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Parents' perspectives on expanded newborn genomic screening in Abu Dhabi, United Arab Emirates

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Abstract

Background Newborn genomic screening offers the potential for early detection and management of genetic disorders. Understanding parental perspectives is essential before integrating genomic testing into standard newborn screening.

Methods This was a descriptive cross-sectional study surveyed 568 parents in Abu Dhabi, United Arab Emirates (UAE). An online self-administered validated and piloted questionnaire was used to gather information on demographic characteristic and perspectives regarding newborn genomic screening. Data were analysed using R version 4.4.3.

Results Most parents (78.2%) supported integrating genomics into newborn screening programs, with 63.5% stating it requires distinct management from standard screening. Females preferred geneticists (38.2% vs. 32.5%, $p < 0.001$) and hospitals (45.1% vs. 39.2%, $p < 0.001$) for discussions, with 74.2% emphasizing explicit consent compared to 68.5% of males ($p < 0.002$). Treatability (82.7%), age of symptom onset (74.1%), and severity (72.2%) were key decision-making factors. Additionally, 66.7% preferred genomic testing to be covered by insurance, and 82.2% supported storing genomic data for future use.

Conclusion Parents participated in the study strongly support genomic newborn screening. Gender-based differences emphasize the need for tailored communication and culturally sensitive strategies to inform policy development and implementation of newborn genomic screening program in the UAE and similar contexts.

Keywords Newborn genomic screening, Parental perspectives, Public health, Testing

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Introduction

Newborn screening programs are foundational public health initiatives designed to detect genetic, metabolic, and other congenital conditions early in life, enabling timely interventions before clinical symptoms emerge [1]. Since their origin in the 1960s with phenylketonuria (PKU) screening, NBS programs have broadened globally to include a wide range of treatable disorders [2].

In the United Arab Emirates (UAE), the National Neonatal Screening Program was launched in January 1995, initially screening for PKU. Over the years, it expanded to include congenital hypothyroidism (1998), sickle cell disease (2002), congenital adrenal hyperplasia (2005), biotinidase deficiency (2010), and, more recently, G6PD deficiency (2019). The program incorporated tandem mass spectrometry in 2011 and initiated a cystic fibrosis screening pilot in 2020 [3, 4]. Currently, the UAE screens for more than 40 conditions, alongside universal newborn hearing and congenital heart disease screening, with over 1.3 million infants screened and approximately 2,700 cases of life-threatening or debilitating conditions identified and treated early [4].

The expansion of newborn screening is particularly crucial in the UAE due to the high consanguinity rates—estimated to exceed 50% among Emiratis—and the resulting elevated burden of autosomal recessive disorders [5, 6]. In this context, incorporating genomic technologies into newborn screening presents a significant opportunity to enhance early detection of rare but serious conditions with a genetic basis.

Genomic newborn screening (gNBS) leverages next-generation sequencing and other genomic tools to identify disease-causing variants before symptom onset [7]. Studies in the United States and China have demonstrated that gNBS shortens the diagnostic odyssey, improves survival and quality of life, and can be cost-effective in high-burden settings [8, 9]. Despite its promise, the implementation of gNBS raises a range of ethical, social, and logistical concerns. These include questions about data privacy, the psychological impact of uncertain findings, informed consent, and the return of results—especially for conditions without proven treatments or with adult-onset manifestations [10–12].

Parental perspectives are central to addressing these concerns, shaping the uptake, design, and trustworthiness of gNBS programs. Studies consistently show that parental knowledge, values, and preferences strongly influence decision-making in genomic screening [13–15]. For instance, while many parents support screening for treatable childhood-onset conditions, they are more cautious about screening for adult-onset disorders or those lacking interventions [16–18]. This reflects parents' desire for actionable results and concern about generating anxiety from uncertain or non-actionable findings [19]. Moreover, engaging parents in the design and implementation of gNBS policies—through educational interventions, consent tools, and advisory boards—has been shown to enhance ethical acceptability and program sustainability [20, 21].

While international studies emphasize the importance of clear communication, culturally sensitive messaging, and prenatal education in building support for gNBS [22–24], little is known about parental perspectives in the UAE. This study aims to explore parental perspectives on expanded newborn genomic screening in Abu Dhabi, UAE, including ethical, social, and logistical concerns, as well as preferences for implementation and communication strategies. The findings are presented to inform future policy decisions, public health education, and the ethical rollout of genomic screening initiatives in the region.

