

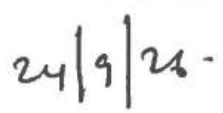
PARENTAL PERCEPTION OF GENOMIC
NEWBORN SCREENING (GNBS)
IN A MALAYSIAN TERTIARY
PAEDIATRIC SETTING



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UNIVERSITI KEBANGSAAN MALAYSIA

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		2025/2026	
Tajuk Tesis / Disertasi (Thesis / Dissertation Title)		PERSEPSI IBU BAPA TERHADAP SARINGAN GENOMIK BAYI BARU LAHIR (GNBS) DI SEBUAH HOSPITAL TERTIER DI MALAYSIA PARENTAL PERCEPTION OF GENOMIC NEWBORN SCREENING (GNBS) IN A MALAYSIAN TERTIARY PAEDIATRIC SETTING	
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PARENTAL PERCEPTION OF GENOMIC NEWBORN SCREENING (GNBS)
IN A MALAYSIAN TERTIARY PAEDIATRIC SETTING

CHIN PEI VOON

THESIS SUBMITTED IN FULFILMENT FOR THE DEGREE OF MASTER OF
MEDICAL SCIENCE (GENETIC COUNSELLING)

FACULTY OF MEDICINE
UNIVERSITI KEBANGSAAN MALAYSIA
KUALA LUMPUR

2026

PERSEPSI IBU BAPA TERHADAP SARINGAN GENOMIK BAYI BARU LAHIR
(GNBS) DI SEBUAH HOSPITAL TERTIER DI MALAYSIA



TESIS YANG DIKEMUKAKAN UNTUK MEMPEROLEH IJAZAH SARJANA
SAINS PERUBATAN (GENETIK KAUNSELING)

FAKULTI PERUBATAN
UNIVERSITI KEBANGSAAN MALAYSIA
KUALA LUMPUR

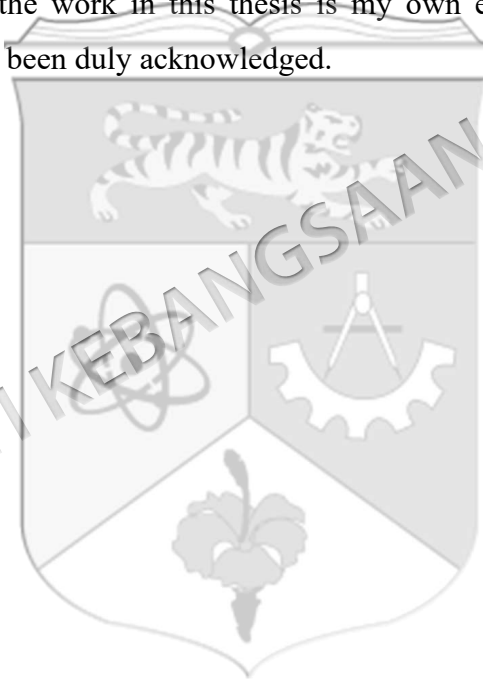
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DECLARATION

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

21 September 2026

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P148737



ACKNOWLEDGEMENT

First and foremost, I would like to express my deepest gratitude to God for His blessings, guidance and strength throughout my Master's journey. His grace has sustained me physically, mentally and emotionally throughout the completion of this thesis. I am thankful for the good health, determination and ample resources I have received throughout this challenging but fulfilling journey.

I would like to express my sincere appreciation to my supervisor, Associate Professor Dr Shareena Binti Ishak, my co-supervisors, Dr Wan Nurulhuda Binti Wan Md Zin, Dr Shyahlini Devi A/P Rajendran and my co-researcher, Ms Sharifah Azween Bintin Syed Omar for their invaluable guidance, continuous support, constructive feedback and encouragement throughout this research. Their expertise, patience and dedication have provided me with clear direction and confidence in completing this study. I am also grateful to Dr Chew Hui Bein, clinical genetic specialist from Hospital Kuala Lumpur (HKL) who generously agreed to review and evaluate my questionnaire as part of the expert content validation process. Special thanks are also extended to the researchers, Dr Azhar Rahma and SN Bernarda Prosenc who kindly granted permission for the adaptation and use of their questionnaires in this study.

My sincere appreciation is extended to Universiti Kebangsaan Malaysia (UKM) and Hospital Tunku Ampuan Besar Tuanku Aishah Rohani (TABTAR) for granting permission to conduct this research. I would also like to thank the staff of the paediatric outpatient clinics for their assistance, cooperation and friendliness throughout the data collection process. Their willingness to help identify eligible participants greatly facilitated the conduct of this study. I am equally grateful to all individuals who participated in the pilot testing and provided valuable feedback that contributed to the improvement of the questionnaire.

I would also like to acknowledge the financial support received throughout my studies. This research was supported by the fundamental grant from the Secretariat for Research & Innovation. I am also grateful for the educational financial assistance provided by the National Higher Education Fund Corporation (PTPTN) and Yayasan Pahang which enabled me to pursue and complete my postgraduate studies.

My heartfelt thanks go to my coursemates Foong Le Hui, Nur Mashitah Binti Mohd Shahrom, Siti Aisyah Binti Syamsul and friends for their companionship, encouragement, knowledge sharing and unwavering support throughout this journey. Their motivation and friendship made the challenges of postgraduate study more manageable and meaningful. I am particularly grateful to those who assisted in the pilot study and contributed their time and effort to support this research. I would also like to express my sincere appreciation to all parents who willingly participated in this study. Their time, cooperation and words of encouragement were invaluable and made this research possible.

Finally, I would like to dedicate my deepest gratitude to my mother, Tien Kok Moi and my uncle, Tien Kok Ping for their unconditional love, patience, understanding and continuous support. They have always been my source of strength and encouragement, standing by me through every challenge and achievement. Their unwavering belief in me has been instrumental in reaching this milestone.

To everyone who has contributed directly or indirectly to this research and my postgraduate journey, including those whose names are not specifically mentioned here, please accept my heartfelt appreciation. Your kindness, support and encouragement will always be remembered.



ABSTRAK

Saringan genomik bayi baru lahir (gNBS) merupakan satu pendekatan baru yang berpotensi untuk mengenal pasti penyakit genetik pada peringkat awal, termasuk keadaan yang tidak dapat dikesan melalui program saringan bayi baru lahir konvensional. Walau bagaimanapun, penerangan mengenai persepsi ibu bapa terhadap gNBS di Malaysia masih terhad. Kajian ini bertujuan untuk menilai perspektif, sikap dan penerimaan ibu bapa terhadap gNBS serta mengenal pasti faktor yang berkaitan dengan kesediaan mereka untuk menyokong pelaksanaannya. Kajian keratan rentas kuantitatif telah dijalankan dalam kalangan 429 ibu bapa yang menghadiri klinik pesakit luar pediatrik di Hospital Tunku Ampuan Besar Tuanku Aishah Rohani (Hospital TABTAR). Peserta melengkapkan soal selidik berstruktur tadbir sendiri selepas diberikan maklumat mengenai saringan bayi baru lahir dan gNBS. Kesedaran terhadap gNBS adalah rendah, dengan hanya 21.0% ibu bapa melaporkan pernah mendengar tentang gNBS sebelum ini. Walaupun demikian, majoriti ibu bapa menyokong pelaksanaan gNBS (74.6%). Manfaat yang paling kerap diamati ialah rawatan awal dan berkesan (88.1%), diagnosis genetik yang awal dan tepat pada masanya (82.3%), serta penyediaan maklumat kesihatan yang tepat dan menyeluruh (78.3%). Perhatian utama yang dibangkitkan termasuk kebimbangan terhadap kesihatan masa depan anak (79.5%), kos kewangan (74.1%) dan risiko salah tafsir keputusan genetik (64.1%). Kos ujian dan rawatan (86.9%), jenis penyakit yang disaring (75.4%) serta ketersediaan rawatan (71.9%) merupakan faktor utama yang mempengaruhi keputusan ibu bapa. Analisis multivariat menunjukkan bahawa kepercayaan bahawa penyakit genetik boleh dirawat (aOR = 1.98, 95% CI: 1.03-3.80, $p = 0.039$) merupakan satu-satunya faktor bebas yang berkaitan dengan sokongan terhadap gNBS. Dapatan kajian ini menunjukkan bahawa ibu bapa di Malaysia secara umumnya menerima gNBS walaupun tahap kesedaran mengenainya masih terhad, sekali gus menekankan kepentingan pendidikan awam, penyediaan sumber kaunseling genetik dan pembangunan rangka kerja tadbir urus yang sesuai bagi menyokong pelaksanaannya.

ABSTRACT

Genomic newborn screening (gNBS) is an emerging approach that offers the potential for early identification of genetic conditions beyond those detected through conventional newborn screening. However, evidence regarding parental perceptions on gNBS in Malaysia remains limited. This study aimed to assess parental perspectives, attitudes and acceptance of gNBS and to identify factors associated with willingness to support its implementation. A quantitative cross-sectional survey was conducted among 429 parents attending the paediatric outpatient clinics at Hospital Tunku Ampuan Besar Tuanku Aishah Rohani (Hospital TABTAR). Participants completed a structured self-administered questionnaire following provision of information regarding newborn screening and gNBS. Awareness of gNBS was low, with only 21.0% of parents reporting prior awareness. Nevertheless, most parents supported the implementation of gNBS (74.6%). The most commonly perceived benefits were early and effective treatment (88.1%), timely genetic diagnosis (82.3%) and provision of comprehensive health information (78.3%). Key concerns included anxiety regarding a child's future health (79.5%), financial costs (74.1%) and misinterpretation of genetic results (64.1%). Cost of testing and treatment (86.9%), diseases included in screening (75.4%) and treatment availability (71.9%) were the main factors influencing parental decision-making. Multivariable analysis identified belief in the treatability of genetic diseases (aOR = 1.98, 95% CI: 1.03-3.80, $p = 0.039$) as the only independent factor associated with support for gNBS. These findings suggest that Malaysian parents are generally receptive towards gNBS despite limited awareness, highlighting the need public education, genetic counselling resources, appropriate governance frameworks to support its implementation.

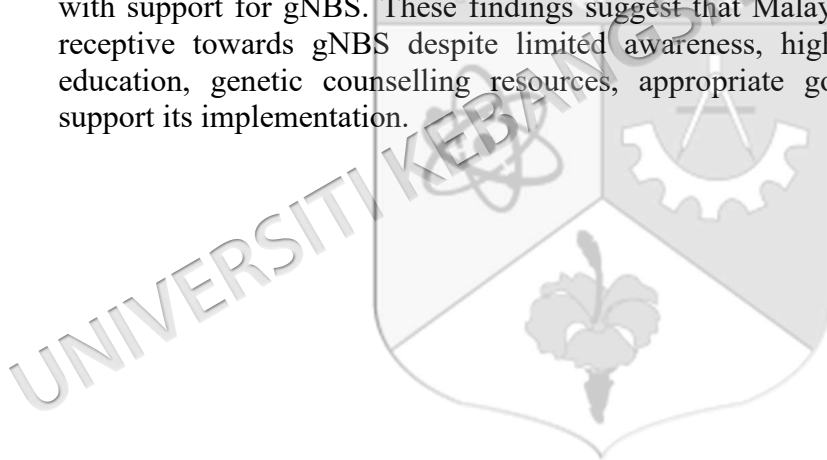


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LIST OF ABBREVIATIONS

aOR	Adjusted Odds Ratio
ARCADIA	Arcadia Life Sciences Sdn. Bhd.
CAATS	Centre for Advanced Analytical Toxicology Services
CI	Confidence Interval
DNA	Deoxyribonucleic Acid
G6PD	Glucose-6-phosphate dehydrogenase
gNBS	Genomic Newborn Screening
I-CVI	Item-Level Content Validity Index
IEM	Inborn Errors of Metabolism
IMR	Institute for Medical Research
JEPUKM	Research Ethics Committee of Universiti Kebangsaan Malaysia
MOH	Malaysia's Ministry of Health
MS/MS	Electrospray Tandem Mass Spectrometry
MYPOPI	Malaysian Patients' Organisation for Primary Immunodeficiencies
NBS	Newborn Screening
PKU	Phenylketonuria
REF	Reference Group
SCID	Severe Combined Immunodeficiency
S-CVI	Scale-Level Content Validity Index
SHINE	Screening for Health, Intervention, and Nurturing of Every Child
SMA	Spinal Muscular Atrophy
TABTAR	Tunku Ampuan Besar Tuanku Aishah Rohani
UKM	Universiti Kebangsaan Malaysia
UMMC	Universiti Malaya Medical Centre
XLA	X-Linked Agammaglobulinaemia

CHAPTER I

INTRODUCTION

1.1 RESEARCH BACKGROUND

Newborn screening (NBS) has long been a vital part of public health systems worldwide that is used to detect certain serious but treatable conditions before symptoms appear in infants. The conditions encompass metabolic, endocrine, haematologic and immunological disorders, many of which may initially be clinically silent despite the potential for significant morbidity if left undiagnosed. By identifying these medical conditions in newborns through biochemical markers such as amino acids, acylcarnitines and organic acids, NBS allows infants to receive timely care even before symptoms appear (Pappas 2023; Shah et al. 2024). In the absence of NBS, affected infants may only be identified after clinical manifestations emerge, which can delay diagnosis and treatment and increase the risk of preventable complications. Thus, the existence of NBS has brought benefits to healthcare sectors, significantly improving early detection, medical management and overall quality of life for affected individuals (Chen et al. 2023; Habib et al. 2025; Kumarasamy et al. 2024).

In Malaysia, NBS has been implemented for decades, but its scope remains limited compared with expanded NBS programmes in many other countries. The national programme currently screens for conditions such as glucose-6-phosphate dehydrogenase (G6PD) deficiency and congenital hypothyroidism (Habib et al. 2025). Unlike regions that have adopted expanded NBS through technologies such as tandem mass spectrometry to detect a broader range of inborn errors of metabolism (IEM), Malaysia's NBS currently focuses on a small number of conditions. Consequently, some genetic conditions may remain undetected through the existing screening programme, particularly those that are not included in the current screening panel.

As technology advances, the field of NBS is also evolving. Genomic medicine is now becoming accessible in many countries, giving rise to genomic newborn screening (gNBS), which uses genome-wide sequencing or targeted gene panels to detect a wider range of genetic conditions soon after birth (Bhattacharya et al. 2014; Bick et al. 2022; La Marca et al. 2023; Levy 2021; Watson, Lloyd-Puryear & Howell 2022). Rather than relying primarily on biochemical signals, gNBS looks directly at an infant's genetic information, offering the possibility of identifying a broader range of genetic conditions that traditional methods may miss.

Several countries including the United States, the United Kingdom, nations in Europe and Africa, as well as Australia, India, and China, have already begun exploring gNBS through pilot projects and national research programmes (Downie et al. 2024; Jiang, Wang & Gu 2023; Jolly & Scaria 2025; Lancet 2023; Therrell et al. 2024). Their experiences show both promise and complexity of implementing gNBS. While gNBS may facilitate earlier diagnosis and improve long-term health outcomes, its broader application also raises important questions about ethical-legal frameworks, psychosocial factors, population factors, economic factors and resource constraints (Bick et al. 2022; Lancet 2023; Remec et al. 2021; Shah et al. 2024; Spiekerkoetter et al. 2023; Watson, Lloyd-Puryear & Howell 2022). These concerns include decisions regarding which conditions should be screened, how uncertain or clinically actionable findings should be managed, how genomic information should be communicated and protected, and whether equitable access to subsequent diagnostic and clinical services can be ensured.

Malaysia remains at an early stage in the adoption and exploration of genomic newborn screening. Public awareness, understanding and involvement in gNBS-related research remain limited while international implementation models may not be directly applicable due to differences in cultural values, population genetics, health literacy and governance structures (Jolly & Scaria 2025; Lynch et al. 2024; Therrell et al. 2024; Ziegler et al. 2024). Prior to considering large-scale implementation, it is essential to understand how Malaysian parents perceive this emerging screening approach, as parental acceptance and engagement are critical determinants of successful programme uptake. To address this knowledge gap, this study explores parental perspectives,

attitudes and acceptance of gNBS within a tertiary paediatric setting. The findings are expected to provide meaningful insights that can guide future research, shape educational efforts and support evidence-based policy development for the responsible integration of gNBS into Malaysia's healthcare system.

1.2 PROBLEM STATEMENT

Malaysia continues to shoulder a considerable burden of congenital and rare diseases, many of which manifest early in life yet remain frequently underdiagnosed, diagnosed only after clinical symptoms become apparent or inadequately managed. Although individual rare diseases are uncommon, collectively they represent an important health burden, especially when affected infants may experience significant morbidity if diagnosis and management are delayed. Existing newborn screening programmes are relatively limited, covering only a small set of conditions. Consequently, some affected infants may not be identified during the newborn period, potentially delaying appropriate medical care and specialist management. Early medical care may also be delayed partly due to insufficient awareness among both healthcare providers and families, particularly regarding the recognition and management of congenital and rare diseases (Habib et al. 2025; Kumarasamy et al. 2024). gNBS has the potential to address these limitations by enabling the early detection of a wider range of conditions, thereby creating opportunities for earlier clinical assessment, intervention and appropriate long-term management, especially for conditions in which early treatment may prevent or reduce disease-related complications (Jiang, Wang & Gu 2023; Lancet 2023; Shah et al. 2024). Despite these benefits, research in Malaysia remains sparse regarding parental perspectives, attitudes and acceptance of gNBS, as well as the factors influencing its acceptance. Understanding these factors is essential for informing its successful adoption and integration into the national healthcare system.

1.3 OBJECTIVE OF RESEARCH

This study aims to investigate parental perception of gNBS and to identify factors influencing parental acceptance of gNBS in a Malaysian tertiary paediatric setting.

1.3.1 General Objective

To assess parental perspectives, attitudes and acceptance of genomic newborn screening (gNBS) among parents attending Hospital Tunku Ampuan Besar Tuanku Aishah Rohani (Hospital TABTAR), UKM Specialist Children's Hospital.

1.3.2 Specific Objectives

1. To determine parents' perspectives, attitudes and acceptance of gNBS.
2. To identify factors influencing with parental perceptions of and willingness to support the implementation of gNBS within the Malaysian healthcare system.

1.4 HYPOTHESIS

1.4.1 Null Hypothesis (H_0)

There is no significant association between parental attitudes toward or acceptance of genomic newborn screening (gNBS), and sociodemographic characteristics including age, gender, educational attainment, or perceptions of genetic disease and genetic testing, among parents at Hospital TABTAR.

1.4.2 Alternative Hypothesis (H_1)

There is a significant association between parental attitudes toward and acceptance of genomic newborn screening (gNBS) and sociodemographic characteristics including age, gender, educational attainment and perception of genetic disease and genetic testing, among parents at Hospital TABTAR.

1.5 SIGNIFICANCE OF THE STUDY

This study seeks to establish baseline evidence on parental awareness, perceptions, attitudes and acceptance of genomic newborn screening (gNBS). As genomic medicine continues to advance globally, understanding parental views toward this emerging technology is essential for assessing readiness and potential acceptability of gNBS within the Malaysian healthcare context. The findings will provide valuable insights

into the factors influencing parental decision-making and willingness to support the adoption of gNBS.

This study also addresses the current limited data on public perceptions of genomic medicine in Malaysia, especially in relation to gNBS. By identifying knowledge gaps, misconceptions and parental concerns, the findings may guide the development of targeted educational initiatives, genetic counselling frameworks and informed consent resources that are responsive to the needs and values of Malaysian families. Furthermore, the results may contribute to the development of policy frameworks and genomic data governance, particularly in addressing ethical, legal, and social implications associated with gNBS implementation. These insights may assist policymakers and healthcare administrators in evaluating the feasibility, acceptability and potential integration of gNBS into existing healthcare services.

Understanding parental acceptance and the sociocultural factors influencing decision-making is essential to ensure that any future implementation of gNBS is ethically sound, culturally appropriate and publicly supported. Beyond immediate policy and clinical implications, this study may serve as a foundation for further research and development in genetics and public health in Malaysia.

