, coordination, and responsiveness across the systems they interact with. Although laws and policy protections remain important, the findings suggest that reducing disadvantage also depends on addressing wider structural barriers, improving awareness of rare diseases, and strengthening support systems. By amplifying parents' voices, this study adds a Malaysian perspective to the growing discussion on genetic discrimination and highlights the need for healthcare, education, social, and policy settings that better recognize and respond to the unique challenges faced by rare disease communities.

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APPENDIX A
QUESTIONNAIRE

Expected Theme	Sub-theme	Initial Codes	Suggested Interview Questions (in Malay)
Demographic questions	-	-	• Umur Ibu/bapa • Jantina • Bangsa / Etnik • Tahap pendidikan tertinggi • Pekerjaan Ibu/bapa • Status perkahwinan (Berkahwin/Ibu/bapa Tunggal) • Bilangan anak dan umur anak • Umur anak yang mempunyai penyakit genetik jarang • Diagnosis penyakit genetik jarang anak • Lokasi tempat tinggal (bandar / luar bandar, negeri) • Anggaran pendapatan isi rumah bulanan
1. Journey to Diagnosis and Understanding	-	Diagnostic delays, lack of provider awareness, uncertainty	• Boleh puan/tuan kongsikan bagaimana pertama kali puan/tuan tahu mengenai keadaan anak? • Boleh ceritakan sedikit pengalaman puan/tuan dengan jangka masa yang diambil untuk dapatkan diagnosis anak? • Apakah yang puan/tuan faham mengenai diagnosis penyakit anak?

2. Genetic Testing and Counselling Experience	—	Fear of knowing, decision-making, counselling quality	• Adakah ujian genetik telah dilakukan? Apakah yang mendorong puan/tuan mempertimbangkan untuk melakukan ujian genetik? • Bagaimana pengalaman puan/tuan semasa membuat ujian genetik atau semasa mendapatkan khidmat kaunseling genetik? • Adakah terdapat sebarang kebimbangan atau persoalan semasa puan/tuan mempertimbangkan ujian genetik atau semasa ingin berkongsi keputusan ujian tersebut?
3. Genetic Discrimination Experiences	3.1 Healthcare	Provider bias, dismissal, labelling, diagnostic overshadowing	• Sepanjang menjalani rawatan, adakah terdapat situasi di mana puan/tuan merasakan kebimbangan puan/tuan tidak diambil serius? • Setelah mendapatkan diagnosis untuk penyakit anak, pernahkah puan/tuan merasa perubahan pada layanan atau rawatan daripada mana-mana doktor? • Pernahkan puan/tuan merasa seperti dilabel atau dilayan secara berbeza oleh profesional kesihatan disebabkan oleh penyakit anak?
	3.2 Education / Schooling	Bullying, peer stigma, lack of accommodations	• Boleh puan/tuan kongsi bagaimana respon pihak sekolah selepas diberitahu mengenai diagnosis anak? • Adakah anak puan/tuan menghadapi sebarang cabaran dari segi sosial atau akademik di sekolah? • Pada pandangan puan/tuan, adakah pihak sekolah faham dan cuba untuk membantu anak menyesuaikan diri dalam situasi ini?

	3.3 Employment (Parent)	Career disruption, reduced support, loss of opportunity	• Adakah diagnosis penyakit anak memberi kesan terhadap kerja atau kerjaya puan/tuan? • Pernahkah puan/tuan merasa majikan atau rakan sekerja tidak memahami peranan puan/tuan sebagai ibu bapa kepada anak dengan penyakit jarang jumpa ini?
	3.4 Insurance / Financial Access	Insurance denial, fear of uninsurability, financial insecurity	• Boleh puan/tuan kongsikan sebarang pengalaman berkaitan masalah dalam permohonan insurans atau perkhidmatan bantuan kewangan selepas anak mendapatkan diagnosis? • Adakah puan/tuan menghadapi kesukaran atau kebimbangan berkaitan dengan perancangan kewangan atau perlindungan insurans?
	3.5 Social / Community Stigma	Social exclusion, judgement, labelling, reduced inclusion	• Bagaimanakah reaksi orang sekeliling seperti jiran, saudara mara, atau rakan terhadap diagnosis anak puan/tuan? • Pernahkah puan/tuan merasa dihakimi, disisihkan atau tidak difahami oleh komuniti atau kenalan terdekat disebabkan oleh penyakit anak?
4. Emotional and Social Impact	—	Anxiety, stress, isolation, emotional fatigue	• Sejak mendapat tahu mengenai diagnosis, bagaimanakah keadaan emosi puan/tuan dan keluarga? Adakah sebarang perubahan dalam kehidupan harian?