Materials and methods

Study design population and sampling

A descriptive cross-sectional online survey was conducted targeting parents residing in Abu Dhabi to assess their perspectives on expanded newborn genomic screening. The sample size was calculated using Raosoft based on an expected proportion of 50% (to allow maximum variability) on support to the development of gNBS, with a confidence level of 95% and a 5% margin of error, the minimum required sample size was estimated to be 385 participants. To account for potential non-completion or ineligible entries, we aimed to reach at least 450 participants. A convenience sampling approach was used. The survey was made available online via digital platforms, including WhatsApp, Facebook, and Instagram.

Information about the study, including a detailed Participant Information Sheet (PIS), was shared publicly through these platforms and parental networks. The PIS explained the study's objectives, inclusion criteria, voluntary nature of participation, confidentiality safeguards, and approximate survey duration. The inclusion criteria were: (1) parent or legal guardian of a child aged less than one month, (2) residing in Abu Dhabi, and (3) able to complete the survey in either English or Arabic. No monetary incentives were provided. Participant contact information was not required or collected. IP addresses were not recorded to ensure anonymity, and the survey platform (SurveyMonkey) was configured to allow only one response per device.

Measures and data collection

An online self-administered questionnaire was developed based on recent literature [13, 17–20] and expert consultation. The literature review informed the structure and content of the tool, while three experts—a clinical geneticist, a public health researcher, and a bioethicist—were

consulted to ensure clinical relevance, cultural appropriateness, and ethical rigor. These experts provided input on question formulation, ordering, and clarity.

The questionnaire was initially drafted in English and translated into Arabic using the forward–backward translation method to ensure linguistic and conceptual equivalence. It included four main domains:

- **Demographic and health-related variables**: gender, marital status, number of children, nationality, attainment of a university level of educational, employment status, monthly income, history of chronic illness, and presence of genetic disorders in the family or children.
- **Parental perspectives on genomic newborn screening**: perceived importance, perceived benefits and risks, and attitudes toward integrating genomic testing into standard screening programs.
- **Implementation preferences**: preferred timing of information provision, responsible personnel (e.g., doctor, nurse, geneticist), preferred setting for discussion (e.g., hospital, clinic), and consent process preferences. Before presenting questions on preferences, a brief description of the roles of healthcare professionals (e.g., geneticists, pediatricians) was included in the survey introduction to assist respondents with informed choices.
- **Ethical and logistical considerations**: data privacy concerns, future use of genetic data, storage and governance of genomic information, and funding responsibility.

A combination of Likert-scale, multiple-choice, and categorical questions was used. The questionnaire was piloted with 30 parents from the target population to assess clarity, cultural appropriateness, and response time. Based on feedback, minor revisions were made. The final version was distributed online using SurveyMonkey. A 'back button' was enabled to allow participants to review and revise answers. Mandatory fields were enforced using the survey tool's built-in validation settings to reduce missing data. This study was conducted and reported in accordance with the Checklist for Reporting Results of Internet E-Surveys (CHERRIES) [25].

Data management and statistical analysis

Survey responses were stored securely within the SurveyMonkey platform and were accessible only to the Principal Investigator. Data were exported and analysed using R version 4.4.3. Descriptive statistics (frequencies and percentages) were used to summarize participant characteristics and responses. The Chi-square test was used to assess gender-based differences in preferences on genomic newborn screening. Statistical significance was set at $p < 0.05$.

Ethical considerations

The study received ethical approval from the Social Science Ethical Review Committee at the United Arab Emirates University (IRB No. ERSC_2024_4422), in accordance with the Declaration of Helsinki. Informed consent was obtained electronically before the survey began. Participants were informed of their right to withdraw at any time, and data confidentiality and anonymity were rigorously maintained throughout the study.

Results

A total of 568 parents completed the survey out of 724 who accessed the questionnaire, yielding a response rate of 78.5%. Of these, 54.8% were female and 45.2% male. Most participants were married (93.1%), while 4.0% were divorced and 2.8% widowed. Emirati nationals accounted for 81.9% of the sample. A majority (85.4%) held a university degree, and 76.6% were employed. Monthly income distribution showed that 42.3% earned < 3,000 AED, while 24.1% earned 3,000–10,000 AED, 17.6% earned 11,000–20,000 AED, and 16.0% earned > 20,000 AED. Approximately quarter of participants (24.8%) reported having children with genetic disorders, while 45.8% had relatives with hereditary conditions. History of chronic illnesses was reported by 37.3% of participants (Table 1).