CHAPTER II

LITERATURE REVIEW

2.1 OVERVIEW AND EVOLUTION OF NEWBORN SCREENING (NBS)

The origins of newborn screening (NBS) date back to the late 1950s and early 1960s, when early programmes enabled the identification of phenylketonuria (PKU) in newborns. The introduction of PKU screening demonstrated that a serious inherited metabolic disorder could be detected before the onset of clinical symptoms and that early dietary treatment could substantially reduce the risk of severe neurological impairment. This provided early evidence that screening apparently healthy newborns for treatable conditions could alter disease outcomes and established an important foundation for the development and expansion of NBS programmes. Initial detection methods relied on urine assays, which were later replaced by more reliable blood spot testing. The effectiveness of these early initiatives led to the adoption of mandatory NBS across the United States (La Marca et al. 2023; McCandless & Wright 2020; Watson, Lloyd-Puryear & Howell 2022). As scientific understanding and diagnostic technologies progressed, the scope of NBS expanded to encompass a wider range of inborn metabolic disorders and genetic conditions, including homocystinuria, maple syrup urine disease, congenital hypothyroidism, cystic fibrosis, sickle cell disease, galactosaemias, hemoglobinopathies, biotinidase deficiency, glucose-6-phosphate dehydrogenase (G6PD) deficiency, congenital adrenal hyperplasia, and severe combined immunodeficiency (SCID) among others (Bhattacharya, Wotton & Wiley 2014). The cumulative evidence consistently demonstrates that NBS facilitates the early detection of otherwise asymptomatic diseases, enabling timely diagnosis and treatment while reducing mortality and morbidity (Bhattacharya, Wotton & Wiley 2014; Bick et al. 2022; Chen et al. 2023; Habib et al. 2025).

2.2 EVOLUTION FROM NBS TOWARDS GENOMIC NEWBORN SCREENING (GNBS)

The evolution of NBS has progressed from the detection of a single metabolic disorder to the identification of a wider range of conditions through the adoption of electrospray tandem mass spectrometry (MS/MS)-based protocols (Bhattacharya, Wotton & Wiley 2014; La Marca et al. 2023). Despite these advances, conventional NBS still largely depends on biochemical markers measured in blood samples and is generally restricted to a predefined panel of conditions, meaning that certain disorders may remain overlooked. Consequently, conditions that are not included in the screening panel or not produce detectable biochemical abnormalities during the newborn period may remain unidentified. Furthermore, expanding conventional NBS to include additional conditions requires consideration of test performance, clinical utility, availability of effective treatment and the resources required for implementation. Developments in genomic science have begun to address these limitations, giving rise to genomic newborn screening (gNBS) (Bhattacharya, Wotton & Wiley 2014; Vears et al. 2023).

gNBS employs genomic sequencing to identify DNA variants associated with medical conditions in infants, markedly expanding detection capabilities to include a wider range of genetic and rare diseases (Bick et al. 2022; Stark & Scott 2023). This genomic approach enhances opportunities for timely care planning, early intervention, informed clinical decision-making and potential long-term reductions in healthcare costs, especially for conditions that may not be readily identified through conventional biochemical screening (Chen et al. 2023; Habib et al. 2025; Kumarasamy et al. 2024). However, gNBS should not be considered a complete replacement for conventional NBS, as genomic sequencing has its own technical, interpretive, ethical and clinical limitations. At present, genomic approaches are used primarily as second-tier confirmatory or diagnostic testing in some settings, while established biochemical screening remains an important first-line option. Nonetheless, several countries, including the United States, the United Kingdom, various European nations, Australia and China, are actively assessing the potential of implementing gNBS as a first-tier screening strategy. Ongoing studies continue to evaluate its feasibility, clinical utility, and practical integration into public health programmes (Chen et al. 2023; La Marca et al. 2023; Lynch et al. 2024; Watson, Lloyd-Puryear & Howell 2022).

2.3 POTENTIAL BENEFITS AND CHALLENGES OF GNBS

The rapid advancement of genomic medicine and molecular diagnostic technologies has significantly expanded the landscape of disease detection. As these technologies become increasingly integrated into clinical practice, there is a growing demand for their application in both healthcare and research settings. This expansion has contributed to improving diagnostic capabilities while simultaneously increasing the need for trained genetic professionals, laboratory personnel, and specialized expertise.

gNBS represents one application within this broader development of genomic-based healthcare. Similar to other genomic technologies, gNBS offers important potential benefits but also presents significant challenges that must be carefully considered. In terms of disease management, gNBS offers the potential to avert delays in treatment for late-onset diseases, thereby improving long-term health outcomes and alleviating strain on healthcare systems (Jiang, Wang & Gu 2023; Lancet 2023; Shah et al. 2024).

However, as with many emerging innovations, gNBS brings not only advantages but also a set of challenges that must be carefully considered prior to widespread adoption. Concerns span ethical and psychological dimensions (such as informed consent, future autonomy, psychological impact, stigma and discrimination), as well as technical and clinical factors (including complexities in data interpretation, limited therapeutic options, risks of false-positive or false-negative results, and the readiness of healthcare infrastructures). In addition, issues related to data privacy, security and potential misuse remain central to ongoing debate (Bick et al. 2022; Chen et al. 2023; Jolly & Scaria 2025; Kelly, Makarem & Wasserstein 2016; Levy 2021; Lynch et al. 2024; Shah et al. 2024; Therrell et al. 2024; Vears et al. 2023; Ziegler et al. 2024).

Addressing these complexities requires coordinated efforts among clinicians, parents, researchers and policymakers to ensure that the benefits of gNBS are realised while minimising risks and promoting ethically sound implementation.

2.4 MALAYSIAN NBS LANDSCAPE

In Malaysia, NBS was first introduced in 1980 with the detection of glucose-6-phosphate dehydrogenase (G6PD) deficiency using cord blood samples. The introduction of G6PD screening was particularly important because affected newborns are at risk of developing neonatal jaundice and related complications if the condition is not identified and appropriately managed (Lee et al. 2022; Leong et al. 2014; Luzzatto, Nannelli & Notaro 2016). Hearing screening followed in 2000, reflecting the importance of early identification of congenital hearing loss to facilitate timely intervention and minimise its potential impact on speech, language and developmental outcomes. By 2003, a mandatory national NBS programme for congenital hypothyroidism had been established. Early detection of congenital hypothyroidism is important because timely treatment can prevent or reduce the risk of serious developmental and neurological consequences (Leong et al. 2014; Rajadurai et al. 2021; Van Trotsenburg et al. 2020).

Gradually, screening initiatives expanded to include selected IEM such as amino acid, fatty acid and organic acid disorders. The expansion of metabolic screening has been facilitated by advances in biochemical technologies, particularly tandem mass spectrometry, which allows multiple metabolic disorders to be screened from a single blood sample. Early identification of these disorders is clinically important because many IEMs may be asymptomatic during the newborn period but can result in severe metabolic decompensation and other complications if diagnosis and treatment are delayed. However, these expanded tests are not routinely offered nationwide and are generally available on an optional basis or for infants with clinical indications. Currently, expanded screening services have been provided through the Institute for Medical Research (IMR), Hospital Kuala Lumpur (HKL), Universiti Malaya Medical Centre (UMMC) and Centre for Advanced Analytical Toxicology Services (CAATS) under Universiti Sains Malaysia, primarily upon request from members of the public and selected private hospitals (Leong et al. 2014; Shafie et al. 2020; Yunus et al. 2016). This results in variability in access to expanded NBS across healthcare settings and limits its availability as part of the routine national screening programme.

Beyond these expanded screening services, several institutions have since introduced expanded newborn screening programmes that provide screening beyond the national panel. For example, UMMC offers expanded screening for more than 30 IEMs, allowing parents to opt for additional testing beyond the national screening panel (Habib et al. 2025). More recently, in 2024, Arcadia Life Sciences Sdn. Bhd. (ARCADIA) launched the Screening for Health, Intervention, and Nurturing of Every Child (SHINE) programme in collaboration with Revvity Singapore Pte. Ltd. and with support from Malaysian Patients' Organisation for Primary Immunodeficiencies (MYPOPI). The programme offers screening for more than 60 conditions, including metabolic disorders, severe combined immunodeficiency (SCID), X-linked agammaglobulinaemia (XLA) and spinal muscular atrophy (SMA) (Nezar 2024).

Despite ongoing efforts, underdiagnosis and inadequate access to treatment for rare diseases and congenital disorders remain significant public health concerns in Malaysia (Habib et al. 2025; Kumarasamy et al. 2024). The availability of expanded screening through selected institutions and programmes demonstrates growing interest in broadening NBS in Malaysia. However, variation in screening availability between the national programme and institution-based or privately supported initiatives highlights the ongoing challenges in achieving broader and more equitable access to expanded newborn screening. This landscape highlights the need to consider approaches that could further expand the range of conditions detectable during the newborn period while ensuring that screening is supported by appropriate diagnostic, treatment and follow-up services.

2.5 READINESS OF GNBS IN MALAYSIA

Malaysia has witnessed a gradual expansion of genetic and molecular diagnostic services over the past decade. Genetic testing is increasingly accessible through a network of local laboratories, international laboratory partnerships and regional branches of overseas testing providers. While certain tests can be performed locally, others continue to be referred to overseas laboratories because of technological, expertise, or database requirements. These services are utilized by both public and

private healthcare institutions, reflecting the growing integration of genetic testing into routine clinical practice.

The increasing availability and utilization of genetic testing services indicate a growing capacity for genomic medicine within the Malaysian healthcare system. However, translating these developments into large-scale population-based programmes such as gNBS remains challenging. Factors including infrastructure requirements, workforce capacity, cost considerations, data governance, ethical concerns and public awareness must be addressed before nationwide implementation can be considered (Chang et al. 2024; Kumarasamy et al. 2024).

As a developing nation, Malaysia has made steady but modest progress in adopting genomic technologies, and the integration of gNBS into the national healthcare system remains a longer-term objective (Habib et al. 2025; Kumarasamy et al. 2024). While international experiences can offer important insights once gNBS implementation begins, contextual differences such as Malaysia's diverse population, cultural values, public awareness and political structures constrain the direct transferability of foreign models (Jolly & Scaria 2025; Lynch et al. 2024; Therrell et al. 2024; Ziegler et al. 2024). Consequently, locally driven research is crucial to inform, guide and support the future development of gNBS within the Malaysian context. Despite this growing capacity for genomic medicine, the readiness of the healthcare system alone does not guarantee successful implementation of gNBS as several structural, regulatory and ethical challenges remain that may influence its feasibility in Malaysia.

2.6 BARRIERS OF GNBS IN MALAYSIA

Recently in 2026, Malaysia's Ministry of Health (MOH) introduced the National Policy for Rare Diseases which aims to establish a structured, equitable and sustainable healthcare framework for individuals with rare diseases. The policy outlines multiple strategic areas of action, including strengthening diagnostic services, improving access to treatment, enhancing workforce capacity and promoting research and data systems.

These initiatives are intended to address existing gaps within the healthcare system, especially in relation to rare disease management and genetic services.

Despite these policy developments, the implementation of gNBS in Malaysia continues to face substantial challenges. The introduction of gNBS in Malaysia is hindered by a range of structural and contextual challenges. Chang et al. (2024) highlight several key barriers, including constrained funding, insufficient laboratory and technological infrastructure, limited numbers of trained healthcare personnel, the absence of standardised regulatory frameworks and limited local evidence to guide policy and practice.

In addition, ethical, legal, and psychosocial considerations remain critical barriers to the implementation of gNBS. Issues such as data privacy, management of incidental findings, cost-effectiveness, and the potential emotional and psychological impact on families require thoughtful attention and systematic evaluation before implementation can proceed responsibly and sustainability into healthcare system (Jolly & Scaria 2025; Lynch et al. 2024; Therrell et al. 2024; Ziegler et al. 2024).

2.7 IMPORTANCE OF PARENTAL PERSPECTIVES

As Malaysia is still in the early stages of adopting gNBS, parents are expected to play a central role as key decision-makers. Understanding their perspectives is therefore necessary prior to large-scale implementation to ensure that the technology is introduced in a responsible and appropriate manner within the local healthcare system.

In this context, examining the factors that influence parental perceptions, attitudes and acceptance of gNBS may provide important insights to guide future healthcare policies, support the development of targeted educational and communication strategies and guide further research on the implementation of gNBS in Malaysia. Such evidence is also important in ensuring that the adoption of gNBS is equitable, ethically sound and socially responsible.

Developing a robust framework for implementing gNBS in Malaysia necessitates coordinated engagement among diverse stakeholders including healthcare

professionals, policymakers, institutions and the public. This study seeks to support that process by exploring parental perceptions of gNBS at Hospital TABTAR. The findings are expected to identify gaps in current knowledge and practice, gauge public readiness, inform effective communication strategies and lay groundwork for future research and policy developments tailored to local context.

2.8 RESEARCH GAP

Adoption of genomic medicine within NBS programmes is an emerging area of interest across many countries. Several international pilot studies and national research initiatives have been conducted to evaluate the feasibility of implementing gNBS in different healthcare settings. A key focus of these efforts is the assessment of parental views, as public perception is recognised as an important factor in shaping governance structures, ethical considerations and implementation frameworks. These studies highlight the importance of generating context-specific evidence to support decision-making that is aligned with local healthcare systems, resources and sociocultural values.

A significant gap in the existing literature is the absence of empirical data on how Malaysian parents perceive gNBS. Understanding their perspectives, attitudes and the factors influencing their acceptance of gNBS is essential for designing policies and implementation strategies that are both effective and socially responsive (Franková et al. 2019; Lynch et al. 2024; Waisbren et al. 2014).

CHAPTER III

METHODOLOGY

3.1 STUDY DESIGN

This study employed a prospective, cross-sectional quantitative survey design to assess parental perspectives, attitudes and acceptance of gNBS. This design enabled the collection of data at a single point in time, providing a snapshot of parental perceptions as well as the factors that may influence their views within a real-world clinical setting.

3.2 STUDY DURATION

The study was conducted over a two-month period, from May to June 2026.

3.3 STUDY SETTING

The study was carried out in the paediatric outpatient clinics of Hospital Tunku Ampuan Besar Tuanku Aishah Rohani, UKM Specialist Children's Hospital (Hospital TABTAR), a tertiary paediatric healthcare centre in Malaysia. Data collection was conducted across multiple paediatric outpatient clinics within the hospital to ensure a broader representation of eligible participants.

3.4 STUDY POPULATION

The target population comprised parents or guardians of paediatric outpatients attending the paediatric outpatient clinics at Hospital TABTAR.

3.4.1 Inclusion Criteria

- Parents/guardians aged 18 years and above

- Malaysian citizens
- Willing to provide informed consent
- Able to read and understand either Bahasa Melayu or English
- Having at least one child receiving outpatient paediatric care at Hospital TABTAR

3.4.2 Exclusion criteria

- Parents who had previously participated in similar studies conducted at the same site
- Parents unable to comprehend the study information due to language barriers or literacy limitations

3.5 SAMPLE SIZE

The minimum required sample size was determined using the single-proportion formula for cross-sectional studies:

$$n = \frac{z^2 p(1-p)}{E^2}$$

In the absence of prior Malaysian data on parental perceptions of gNBS, the calculation assumed a 95% confidence level ($Z = 1.96$), a 5% margin of error ($E = 0.05$) and a conservative estimated proportion ($p = 0.5$). This yielded a minimum required sample size of 385 participants.

To account for potential incomplete responses, missing data or invalid entries, an additional 10% was added, resulting in a final target sample size of 423 participants.

A total of 429 valid responses were obtained, exceeding the minimum required sample size.

3.6 STUDY CONDUCT

Ethical approval for this study was obtained from the Research Ethics Committee of Universiti Kebangsaan Malaysia (JEPUKM) (Appendix A) prior to the commencement of data collection (Reference No. JEP-2026-109). Research conduct approval (Appendix B) was granted under project code FF-2026-159. Ethical approval ensured that the study was conducted in accordance with established ethical principles, legal requirements and professional standards for research involving human participants.

A convenience sampling approach was used during routine operating clinic hours. Patient appointment lists were obtained from clinic staff to facilitate the identification of potentially eligible participants. Parents who were present in the waiting area were approached and screened for eligibility. Those who met the inclusion criteria were provided with a brief explanation of the study, along with a parent information sheet (Appendix C), informed consent form (Appendix D) and an introductory pamphlet on NBS and gNBS (Appendix E). Eligible participants were then invited to participate in the study and written informed consent was obtained prior to participation.

Participants were subsequently given a structured, self-administered questionnaire (Appendix F), available in either paper-based or online format depending on their preference. Participation was entirely voluntary and anonymous, and non-participation had no impact on the clinical care received. The questionnaire required approximately 20 to 30 minutes to complete.

3.7 STUDY TOOL

Data were collected using an adapted structured questionnaire available in both English and Bahasa Melayu. The questionnaire was adapted from instruments developed by Elhadi et al. (2025) and Prosenc et al. (2023), and permission to use and adapt these instruments were obtained from the original authors.

The questionnaire was reviewed and modified to ensure its relevance to the Malaysian healthcare context and cultural setting. A forward-backward translation

process was then conducted to produce the Bahasa Melayu version. Two independent bilingual language educators performed the translation and back-translation while blinded to the original questionnaire. Any discrepancies identified were discussed and resolved through consensus to ensure conceptual and linguistic equivalence.

The translated questionnaire subsequently underwent pilot testing, face validation, bilingual validation and content validation. Feedback from these processes was used to refine the wording, structure and response options. Minor revisions were made prior to finalisation. The final bilingual questionnaire was made available in both paper-based and online formats for data collection.

3.7.1 Questionnaire Design and Structure

The questionnaire consisted of five sections with a total of 37 items, as shown in Table 3.1. Most questions were close-ended with response options including "Yes", "No" and "Not sure", while several items allowed multiple responses. The full questionnaire is provided in the Appendix F.

Table 3.1 Questionnaire Design and Structure

Section	Content	Number of Items
A	Demographics	13
B	Perception of Genetic Disease and Genetic Testing	4
C	Parental Perspectives on gNBS	6
D	gNBS Implementation Preferences	8
E	Ethical and Logistical Considerations	6

3.7.2 Pilot Study and Face Validation

A pilot study involving 12 participants was conducted to evaluate the clarity, comprehensibility, cultural appropriateness and feasibility of the questionnaire. The pilot study also assessed the time required for completion and identified any ambiguous or problematic items.

Each participant completed both the English and Bahasa Melayu versions of the questionnaire on separate occasions with at least a one-day interval between

administrations. Following completion, cognitive debriefing interviews were conducted individually to explore participants' understanding of the items and the clarity of the bilingual translations.

The cognitive debriefing guide was adapted from recommendations by Schaefer (2021). Participants were first asked to provide their overall impressions of the questionnaire, followed by a structured item-by-item review to identify areas requiring improvement.

Feedback obtained during the pilot testing and face validation process was systematically reviewed and used to refine the wording, structure, response options and language of the questionnaire. Minor revisions were made accordingly. The average completion time, including reading and responding was 23.08 minutes, ranging from 18 to 30 minutes.

3.7.3 Bilingual Validation

Agreement between the English and Bahasa Melayu versions were assessed by comparing participants' responses to corresponding items. An agreement score of 1 was assigned when responses were identical and 0 when responses differed. Percentage agreement was then calculated for each item and averaged across all items to determine overall bilingual equivalence. The bilingual validation demonstrated an average agreement of 87.92% which falls within an acceptable range, indicating good equivalence between the two versions.

3.7.4 Content Validation

Content validity was assessed by a panel of three experts comprising a clinical geneticist, a neonatologist and a genetic counsellor. The experts evaluated each item for relevance, clarity and representativeness using a four-point rating scale.

