Enhancing newborn genetic screening via prenatal carrier screening

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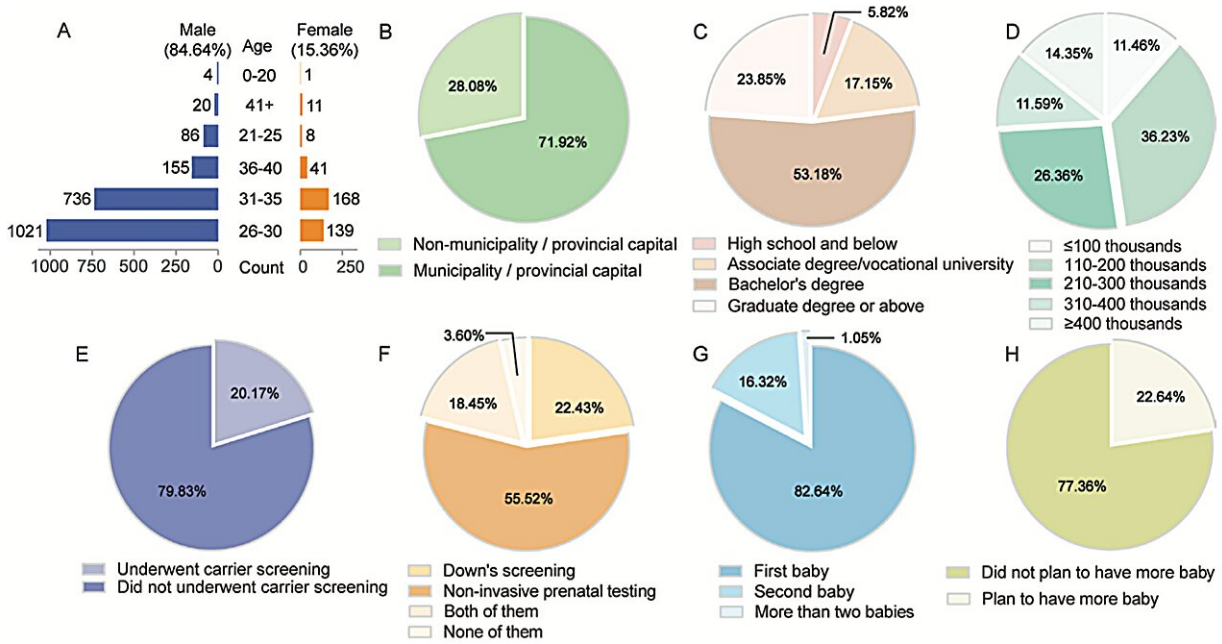

Figure 1. Demographic characteristics of respondents. **Panel A.** Age distribution of respondents. **Panel B.** Geographical distribution of respondents. **Panel C.** Educational level of respondents. **Panel D.** Annual family income of respondents. **Panel E.** Whether they underwent carrier screening. **Panel F.** Whether they underwent Down syndrome screening. **Panel G.** Which baby this was for the respondents (first, second, etc.). **Panel H.** Reproductive plan for respondents.

Demographic characteristics of respondents. Credit: *Journal of Global Health* (2024). DOI: 10.7189/jogh.14.04044

Newborn genetic screening (NBGS) seeks to assist in the early identification of newborns at risk for severe genetic disorders. The integration of genomics into newborn screening represents a significant

advancement, enhancing disease detection while reducing false positive rates. However, sharing results about genetic carriers is a delicate balance between sharing important information and not causing unnecessary worry for families.

To navigate these complex considerations, a joint study led by Nanjing Women and Children's Healthcare Hospital and Jing-Yu Zhao at BGI Genomics, recently published in the *Journal of Global Health*, surveyed parents of newborns identified as carriers through genetic screening. This research aimed to gather their perspectives and inform the development of these vital screening programs.

Research methodology

The study focused on parents of newborns identified as carriers through NBGS at the Nanjing Women and Children's Healthcare Hospital. A comprehensive survey comprising 26 questions was distributed, collecting responses from 2,390 participants. The survey delved into various aspects, including demographic information, understanding of genetic diseases, and the perceived impact of NBGS results.