The majority of respondents (78.5%) supported integrating genomic testing into newborn screening program, while 59.2% agreed that the consent process should differ from that of traditional medical screening for newborns. Despite the high level of support, approximately 21.5% of respondents expressed uncertainty or opposition (Fig. 1).

Table 2. shows that most parents (74.6%) preferred genomic screening to be discussed at birth, with 13.0% favouring discussion during pregnancy and 12.3% during early childhood and most parents support different management from the traditional medical examination (63.5%). Pertaining to the appropriate personnel to inform parents, 36.1% preferred geneticists, 24.8% general practitioners, 21.5% obstetricians, 15.1% paediatricians, and 2.5% preferred midwives. The hospital (42.3%) was the preferred location for these discussions, followed by the general practitioner's clinic (27.6%) and obstetrics clinic (27.8%). Consent for genomic testing was considered essential by 71.8%, while 28.2% believed testing should be automatic standard practice, and the majority preferred to consent during pregnancy (81.3%). Most parents (80.5%) supported offering genomic testing to all newborns, while smaller groups preferred selective testing based on clinical need at doctors' discretion or family history (Fig. 2).

Table 1 Demographic characteristics of participants ($n = 568$)

Socio-demographic variables		*N*	(%)
Gender	Male	257	(45.2)
	Female	311	(54.8)
Number of children	1	124	(22.4)
	2	95	(17.1)
	3	86	(15.5)
	4	80	(14.4)
	5	65	(11.7)
	6	52	(9.4)
	> 6	52	(9.4)
Marital status	Married	529	(93.1)
	Divorced	23	(4.0)
	Widowed	16	(2.8)
Nationality	Emirati	465	(81.9)
	Non-Emirati	103	(18.1)
University degree attainment	Yes	485	(85.4)
	No	83	(14.6)
Employment status	Employed	435	(76.6)
	Not employed	133	(23.4)
Monthly income	Less than 3,000 AED	240	(42.3)
	3000-10,000 AED	137	(24.1)
	11.000–20,000 AED	100	(17.6)
	> 20,000 AED	91	(16.0)
Children with genetic disorders	Yes	141	(24.8)
	No	427	(75.2)
Relatives with hereditary genetic disorders	Yes	260	(45.8)
	No	308	(54.2)
Chronic illness	Yes	212	(37.3)
	No	336	(59.2)
	I do not know	20	(3.5)

Table 3 shows parental preferences for communication, consent, and data use in gNBS. Most participants (67.4%) preferred receiving information about newborn genomic testing through personal discussions, and a majority (86.8%) expressed a desire to know detailed information about the genetic conditions before pregnancy. Moreover, 66.7% believed health insurance should cover genomic testing, while 31.2% advocated for government funding. Furthermore, 82.2% supported the storage of genomic data for future use, and 82.4% wanted updates on new findings derived from stored samples.

Participants prioritized treatability (82.7%), the age of symptom onset (74.1%), and symptom severity (72.2%) as critical factors in deciding on gNBS. Additionally, the certainty of symptom manifestation during childhood (74.3%) and clarity regarding the specific symptoms (73.8%) were deemed important (Table 4).

Figure 3 lists factors influencing decision-making, these include; whether test costs are covered by insurance (63.0%); which diseases were tested for (59.7%); and the cost of testing (47.9%). Additional considerations include treatment availability (47.7%) and the possibility of future re-contact with updated findings (41.2%).

Parents considered early detection (90.0%), early effective treatment (63.0%), and avoidance of complications (52.3%) as the top benefits of integrating genomics into newborn screening. However, they also reported risks including fear of negative results (55.1%), misinterpretation of information (39.6%), and decision-making hesitation (38.9%), indicating a dual perception of opportunity and concern surrounding genomic newborn screening (Fig. 4).

Gender-Based differences in perspectives

A post-hoc gender-stratified analysis revealed statistically significant gender-based differences in several gNBS preferences (Table 5). Female respondents more frequently selected geneticists as the preferred personnel to communicate genomic screening information (38.2% vs. 32.5%, $p = 0.001$), and showed a stronger preference for hospital settings (45.1% vs. 39.2%, $p = 0.001$) compared to males, suggesting a greater inclination toward institutional and specialized support. Furthermore, a significantly higher proportion of females emphasized the need for explicit parental consent (74.2% vs. 68.5%, $p = 0.002$), while males were more open to automatic testing.

Discussion

The integration of next-generation sequencing into newborn screening has advanced the early detection of genetic conditions, offering diagnostic insights beyond the capabilities of traditional biochemical assays [26]. Unlike conventional screening, gNBS enable the identification of a wider array of disorders, including those without biochemical markers or immediate treatments [27, 28]. While the benefits are significant, understanding parental perspectives is essential for ethically sound implementation.