The item-level content validity index (I-CVI) was calculated as follows: $I-CVI = (\text{Number of experts rating the item as 3 or 4}) / (\text{Total number of experts})$. An I-CVI value of ≥ 0.78 was considered acceptable. The scale-level content validity index (S-

CVI) was calculated as the average of all I-CVI values across items, with values ≥ 0.90 indicating excellent content validity.

All items achieved an I-CVI of 1.00, while the overall S-CVI was also 1.00, indicating excellent content validity of the instrument.

3.8 DATA ANALYSIS

Data was entered, cleaned and coded prior to analysis using SPSS Statistics version 32.0.0.0 (134). Data cleaning procedures included checking for missing values, outliers and inconsistencies to ensure data accuracy and completeness.

Descriptive statistics including frequencies and percentages for categorical variables and means with standard deviations for continuous variables, were used to summarize participants' demographic characteristics as well as their responses regarding perceptions, attitudes and acceptance of gNBS.

Selected categorical variables were recategorised prior to inferential analysis to ensure adequate cell counts and facilitate statistical analysis. Specifically, educational level was dichotomised into low educational level (no formal, primary and secondary education) and high educational level (tertiary education).

Inferential analyses were conducted to examine associations between sociodemographic factors and parental responses. Pearson's chi-square test or Fisher-Freeman-Halton Exact Test, as appropriate, was used for categorical variables. Variables that demonstrated statistically significant associations in bivariate analyses were subsequently included in logistic regression models to identify factors independently associated with willingness to support the implementation of gNBS.

A p-value of less than 0.05 was considered statistically significant.

CHAPTER IV

RESULTS

4.1 DEMOGRAPHIC PROFILE OF PARTICIPANTS

A total of 429 parents participated in this study. The demographic characteristics of the respondents are summarised in Table 4.1. Overall, the respondents comprised individuals from diverse age groups, ethnic backgrounds, educational levels, occupations and income categories, providing a broad representation of parents attending Hospital TABTAR.

As shown in Table 4.1, nearly half of the respondents were aged 39 - 48 years (49.9%), followed by those aged 29 - 38 years (36.6%). Female respondents constituted the majority of the sample (64.6%), while males accounted for 35.4%.

Most respondents were of Malay ethnicity (82.5%) and practised Islam (83.4%). In terms of educational attainment, the majority had completed tertiary education (80.2%), whereas 18.4% had attained secondary education and only a small proportion reported primary (1.2%) or no formal education (0.2%). Most respondents were employed (85.5%), with government sector employees representing the largest occupational group (47.3%), followed by those working in the private sector (25.6%). With respect to household income, 43.1% of respondents belonged to the M40 income category (RM5250 - RM11819), while 38.2% were classified as B40 (< RM5250) and 18.6% as T20 ($\geq$ RM11820).

Regarding health- and genetic-related characteristics, 21.4% of respondents reported having a chronic disease, while 22.4% reported a family history of genetic disorder. Previous experience with genetic testing was reported by 23.8% of

respondents. Only 12.1% had previously attended a genetic clinic, whereas the majority (87.9%) had no prior experiences with genetic clinic services.

Table 4.1 Demographic Profile of Participants, N = 429

Demographic		Frequency (n)	Percentage (%)
Age (years), (n, %)	18 - 28	10	2.3
	29 - 38	157	36.6
	39 - 48	214	49.9
	49 - 58	43	10.0
	59 - 68	5	1.2
Gender (n, %)	Male	152	35.4
	Female	277	64.6
Ethnicity (n, %)	Malay	354	82.5
	Chinese	55	12.8
	Indian	14	3.3
	Other	6	1.4
Religion (n, %)	Islam	358	83.4
	Buddhism	45	10.5
	Christianity	10	2.3
	Hinduism	13	3.0
	Other	3	0.7
	No formal education	1	0.2
Education (n, %)	Primary	5	1.2
	Secondary	79	18.4
	Tertiary	344	80.2
	Other	1	0.2
Occupation (n, %)	Private	110	25.6
	Government	203	47.3
	Self-employed	54	12.6
	Unemployed	54	12.6
	Student	2	0.5
	Other	6	1.4
Income (n, %)	< RM5250 (B40)	164	38.2
	RM5250–RM11819 (M40)	185	43.1
	≥ RM11820 (T20)	80	18.6

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Marital status (n, %)	Single	7	1.6
	Married	403	93.9
	Divorced	13	3.0
	Widowed	4	0.9
	Separated	2	0.5
Number of children (n, %)	1	78	18.2
	2	133	31.0
	3	113	26.3
	≥ 4	105	24.5
Chronic illness (n, %)	Yes	92	21.4
	No	337	78.6
Family with genetic disorder (n, %)	Yes	96	22.4
	No	333	77.6
Prior genetic testing experience (n, %)	Yes	102	23.8
	No	327	76.2
Visited genetic clinic (n, %)	Yes	52	12.1
	No	377	87.9

4.2 PARENTAL PERSPECTIVES, ATTITUDES AND ACCEPTANCE OF GNBS

4.2.1 Parental Awareness and Perceptions of Genetics and gNBS

This section describes parental awareness and perceptions related to genetics, NBS, and gNBS. The findings are presented in Table 4.2.

Regarding perceptions of genetic disease risk, over half of the respondents (51.5%) perceived the likelihood of a child being born with a genetic disease as low, while 28.7% considered the risk moderate and 19.8% perceived it as very low. When asked about the treatment of genetic conditions, 47.3% believed that modern medicine can treat genetic diseases, whereas 16.8% disagreed and 35.9% were uncertain. Most respondents (74.6%) reported that prenatal testing could reduce the risk of having a child with a genetic condition, while 8.9% disagreed and 16.6% were unsure.

Awareness levels varied across the various forms of genetic testing and screening. Less than half of the respondents (44.5%) were aware that genetic testing can be performed in newborns, while 29.1% reported no awareness and 26.3% were unsure. Awareness of conventional NBS was slightly higher, with 48.0% reporting awareness, compared with 42.9% were unaware and 9.1% who were uncertain. In contrast, awareness of gNBS was considerably lower. Only 21.0% of respondents reported being aware of gNBS, while 71.3% were unaware of its existence and 7.7% were unsure. Compared with NBS, awareness of gNBS was substantially lower among the study population.

Table 4.2 Awareness and Perceptions of Genetics and gNBS

Variables	Response	Frequency (n)	Percentage (%)
Likelihood of genetic disease	Very small	85	19.8
	Small	221	51.5
	Moderate	123	28.7
Modern medicine treats genetic disease	Yes	203	47.3
	No	72	16.8
	Not sure	154	35.9
Prenatal testing reduces risk	Yes	320	74.6
	No	38	8.9
	Not sure	71	16.6
Know genetic testing exists in newborns	Yes	191	44.5
	No	125	29.1
	Not sure	113	26.3
NBS awareness	Yes	206	48.0
	No	184	42.9
	Not sure	39	9.1
gNBS awareness	Yes	90	21.0
	No	306	71.3
	Not sure	33	7.7

4.2.2 Parental Perceptions of gNBS

This section describes parental perceptions of gNBS in terms of its perceived benefits (Figure 4.1) and potential risks (Figure 4.2).

Overall, respondents reported several perceived benefits of gNBS. The most frequently selected benefit was the ability to provide early and effective treatment (88.1%), followed by facilitating an early and timely genetic diagnosis (82.3%) and providing accurate and comprehensive health information (78.3%). Parents viewed gNBS favourably, especially for its potential clinical benefits.

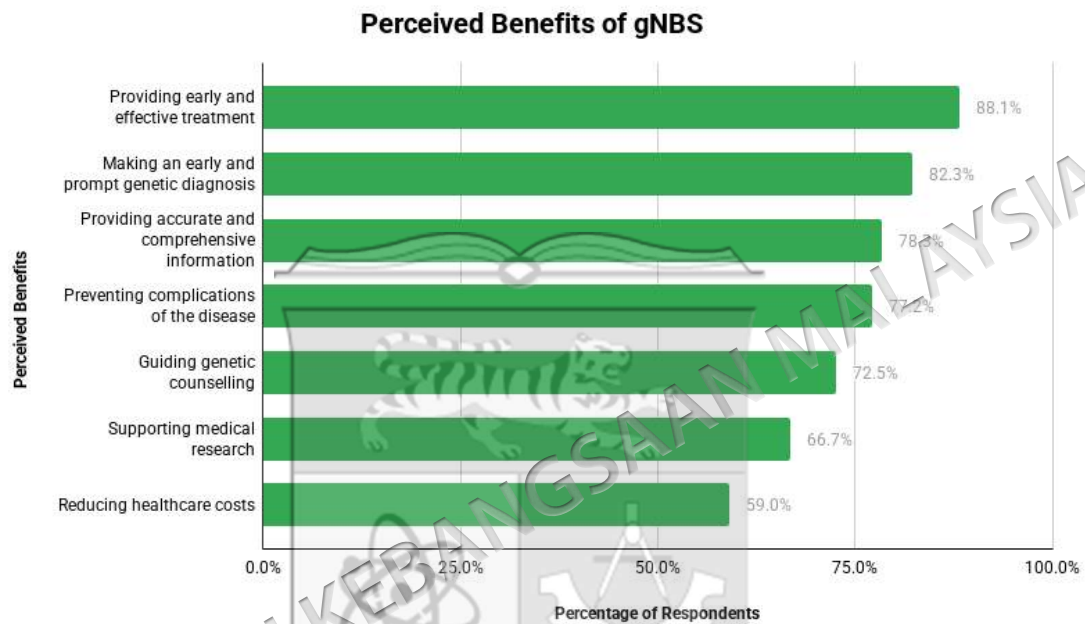


Figure 4.1 Perceived Benefits of gNBS

Figure 4.2 illustrates respondents' perceived risks and concerns regarding gNBS. The most frequently reported concern was anxiety about a child's future health following the disclosure of genetic results (79.5%). Concerns regarding the cost of genetic testing and subsequent treatment (74.1%). In addition, 64.1% expressed concern about the possibility of misinterpreting genetic results and making inappropriate decisions. Other reported concerns included issues related to privacy and confidentiality of genetic information, potential discrimination and the uncertainty associated with some genetic findings. Overall, concerns related to psychological impact, financial burden and interpretation of genetic information were among the most commonly reported perceived risks of gNBS.

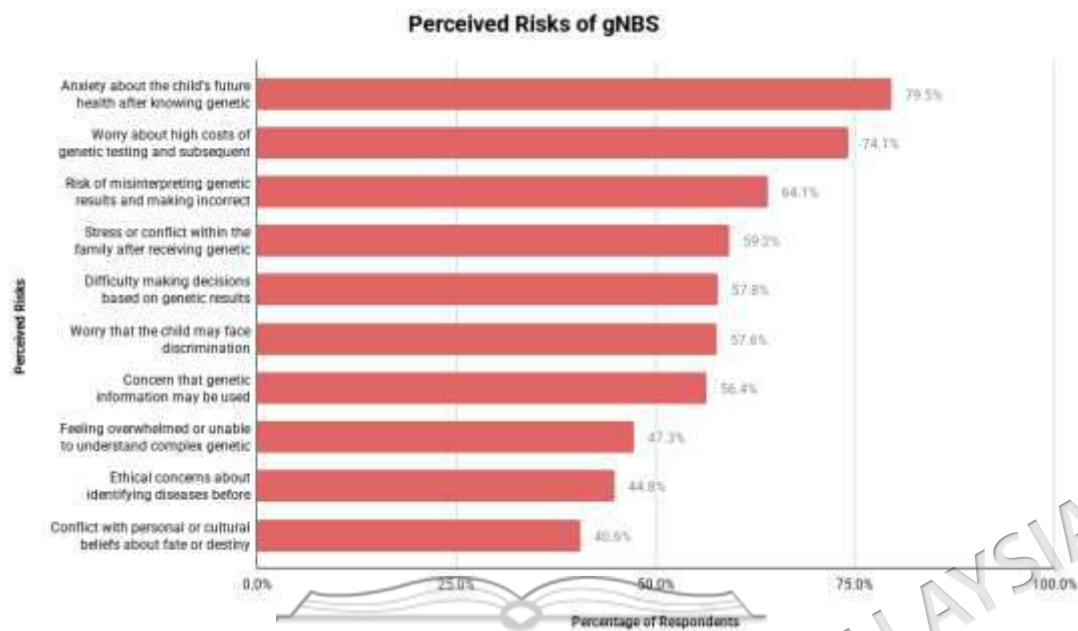


Figure 4.2 Perceived Risks of gNBS

4.2.3 Parental Attitudes Toward and Acceptance of gNBS

This section describes parental attitudes towards and acceptance of gNBS, as presented in Figure 4.3.

The majority of respondents (74.6%) supported the implementation of gNBS for all newborns, while 25.4% were either opposed or uncertain. Acceptance of genetic screening was further reflected by the finding that 82.5% of respondents preferred to know about detectable genetic conditions before pregnancy. Respondents also expressed strong support for informed consent in the context of gNBS. The majority (90.2%) believed that parental consent should be obtained before conducting gNBS, whereas only 9.8% felt that gNBS should be performed routinely without parental consent.

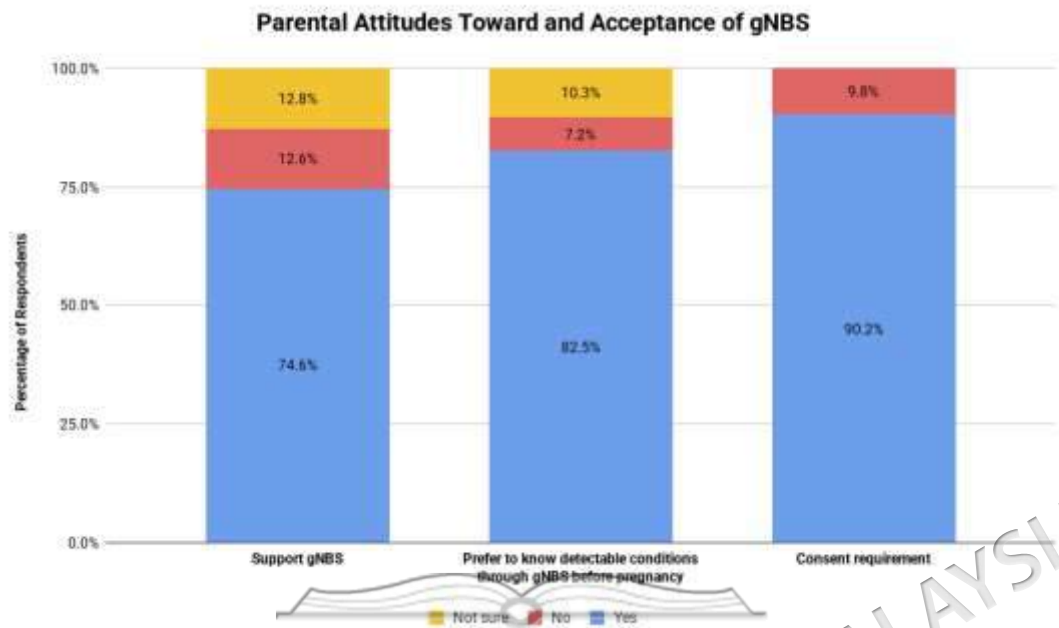


Figure 4.3 Parental Attitudes Toward and Acceptance of gNBS

4.2.4 Parental Implementation Preferences for gNBS

This section describes parental preferences regarding the implementation of gNBS, as presented in Table 4.3.

With regard to the timing of discussions about gNBS, equal proportions of respondents preferred these discussions to occur before (37.5%) and during pregnancy (37.5%). Smaller proportions preferred discussions at birth (8.9%) or after birth (5.4%), while 10.7% were unsure. Regarding the provision of information, respondents most frequently selected paediatricians (64.8%) and obstetricians/gynaecologists (64.6%) as the preferred healthcare professionals to explain gNBS. Clinical geneticist (43.4%) and genetic counsellors (39.9%) were selected less frequently, followed by general practitioners (31.5%), nurses (19.8%) and midwives (14.0%). The most preferred setting for discussing gNBS was the obstetric or antenatal clinic (57.6%), followed by a dedicated genetic counselling unit (55.0%) and hospital-based services (50.3%).

Face-to-face discussion was the most preferred method of information delivery (76.2%), followed by written materials such as pamphlets and brochures (67.4%) and online resources including websites, videos and mobile applications (62.9%).

Most respondents preferred that consent for gNBS be obtained during pregnancy (73.7%), whereas 17.0% preferred consent to be obtained at birth and 9.3% were unsure. With regard to eligibility for gNBS, the most frequently selected groups were newborns with a family history of genetic disease (68.5%), newborns with a parent or sibling affected by a known genetic condition (67.4%) and all newborns regardless of family history (61.8%). Other groups identified included newborns considered high risk by their doctor (59.7%) and newborns admitted to neonatal intensive care unit (40.3%).

Table 4.3 Parental Implementation Preference of gNBS

Variables	Response	Frequency (n)	Percentage (%)
Timing for discussing gNBS	Before pregnancy	161	37.5
	During pregnancy	161	37.5
	At birth	38	8.9
	After birth	23	5.4
	Not sure	46	10.7
Personnel to provide gNBS information	Paediatrician	278	64.8
	Obstetrician / Gynaecologist	277	64.6
	Clinical geneticist	186	43.4
	Genetic counsellor	171	39.9
	General practitioner	135	31.5
	Nurse	85	19.8
	Midwife	60	14.0
	Not sure	35	8.2
Setting for discussing gNBS	Obstetrics / antenatal clinic	247	57.6
	Dedicated genetic counselling unit	236	55.0
	Hospital	216	50.3
	Via written materials	146	34.0
	General practitioner's clinic	134	31.2
	Via telehealth / video consultation	88	20.5
	Not sure	41	9.6
Mode of gNBS information delivery	Face-to-face discussion	327	76.2
	Written materials	289	67.4
	Online resources	270	62.9
	Group information session	186	43.4
	Telehealth / video consultation	142	33.1

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	A combination of the above	176	41.0
Timing for consent	During pregnancy	316	73.7
	At birth	73	17.0
	Not sure	40	9.3
Eligibility for gNBS	Newborns with a family history of genetic disease (any relative)	294	68.5
	Newborns with a parent or sibling with a known genetic condition	289	67.4
	All newborns	265	61.8
	Newborns identified as high-risk by their doctor	256	59.7
	Newborns admitted to the neonatal intensive care unit (NICU)	173	40.3
	Not sure	37	8.6

4.2.5 Factors Influencing Parental Decisions on gNBS

This section describes the factors influencing parental decisions regarding gNBS, as presented in Figure 4.4.

The most frequently selected factor influencing decision-making was the cost of testing and treatment was (86.9%). This was followed by the diseases being tested for (75.4%) and the availability of treatment for identified conditions (71.9%). Other commonly selected factors included disease severity (66.3%), insurance coverage (65.8%), and the likelihood and timing of symptom onset (62.3%).

In contrast, research-related and family-related considerations were selected less frequently. These included the storage of samples for research purposes (48.9%), whether the results would apply to other children in the family (47.5%) and the perceived impact of results on other children (35.4%). The least frequently selected factor was the location of treatment (0.2%).

Generally, the findings indicate that parental decisions were primarily driven by financial burden and clinical relevance, while research-related and logistical factors played a secondary role.