			• Boleh puan/tuan ceritakan sebarang perubahan dalam hubungan sosial dengan orang sekeliling selepas menerima diagnosis? • Apakah perkara yang paling mencabar dari segi emosi untuk puan/tuan atau anak?
5. Coping Strategies and Support	—	Self-advocacy, avoidance, support group use, faith	• Bagaimanakah puan/tuan dan keluarga menghadapi cabaran-cabaran yang datang bersama diagnosis ini? • Apakah yang membantu puan/tuan untuk terus tabah menghadapi situasi yang sukar seperti ini? • Adakah puan/tuan menerima sebarang bentuk sokongan atau sumber dari mana mana pihak yang membantu selama ini?
6. Suggestions and Unmet Needs	—	Lack of awareness, system gaps, policy suggestions	• Pada pandangan puan/tuan, apakah perubahan atau bentuk bantuan yang boleh membantu keluarga lain yang mengalami situasi serupa? • Adakah terdapat keperluan atau sokongan yang puan/tuan rasa tidak dipenuhi sepanjang pengalaman mencabar ini? • Sekiranya ada peluang untuk menyampaikan cadangan kepada doktor, guru, atau pihak yang bertanggungjawab menjaga kebajikan komuniti penyakit jarang jumpa ini, apa yang puan/tuan ingin mereka faham tentang situasi keluarga seperti puan/tuan?

APPENDIX B

PATIENT INFORMATION SHEET

Lembaran Maklumat Peserta

Tajuk Kajian

Pengkajian Diskriminasi Genetik dalam kalangan Ibu Bapa Kanak-Kanak yang menghidap Penyakit Jarang Jumpa di Malaysia: Satu Kajian Kualitatif

Exploring Genetic Discrimination Experienced by Parents of Children with Rare Diseases in Malaysia: A Qualitative Study

Pendahuluan

Anda adalah dijemput untuk menyertai kajian. Sebelum anda mengambil bahagian dalam kajian ini, adalah penting untuk anda mengambil masa untuk membaca dan memahami Lembaran Maklumat Peserta ini.

Tujuan Kajian

Kajian ini bertujuan untuk memahami pengalaman ibu bapa yang mempunyai anak dengan penyakit genetik jarang jumpa, khususnya dari segi diskriminasi berdasarkan maklumat genetik. Kajian ini akan melihat bagaimana diskriminasi ini berlaku dalam konteks penjagaan kesihatan, pendidikan, insurans, pekerjaan dan kehidupan sosial.

Bagaimanakah kajian ini dijalankan?

Anda akan diminta untuk menandatangani surat persetujuan yang menyatakan kesediaan anda untuk mengambil bahagian dalam kajian ini. Maklumat seperti sejarah perubatan dan data demografi akan dicatat dan direkodkan.

Apa yang Perlu Anda Lakukan?

- Anda akan ditemu bual secara **bersemuka atau dalam talian**, selama lebih kurang **30–60 minit**.
- Temu bual akan dijalankan dalam **Bahasa Melayu**.
- Temu bual akan **dirakam audio** dengan keizinan anda.
- Anda boleh menolak untuk menjawab sebarang soalan, atau berhenti bila-bila masa tanpa sebarang akibat.

Kenapa Anda Dialu-alukan untuk Menyertai Kajian Ini?

Anda dialu-alukan untuk menyertai kajian ini kerana anda merupakan ibu bapa atau penjaga utama kepada seorang kanak-kanak yang menghidap penyakit genetik jarang jumpa dan pengalaman anda sangat berharga untuk membantu memperbaiki sistem sokongan sedia ada.

Risiko Penyertaan dan Sokongan Tersedia

Dengan menyertai kajian ini, anda mungkin akan berasa emosi atau tidak selesa apabila berkongsi sebahagian pengalaman lalu yang berkaitan. Anda bebas untuk

berehat atau menarik diri dari kajian ini pada bila-bila masa. Sekiranya anda merasa sedih, tertekan atau tidak selesa semasa atau selepas temu bual, anda boleh menghubungi Kaunselor Genetik melalui maklumat yang tertera di bawah bagi mendapatkan sokongan. Anda juga boleh menghubungi Kaunselor Genetik sekiranya anda memerlukan sokongan sosial disebabkan oleh diskriminasi genetik.