The findings revealed that 78.5% of participants supported the development of gNBS, indicating broad public acceptance and awareness of its potential benefits. However, the 21.5% who opposed or were uncertain about genomic testing reflect important knowledge gaps or concerns that must be addressed through targeted health communication. These findings mirror those from previous international studies—including in Arab and MENA countries—which have emphasized the role of prenatal education, informed consent, and trust-building in increasing parental acceptance [4, 17, 29–32]. The high level of support in this study is consistent with a U.S.-based study where participants demonstrated strong support for newborn screening regardless of the method used [17].

Preferences for discussing gNBS at birth (74.6%), receiving information from geneticists (36.1%), and doing

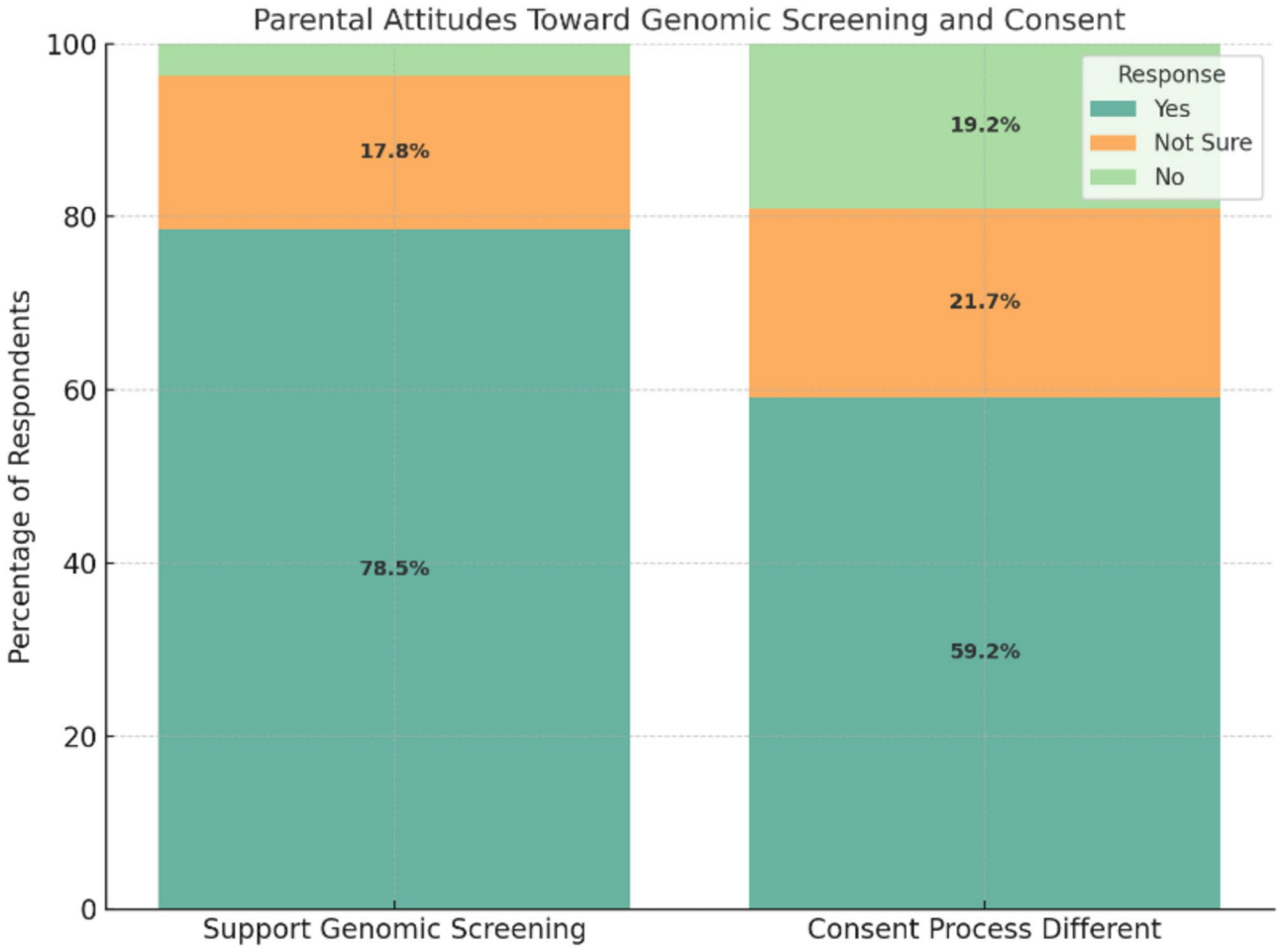

Fig. 1 Parental attitudes toward genomic screening and consent

so within hospital settings (42.3%) highlight parents' desire for expert guidance in credible healthcare environments. These preferences point to a need for accessible and structured genomic counselling pathways. These findings are in line with existing evidence that supports using trained personnel in trusted settings to deliver complex genetic information [33–35].