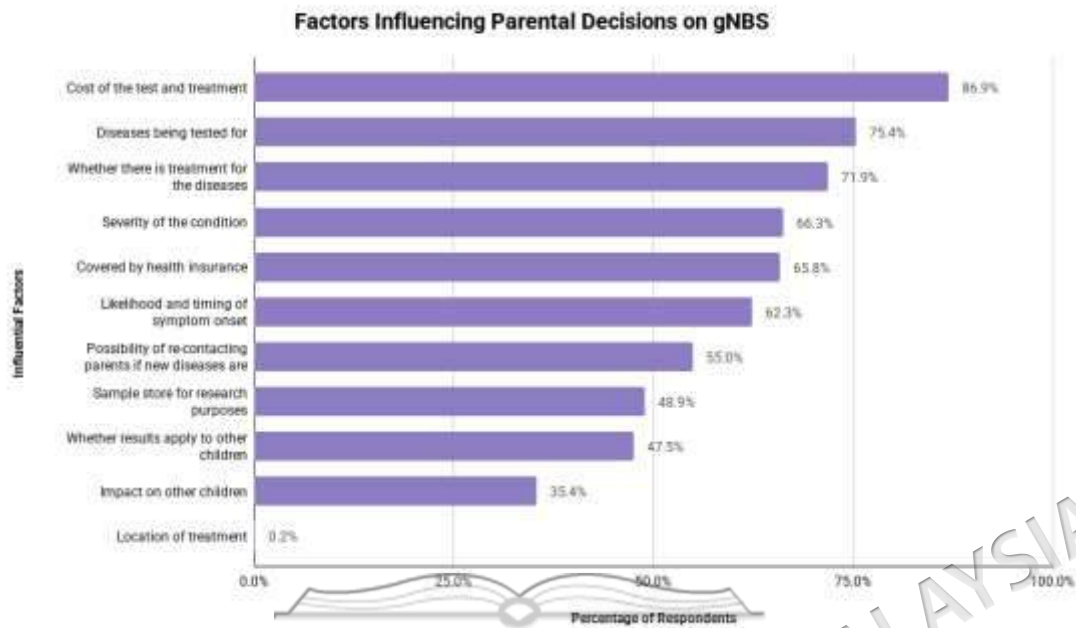


Figure 4.4 Factors Influencing Parental Decisions on gNBS

4.2.6 Parental Ethical and Logistical Considerations

This section describes parental ethical and logistical considerations regarding gNBS, as presented in Table 4.4.

Most respondents preferred that gNBS be subsidised by the government (68.3%), while smaller proportions preferred funding through insurance coverage (16.6%) or individual self-payment (15.2%).

In terms of data governance, the majority of respondents (80.7%) supported the storage of genomic data within hospital systems for future clinical care. For the use of stored genomic data in research, the most frequently selected consent model was explicit consent each time data is used (62.9%), followed by one-time consent at the point of testing (19.3%).

Preferences regarding data storage duration varied. The most commonly selected option was storage until the child reaches adulthood (30.3%), followed by lifetime storage (22.8%) and indefinite storage without time limits (19.3%). A smaller

proportion preferred storage only until the initial results had been reported (8.4%), while 19.1% were unsure.

Most respondents (86.7%) expressed interest in receiving future findings resulting from the reanalysis of stored genomic data. Only 0.7% indicated no interest, while 12.6% were unsure.

When asked whether gNBS requires different governance and management compared with conventional newborn screening, 69.0% of respondents agreed, while 8.6% disagreed and 22.4% were uncertain.

Table 4.4 Parental Ethical and Logistical Considerations

Variables	Response	Frequency (n)	Percentage (%)
Payer for testing	Government	293	68.3
	Insurance	71	16.6
	Individuals	65	15.2
Support data storage for clinical care	Yes	346	80.7
	No	23	5.4
	Not sure	60	14.0
Support data storage for research	Explicit consent each time	270	62.9
	One-time consent	83	19.3
	No	36	8.4
	Not sure	40	9.3
Data storage duration	18 years	130	30.3
	Lifetime	98	22.8
	Indefinite	83	19.3
	Only initial result	36	8.4
	Not sure	82	19.1
Interest in future findings	Yes	372	86.7
	No	3	0.7
	Not sure	54	12.6
gNBS requires different management from conventional NBS	Yes	296	69.0
	No	37	8.6
	Not sure	96	22.4

4.3 FACTORS ASSOCIATED WITH PERCEPTIONS AND WILLINGNESS TO SUPPORT GNBS

4.3.1 Association Between Selected Sociodemographic Factors and Selected Perceived Benefits and Risks of gNBS

Table 4.5 and Table 4.6 present the associations between selected sociodemographic factors and selected perceived benefits and risks of gNBS. The sociodemographic factors examined were gender, educational attainment and monthly household income. The selected perceived benefits included providing early and effective treatment, facilitating early and prompt genetic diagnosis, and providing accurate and comprehensive health information. The selected perceived risks included anxiety regarding a child's future health after receiving genetic results, concerns about the cost of genetic testing and subsequent treatment, and the risk of misinterpreting genetic results and making incorrect decisions. These perceived benefits and risks were selected based on the most frequently reported responses and were consistently used throughout Sections 4.3.1 and 4.3.2.

Among the perceived benefits, gender was significantly associated with the perception that gNBS provides early and effective treatment ($p = 0.005$). Female respondents were more likely than male respondents to identify this benefit (91.3% vs. 82.2%). Educational attainment was also significantly associated with this perceived benefit ($p < 0.001$), with respondents who had completed higher education reporting this benefit more frequently than those with lower educational attainment. No significant association was observed between household income and this perceived benefit ($p = 0.240$). Furthermore, no significant associations were found between gender, educational attainment or household income and the perceived benefits of facilitating an early and prompt genetic diagnosis or providing accurate and comprehensive health information (all $p > 0.05$).

Table 4.5 Association Between Selected Sociodemographic Factors and Selected Perceived Benefits of gNBS

Variables	Response	Providing early and effective treatment		P value
		Agree n (%)	Disagree n (%)	
Gender	Male	125 (82.2)	27 (17.8)	0.005
	Female	253 (91.3)	24 (8.7)	
Education level	Low level	66 (77.6)	19 (22.4)	< 0.001
	High level	312 (90.7)	32 (9.3%)	
Income	B40	139 (84.8)	25 (15.2)	0.240
	M40	167 (90.3)	18 (9.7)	
	T20	72 (90.0)	8 (10.0)	
Variables	Response	Making an early and prompt genetic diagnosis		P value
		Agree n (%)	Disagree n (%)	
Gender	Male	124 (81.6)	28 (18.4)	0.777
	Female	229 (82.7)	48 (17.3)	
Education level	Low level	69 (81.2)	16 (18.8)	0.765
	High level	284 (82.6)	60 (17.4)	
Income	B40	130 (79.3)	34 (20.7)	0.434
	M40	156 (84.3)	29 (15.7)	
	T20	67 (83.8)	13 (16.3)	
Variables	Response	Providing accurate and comprehensive information		P value
		Agree n (%)	Disagree n (%)	
Gender	Male	120 (78.9)	32 (21.1)	0.816
	Female	216 (78.0)	61 (22.0)	
Education level	Low level	72 (84.7)	13 (15.3)	0.111
	High level	264 (76.7)	80 (23.3)	
Income	B40	127 (77.4)	37 (22.6)	0.066
	M20	153 (82.7)	32 (17.3)	
	T20	56 (70.0)	24 (30.0)	

Chi-square test was used. $P < 0.05$ considered statistically significant.

Regarding perceived risks, educational attainment was significantly associated with concern about the risk of misinterpreting genetic results and making inappropriate decisions ($p = 0.002$). The proportion of respondents selecting this concern was lower among those with a low educational level (no formal, primary or secondary education; 49.4%) than among those with a high educational level (tertiary education; 67.7%). No

significant associations were observed between gender or household income and this perceived risk (both $p > 0.05$). In addition, no significant associations were found between the selected sociodemographic factors and concerns regarding anxiety about a child's future health following disclosure of genetic results or concerns about the high cost of genetic testing and subsequent treatment (all $p > 0.05$).

Table 4.6 Association Between Selected Sociodemographic Factors and Selected Perceived Risks of gNBS

Variables	Response	Anxiety about the child's future health after knowing genetic results		P value
		Agree n (%)	Disagree n (%)	
Gender	Male	114 (75.0)	38 (25.0)	0.088
	Female	227 (81.9)	50 (18.1)	
Education level	Low level	64 (75.3)	21 (24.7)	0.285
	High level	277 (80.5)	67 (19.5)	
Income	B40	132 (80.5)	32 (19.5)	0.545
	M40	149 (80.5)	36 (19.5)	
	T20	60 (75.0)	20 (25.0)	
Variables	Response	Worry about high costs of genetic testing and subsequent treatment		P value
		Agree n (%)	Disagree n (%)	
Gender	Male	108 (71.1)	44 (28.9)	0.282
	Female	210 (75.8)	67 (24.2)	
Education level	Low level	60 (70.6)	25 (29.4)	0.406
	High level	258 (75.0)	86 (25.0)	
Income	B40	120 (73.2)	44 (26.8)	0.056
	M40	146 (78.9)	39 (21.1)	
	T20	52 (65.0)	28 (35.0)	
Variables	Response	Risk of misinterpreting genetic results and making incorrect decisions		P value
		Agree n (%)	Disagree n (%)	
Gender	Male	93 (61.2)	59 (38.8)	0.351
	Female	182 (65.7)	95 (34.3)	
Education level	Low level	42 (49.4)	43 (50.6)	0.002
	High level	233 (67.7)	111 (32.3)	
Income	B40	102 (62.2)	62 (37.8)	0.765
	M40	122 (65.9)	63 (34.1)	

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T20	51(63.7)	29 (36.3)
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Chi-square test was used. $P < 0.05$ considered statistically significant.

Association Between Genetics-Related Perceptions Factors and Selected Perceived Benefits and Risks of gNBS

Table 4.7 and Table 4.8 present the associations between selected genetics-related perception factors and selected perceived benefits and risks of gNBS. The factors examined were perceived likelihood of genetic disease, belief that modern medicine can treat genetic diseases, and perception that prenatal testing can reduce the risk of having a child with a genetic condition.

Among the perceived benefits, the perception that prenatal testing can reduce the risk of having a child with a genetic condition was significantly associated with the perception that gNBS provides early and effective treatment ($p = 0.016$). Respondents who agreed with this statement were more likely to identify early and effective treatment as a benefit of gNBS (90.6%) compared with those who disagreed (84.2%) or were unsure (78.9%).

The same perception factor was also significantly associated with the belief that gNBS provides accurate and comprehensive information ($p < 0.001$). A higher proportion of respondents who agreed that prenatal testing reduces risk selected this benefit (84.1%) compared with those who disagreed (63.2%) or were unsure (60.6%).

No significant associations were observed between perceived likelihood of genetic disease or belief that modern medicine can treat genetic diseases and any of the selected perceived benefits of gNBS (all $p > 0.05$).

Table 4.7 Association Between Genetics-Related Perceptions Factors and Selected Perceived Benefits of gNBS

Variables	Response	Providing early and effective treatment		P value
		Agree n (%)	Disagree n (%)	
Likelihood of genetic disease	Very small	73 (85.9)	12 (14.1)	0.777
	Small	196 (88.7)	25 (11.3)	
	Moderate	109 (88.6)	14 (11.4)	
Modern medicine treats genetic disease	Yes	184 (90.6)	19 (9.4)	0.075
	No	58 (80.6)	14 (19.4)	
	Not sure	136 (88.3)	18 (11.7)	
Prenatal testing reduces risk	Yes	290 (90.6)	30 (9.4)	0.016
	No	32 (84.2)	6 (15.8)	
	Not sure	56 (78.9)	15 (21.1)	
Variables	Response	Making an early and prompt genetic diagnosis		P value
		Agree n (%)	Disagree n (%)	
Likelihood of genetic disease	Very small	70 (82.4)	15 (17.6)	0.643
	Small	185 (83.7)	36 (16.3)	
	Moderate	98 (79.7)	25 (20.3)	
Modern medicine treats genetic disease	Yes	163 (80.3)	40 (19.7)	0.584
	No	61 (84.7)	11 (15.3)	
	Not sure	129 (83.8)	25 (16.2)	
Prenatal testing reduces risk	Yes	270 (84.4)	50 (15.6)	0.129
	No	30 (78.9)	8 (21.1)	
	Not sure	53 (74.6)	18 (25.4)	
Variables	Response	Providing accurate and comprehensive information		P value
		Agree n (%)	Disagree n (%)	
Likelihood of genetic disease	Very small	68 (80.0)	17 (20.0)	0.531
	Small	176 (79.6)	45 (20.4)	
	Moderate	92 (74.8)	31 (25.2)	
Modern medicine treats genetic disease	Yes	163 (80.3)	40 (19.7)	0.642
	No	55 (76.4)	17 (23.6)	
	Not sure	118 (76.6)	36 (23.4)	
Prenatal testing reduces risk	Yes	269 (84.1)	51 (15.9)	< 0.001
	No	24 (63.2)	14 (36.8)	
	Not sure	43 (60.6)	28 (39.4)	

Chi-square test was used. $P < 0.05$ considered statistically significant.

In addition, no significant associations were observed between any of the selected genetics-related perception factors and the perceived risks of anxiety regarding a child's future health following disclosure of genetic results, concerns about the cost of genetic testing and subsequent treatment, or the risk of misinterpreting genetic results and making inappropriate decisions (all $p > 0.05$).

Table 4.8 Association Between Genetics-Related Perceptions Factors and Selected Perceived Risks of gNBS

Variables	Response	Anxiety about the child's future health after knowing genetic results		P value
		Agree n (%)	Disagree n (%)	
Likelihood of genetic disease	Very small	64 (75.3)	21 (24.7)	0.137
	Small	184 (83.3)	37 (16.7)	
	Moderate	93 (75.6)	30 (24.4)	
Modern medicine treats genetic disease	Yes	160 (78.8)	43 (21.2)	0.923
	No	57 (79.2)	15 (20.8)	
	Not sure	124 (80.5)	30 (19.5)	
Prenatal testing reduces risk	Yes	254 (79.4)	66 (20.6)	0.799
	No	29 (76.3)	9 (23.7)	
	Not sure	58 (81.7)	13 (18.3)	
Variables	Response	Worry about high costs of genetic testing and subsequent treatment		P value
		Agree n (%)	Disagree n (%)	
Likelihood of genetic disease	Very small	63 (74.1)	22 (25.9)	0.284
	Small	170 (76.9)	51 (23.1)	
	Moderate	85 (69.1)	38 (30.9)	
Modern medicine treats genetic disease	Yes	148 (72.9)	55 (27.1)	0.209
	No	49 (68.1)	23 (31.9)	
	Not sure	121 (78.6)	33 (21.4)	
Prenatal testing reduces risk	Yes	239 (74.7)	81 (25.3)	0.874
	No	27 (71.1)	11 (28.9)	
	Not sure	52 (73.2)	19 (26.8)	
Variables	Response	Risk of misinterpreting genetic results and making incorrect decisions		P value
		Agree n (%)	Disagree n (%)	
Likelihood of genetic disease	Very small	54 (63.5)	31 (36.5)	0.887
	Small	144 (65.2)	77 (34.8)	

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	Moderate	77 (62.6)	46 (37.4)	
Modern medicine treats genetic disease	Yes	130 (64.0)	73 (36.0)	0.183
	No	40 (55.6)	32 (44.4)	
	Not sure	105 (68.2)	49 (31.8)	
Prenatal testing reduces risk	Yes	206 (64.4)	114 (35.6)	0.889
	No	23 (60.5)	15 (39.5)	
	Not sure	46 (64.8)	25 (35.2)	

Chi-square test was used. $P < 0.05$ considered statistically significant.

4.3.2 Association Between Selected Sociodemographic Factors and Willingness to Support gNBS

Table 4.9 presents the associations between selected sociodemographic factors and willingness to support the implementation of gNBS for all newborns. The sociodemographic factors examined were gender, educational attainment and monthly household income, consistent with those analysed in Section 4.3.1.

Monthly household income was significantly associated with willingness to support the implementation of gNBS ($p = 0.026$). The proportion of respondents supporting gNBS increased across income categories, from 70.1% among respondents in the B40 group to 76.8% in the M40 group and 78.8% in the T20 group. In contrast, no significant associations were observed between willingness to support gNBS and gender ($p = 0.356$) or educational attainment ($p = 0.069$).

Table 4.9 Association Between Selected Sociodemographic Factors and Willingness to Support gNBS

Variables	Response	Support gNBS			P value
		Yes n (%)	No n (%)	Not sure n (%)	
Gender	Male	111 (73.0)	17 (11.2)	24 (15.8)	0.356
	Female	209 (75.5)	37 (13.4)	31 (11.2)	
Education level	Low level	60 (70.6)	8 (9.4)	17 (20.0)	0.069
	High level	260 (75.6)	46 (13.4)	38 (11.0)	
Income	B40	115 (70.1)	18 (11.0)	31 (18.9)	0.026

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M40	142 (76.8)	23 (12.4)	20 (10.8)
T20	63 (78.8)	13 (16.3)	4 (5.0)

Chi-square test was used. $P < 0.05$ considered statistically significant.

4.3.3 Association Between Genetics-Related Perceptions Factors and Willingness to Support gNBS

Table 4.10 presents the associations between selected genetics-related perception factors and willingness to support the implementation of gNBS for all newborns. The factors examined were perceived likelihood of genetic disease, belief that modern medicine can treat genetic diseases, and perception that prenatal testing can reduce the risk of having a child with a genetic condition, consistent with those analysed in Section 4.3.2.

Belief that modern medicine can treat genetic diseases was significantly associated with willingness to support gNBS ($p = 0.003$). Respondents who agreed that genetic diseases can be treated with modern medicine were more likely to support gNBS (82.3%) than those who disagreed (65.3%) or were unsure (68.8%).

Similarly, the perception that prenatal testing can reduce the risk of having a child with a genetic condition was significantly associated with willingness to support gNBS ($p < 0.001$). Support for gNBS was highest among respondents who agreed with this statement (80.9%), whereas 56.0% of those who disagreed or were unsure supported gNBS. In contrast, perceived likelihood of genetic disease was not significantly associated with willingness to support gNBS ($p = 0.461$).

Table 4.10 Association Between Genetics-Related Perceptions Factors and Willingness to Support gNBS

Variables	Response	Support gNBS			P value
		Yes n (%)	No n (%)	Not sure n (%)	
Likelihood of genetic disease	Very small	60 (70.6)	13 (15.3)	12 (14.1)	0.461
	Small	162 (73.3)	31 (14.0)	28 (12.7)	
	Moderate	98 (79.9)	10 (8.1)	15 (12.2)	
Modern medicine treats genetic disease	Yes	167 (82.3)	17 (8.4)	19 (9.4)	0.003
	No	47 (65.3)	16 (22.2)	9 (12.5)	
	Not sure	106 (68.8)	21 (13.6)	27 (17.5)	
Prenatal testing reduces risk	Yes	259 (80.9)	33 (10.3)	28 (8.8)	< 0.001*
	No	26 (68.4)	9 (23.7)	3 (7.9)	
	Not sure	35 (49.3)	12 (16.9)	24 (33.8)	

Chi-square test was used. * indicated Fisher-Freeman-Halton-Exact Test was applied where appropriate. $P < 0.05$ considered statistically significant.

4.3.4 Predictors of Willingness to Support gNBS

Binary logistic regression analysis was performed to identify factors independently associated with willingness to support gNBS. Variables with a p-value of < 0.25 in the bivariate analysis were entered into the multivariable regression model. The results are presented in Table 4.11.

After adjustment for potential confounding factors, monthly household income was not significantly associated with willingness to support gNBS. Similarly, although the perception that prenatal testing can reduce the risk of having a child with a genetic condition was significantly associated with willingness to support gNBS in the bivariate analysis, neither comparison with the reference category remained statistically significant in the multivariable model.

In contrast, belief that genetic diseases can be treated with modern medicine remained significantly associated with willingness to support gNBS. Respondents who believed that genetic diseases can be treated with modern medicine had approximately twice the odds of supporting the implementation of gNBS compared with those who did not (aOR = 1.98, 95% CI: 1.03 - 3.80, $p = 0.039$).

Overall, belief in the treatability of genetic diseases using modern medicine was the only independent factor associated with willingness to support the implementation of gNBS in this study.