Faedah Kajian

Penyertaan dalam kajian ini tidak memberi sebarang manfaat secara langsung kepada anda, tetapi dapatan daripada kajian ini boleh membantu menambah baik dasar, sistem sokongan dan amalan kaunseling genetik untuk keluarga lain pada masa akan datang.

Adakah anda perlu menyertai kajian ini?

Penyertaan dalam kajian ini adalah secara sukarela. Sekiranya anda bersetuju untuk mengambil bahagian, anda akan diminta untuk menandatangani “Surat Kebenaran”. Anda akan diberikan salinan surat kebenaran tersebut dan Lembaran Maklumat Peserta ini. Sekiranya anda telah mengambil bahagian, anda masih boleh menarik diri daripada kajian tanpa sebarang denda atau implikasi kepada perkhidmatan kesihatan dan sokongan yang anda terima. Data anda tidak akan digunakan dan akan dihapuskan.

Maklumat Sulit

Maklumat dalam kajian ini akan dibuat dalam bentuk laporan yang akan diterbitkan. Maklumat-maklumat ini hanya boleh diakses oleh para pengkaji dan Jawatankuasa Etika Penyelidikan Universiti Kebangsaan Malaysia. Maklumat ini akan dilaporkan dalam bentuk kolektif dan tidak akan merujuk kepada individu tertentu. Dengan ini maklumat anda adalah sulit dan terpelihara.

- Identiti anda akan dirahsiakan sepenuhnya.
- Nama sebenar tidak akan digunakan dalam sebarang laporan.
- Hanya penyelidik akan mempunyai akses kepada data.

Kepada siapakah yang saya boleh bertanya tentang kajian ini?

Sekiranya anda mempunyai sebarang persoalan, anda boleh merujuk kepada Kumpulan Penyelidik. Anda juga boleh menghubungi Jawatankuasa Etika Penyelidikan UKM untuk pengesahan.

Kumpulan Penyelidik

Dr Loh C-Khai
Jabatan Pediatrik
Hospital Tengku Ampuan Besar Tuanku Aishah Rohani (TABTAR)
Hospital Pakar Kanak Kanak UKM
Nombor Telefon: 012- 4755080

Siti Aisyah Binti Syamsul
Pelajar Sarjana Kaunseling Genetik
Universiti Kebangsaan Malaysia (UKM)
Nombor Telefon: 0112-5613109

Kaunselor Genetik

Sharifah Azween Binti Syed Omar

Klinik Genetik

Hospital Tengku Ampuan Besar Tuanku Aishah Rohani (TABTAR)

Hospital Pakar Kanak Kanak UKM

Nombor Telefon: 03-91912207/ 019-6240344



APPENDIX C

CONSENT FORM

BORANG KEIZINAN UNTUK MENYERTAI KAJIAN

Tajuk Kajian: Pengkajian Diskriminasi Genetik dalam kalangan Ibu Bapa Kanak-Kanak yang menghidap Penyakit Jarang Jumpa di Malaysia: Satu Kajian Kualitatif
Exploring Genetic Discrimination Experienced by Parents of Children with Rare Diseases in Malaysia: A Qualitative Study

Nama Penyelidik: Dr Loh C-Khai, Dr Nur Azah Binti Mohd Isa, Pn Sharifah Azween Binti Syed Omar dan Siti Aisyah Binti Syamsul

Saya,, Nombor Kad Pengenalan:

- telah membaca maklumat dalam Lembaran Maklumat Peserta
- telah diberikan masa untuk berfikir mengenainya dan semua soalan-soalan saya telah dijawab dengan memuaskan.
- memahami bahawa saya bebas boleh memilih untuk menarik diri daripada kajian ini pada bila-bila masa tanpa sebab dan tanpa apa-apa akibat
- bersetuju untuk temu bual ini dirakam audio.
- memahami yang nama saya tidak akan disiarkan di dalam penulisan kajian.

Saya secara sukarela bersetuju untuk menjadi sebahagian daripada kajian penyelidikan ini, mematuhi prosedur kajian, dan untuk menyediakan maklumat yang diperlukan untuk doktor, penyelidik, atau ahli-ahli kakitangan lain, seperti yang diminta.