Ethical considerations were central to parental decision-making. Most participants (71.8%) believed explicit consent was essential, with 81.3% preferring it be obtained during pregnancy. These results align with ethical frameworks emphasizing transparency, autonomy, and informed decision-making in genomic medicine [36]. They also resonate with studies where parents expressed a desire to receive information prenatally and emphasized the importance of tailored consent protocols for genomic screening [18, 20]. The strong support for data storage (82.2%) and receiving future updates (82.4%) suggests a general trust in the long-term medical utility of genomic information, especially when safeguards are in place.

Parents prioritized treatability (82.7%), age of symptom onset (74.1%), and symptom severity (72.2%) when evaluating which conditions should be included in newborn genomic screening. These findings are similar to previous studies showing that parents are more supportive of screening for actionable or early-onset disorders and more hesitant when the prognosis is unclear or untreatable [18, 20, 23]. These preferences underscore the need to tailor genomic screening panels to align with parental expectations and ethical thresholds for actionable outcomes. Moreover, perceived risks such as privacy concerns and psychological burden were reported in the current study as well as in many other studies identified increased anxiety as a key challenge to gNBS program sustainability [37–43]. These insights further reinforce the need for robust pre- and post-test genetic counselling.

Gender-based differences emerged in several aspects of genomic testing preferences. Female participants showed a stronger preference for geneticists (40.8% vs. 30.4%) and hospitals (51.4% vs. 31.1%) as sources and settings for discussion. They were also more supportive

Table 2 Parental preferences and key considerations for newborn genomic testing ($n = 568$)

Variable	Response	*N*	(%)
gNBS requires different management from traditional screening	Yes	360	(63.5)
	No	43	(7.6)
	I am not sure	164	(28.9)
Preferred timing for screening discussion	During pregnancy	74	(13.0)
	At birth	424	(74.6)
	During childhood (3–6 months)	70	(12.3)
Preferred personnel to provide information	General practitioner	141	(24.8)
	Obstetrician	122	(21.5)
	Midwife	14	(2.5)
	Geneticist	205	(36.1)
	Pediatrician	86	(15.1)
Preferred setting for discussing gNBS	General practitioner's clinic	157	(27.6)
	Obstetrics clinic	158	(27.8)
	In the hospital	240	(42.3)
	Online	13	(2.3)
Consent requirement	Consent required	408	(71.8)
	Automatic testing	160	(28.2)
Preferred timing for consent	During pregnancy	462	(81.3)
	At birth	106	(18.7)

gNBS: Genomic newborn screening

Table 3 Parental preferences for communication, consent, and data use in newborn genomic screening ($n = 568$)

Variable	Response	*N*	(%)
Preferred mode of information delivery	Telemedicine (online)	130	(23.0)
	In writing	54	(9.6)
	Personal discussion	380	(67.4)
Desire for pre-pregnancy information	Yes	493	(86.8)
	No	39	(6.9)
	I do not know	36	(6.3)
Preferred payer for testing	Health insurance	379	(66.7)
	Government	177	(31.2)
	Individuals	12	(2.1)
Support for data storage	Yes	467	(82.2)
	No	45	(7.9)
	I do not know	56	(9.9)
Interest in future findings	Yes	468	(82.4)
	No	49	(8.6)
	I do not know	51	(9.0)

of prenatal consent (82.6% vs. 79.8%) and explicit consent overall (66.6% vs. 78.2%). These differences may reflect women's closer engagement with maternal and child health services and heightened sensitivity to ethical decision-making. Similar patterns have been observed in other studies, where women demonstrated greater engagement and decisional caution in genomic screening contexts [24].