Table 4.11 Binary Logistic Regression Analysis for Factors Associated with Willingness to Support gNBS

Variable	P value	aOR (95% CI)
Income (Ref: B40)	0.374	
M40 vs. B40	0.258	1.34 (0.81 – 2.21)
T20 vs. B40	0.233	1.50 (0.77 – 2.92)
Modern medicine treats genetic disease (Ref: No)	0.074	
Yes vs. No	0.039	1.98 (1.03 – 3.80)
Not sure vs. No	0.513	1.24 (0.65 – 2.36)
Prenatal testing reduces risk (Ref: No)	< 0.001	
Yes vs. No	0.190	1.69 (0.77 – 3.72)
Not sure vs. No	0.076	0.45 (0.19 – 1.09)

aOR = Adjusted Odds Ratio; CI = Confidence Interval; Ref = reference group.
Variables: $p < 0.25$ in bivariate analysis. $P < 0.05$ considered statistically significant.

CHAPTER V

DISCUSSION

5.1 OVERVIEW OF FINDINGS

This study explored parental perceptions, attitudes and acceptance and implementation preferences regarding gNBS among parents attending paediatric outpatient clinics at Hospital TABTAR. Overall, the findings indicate that Malaysian parents demonstrated generally positive attitudes towards gNBS and strong support for its implementation despite relatively low levels of awareness. While awareness of conventional NBS was moderate, awareness of gNBS was considerably lower, indicating a significant knowledge gap regarding emerging genomic technologies. Nevertheless, most parents recognised the potential value of gNBS and agreed that it requires distinct governance, consent processes and long-term management compared with conventional NBS.

Parents perceived several important benefits of gNBS, especially its ability to enable early and effective treatment, facilitate early and prompt genetic diagnosis and provide accurate and comprehensive health information. However, they also expressed concerns regarding the potential psychological impact of genetic findings, the risk of misinterpreting results and the financial burden associated with testing and subsequent treatment. These concerns were reflected in parental decision-making, with the cost of testing and treatment, the diseases included in the screening panel and the availability of effective treatments identified as key factors affecting parental support.

The findings from this study suggest that Malaysian parents are generally receptive towards gNBS but highlight the need for public education, appropriate counselling strategies to improve genetic literacy and support informed decision-

making as well robust ethical and regulatory frameworks to support its future implementation.

5.2 PARENTAL PERSPECTIVES, ATTITUDES TOWARD AND ACCEPTANCE OF GNBS

5.2.1 Parental Awareness and Perceptions of Genetics and gNBS

This study found relatively low awareness of gNBS among Malaysian parents, with only 21.0% reporting prior awareness of gNBS. Despite limited awareness of genomic screening, most respondents demonstrated a basic understanding of genetic health concepts, with the majority recognising that prenatal testing can reduce the risk of having a child with a genetic condition. This indicates that parents may be familiar with established genetic services such as prenatal testing, while they may have limited exposure to newer genomic screening approaches. Similar findings have been reported internationally, where awareness of genomic technologies among parents and the general public remains limited despite increasing interest in genomic medicine and precision healthcare (Casauria et al. 2023; Doll et al. 2026).

Several factors may explain the low level of awareness observed in this study. Unlike conventional NBS, gNBS has not yet been implemented as part of routine healthcare services in Malaysia, resulting in limited public exposure to its purpose, benefits and implications. Public education initiatives related to genomic medicine remain relatively limited compared with more established health screening programmes. In addition, access to specialised genetic services in Malaysia is largely concentrated within tertiary healthcare centres such as Hospital TABTAR, HKL, Penang General Hospital and Universiti Malaya Medical Center. Furthermore, the limited number of trained genetics professionals may restrict opportunities for public engagement, genetic counselling and dissemination of genomic information (Codeblue 2021; Genetic Counselling Society of Malaysia n.d.). As genomic medicine continues to develop within Malaysia, awareness among both the public and healthcare providers is likely to vary considerably depending on prior exposure and access to genetic services.

Despite the low level of awareness, most parents expressed support for the implementation of gNBS. This finding suggests that parental acceptance may be driven more by the perceived clinical benefits of early diagnosis and treatment rather than prior familiarity with genomic screening itself (Casauria et al. 2023; Timmins et al. 2022). The discrepancy between awareness and acceptance observed in this study highlights the importance of comprehensive public education, healthcare professional training and community engagement initiatives to improve genomic literacy and support informed decision-making before the wider implementation of gNBS in Malaysia.

5.2.2 Parental Perceptions of gNBS

The study found that the three most frequently perceived benefits of gNBS were providing early and effective treatment, enabling early and prompt genetic diagnosis, and providing accurate and comprehensive information. These findings suggest that parents strongly recognised the clinical value of gNBS, especially its potential to facilitate early diagnosis and intervention before the onset of symptoms. This aligns with the primary aim of genomic screening, which is to identify actionable genetic conditions at an early stage, allowing timely management and potentially improving health outcomes. Previous studies have similarly reported that parents value the ability of genomic screening to reduce diagnostic delays, facilitate early access to treatment and support informed healthcare and reproductive decision-making for families (Christie et al. 2013; Tluczek, Ersig & Lee 2022).

Despite recognising these benefits, parents also expressed concerns regarding several potential risks associated with gNBS. The most frequently selected concern was anxiety about the child's future health after receiving genetic results. This finding is consistent with previous studies showing that parents may experience emotional distress when confronted with information about a genetic condition, especially when they feel unprepared to understand the diagnosis or its implications (Grob, 2008; Jessup et al., 2015). The disclosure of genetic findings may also alter parents' perceptions of their child's health and future, creating uncertainty regarding prognosis, caregiving responsibilities and family dynamics. Concerns about potential stigma or discrimination

associated with genetic information have also been reported in the literature and may contribute to parental anxiety (Tluczek, Ersig & Lee 2022; Wang et al. 2024).

Financial concerns were another important perceived risk, with many parents expressing worries about the costs of genetic testing and subsequent treatment. This finding may reflect concerns regarding the long-term financial burden associated with managing genetic conditions, especially when specialised treatments, long-term monitoring or orphan drugs are required. Previous publications have highlighted that the affordability of genetic services and treatment remains a significant concern for families, especially in settings where funding support and insurance coverage are limited (Thong & Ahmad Annuar, 2018; Wang et al., 2024). Such concerns may also raise issues of health equity, as economically disadvantaged families may face greater barriers to accessing genomic services and follow-up care.

Another commonly perceived risk was the possibility of misinterpreting genetic results and making incorrect decisions. Given the complexity of genomic information, parents may experience confusion or uncertainty when interpreting test results, especially in the absence of adequate counselling and support. Previous studies have shown that misunderstandings of genetic information may contribute to anxiety, influence parenting behaviours and affect decisions related to future reproductive planning and healthcare management (Tluczek, Ersig & Lee, 2022; Wang et al., 2024). This highlights the importance of providing clear communication and appropriate genetic counselling to ensure that parents can understand and appropriately use genomic information.

Overall, the findings indicate that parents recognised both the potential benefits and challenges of gNBS. While they valued the opportunities for early diagnosis, treatment and informed decision-making, concerns regarding psychological impact, financial burden and result interpretation remained important considerations. These findings underscore the need for comprehensive education, accessible genetic counselling services and equitable healthcare policies to support the responsible implementation of gNBS in Malaysia.

5.2.3 Parental Attitudes Toward and Acceptance of gNBS

This study demonstrated a high level of parental acceptance of gNBS with the majority of parents expressing support for its implementation. This finding is consistent with previous studies that reported generally favourable parental attitudes towards gNBS and their potential benefits for early disease detection and intervention (Elhadi et al. 2025; Bombard et al. 2014). The high level of support observed in this study may reflect parents' recognition of the potential value of obtaining genetic information that could improve health outcomes through earlier diagnosis, treatment and preventive care.

In addition to support the implementation of gNBS, most parents agreed that parental consent should be obtained before testing rather than adopting a routine screening approach without explicit consent. This finding suggests that parents recognise the ethical and personal implications of genomic information and wish to remain actively involved in decisions regarding genetic testing for their children. Unlike conventional newborn screening, which primarily targets a limited number of treatable conditions, gNBS may generate broader and more complex information with potential implications for the child and family. Consequently, parents may perceive informed consent as an important safeguard to ensure understanding, autonomy and voluntary participation in the screening process (Elhadi et al. 2025; Tluczek, Ersig & Lee 2022).

The majority of parents also indicated a preference for knowing detectable genetic conditions before pregnancy. Although studies specifically examining this preference in the context of gNBS are limited, findings from prenatal genetic screening research suggest that parents value earlier access to genetic information to support reproductive planning and decision-making (Ersig, Jaja & Tluczek 2023). Early knowledge of genetic risks may provide parents with additional time to consider available reproductive options, prepare emotionally and practically for the possibility of having a child with a genetic condition and seek appropriate medical support when required. Previous research has further shown that access to genetic information before or during pregnancy may reduce uncertainty, facilitate informed decision-making and improve preparedness for future healthcare needs (Doll et al. 2025; Munro et al. 2018; Siranosian et al. 2023).

These findings suggest that Malaysian parents are generally receptive towards gNBS while emphasising the importance of informed choice and parental involvement in decision-making. The preference for parental consent and earlier access to genetic information highlights the need for effective education and counselling strategies to support informed and autonomous decisions regarding genomic screening.

5.2.4 Parental Implementation Preferences for gNBS

This study also provides valuable insights into parental preferences regarding the future implementation of gNBS in Malaysia. Overall, parents expressed preference for receiving information about gNBS before or during pregnancy, with many also indicating that consent should be obtained during the antenatal period. These findings suggest that parents prefer early engagement with genomic information, allowing sufficient time to understand the purpose, benefits and limitations of gNBS before making decisions regarding testing. Previous systematic review studies have similarly reported that parents prefer receiving information about genetic testing before the birth of their child, when they are better able to process information and participate in informed decision-making (Ersig, Jaja & Tluczek 2023).

Parents also preferred discussions regarding gNBS to be conducted by paediatricians and obstetricians/gynaecologists, with antenatal clinics, genetic counselling units and maternity-related healthcare settings recognized as the ideal place for sharing information. This preference indicates the confidence parents have in healthcare professionals who engage with maternal and child healthcare. The findings further highlight the importance of integrating gNBS education into existing maternal and child health services and ensuring collaboration between paediatric, obstetric and genetic healthcare providers.

In terms of communication methods, face-to-face discussions were highly favoured, although educational resources like pamphlets and brochures were also positively perceived. This suggests that parents value opportunities for direct interaction with healthcare professionals while also having access to supplementary materials that can be reviewed at their own pace. Due to the complexity of genomic information,

combining verbal counselling with written materials might enhance comprehension and facilitate informed choices (Genetic Alliance 2009; Suckiel et al. 2016).

Regarding eligibility for screening, parents showed the strongest support for offering gNBS to newborns with a family history of genetic conditions or those with affected first-degree relatives, although support for universal screening of all newborns was also observed. This result suggests that parents see more advantages and reasons for genomic screening in those with increased genetic risk, while still being generally open to broader population-based implementation. Collectively, these results offer significant insights for the development of future gNBS programs and indicate that execution strategies ought to focus on early education, engagement of reliable healthcare professionals and clear communication methods.

5.2.5 Factors Influencing Parental Decisions on gNBS

This study showed that the most influential factors affecting parental decisions on gNBS were the cost of testing and treatment, the diseases included in the screening programme and the availability of treatment for the conditions detected. Rather than focusing solely on the technology itself, these findings suggest that parents evaluate gNBS based on its perceived practical value and clinical usefulness.

When considered collectively, these factors indicate that parents are concerned not only with whether genetic information can be obtained, but also whether the information generated is meaningful, actionable and beneficial for their child and family. Parents may be more willing to support gNBS when the conditions being screened are clearly defined, when effective treatments or interventions are available and when the potential health benefits justify the financial costs associated with testing and subsequent care. Similar considerations have been reported in previous studies, where the perceived utility and actionability of genomic information were important determinants of public acceptance of genomic screening programmes (DeLuca et al. 2017; Ersig, Jaja & Tluczek, 2023).

The significance of cost as a factor in decision-making may also indicate worries about the affordability of genetic testing and the ongoing management of genetic

conditions, especially if these services lack comprehensive coverage from healthcare systems or insurance providers. Previous studies have reported that parents favour financial assistance methods such as insurance coverage to alleviate the financial economic burden associated with genomic testing and follow-up care (Wang et al. 2024). Overall, these findings suggest that parental support for gNBS is influenced not only by the potential benefits of screening but also by perceptions of value, affordability and clinical relevance. Therefore, future implementation strategies should ensure that the purpose, scope and expected benefits of gNBS are clearly communicated to parents while addressing concerns regarding expenses and care accessibility.

5.2.6 Parental Ethical and Logistical Considerations

The findings of this study provide significant understanding of parental expectations for the ethical and practical application of gNBS in Malaysia. Most parents preferred that gNBS be subsidised by the government, indicating concerns regarding affordability and a belief that access to genomic screening should be equitable rather than dependent on an individual's financial capacity. This result aligns with the significance of cost identified in other sections in this study and emphasizes the necessity for policymakers to explore sustainable funding strategies that reduce financial barriers to access.

The majority of parents also supported the storage of genomic data within hospital systems for future clinical care and in research databases that may be shared with other researchers. These findings suggest that parents generally recognise the possible benefits of genomic data beyond initial screening such as its application for future health management and research. However, such acceptance also underscores the importance of establishing robust governance frameworks that address data privacy, security, transparency and appropriate data-sharing practices (Ersig, Jaja & Tluczek 2023). Public trust is likely to be essential for the successful implementation of gNBS, especially considering the sensitive and long-term nature of genomic information.

Parental opinions on the duration of genomic data storage varied significantly, with no single option receiving overwhelming support. This variation may indicate differing viewpoints on the balance between the possible future advantages of keeping

genomic data and concerns regarding privacy, confidentiality and long-term data management. The lack of a definitive agreement indicates that policies regarding genomic data retention should be developed carefully, taking into account public values and expectations.

A large majority of parents expressed interest in receiving updated findings from future reanalysis of stored genomic data. This finding indicates that parents view genomic information as dynamic rather than static and recognise the potential value of ongoing reinterpretation as scientific knowledge advances. The findings also underscore the necessity of addressing policies concerning recontact, result re-disclosure and long-term follow-up in developing future gNBS programmes.

Significantly, most parents agreed that gNBS should be managed differently from existing newborn screening programmes. This perception may reflect an awareness that genomic screening involves broader ethical, legal and social considerations than existing newborn screening, including issues related to informed consent, data governance, result interpretation, reanalysis and long-term follow-up. These findings suggest that successful implementation of gNBS will require not only technical and clinical readiness but also the development of comprehensive governance frameworks that ensure the programme remains ethically acceptable, transparent and responsive to parental expectations.

5.3 FACTORS INFLUENCING PARENTAL PERCEPTIONS AND WILLINGNESS TO SUPPORT GNBS

5.3.1 Sociodemographic Factors and Perception

Several sociodemographic factors were significantly associated with selected perceived benefits and risks of gNBS. Gender was significantly associated with perceiving gNBS as providing early and effective treatment. Female parents were more likely than male parents to identify this benefit. This finding may reflect the greater involvement of mothers in child healthcare and preventive health decision-making, which may increase their appreciation of the potential clinical value of early detection through genomic screening.

Education level was significantly associated with perceiving early and effective treatment as a benefit of gNBS. However, parents across all educational backgrounds generally recognised this benefit, suggesting a broad appreciation of the potential role of gNBS in improving health outcomes through earlier diagnosis and intervention. This suggests that support for the clinical utility of gNBS extends across diverse educational backgrounds.

In addition, education level was significantly associated with concerns regarding the misinterpretation of genetic results and the possibility of making incorrect decisions. Parents with higher educational levels may have greater awareness of the complexity of genomic information and the challenges associated with interpreting genetic findings. This finding highlights the importance of providing clear and accurate information, as well as access to appropriate genetic counselling, to support informed decision-making among parents considering gNBS.

5.3.2 Genetics-Related Perceptions Factors and Perception

This study found that parents who believed that prenatal testing could reduce the risk of having a child with a genetic condition were significantly more likely to perceive providing early and effective treatment and providing accurate and comprehensive information as benefits of gNBS. This finding suggests that parents with a greater appreciation of the role of genetic testing may be more likely to recognise the potential clinical and informational benefits of gNBS. Similar to prenatal genetic testing, gNBS offers genetic insights that can enable earlier interventions, enhance disease management and assist in making informed healthcare choices. Consequently, parents who recognize the importance of prenatal testing may also be more open to the advantages provided by genomic screening postnatally.

No significant associations were observed between the genetics-related perceptions factors examined and the selected perceived risks of gNBS. This finding suggests that concerns regarding the potential risks of gNBS may be shared across parents regardless of their level of genetic knowledge or awareness.

5.3.3 Sociodemographic Factors and Willingness to Support gNBS

Although a significant association was observed between household income and willingness to support gNBS, the proportion of parents supporting gNBS was relatively high across all income groups. This suggests that support for gNBS was generally favourable regardless of socioeconomic status.

However, previous studies have reported inconsistent findings regarding the relationship between demographic characteristics and interest in acceptance of gNBS. A review by Smits et al. (2026) found that associations between income and support for gNBS varied across studies, with some reporting significant relationships while others found no association. This may suggest that socioeconomic status alone may not fully explain parental willingness to support gNBS.

5.3.4 Genetics-Related Perceptions Factors and Willingness to Support gNBS

This study found significant associations between genetics-related perceptions factors and willingness to support gNBS. Parents who believed that modern medicine can treat genetic diseases and those who believed that prenatal testing can reduce the risk of having a child with a genetic condition were more likely to support the implementation of gNBS.

The association between perceived treatability of genetic conditions and support for gNBS aligns with earlier research indicating higher acceptance of genomic screening when participants acknowledged possible clinical advantages and available interventions (Bombard et al. 2014). Parents may see more value in genomic screening when the information obtained can lead to treatment, preventive measures or improved health outcomes. Likewise, parents who believed that prenatal testing can reduce genetic disease risk may value genetic information more in healthcare decision-making and disease management, resulting in more positive views of gNBS.

These findings are also supported by previous research demonstrating that positive attitudes towards genomic technologies are more common among individuals with higher genetic knowledge and comprehension (Prosenc et al. 2023). The findings

collectively indicate that knowledge and perceptions regarding the usefulness of genetic information may play an important role in shaping parental acceptance of gNBS.

5.3.5 Predictors of Willingness to Support gNBS

The logistic regression analysis identified genetic knowledge-related factors rather than sociodemographic characteristics as significant predictors of willingness to support gNBS. While household income was associated with willingness to support gNBS in the bivariate analysis, it was not retained as a significant predictor after adjustment for other variables. This finding suggests that parental support for gNBS may be influenced more by perceptions and understanding of genetic testing than by demographic characteristics alone.

Parents who believed that modern medicine can treat genetic diseases were significantly more likely to support the implementation of gNBS. This finding may reflect the importance of perceived clinical utility in shaping acceptance of genomic screening. Parents tend to have a more positive view of gNBS when they think that identifying genetic conditions can result in significant interventions, enhanced disease management, or improved health results. Bombard et al. (2014) have also reported similar findings, indicating that the perceived actionability of genomic information significantly influenced support for genomic screening programs.

5.4 IMPLICATIONS

The findings of this study have several implications for the future implementation of gNBS in Malaysia. First, the relatively low awareness of genetic testing in newborns and gNBS, despite generally positive parental acceptance, highlights the importance of strengthening genetic education and public awareness initiatives. Healthcare professionals involved in maternal, neonatal and paediatric care may play a key role in providing accurate and accessible information to support informed decision-making. The findings also emphasise the need for the development of appropriate educational and genetic counselling resources tailored to the needs of Malaysian parents (Genetic Alliance 2009).

From a policy perspective, the findings indicate that the successful implementation of gNBS will require robust ethical and governance frameworks. Parents demonstrated strong support for informed consent, appropriate management of genomic information and the return of updated findings from future reanalysis (Friedman 2024; Levy 2021). These expectations highlight the importance of establishing clear policies regarding consent processes, genomic data storage, privacy protection, data sharing and long-term follow-up. In addition, parental preference for government subsidisation suggests that considerations of affordability and equitable access should be incorporated into future funding strategies.