Peserta

Tandatangan:

NRIC:

Tel no:

Tarikh:

.....	
Nama Saksi	Nama Penyelidik
.....	
(Tandatangan)	(Tandatangan)
.....	
(No. IC)	No. IC)
.....	
(Tarikh)	(Tarikh)

APPENDIX D

UKM'S ETHICS APPROVAL



FAKULTI PERUBATAN • FACULTY OF MEDICINE

Reference: UKM.FPR.SPI 800-2/28

Date : 17 December 2025

Dr Loh C-Khai
Department of Paediatrics
Faculty of Medicine, UKM

Dear Dr,

APPROVAL TO CONDUCT RESEARCH IN HOSPITAL TUNKU AMPUAN BESAR TUANKU AISHAH ROHANI, HOSPITAL PAKAR KANAK-KANAK UKM

Title : Exploring Genetic Discrimination Experienced by Parents of Children with Rare Diseases in Malaysia: A Qualitative Study

Project Code : FF-2025-510

With reference to the above, the Faculty of Medicine UKM and Hospital Tunku Ampuan Besar Tuanku Aishah Rohani, Hospital Pakar Kanak-Kanak UKM (HTABTAR) has approved the above study based on the following information:

Duration of Research	27 November 2025 – 26 August 2026
Financial Support	RM 400.00 from Faculty of Medicine Fundamental Grants (GFFP) (Please refer Appendix 1 and 2)

A Progress Report should be submitted to the committee every six (6) months from the date of approval and a Final Report within three (3) months after expiration of the study approval. Failure to submit the Final Report will affect the application of research and financial support in the future.

The Secretariat reserves the right to withdraw the approved financial assistance if it is not spent within six (6) months from the date of approval. A show cause letter must be submitted to the Secretariat if the financial assistance is not spent according to the approved expenses. All researchers are hereby required to acknowledge the GFFP Grant as the primary (first) funding source in all related publications and to comply with the publishing KPIs as stipulated in Appendix 1.

Application for additional budget should be submitted to the Secretariat within one (1) month before the expiration of approval. If the Principle Investigator leaves UKM, he/she must inform the Secretariat and name the replacement. Please visit our website (<https://www.ukm.my/spifper/forms/>) for more information and guidelines.

Please register the research at the National Medical Research Register (www.nmrr.gov.my) if involved any use of infrastructure/organization/facility/staff/students/patients in the hospital/clinic/organization under the Ministry of Health Malaysia. Researchers must also ensure that notification to the Ministry of Health is made if any results of this research project are to be published in a journal or presented at a conference.



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Telefon: +603-9145 5003/9481/9497/8798 Emel: sppl@ppukm.ukm.edu.my Laman Web: <https://www.ukm.my/fper>

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For your information, a copy of the approval letter for co-researchers and students will be send via e-mail. Please to be reminded of these project can only be carried out after obtaining a certificate and letter of approval by UKM Ethics Committee.

Thank you.

Your sincerely,



PROFESSOR DATIN DR MARINA MAT BAKI
Dean
Faculty of Medicine, UKM



PROFESSOR DATUK DR ISMAIL SAGAP
Director
Hospital Tunku Ampuan Besar Tuanku Aishah Rohani
Hospital Pakar Kanak-Kanak (HTABTAR)

- s.k - **Professor Dr. Mohamad Nasir Shafiee**
Deputy Dean (Research & Innovation)
Faculty of Medicine, UKM
- **Chairman Research Ethics Committee UKM**
Secretariat of Research Ethics UKM
UKM Medical Centre
- **Mrs Hamidah Nayan**
Chief of Co-Treasurer
Cheras Campus Financial Zone
Treasurer's Department, UKM
(Please activate an account in the financial system for the distribution of grant to the project code as above. Thank you)
- **Head Department of Paediatrics**
Faculty of Medicine, UKM
- **Dr Nur Azah Mohd Isa**
Department of Paediatrics
Hospital Tunku Ampuan Besar Tuanku Aishah Rohani (HTABTAR)
- **Mrs Sharifah Azween Syed Omar**
Dr Siti Aisyah Syamsul
Department of Paediatrics
Faculty of Medicine, UKM
- **FF-2025-510 File**
MMBN/FF-2025-510



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