This study has several limitations. The convenience sampling method limits the generalizability of the findings beyond the Abu Dhabi context. The online nature of the survey may have excluded parents with limited internet access or digital literacy, leading to potential selection

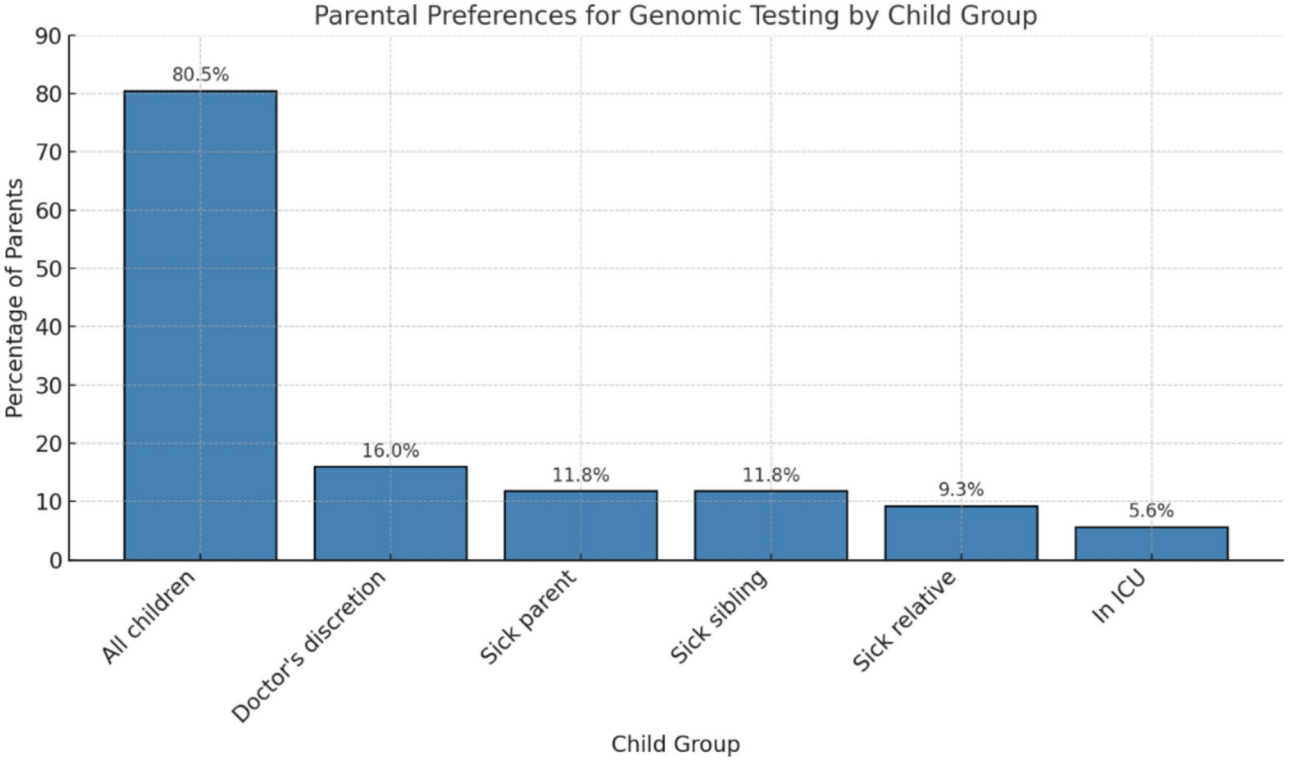

Fig. 2 Parental preferences for genomic testing by child group

Table 4 Importance of case attributes in parental decision-making for newborn genomic testing

Case attributes	Yes (*N* (%)	No (*N* (%)	I don't know *N* (%)
Treatability (treatable or untreatable)	470 (82.7)	38 (6.7)	60 (10.6)
Age of symptom onset	421 (74.1)	35 (6.2)	112 (19.7)
Severity of the condition	410 (72.2)	37 (6.5)	121 (21.3)
Certainty of childhood onset of symptoms	422 (74.3)	34 (6.0)	112 (19.7)
Certainty about the type of symptoms	419 (73.8)	36 (6.3)	113 (19.9)

bias. Additionally, the self-reported format may be influenced by social desirability bias. Future studies should consider longitudinal or mixed-method designs, incorporating qualitative interviews to gain deeper insights into the evolving attitudes and lived experiences of parents navigating genomic screening decisions.

Conclusion

Most of participants supported integrating genomic testing into the standard newborn screening program, emphasizing treatability, age of symptom onset, and severity of conditions as key decision-making factors. Parents expressed strong preferences for discussing genomic testing at birth with geneticists in hospital settings. Most parents favored explicit consent processes, particularly during pregnancy, and supported the storage of genomic data for future use, reflecting trust in the potential benefits of genomic advancements. However, the significant proportion of parents who were uncertain or opposed to genomic testing prompt the need for targeted education and communication strategies to address concerns such as privacy and psychological impacts.