The findings further suggest that successful implementation of gNBS should be accompanied by ongoing and broader efforts to strengthen genomic literacy and stakeholder involvement in genomic medicine. As gNBS represents one application of genomic medicine, improving public understanding of genomic concepts through engagement with parents, healthcare professionals, policymakers and other stakeholders may facilitate informed decision-making and enhance public acceptance of current and future genomic healthcare initiatives. Given the complexity of genomic information and the ethical considerations surrounding genomic screening, such engagement is essential to ensure that implementation remains responsive to public expectations and societal values. Specifically for gNBS, pilot implementation initiatives could provide an opportunity to evaluate its feasibility, acceptability and operational requirements of gNBS prior to broader integration into the Malaysian healthcare system (Boemer & Servais 2025; Kariyawasam et al. 2024; Lunke et al. 2025; Schnabel-Besson et al. 2026; Smit et al. 2020).

5.5 LIMITATIONS

This study has several limitations that should be considered when interpreting the findings. First, the study used a convenience sampling method and was conducted at a single tertiary paediatric centre. Therefore, the findings may not be representative of all Malaysian parents, limiting the generalisability of the results to the wider population.

Second, although a range of sociodemographic, awareness and genetic knowledge factors were examined, not all potential determinants of parental perceptions and willingness to support gNBS were included in the analysis. Factors such as health literacy, previous experiences with genetic conditions, trust in healthcare systems, cultural beliefs and religious considerations were not assessed and may have influenced parental decision-making regarding gNBS.

Third, this study primarily focused on parental perceptions, attitudes and willingness to support the implementation of gNBS. Several implementation-related aspects explored in the questionnaire, including preferred healthcare personnel, communication methods, settings and timing for information delivery and consent, were not examined in depth. As a result, the study was unable to comprehensively evaluate how these factors may influence the acceptability and implementation of gNBS.

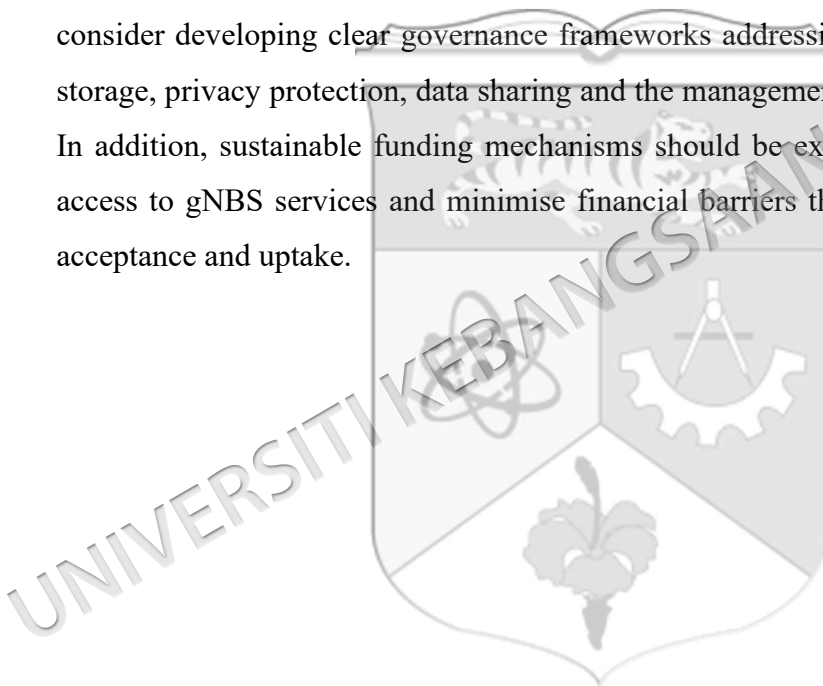
Finally, the cross-sectional design captured parental views at a single point in time and did not allow assessment of changes in perceptions, attitudes or willingness to support gNBS over time. Consequently, causal relationships between the factors examined and parental support for gNBS could not be established.

5.6 RECOMMENDATIONS

Based on the findings of this study, future research can be improved by involving larger and more diverse samples recruited from multiple healthcare settings and regions across Malaysia to improve the representativeness and generalisability of the findings. Additional factors that may influence parental perceptions and acceptance of gNBS such as health literacy, cultural and religious beliefs, previous experiences with genetic conditions and trust in healthcare systems should also be explored. Furthermore, qualitative studies may provide a deeper understanding of parental concerns, expectations and decision-making processes, while longitudinal studies could assess how perceptions and acceptance of gNBS evolve over time. Future study also may consider exploring the perspectives of healthcare providers regarding the implementation of gNBS within the Malaysian healthcare system (Abdullahi et al. 2026).

From a clinical perspective, the relatively low awareness of genetic testing in newborns and gNBS observed in this study highlights the need for educational initiatives to improve public understanding of genomic medicine. Educational materials and counselling resources should be developed to support parents in making informed decisions regarding gNBS. Healthcare professionals involved in maternal, neonatal and paediatric care should also be equipped with appropriate knowledge and communication skills to effectively discuss the benefits, limitations and implications of genomic screening with parents.

At the policy level, the generally positive parental acceptance of gNBS suggests potential readiness for future implementation in Malaysia. Policymakers should consider developing clear governance frameworks addressing informed consent, data storage, privacy protection, data sharing and the management of genomic information. In addition, sustainable funding mechanisms should be explored to ensure equitable access to gNBS services and minimise financial barriers that may influence parental acceptance and uptake.



CHAPTER VI

CONCLUSION

This study revealed that Malaysian parents generally demonstrated positive attitudes towards gNBS, with the majority expressing support for its implementation. Despite the limited awareness of genetic testing in newborns and gNBS was relatively low, parents recognised several potential advantages, especially in enabling early and effective treatment, facilitating timely genetic diagnosis, and providing accurate and comprehensive genetic information that may support clinical management and informed decision-making. Cost, treatment availability and the scope of conditions screened were among the main factors influencing decision-making. Parents also expressed strong support for informed consent, government subsidisation and receiving updates from future reanalysis of genomic data.

Several sociodemographic and genetic knowledge factors were associated with parental perceptions and willingness to support gNBS. In particular, beliefs regarding the treatability of genetic conditions and the role of prenatal testing were significantly associated with support for gNBS. Logistic regression analysis further identified perception that modern medicine can treat genetic diseases as the only independent predictor of willingness to support the implementation of gNBS.

Overall, this study demonstrated that Malaysian parents generally showed positive perceptions and acceptance towards gNBS despite limited prior awareness of the technology. The findings suggest that parents recognise the potential of gNBS to facilitate earlier diagnosis and intervention, improve health outcomes and quality of life, and support informed healthcare and reproductive decision-making. Beyond its potential contribution to reducing morbidity and mortality associated with genetic conditions, gNBS may also enhance awareness of genomic medicine, precision

medicine and rare diseases among the public. These findings provide valuable evidence to support future planning, policy development and the responsible implementation of gNBS within the Malaysian healthcare system.



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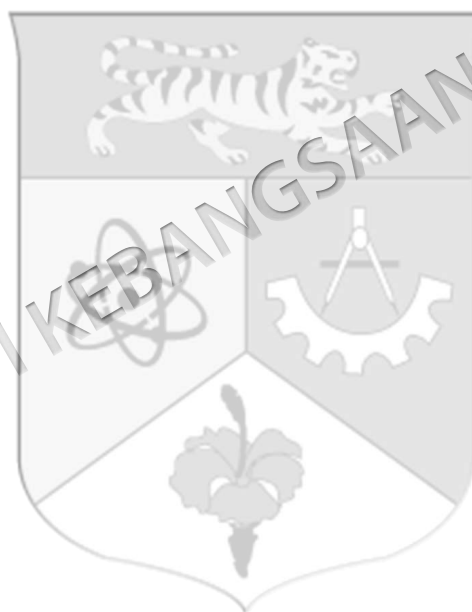
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APPENDIX A

ETHICS APPROVAL LETTER



SEKRETARIAT ETIKA PENYELIDIKAN UKM • UKM RESEARCH ETHICS SECRETARIAT

Reference: UKM PPI/111/8
Date: 17 April 2026

Associate Professor Dr. Shareena Binti Ishak
Department of Paediatrics
Faculty of Medicine UKM

Dear Professor/Datuk/Dato'/Datin/Sir/Madam,

ETHICAL APPROVAL TO CONDUCT RESEARCH IN THE NATIONAL UNIVERSITY OF MALAYSIA

Research title : *Persepsi Ibu Bapa Terhadap Saringan Genomik Bayi Baru-Lahir (GNBS) Di Sebuah Hospital Tertier Di Malaysia*

Ethics Ref. No. : JEP-2026-109

Approval period : 09 April 2026 – 08 April 2027

Study Site : Hospital Tunku Ampuan Besar Tuanku Aishah Rohani Hospital, Pakar Kanak-Kanak-Kanak (HPKK UKM)

Sample Size : 423 Subjects

With reference to the above:

2. The Research Ethics Committee (Health and Life Sciences), The National University of Malaysia has provided ethical approval for above study. **Please be reminded permission from the Deputy Dean of research of the faculty or Director of Institute/Centre and all relevant heads of departments / units where the study will be carried out must be obtained prior to the study.** You are required to follow and comply with their decision and all other relevant regulations.

3. Please note that the investigator's responsibility is to ensure that:

- i. All Adverse Events should be reported to REC (Health and Life Sciences) UKM as soon as possible.
- ii. Progress report should be submitted every 6 Months.
- iii. All changes / amendments to the study documents / study sites / study team must be notified to the RECUKM. All changes must be approved by REC (Health and Life Sciences) UKM before continuation of the study.
- iv. Application for renewal of the approval has to be submitted to REC (Health and Life Sciences) UKM within one month (1 month) prior to the expiry of ethical approval.
- v. Final Report should be provided to REC (Health and Life Sciences) UKM when the project is complete.
- vi. Please take note that all records and data are to be kept strictly CONFIDENTIAL and can only be used for the purpose of this study. All precautions are to be taken to maintain data confidentiality.

Required forms can be obtained from the The Research Ethics Committee (Health and Life Sciences), The National University of Malaysia website: <https://www.ukm.my/jepukm>

Thank you.

Sekretariat Etika Penyelidikan Universiti Kebangsaan Malaysia
Tingkat 1, Blok Klinik, Hospital Canselor Tuanku Muhriz, Pusat Perubatan UKM, Jalan Yacob Latif, Bandar Tun Razak, 68000 Cheras Kuala Lumpur.
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Email: gapukm@ukm.edu.my Web: <https://www.ukm.my/jepukm/>
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SEKRETARIAT ETIKA PENYELIDIKAN UKM • UKM RESEARCH ETHICS SECRETARIAT

Yours sincerely,

PROFESOR DR MOHD SHAHRIR MOHAMED SAID
Chairman
Research Ethics Committee (Health and Life Sciences)
The National University Of Malaysia

- C.C. - **Director**
Hospital Canselor Tuanku Muhriz
- **Director**
Hospital Tunku Ampuan Besar Tuanku Aishah Rohani Hospital,
Pakar Kanak-Kanak-Kanak (HPKK UKM)
- **Deputy Dean (Research & Innovation)**
Faculty of Medicine, UKM
- **Head Department of Paediatrics**
Faculty of Medicine UKM
- **Dr. Wan Nurulhuda Binti Wan Md Zin**
Department of Paediatrics
Hospital Canselor Tuanku Muhriz
- **Dr. Shyahlini Devi A/P Rajendran**
Department of Paediatrics
- **Mrs. Sharifah Azween Binti Syed Omar**
Dr. Chin Pei Voon (P148737 – Master Candidate)
Department of Paediatrics
Faculty of Medicine UKM
- Approval letter File 2026

Sekretariat Etika Penyelidikan Universiti Kebangsaan Malaysia
Tingkat 1, Blok Klinik, Hospital Canselor Tuanku Muhriz, Pusat Perubatan UKM, Jalan Yaacob Latif, Bender Tun Razak, 58000 Cheras Kuala Lumpur.
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SEKRETARIAT ETIKA PENYELIDIKAN UKM • UKM RESEARCH ETHICS SECRETARIAT

NAME OF ETHICS COMMITTEE/IRB: Research Ethics Committee, The National University of Malaysia	ETHICS COMMITTEE/IRB REF NO : JEP-2026-109
PROTOCOL TITLE: <i>Persepsi Ibu Bapa Terhadap Saringan Genomik Bayi Baru Lahir (GNBS) Di Sebuah Hospital Tertier Di Malaysia</i>	
PRINCIPAL INVESTIGATOR: Associate Professor Dr. Shareena Binti Ishak Department of Paediatrics Faculty of Medicine UKM	

The following items ☒ have been received and reviewed in connection with the above study to be conducted by the above investigator.

Documents

- ☒ Research Application Form (UKM-JEP-BO01)
☒ Review Form (UKM-JEP-BO02)
☒ Non-Disclosure Agreement (UKM-JEP-BO03)
☒ Conflict Of Interest Form
☒ Research Proposal / Protocol
☒ Participant Information Sheet:
☒ Malay ☒ English ☐ Others
☒ Participant Consent Form:
☒ Malay ☒ English ☐ Others
☒ Questionnaire:
☒ Malay ☒ English ☐ Others
☒ Curriculum Vitae of Researchers:
☒ Principal ☒ Co-researcher ☒ Student
☒ Good Clinical Practice Certificate (GCP)
☐ Project Agreement

The Research Ethics Committee (Health and Life Sciences), The National University of Malaysia operates in accordance to the International Conference of Harmonization Good Clinical Practice Guidelines.

Comments (if any):

Date of Approval: 09 April 2026


PROFESSOR DR. MOHD SHAHRIR MOHAMED SAID
 Chairman
 Research Ethics Committee (Health and Life Sciences)
 The National University of Malaysia

APPENDIX B

RESEARCH APPROVAL LETTER



FAKULTI PERUBATAN • FACULTY OF MEDICINE

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

Date : 12 May 2026

Associate Professor Dr Shareena Ishak
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 (HTABTAR)

Title Parental Perception of Genomic Newborn Screening (gNBS) in a Malaysian Tertiary Paediatric Setting

Project Code FF-2026-159

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	09 April 2026 – 08 April 2027
Financial Support	RM 1,190.00 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/spifperforms/>) for more information and guidelines.

Please register the research at the National Medical Research Register (www.nmr.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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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 DATO' DR MARINA MAT BAKI
 Dean
 Faculty of Medicine, UKM


PROFESSOR DR HAMZAINI ABDUL HAMID
 Director
 Hospital Tunku Ampuan Besar Tuanku Aishah Rohani
 Hospital Pakar Kanak-Kanak (HTABTAR)

- s.k
- **Professor Dr Syahrul Sazliyana Shaharir**
 Deputy Dean (Research & Innovation)
 Faculty of Medicine, UKM
 - **Chairman Research Ethics Committee UKM**
 Secretariat of Research Ethics UKM
 UKM Medical Centre
 - **Mr Abdul Muiz Sheafi**
 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 Wan Nurulhuda Wan Muz Zin**
 Department of Paediatrics
 Hospital Gengseng Tuanku Muhriz (HCTM), UKM
 - **Dr Shyahlini Devi a/p Rajendran**
 Department of Paediatrics
 Hospital Tunku Ampuan Besar Tuanku Aishah Rohani
 Hospital Pakar Kanak-Kanak (HTABTAR), UKM
 - **Mrs Sharifah Azween Syed Omar**
 Dr Chin Pei Voon
 Department of Paediatrics
 Faculty of Medicine, UKM
 - **FF-2026-159 File**

MBR/FF-2026-159



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Appendix 1

TERMS OF PUBLICATION OF FUNDAMENTAL GRANTS OF THE FACULTY OF MEDICINE

FUND CATEGORY	PUBLISHING KPIs
RM 25,000.00 - RM 30,000.00	Researchers must publish at least: • one (1) article in the WOS Indexed Journal (Q1-Q2)
RM 15,000.00 - RM 24,999.99	Researchers must publish at least: • one (1) article in the WOS Indexed Journal (Q3-Q4)
RM5,000.00 - RM 14,999.99	Researchers must publish at least : • one (1) article in an Indexed Journal (at least ESCI or SCOPUS)
RM100.00 - RM4,999.99	Researchers must publish at least: • one (1) article in a Journal Not Indexed OR • two (2) other publications
*Other publications are such as case studies, technical reports, comprehensive reports, magazine articles, newspapers, original works, publications from conferences, digital or print media. * Each publication should place GFFP as an acknowledgement in the first position *Publications must be registered in the e-repository system within 6 months after the end date of the research project.	



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Appendix 2

DETAILED LIST OF APPROVED EXPENSES

Title : Parental Perception of Genomic Newborn Screening (gNBS) in a Malaysian Tertiary Paediatric Setting

Project Code : FF-2026-159

NO.	ITEM	NOTES	VOTE	TOTAL APPROVED (RM)
1.	Travel and Transportation: - Transportation expenses		21000	350.00
2.	Specialized Services: - Translation process fee		29000	840.00
TOTAL BUDGET APPROVED :				RM 1,190.00



Endorsed by

Profesor Dr Syahrul Sazliyana Shaharir
Deputy Dean (Research & Innovation)
Faculty of Medicine UKM
Date : 27 April 2026



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APPENDIX C

PARENT INFORMATION SHEET

PARENT INFORMATION SHEET

Research Title:

Tajuk Kajian:

Parental Perception of Genomic Newborn Screening (gNBS) in a Malaysian Tertiary Paediatric Setting

Persepsi Ibu Bapa terhadap Saringan Genomik Bayi Baru Lahir (gNBS) di Sebuah Hospital Tertier di Malaysia

Dear Parents,

Ibu Bapa yang dihormati,

You are invited to participate in a research study. Before agreeing to take part, please take some time to read and understand the information provided in this information sheet.

Anda dijemput untuk mengambil bahagian dalam kajian penyelidikan ini. Sebelum bersetuju untuk mengambil bahagian, sila luangkan masa untuk membaca dan memahami maklumat yang disediakan dalam lembaran ini.

Introduction:

Pengenalan:

Newborn screening is a health test done for babies soon after birth to detect certain serious but treatable medical conditions before symptoms appear. Early detection allows treatment to be started early and can improve a child's health and development.

Saringan bayi baru lahir ialah satu ujian kesihatan yang dilakukan ke atas bayi sejurus selepas kelahiran untuk mengesan beberapa penyakit serius tetapi boleh dirawat sebelum gejala muncul. Pengesanan awal membolehkan rawatan dimulakan lebih awal dan dapat meningkatkan kesihatan serta perkembangan kanak-kanak.

With advances in medical technology, newborn screening has expanded over the years to include more conditions. Recently, genomic technology has made it possible to screen for some genetic conditions by analysing selected genes. This is known as genomic newborn screening (gNBS). This type of screening may help identify genetic conditions that could affect a child's health during childhood and provide information about future health risks.

Seiring dengan kemajuan teknologi perubatan, saringan bayi baru lahir telah diperluaskan dari masa ke semasa untuk merangkumi lebih banyak penyakit. Kini, teknologi genomik membolehkan saringan bagi beberapa keadaan genetik melalui analisis gen terpilih. Ini

dikenali sebagai saringan genomik bayi baru lahir (gNBS). Saringan ini berpotensi mengenal pasti penyakit genetik yang boleh menjejaskan kesihatan kanak-kanak semasa kecil dan juga memberi maklumat tentang risiko kesihatan pada masa hadapan.

Several countries have started to study and test genomic newborn screening in research projects. However, in Malaysia, this approach is still new and there is limited local information about parents' views, understanding and acceptance of this type of screening.

Beberapa negara telah mula mengkaji dan menguji saringan genomik bayi baru lahir melalui projek penyelidikan. Walau bagaimanapun, di Malaysia, pendekatan ini masih baru dan maklumat tempatan mengenai pandangan, kefahaman dan penerimaan ibu bapa terhadap jenis saringan ini adalah terhad.

Therefore, this study aims to understand parents' opinions and perspectives on genomic newborn screening to help to guide future research and planning in Malaysia.

Oleh itu, kajian ini bertujuan untuk memahami pendapat dan perspektif ibu bapa mengenai saringan genomik bayi baru lahir bagi membantu perancangan dan penyelidikan pada masa hadapan di Malaysia.

Purpose of Study:

Tujuan Kajian:

The aim of this study is to assess parents' perspectives, attitudes and acceptance of genomic newborn screening (gNBS) among parents at Hospital Tunku Ampuan Besar Tuanku Aishah Rohani, UKM Specialist Children's Hospital (Hospital TABTAR). In addition, this study aims to identify the factors that influence parents' perceptions and their willingness to support the implementation of genomic newborn screening (gNBS) within the healthcare system.

Kajian ini bertujuan untuk menilai pandangan, sikap dan penerimaan ibu bapa terhadap saringan genomik bayi baru lahir (gNBS) di Hospital Tunku Ampuan Besar Tuanku Aishah Rohani, Hospital Pakar Kanak-Kanak UKM (Hospital TABTAR). Selain itu, kajian ini juga bertujuan untuk mengenal pasti faktor-faktor yang mempengaruhi persepsi ibu bapa dan kesediaan mereka untuk menyokong pelaksanaan gNBS dalam sistem penjagaan kesihatan.

What will the study involve?

Apakah yang akan terlibat di dalam kajian ini?

You will be asked to sign a consent form indicating your voluntary agreement to participate in this study. Some personal information will be required when completing the consent form. Once written consent is obtained, you will be included in the study.

Anda akan diminta untuk menandatangani borang persetujuan yang menunjukkan persetujuan sukarela anda untuk mengambil bahagian dalam kajian ini. Beberapa maklumat peribadi akan diperlukan semasa melengkapkan borang persetujuan tersebut. Setelah persetujuan bertulis diperoleh, anda akan dimasukkan sebagai peserta dalam kajian ini.

You will be given a pamphlet to read and understand before proceeding to the questionnaire, which can be completed either in paper form or online via Google Forms based on your preference. After you have completed the questionnaire, the data will be analysed and no further information will be required from you.

Anda akan diberikan risalah untuk dibaca dan difahami sebelum meneruskan kepada soal selidik, yang boleh dilengkapkan sama ada dalam bentuk bercetak atau secara dalam talian melalui Google Forms, bergantung kepada pilihan anda. Setelah anda melengkapkan soal selidik tersebut, data akan dianalisis dan tiada maklumat lanjut akan diperlukan daripada anda.

Benefits:

Faedah:

The data collected from your responses will be analysed and the findings from this study are expected to provide valuable insights that may inform future research, guide implementation strategies, support educational planning and contribute to the development of national policies related to genomic newborn screening (gNBS) in Malaysia.

Data yang dikumpul daripada maklum balas anda akan dianalisis dan hasil kajian ini dijangka dapat memberikan pandangan berharga untuk penyelidikan pada masa hadapan, membantu perancangan pelaksanaan, menyokong perancangan pendidikan, serta menyumbang kepada pembangunan dasar kebangsaan berkaitan dengan saringan genomik bayi baru lahir (gNBS) di Malaysia.

Risks:

Risiko:

There is no risk identified in this study. *Tiada risiko yang dikenal pasti dalam kajian ini.*

Do you have to take part?

Adakah anda perlu menyertai kajian ini?

Participation in this study is entirely voluntary. You have the right to refuse to participate. If you decide to take part, you may still withdraw from the study at any time without any penalty or loss of benefits. If you withdraw, your data will not be used and will be discarded.

Penyertaan dalam kajian ini adalah secara sukarela. Anda berhak untuk menolak penyertaan. Jika anda bersetuju untuk mengambil bahagian, anda masih boleh menarik diri daripada kajian ini pada bila-bila masa tanpa sebarang penalti atau kehilangan sebarang faedah. Jika anda menarik diri, data anda tidak akan digunakan dan akan dihapuskan.

The researcher may also withdraw you from the study if necessary. In such an event, you will not be penalised. Your decision to participate or not will not affect the care or services provided to you or your family members.

Penyelidik juga boleh menarik diri anda daripada kajian ini jika perlu. Dalam keadaan sedemikian, anda tidak akan dikenakan sebarang penalti. Keputusan anda untuk mengambil bahagian atau tidak mengambil bahagian dalam kajian ini tidak akan menjejaskan penjagaan atau perkhidmatan yang diberikan kepada anda atau ahli keluarga anda.

Data & Confidentiality:

Data dan Kerahsiaan:

All information obtained in this study will be kept strictly confidential. The data collected will be analysed and presented in the form of a report and may be published. All results will be reported in an aggregated manner, with no information that can identify individual participants. Your name or any personal identifiers will not appear in any report or publication. Access to the data will be limited to the research team only.

Semua maklumat yang diperolehi dalam kajian ini akan dirahsiakan. Data yang dikumpulkan akan dianalisis dan dibentangkan dalam bentuk laporan dan boleh diterbitkan. Semua data akan dilaporkan secara kolektif tanpa merujuk kepada individu. Nama atau sebarang maklumat pengenalan diri anda tidak akan muncul dalam sebarang laporan atau penerbitan. Akses kepada data adalah terhad kepada pasukan penyelidik sahaja.

Payment and compensation:

Bayaran dan Pampasan:

There is no payment or compensation for participating in this study. You will not be charged any additional fees for participating. However, you will still be required to pay the usual hospital charges for your medical care, as per standard practice.

Tiada bayaran atau pampasan akan diberikan untuk penyertaan dalam kajian ini. Anda juga tidak akan dikenakan sebarang bayaran tambahan untuk menyertai kajian ini. Walau bagaimanapun, anda masih perlu membayar caj hospital seperti biasa untuk rawatan perubatan anda, mengikut amalan standard.

Who can I ask about the study?

Siapakah yang boleh dihubungi jika saya mempunyai pertanyaan?

If you have any questions or require further information about this study, please contact:

Sekiranya anda mempunyai sebarang soalan atau memerlukan maklumat lanjut mengenai kajian ini, sila hubungi:

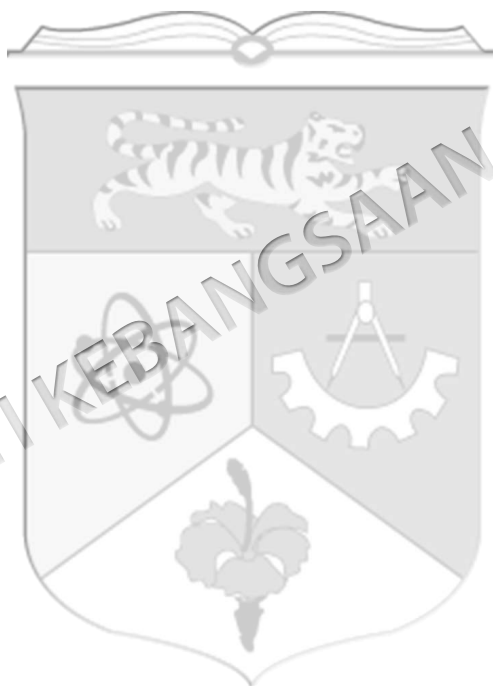
Chin Pei Voon

Genetic Counselling Programme *Program Kaunseling Genetik*

Faculty of Medicine, UKM *Fakulti Perubatan, UKM*

Email Emel: p148737@siswa.ukm.edu.my

Phone Number Nombor telefon: 011-10875984



APPENDIX D

INFORMED CONSENT FORM

INFORMED CONSENT FORM BORANG PERSETUJUAN MAKLUM

Research Title/Tajuk Penyelidikan:

Parental Perception of Genomic Newborn Screening (gNBS) in a Malaysian Tertiary Hospital
Persepsi Ibu Bapa Terhadap Saringan Genomik Bayi Baru Lahir (gNBS) Di Hospital Tertier Di Malaysia

I/Saya
IC Number/Nombor KP:

- have read the information in the Parent Information Sheet
telah membaca maklumat dalam Lembaran Maklumat Ibu Bapa
- have been given sufficient time to consider participation and all of my questions have been answered to my satisfaction
telah diberikan masa yang mencukupi untuk berfikir mengenainya dan semua soalan saya telah dijawab dengan memuaskan
- understand that I may freely choose to withdraw from this study at any time without giving any reason and without any repercussions
memahami bahawa saya bebas untuk menarik diri daripada kajian ini pada bila-bila masa tanpa perlu memberikan sebarang sebab dan tanpa sebarang akibat
- understand that my anonymity will be ensured in the write-up.
memahami bahawa identiti saya akan dirahsiakan dalam penulisan kajian.

I voluntarily agree to take part in this research study, to follow the study procedures and to provide the necessary information to the doctor, nurses, or other staff members, as requested.

Saya dengan sukarela bersetuju untuk mengambil bahagian dalam kajian penyelidikan ini, mematuhi prosedur kajian dan memberikan maklumat yang diperlukan kepada doktor, jururawat, atau kakitangan lain seperti yang diminta.

Signature Tandatangan:

Date/Tarikh:

Name of Witness (if any)

Nama Saksi (sekiranya ada)

Name of Researcher

Nama Penyelidik

Signature Tandatangan

Signature Tandatangan

IC Number Nombor IC

IC Number Nombor IC

Date Tarikh

Date Tarikh

APPENDIX E

INTRODUCTORY PAMPHLET

How many types of newborn screening are there?

There are **THREE** types of newborn screening: Standard Newborn Screening, Expanded Newborn Screening (ENS) and Genomic Newborn Screening (gNBS).

**Standard Newborn Screening**

- Tests for a small number of serious conditions
- Uses a few drops of blood from your baby's heel or umbilical cord
- Performed in the first few days after birth, or at birth using umbilical cord blood
- Part of routine care in most hospitals

E.g., G6PD Deficiency, Cord TSH

Purpose:

To identify serious conditions requiring immediate treatment to prevent disability or life-threatening complications.

UNDERSTANDING NEWBORN SCREENING

A Parent's Guide

What is NEWBORN SCREENING?

Newborn screening is a simple health test performed shortly after your baby is born. It checks for certain health conditions that may not be visible at birth but can affect your child's development and health. Early detection means early treatment, giving your baby the best possible start in life.

Expanded Newborn Screening (ENS)

- Tests for more conditions than standard screening
- Uses a blood sample from your baby's heel
- Focuses on conditions that benefit from early intervention

E.g., Maple Syrup Urine Disease (MSUD), Severe combined immunodeficiency (SCID), Homocystinuria

Important points to know:

- Not all conditions can be detected through screening
- An abnormal result does not always mean your baby has the condition
- Confirmatory test is usually needed for abnormal results
- Your healthcare provider will guide you the next steps



Genomic Newborn Screening (gNBS)

- Analyses selected genes in your baby's DNA
- Uses your baby's blood or saliva
- Screens for genetic conditions that may appear during childhood
- Provides information about future health risks

Important points to know:

- Some results may be uncertain or difficult to interpret
 - Not all detected conditions have available treatment
 - Results require explanation from genetic specialists
 - May reveal information about future health conditions
- Genetic counselling is recommended to understand results

What Makes Genomic Screening Different?

Unlike standard and expanded screening that look for active health problems in your newborn, genomic screening examines your baby's genetic code. This can identify risks for conditions that might not appear until later in childhood or even adulthood.

Benefits: Early awareness allows for preventive care, lifestyle adjustments, and prompt treatment if symptoms develop.

Considerations: Results may reveal information you weren't expecting, including conditions without current treatments. Genetic counselling helps families understand and prepare for these possibilities.



Comparison of Screening Types

Item	Standard Screening	Expanded Screening (ENS)	Genomic Screening (gNBS)
When it is performed?	After birth	After birth	After birth
Sample	Blood	Blood	Blood or Saliva
Number of conditions tested	Few	More	Selected genetic conditions
Uses DNA testing	No	No	Yes
Types of conditions detected	Conditions included in standard program	Additional conditions that may benefit from early treatment	Genetic conditions that may appear in childhood
Results	Normal or abnormal	Normal or abnormal	Positive, negative, uncertain
Follow-up tests needed	Sometimes	Sometimes	Often
Genetic counselling	Not usually	Sometimes	Yes

MEMAHAMI SARINGAN BAYI BARU LAHIR

Panduan untuk Ibu Bapa



Apakah itu SARINGAN BAYI BARU LAHIR?

Saringan bayi baru lahir ialah ujian kesihatan ringkas yang dilakukan seurus selepas bayi dilahirkan. Ia mengesan masalah kesihatan tertentu yang mungkin tidak kelihatan semasa lahir tetapi boleh menjejaskan perkembangan dan kesihatan anak. Pengesanan awal membolehkan rawatan awal diberikan, dan bayi anda dapat permulaan hidup yang terbaik.

Ada berapa jenis saringan bayi baru lahir?

Terdapat TIGA jenis saringan bayi baru lahir: Saringan Bayi Baru Lahir Biasa, Saringan Bayi Baru Lahir Terluas (ENS), dan Saringan Genomik Bayi Baru Lahir (gNBS).



Saringan Bayi Baru Lahir Biasa

- Menguji sebilangan kecil penyakit yang serius
- Menggunakan beberapa titis darah dari tumit atau tali pusat
- Dilakukan dalam beberapa hari pertama selepas kelahiran, atau semasa kelahiran menggunakan darah tali pusat
- Merupakan sebahagian daripada penjagaan rutin di kebanyakan hospital

Contoh: Kekurangan G6PD, Kord TSH

Tujuan:

Untuk mengenal pasti penyakit serius yang memerlukan rawatan segera bagi mengelakkan hilang upaya atau komplikasi yang mengancam nyawa.

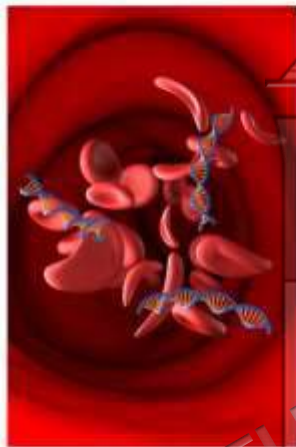
Saringan Bayi Baru Lahir Terluas (ENS)

- Menguji lebih banyak penyakit berbanding saringan biasa
- Menggunakan sampel darah daripada tumit bayi
- Memberi tumpuan kepada penyakit yang mendapat manfaat sekiranya dirawat awal

Contoh: Penyakit urin sirap maple (MSUD), Keimmunokurangan bercampur teruk (SCID), Homosistinuria

Perkara penting untuk diketahui:

- Tidak semua penyakit boleh dikesan melalui saringan
- Keputusan yang tidak normal tidak semestinya bermaksud bayi anda menghadapi penyakit tersebut
- Ujian pengesahan biasanya diperlukan jika keputusan tidak normal
- Kakitangan kesihatan membimbing langkah yang seterusnya



Saringan Genomik Bayi Baru Lahir (gNBS)

- Menganalisa gen-gen terpilih dalam DNA bayi
- Menggunakan sampel darah atau air liur bayi
- Menyaring penyakit genetik yang mungkin muncul semasa zaman kanak-kanak
- Memberikan maklumat tentang risiko kesihatan pada masa hadapan

Perkara penting untuk diketahui:

- Sesetengah keputusan mungkin tidak pasti atau sukar ditafsirkan
- Tidak semua penyakit yang dikesan mempunyai rawatan
- Keputusan memerlukan penjelasan daripada pakar genetik
- Mungkin mendedahkan maklumat tentang risiko kesihatan pada masa hadapan
- Kaunseling genetik disarankan untuk membantu memahami keputusan

Apa yang Membuatkan Saringan Genomik Berbeza?

Tidak seperti saringan biasa dan terluas yang mengesan masalah kesihatan yang sedang berlaku pada bayi, saringan genomik mengkaji kod genetik bayi. Ia boleh mengenal pasti risiko penyakit yang mungkin hanya akan dihadapi ketika zaman kanak-kanak atau dewasa.

Kebaikan: Kesedaran awal membolehkan langkah pencegahan, perubahan gaya hidup dan rawatan awal jika gejala muncul.

Pertimbangan: Keputusan mungkin mendedahkan maklumat yang tidak dijangka, termasuk penyakit yang belum mempunyai rawatan. Kaunseling genetik membantu keluarga memahami dan membuat persediaan terhadap kemungkinan ini.



Perbandingan Jenis Saringan

Perkara	Saringan Biasa	Saringan Terluas (ENS)	Saringan Genomik (gnBS)
Bila ujian dijalankan	Selepas kelahiran	Selepas kelahiran	Selepas kelahiran
Sampel	Darah	Darah	Darah atau air liur
Bilangan penyakit yang diuji	Sedikit	Banyak	Keadaan genetik tertentu
Penggunaan ujian DNA	Tidak	Tidak	Ya
Jenis penyakit yang dikesan	Keadaan yang termasuk dalam program biasa	Keadaan tambahan yang mungkin mendapat manfaat daripada rawatan awal	Keadaan genetik yang mungkin muncul pada zaman kanak-kanak
Keputusan yang diperolehi	Normal atau tidak normal	Normal atau tidak normal	Positif, negatif, tidak pasti
Ujian susulan diperlukan	Kadang-kadang	Kadang-kadang	Selalunya
Kaunseling genetik diperlukan	Tidak selalunya	Kadang-kadang	Ya

APPENDIX F

QUESTIONNAIRE

QUESTIONNAIRE SOAL SELIDIK

General instruction Arahan umum

1. Please read the pamphlet before answering the following questions.
Sila baca risalah terlebih dahulu sebelum menjawab soalan berikut.
2. Please answer each question based on your own views and select only one answer unless stated in the instructions.
Sila jawab setiap soalan berdasarkan pandangan anda dan pilih satu jawapan sahaja melainkan dinyatakan dalam arahan.

Section A: Demographic Bahagian A: Demografi

This section collects your basic information to help us understand the background of participants taking part in this study.

Bahagian ini mengumpul maklumat asas anda untuk membantu kami memahami latar belakang peserta yang mengambil bahagian dalam kajian ini.

1. Age: _____ years old
Umur: _____ tahun

2. Gender *Jantina*
- ☐ Male *Lelaki*
- ☐ Female *Perempuan*

3. Ethnicity *Etnik*
- ☐ Malay *Melayu*
- ☐ Chinese *Cina*
- ☐ Indian *India*
- ☐ Other *Lain-lain*: _____

4. Religion *Agama*

- ☐ Islam *Islam*
☐ Buddhism *Buddha*
☐ Christianity *Kristian*
☐ Hinduism *Hindu*
☐ Other *Lain-lain*: _____

5. Highest Education Level *Tahap Pendidikan Tertinggi*

- ☐ No formal education
Tiada pendidikan normal
☐ Primary *Rendah*
☐ Secondary (SPM / O-Level or equivalent)
Menengah (SPM / O-Level atau setara)
☐ Tertiary (Certificate / Diploma / Degree or equivalent)
Pengajian Tinggi (Sijil / Diploma / Ijazah atau setara)

6. Occupation *Pekerjaan*

- ☐ Private sector employee *Pekerja sektor swasta*
☐ Government employee *Pekerja kerajaan*
☐ Self-employed *Beperja sendiri*
☐ Unemployed *Tidak bekerja*
☐ Student *Pelajar*
☐ Other *Lain-lain*: _____

7. Monthly Household Income *Pendapatan Isi Rumah Bulanan*

- ☐ Below RM5250
Bawah RM5250
☐ RM5250 - RM11819
RM5250 - RM11819
☐ RM11820 and above
RM11820 dan ke atas

8. Marital status *Status Perkahwinan*

- ☐ Single *Bujang*
☐ Married *Berkahwin*
☐ Divorced *Bercerai*
☐ Widowed *Balu / duda*
☐ Separated *Berpisah*

9. Number of Children *Bilangan Anak*

- ☐ 1
☐ 2
☐ 3
☐ 4 or more *4 atau lebih*

10. Do you have any chronic illness or long-term medical condition (e.g. diabetes, hypertension, heart disease, cancer, etc.)?