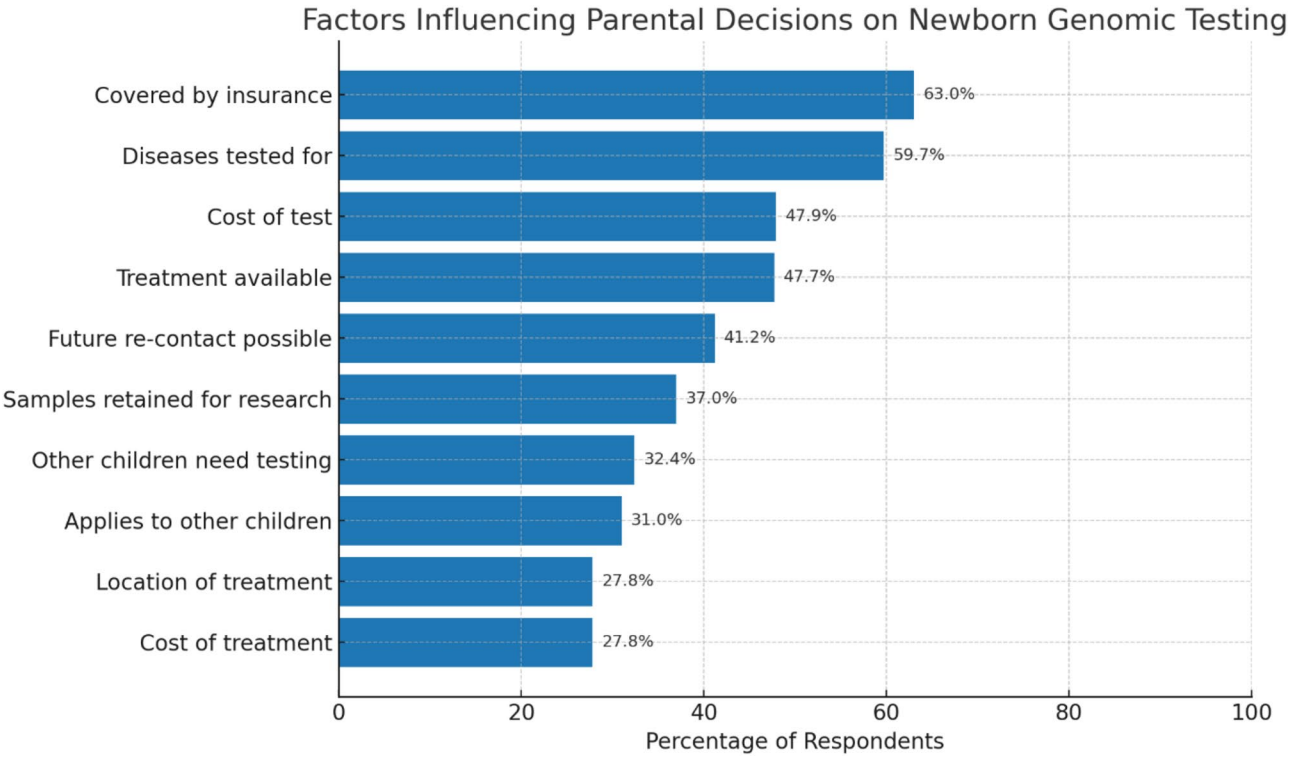

Fig. 3 Factors influencing parental decisions on newborn genomic testing

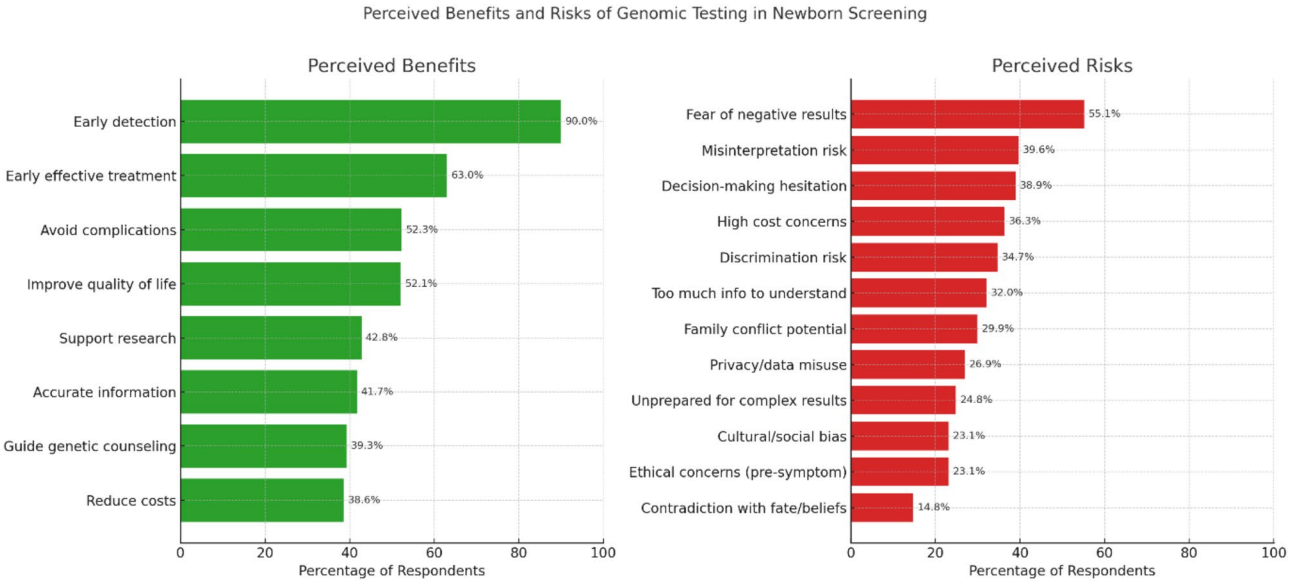

Fig. 4 Parents' perceived benefits and risks of genomic testing in newborn screening

Table 5 Comparison of parental views on key aspects of newborn genomic testing by gender

Variables and responses		Male		Female		P
		N	(%)	N	(%)	
Support for integrating genomic testing in newborn screening	Yes	209	(81.3)	237	(76.2)	0.279
	No	7	(2.70)	14	(4.5)	
	Not sure	41	(16.0)	60	(19.3)	
gNBS requires different management from traditional screening	Yes	164	(64.1)	196	(63.0)	0.395
	No	23	(9.0)	20	6.40)	
	Not sure	69	(27.0)	95	(30.5)	
Timing of consent discussion	During pregnancy	34	(13.2)	40	(12.9)	0.672
	At birth	188	(73.2)	236	(75.9)	
	During childhood (3–6 months)	35	(13.6)	35	(11.3)	
Preferred personnel to provide information	The general practitioner	91	(35.4)	50	(16.1)	< 0.001*
	Obstetrician	53	(20.6)	69	(22.2)	
	Midwife	5	(1.9)	9	(2.9)	
	Geneticist	78	(30.4)	127	(40.8)	
	Pediatrician	30	(11.7)	56	(18.0)	
Preferred setting for discussing gNBS	General practitioner's clinic	103	(40.1)	54	(17.4)	< 0.001*
	Obstetrics clinic	67	(26.1)	91	(29.3)	
	In the hospital	80	(31.1)	160	(51.4)	
	Online	7	(2.7)	6	(1.9)	
Consent requirement	Consent required	201	(78.2)	207	(66.6)	0.002*
	Automatic testing	56	(21.8)	104	(33.4)	
Different gNBS consent from standard medical examination for newborns	Yes	154	(59.9)	182	(58.5)	0.773
	No	46	(17.9)	63	(20.30)	
	Not sure	57	(22.2)	66	(21.20)	
Preferred timing for consent	During pregnancy	205	(79.8)	257	(82.6)	0.382
	At birth	52	(20.2)	54	(17.4)	

gNBS: Genomic newborn screening

*Chi-square test statistically significant at p less than 0.05

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Author contributions
ATR conceptualized the study, designed the questionnaire, supervised data collection, and revised the manuscript. YAME and EM analyzed study's data and critically revised the manuscript. YAME interpreted the findings and draft the manuscript. MA, MAA, FA, TS, YA, AJ, MS, MAS, MSN, KA, KS, NA, OA, SAT, and MSA assisted with data collection and provided critical revisions to the manuscript. All authors read and approved the final manuscript.

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Data availability
The datasets generated and analysed during the current study are available from the corresponding authors upon reasonable request.

Declarations

Ethics approval and consent to participate
This study adhered to the ethical principles outlined in the Declaration of Helsinki. Ethical approval was granted by the Social Science Ethical Review Committee at the United Arab Emirates University. Informed written consent was obtained from all participants before data collection commenced.

Consent for publication
Not applicable.

Competing interests
The authors declare no competing interests.

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