Adakah anda mempunyai sebarang penyakit kronik atau keadaan perubatan jangka panjang (contoh: diabetes, hipertensi, penyakit jantung, kanser dan lain-lain)?

- ☐ Yes *Ya*
☐ No *Tidak*

11. Have you, your children, or any relatives been diagnosed with a genetic disorder (e.g. Down syndrome, thalassemia, muscular dystrophy, etc.)?

Adakah anda, anak-anak anda, atau mana-mana ahli keluarga pernah didiagnosis (dengan penyakit genetik (Contoh: Sindrom Down, talasemia, distrofi otot dan lain-lain)?

- ☐ Yes *Ya*
☐ No *Tidak*

12. Have you had any prior experience with genetic testing (e.g. newborn screening, preconception genetic testing, or prenatal testing)?

Adakah anda pernah mempunyai pengalaman dengan ujian genetik (contohnya saringan bayi baru lahir, ujian genetik pra-konsepsi, atau ujian pranatal)?

- ☐ Yes *Ya*
☐ No *Tidak*

13. Have you or your children ever visited a genetic clinic or had follow-up appointments at a genetic clinic?

Adakah anda atau anak-anak anda pernah mengunjungi klinik genetik atau menjalani temujanji susulan di klinik genetik?

- ☐ Yes *Ya*
☐ No *Tidak*

Section B: Perception of Genetic Disease and Genetic Testing

Bahagian B: Tanggapan terhadap Penyakit Genetik dan Ujian Genetik

This section explores participants' perceptions and awareness of genetic diseases and genetic testing, including perceived risk, beliefs about treatment, and awareness of prenatal and postnatal genetic diagnostic procedures.

Bahagian ini meneroka tanggapan dan tahap kesedaran peserta terhadap penyakit genetik dan ujian genetik, termasuk risiko anggapan, kepercayaan terhadap rawatan, dan kesedaran mengenai prosedur diagnostik genetik pratinatal dan postnatal.

Prenatal: Events or conditions affecting the mother or fetus from the beginning of pregnancy until the baby is born.

Prenatal: *Peristiwa atau keadaan yang berlaku kepada ibu atau fetus dari permulaan kehamilan sehingga bayi dilahirkan.*

Postnatal: Events or conditions affecting the mother or baby after birth.

Postnatal: *Peristiwa atau keadaan yang berlaku kepada ibu atau bayi selepas kelahiran.*

1. How likely is it for a child to be born with a genetic condition?

Apakah kebarangkalian seorang kanak dilahirkan dengan penyakit genetik?

- ☐ Very small (less than one affected child per million births)
Sangat kecil (kurang daripada satu bayi terjejas bagi setiap sejuta kelahiran)
- ☐ Small (one affected child per 10,000 - 100,000 births)
Kecil (satu bayi terjejas bagi setiap 10,000 – 100,000 kelahiran)
- ☐ Moderate (one affected child per 100 - 1,000 births)
Sederhana (satu bayi terjejas bagi setiap 100 – 1,000 kelahiran)

2. Most genetic diseases have been identified within the past 30 years. Do you think modern medicine can treat genetic diseases?

Kebanyakan penyakit genetik telah dikenal pasti dalam tempoh 30 tahun yang lalu. Adakah anda rasa perubatan moden boleh merawat penyakit genetik?

- ☐ Yes Ya
☐ No Tidak
☐ Not sure Tidak pasti

3. Genetic diagnostics are important for identifying genetic diseases. Do you think prenatal genetic diagnostic procedures can help reduce the risk of having a child with a genetic condition?

Diagnostik genetik adalah penting untuk mengenal pasti penyakit genetik. Adakah anda rasa prosedur diagnostik genetik pranal boleh membantu mengurangkan risiko mendapat anak berpenyakit genetik?

- ☐ Yes Ya
☐ No Tidak
☐ Not sure Tidak pasti

4. Do you know of genetic tests that are performed in the newborn period that can identify diseases that manifest later in life (e.g., metabolic disease, epilepsy, etc.)?

Adakah anda mengetahui tentang ujian genetik bayi baru lahir yang dapat mengesan penyakit yang hanya muncul kemudian dalam kehidupan (contohnya, penyakit metabolik, epilepsi dan lain-lain)?

- ☐ Yes Ya
☐ No Tidak
☐ Not sure Tidak pasti

Section C: Parental Perspectives on Genomic Newborn Screening (gNBS)

Bahagian C: Perspektif Ibu Bapa terhadap Saringan Genomik Bayi Baru Lahir (gNBS)

Genomic newborn screening (gNBS) is a test using genomic sequencing to identify DNA changes and detect medical conditions in newborns who are asymptomatic.

Saringan genomik bayi baru lahir (gNBS) ialah satu ujian yang menggunakan penjujukan genom untuk mengenal pasti perubahan DNA dan mengesan keadaan perubahan pada bayi baru lahir yang tidak mempunyai sebarang gejala.

This section explores parents'/guardians' attitudes, perceived benefits and risks, and overall perceptions of gNBS. The information will help understand parents'/guardians' perspectives and guide strategies to communicate the importance and implications of gNBS.

Bahagian ini meneroka sikap, anggapan ibu bapa / penjaga terhadap manfaat dan risiko, serta tanggapan secara menyeluruh terhadap gNBS. Maklumat ini akan membantu memahami perspektif ibu bapa / penjaga dan memberi panduan tentang strategi komunikasi berkenaan kepentingan dan implikasi gNBS.

1. Before reading the pamphlet, have you heard of newborn screening programmes before?

Sebelum membaca risalah ini, pernahkah anda mendengar tentang program saringan bayi baru lahir sebelum ini?

- ☐ Yes Ya
☐ No Tidak
☐ Not sure Tidak pasti

2. Before reading the pamphlet, have you heard of genomic newborn screening (gNBS) before?

Sebelum membaca risalah ini, pernahkah anda mendengar tentang saringan genomik bayi baru lahir (gNBS)?

- ☐ Yes Ya
☐ No Tidak
☐ Not sure Tidak pasti

3. Based on your current understanding from the pamphlet provided, do you think that genomic newborn screening (gNBS) should be used in testing all healthy newborns?

Berdasarkan pemahaman anda sekarang daripada risalah yang diberikan, adakah anda fikir bahawa saringan genomik bayi baru lahir (gNBS) patut dilakukan kepada semua bayi baru lahir yang sihat?

- ☐ Yes Ya
☐ No Tidak
☐ Not sure Tidak pasti

4. Would you prefer to know about the conditions detectable through gNBS before becoming pregnant?

Adakah anda lebih suka mengetahui tentang keadaan yang boleh dikesan melalui gNBS sebelum hamil?

- ☐ Yes *Ya*
☐ No *Tidak*
☐ Not sure *Tidak pasti*

5. In your opinion, what are the possible advantages of genomic newborn screening (gNBS)? (Select all that apply)

Pada pendapat anda, apakah kelebihan yang mungkin ada pada saringan genomik bayi baru lahir (gNBS)? (Pilih semua yang berkenaan)

- ☐ Providing accurate and comprehensive information
Memberikan maklumat yang tepat dan menyeluruh
☐ Providing early and effective treatment
Memberikan rawatan awal dan berkesan
☐ Making an early and prompt genetic diagnosis
Melakukan diagnosis genetik awal dan tepat pada masanya
☐ Preventing complications of the disease
Mengelakkan komplikasi penyakit
☐ Supporting medical research
Menyokong penyelidikan perubatan
☐ Guiding genetic counselling
Memberi panduan dalam kaunseling genetik
☐ Reducing healthcare costs
Mengurangkan kos penjagaan kesihatan

6. In your opinion, what are the potential risks of genomic newborn screening (gNBS)? (Select all that apply)

Pada pendapat anda, apakah risiko yang mungkin berlaku dalam saringan genomik bayi baru lahir (gNBS)? (Pilih semua yang berkenaan)

- ☐ Concern that genetic information may be used without consent or breached
Kebimbangan bahawa maklumat genetik mungkin digunakan tanpa persetujuan atau berlaku kebocoran data
- ☐ Anxiety about the child's future health after knowing genetic results
Kerisauan tentang kesihatan masa depan anak selepas mengetahui keputusan genetik
- ☐ Worry about high costs of genetic testing and subsequent treatment
Kebimbangan tentang kos tinggi ujian genetik dan rawatan susulan
- ☐ Difficulty making decisions based on genetic results
Kesukaran membuat keputusan berdasarkan keputusan genetik
- ☐ Worry that the child may face discrimination
Kebimbangan bahawa anak mungkin akan didiskriminasi
- ☐ Feeling overwhelmed or unable to understand complex genetic information
Rasa terbeban atau tidak dapat memahami maklumat genetik yang kompleks
- ☐ Risk of misinterpreting genetic results and making incorrect decisions
Risiko tersalah tafsir keputusan genetik dan membuat keputusan yang salah
- ☐ Ethical concerns about identifying diseases before symptoms appear
Kebimbangan etika tentang mengenal pasti penyakit sebelum gejala muncul
- ☐ Stress or conflict within the family after receiving genetic results
Tekanan atau konflik dalam keluarga selepas menerima keputusan genetik
- ☐ Conflict with personal or cultural beliefs about fate or destiny
Bertentangan dengan kepercayaan peribadi atau budaya tentang takdir atau ketentuan hidup

Section D: gNBS Implementation Preferences

Bahagian D: Keutamaan Pelaksanaan gNBS

This section examines parents'/guardians' preferences regarding the timing, setting, personnel and consent process for gNBS. The information will help inform parent-centred approaches for delivering gNBS services.

Bahagian ini menilai pilihan ibu bapa/penjaga mengenai masa, tempat, pihak yang terlibat dan proses mendapatkan persetujuan untuk gNBS. Maklumat ini akan membantu memaklumkan pendekatan yang berfokuskan ibu bapa dalam penyampaian perkhidmatan gNBS.

1. When should genomic newborn screening (gNBS) be discussed with parents?

Bilakah saringan genomik bayi baru lahir (gNBS) sepatutnya dibincangkan dengan ibu bapa?

- ☐ Before pregnancy (preconception)
Sebelum kehamilan (prakonsepsi)
- ☐ During pregnancy (antenatal)
Semasa kehamilan (antenatal)
- ☐ At birth (before discharge from the hospital)
Semasa kelahiran (sebelum discaj dari hospital)
- ☐ After birth (e.g. at the 6-week postnatal check)
Selepas kelahiran (contohnya pemeriksaan postnatal pada minggu ke-6)
- ☐ I am not sure
Saya tidak pasti

2. Who do you think should inform parents about genomic newborn screening (gNBS)?
(Select all that apply)

Siapakah yang patut memaklumkan ibu bapa tentang saringan genomik bayi baru lahir (gNBS)? (Pilih semua yang berkenaan)

- ☐ Midwife
Bidan
- ☐ Obstetrician / Gynaecologist
Pakar obstetrik / ginekologi
- ☐ Paediatrician (children's specialist doctor)
Pakar pediatrik (doktor pakar kanak-kanak)
- ☐ General practitioner
Doktor umum
- ☐ Genetic counsellor
Kaunselor genetik
- ☐ Clinical geneticist
Pakar genetik klinikal
- ☐ Nurse
Jururawat

☐ I am not sure

Saya tidak pasti

3. Where should the discussion about genomic newborn screening (gNBS) take place?

(Select all that apply)

Di manakah perbincangan tentang saringan genomik bayi baru lahir (gNBS) sepatutnya dijalankan? (Pilih semua yang berkenaan)

☐ General practitioner's clinic

Klinik doktor umum

☐ Obstetrics / antenatal clinic

Klinik obstetrik / antenatal

☐ Hospital (inpatient ward or maternity unit)

Hospital (wad pesakit dalam atau unit bersalin)

☐ Dedicated genetic counselling unit

Unit kaunseling genetik khusus

☐ Via telehealth / video consultation

Melalui telekesihatan / perundingan video

☐ Via written materials (pamphlet, websites, apps)

Melalui bahan bertulis (risalah, laman web, aplikasi)

☐ I am not sure

Saya tidak pasti

4. Do you think parents should give consent for genomic newborn testing?

Adakah anda fikir bahawa ibu bapa perlu memberi persetujuan untuk ujian genomik bayi baru lahir?

☐ Yes, parents need to give their consent for genetic testing for newborns

Ya, ibu bapa perlu memberikan persetujuan mereka untuk ujian genetik bagi bayi baru lahir

☐ No, the testing should be done routinely for newborns without parental consent

Tidak, ujian tersebut sepatutnya dijalankan secara rutin bagi bayi baru lahir tanpa persetujuan ibu bapa

5. When do you think parents should be asked to give their consent for newborn genomic testing?

Bilakah anda fikir bahawa ibu bapa patut diminta memberikan persetujuan untuk saringan genomik bayi baru lahir?

- ☐ During pregnancy *Semasa kehamilan*
- ☐ At birth *Semasa kelahiran*
- ☐ Not sure *Tidak pasti*

6. How should information about genomic newborn screening (gNBS) be provided to parents? (Select all that apply)

Bagaimanakah maklumat tentang saringan genomik bayi baru lahir (gNBS) patut diberikan kepada ibu bapa? (Pilih semua yang berkenaan)

- ☐ Written materials (e.g. pamphlets, brochures)
Bahan bertulis (contohnya risalah, brosur)
- ☐ Online resources (e.g. websites, videos, apps)
Sumber dalam talian (contohnya laman web, video, aplikasi)
- ☐ Face-to-face discussion with a healthcare provider
Perbincangan secara bersemuka dengan penyedia penjagaan kesihatan
- ☐ Telehealth / video consultation
Telekesihatan / perundingan video
- ☐ Group information session (e.g. antenatal class)
Sesi maklumat berkumpulan (contohnya kelas antenatal)
- ☐ A combination of the above
Gabungan pilihan di atas

7. In your opinion, who should be offered genomic newborn screening (gNBS)? (Select all that apply)

Pada pendapat anda, siapakah yang patut ditawarkan saringan genomik bayi baru lahir (gNBS)? (Pilih semua yang berkenaan)

- ☐ All newborns, regardless of family history
Semua bayi baru lahir, tanpa mengira sejarah keluarga

- ☐ Newborns with a parent or sibling with a known genetic condition
Bayi baru lahir yang mempunyai ibu bapa atau adik-beradik dengan penyakit genetik yang diketahui
- ☐ Newborns with a family history of genetic disease (any relative)
Bayi baru lahir yang mempunyai sejarah keluarga penyakit genetik (mana-mana ahli keluarga)
- ☐ Newborns admitted to the neonatal intensive care unit (NICU)
Bayi baru lahir yang dimasukkan ke unit rawatan rapi neonatal (NICU)
- ☐ Newborns identified as high-risk by their doctor
Bayi baru lahir yang dikenal pasti berisiko tinggi oleh doktor
- ☐ I am not sure
Saya tidak pasti

8. What information is important for parents to know when deciding on newborn genomic testing? (Select all that apply)

Apakah maklumat yang penting untuk diketahui oleh ibu bapa semasa membuat keputusan tentang ujian genomik bayi baru lahir? (Pilih semua yang berkenaan)

- ☐ Cost of the test and treatment
Kos ujian dan rawatan
- ☐ Whether the test is covered by health insurance
Sama ada ujian diadungi oleh insurans kesihatan
- ☐ Location of treatment
Lokasi rawatan
- ☐ Diseases being tested for
Penyakit yang diuji
- ☐ Severity of the condition
Keterukan keadaan penyakit
- ☐ Likelihood and timing of symptom onset
Kebarangkalian dan masa gejala bermula
- ☐ Whether there is treatment for the diseases
Sama ada terdapat rawatan untuk penyakit tersebut
- ☐ Whether results apply to other children
Sama ada keputusan ujian terpakai kepada anak lain

- ☐ Impact on other children (e.g., need for testing)
Kesan kepada anak lain (contohnya keperluan untuk ujian)
- ☐ Possibility of re-contacting parents if new diseases are discovered
Kemungkinan ibu bapa dihubungi semula sekiranya terdapat penemuan penyakit baharu
- ☐ Whether my child's samples will be retained for research purposes
Sama ada sampel anak saya akan disimpan untuk tujuan penyelidikan

Section E: Ethical and Logistical Considerations

Bahagian E: Pertimbangan Etika dan Logistik

This section assesses parents'/guardians' views on data privacy, storage and future use of genomic information and funding of gNBS. The information will help guide policies to ensure ethical and acceptable implementation of gNBS.

Bahagian ini menilai pandangan ibu bapa/penjaga terhadap privasi data, penyimpanan dan penggunaan maklumat genomik pada masa hadapan serta pembiayaan gNBS. Maklumat ini akan membantu dalam penggubalan dasar bagi memastikan pelaksanaan gNBS beretika dan boleh diterima.

1. Who should pay for genetic testing for newborns?

Siapakah yang patut membayar ujian genetik untuk bayi baru lahir?

- ☐ Individuals *Individu*
- ☐ Government *Kerajaan*
- ☐ Health insurance *Insurans kesihatan*

2. Do you think your newborn's genomic data should be stored in the hospital system for future clinical care?

Adakah anda fikir bahawa data genomik bayi baru lahir anda patut disimpan dalam sistem hospital untuk kegunaan penjagaan klinikal pada masa hadapan?

- ☐ Yes *Ya*
- ☐ No *Tidak*
- ☐ Not sure *Tidak pasti*

3. Do you think your newborn's genomic data should be stored in a research database (potentially shared with other researchers)?

Adakah anda fikir bahawa data genomik bayi baru lahir anda patut disimpan dalam pangkalan data penyelidikan (berkemungkinan dikongsi dengan penyelidik lain)?

- ☐ Yes, with my explicit consent each time it is used
Ya, dengan persetujuan jelas saya setiap kali data tersebut digunakan
- ☐ Yes, with a one-time general consent at the time of testing
Ya, dengan persetujuan umum sekali pada masa ujian
- ☐ No, I would not want it shared for research purposes
Tidak, saya tidak mahu data tersebut dikongsi untuk tujuan penyelidikan
- ☐ Not sure
Tidak pasti

4. If stored, how long should your child's genomic data be stored?

Jika disimpan, berapa lama data genomik anak anda patut disimpan?

- ☐ Until the child reaches adulthood (18 years)
Sehingga anak mencapai usia dewasa (18 tahun)
- ☐ For the child's lifetime
Sepanjang hayat anak
- ☐ Indefinitely (no time limit)
Tiada tempoh tertentu (tiada had masa)
- ☐ Only until the initial test results are reported
Hanya sehingga keputusan ujian awal dilaporkan
- ☐ I am not sure
Saya tidak pasti

5. If new health or genetic findings are discovered from reanalysis of your child's stored sample in the future, would you want to receive the updated results?

Sekiranya penemuan baharu kesihatan atau genetik ditemui daripada analisis semula sampel anak anda pada masa hadapan, adakah anda ingin menerima keputusan kemas kini?

- ☐ Yes *Ya*
☐ No *Tidak*
☐ Not sure *Tidak pasti*

6. Do you think genomic newborn screening (gNBS) should be managed differently from standard or expanded newborn screening in terms of guidelines, consent, procedures, policies, or follow-up?

Adakah anda fikir bahawa saringan genomik bayi baru lahir (gNBS) perlu diuruskan secara berbeza daripada saringan bayi baru lahir biasa atau terluas dari segi garis panduan, proses persetujuan, prosedur, polisi atau susulan?

- ☐ Yes *Ya*
☐ No *Tidak*
☐ Not sure *Tidak pasti*

The survey has ended. Thank you for your contribution to this study.

Soal selidik ini telah tamat. Terima kasih atas sumbangan anda dalam kajian ini.