Table 4. Impact of being a carrier on the family*

Characteristics	Think it is necessary to perform NBGS			The impact of NBGS results to your family			
	Yes, n = 2360 (98.74)	No, n = 30 (1.26)	*P*-value	Positive, n = 1749 (73.18)	Negative, n = 458 (19.16)	None, n = 143 (5.98)	*P*-value
Age			0.865				0.577
<35 y	2032 (98.78)	25 (1.22)		1505 (73.16)	397 (19.30)	119 (5.79)	
≥35 y	328 (98.50)	5 (1.50)		244 (73.27)	61 (18.32)	24 (7.21)	
Gender			1				<0.001
Male	1998 (98.76)	25 (1.24)		1496 (73.95)	396 (19.57)	98 (4.84)	
Female	362 (98.64)	5 (1.36)		253 (68.94)	62 (16.89)	45 (12.26)	
Household registration			0.396				0.506
Municipality/provincial capital	1700 (98.89)	19 (1.11)		1257 (73.12)	327 (19.02)	109 (6.34)	
Non-municipality/provincial capital	660 (98.36)	11 (1.64)		492 (73.32)	131 (19.52)	34 (5.07)	
Education			0.650				<0.001
High school degree or below	138 (99.28)	1 (0.72)		88 (63.31)	32 (23.02)	14 (10.07)	
College degree	1661 (98.81)	20 (1.19)		1244 (74.00)	331 (19.69)	79 (4.70)	
Master degree or above	561 (98.42)	9 (1.58)		417 (73.16)	95 (16.67)	50 (8.77)	
Family income in CNY			0.346				0.734
≤200 000	1129 (99.04)	11 (0.96)		828 (72.63)	227 (19.91)	62 (5.44)	
210 000–300 000	622 (98.73)	8 (1.27)		469 (74.44)	114 (18.10)	40 (6.35)	
≥310 000	609 (98.23)	88 (14.19)		452 (72.90)	117 (18.87)	41 (6.61)	
Carrier screening			0.801				0.064
Yes	477 (98.96)	5 (1.04)		361 (74.90)	79 (16.39)	37 (7.68)	
No	1883 (98.69)	25 (1.31)		1388 (72.75)	379 (19.86)	106 (5.56)	
Pregnancy check			0.741				<0.001
Down screening	527 (98.32)	9 (1.68)		393 (73.32)	102 (19.03)	32 (5.97)	
Non-invasive prenatal testing	1313 (98.94)	14 (1.06)		965 (72.72)	273 (20.57)	66 (4.97)	
Both of them	435 (98.64)	6 (1.36)		332 (75.28)	71 (16.10)	31 (7.03)	
Unchecked	85 (98.84)	1 (1.16)		59 (68.60)	12 (13.95)	14 (16.28)	
Which baby is this			<0.001				0.111
First	1957 (99.09)	18 (0.91)		1455 (73.67)	368 (18.63)	118 (5.97)	
Second	382 (97.95)	8 (2.05)		282 (72.31)	81 (20.77)	23 (5.90)	
More	21 (84.00)	4 (16.00)		12 (48.00)	9 (36.00)	2 (8.00)	
Plan to have more baby			0.898				0.080
Yes	535 (98.89)	6 (1.11)		400 (73.94)	88 (16.27)	43 (7.95)	
No	1825 (98.70)	24 (1.30)		1349 (72.96)	370 (20.01)	100 (5.41)	

NBGS – newborn genetic screening
*Presented as n (%) unless specified otherwise.

Impact of being a carrier on the family. Credit: *Journal of Global Health* (2024). DOI: 10.7189/jogh.14.04044

Key fndings

The average age of the respondents was 30.7 years, with a predominance of positive perceptions towards NBGS. Specifically:

- Parental knowledge and attitude towards NBGS: A considerable majority of parents demonstrate an admirable level of awareness regarding NBGS, acknowledging its essential role in the early detection of diseases. A significant 73.18% of the participants are of the opinion that NBGS will positively affect their future by enhancing the understanding of their child's health conditions.
- The dilemma of reporting carrier status: Despite the potential for family anxiety, an overwhelming 98.95% of parents expressed a preference for being informed about carrier results. This indicates a strong parental desire for comprehensive genetic information regarding their children, underlining the necessity for transparency within NBGS processes.
- Impact of prenatal carrier screening: The study highlights a critical observation that families subjected to prenatal carrier screening possess a superior comprehension of NBGS. This suggests that such screenings are instrumental in promoting the implementation of NBGS. Additionally, NBGS carrier reports are seen to potentially encourage parents to pursue carrier screening before subsequent pregnancies, particularly for conditions with higher incidence rates. The revelation that their baby is a carrier prompted 98.83% of couples to consider prenatal counseling before planning another pregnancy, and 91.63% resolved to inform their child about their carrier status upon reaching adulthood, notably among high-income families and those previously screened.
- Socioeconomic and educational influences: The study sheds light on the socioeconomic and educational variances in the awareness and attitudes towards NBGS. It was observed that higher education levels and incomes correlate with an improved understanding, indicating the impact of socio-economic factors on health literacy.

These findings show if people are more aware of and take part in

prenatal carrier screening, it could greatly improve implementation and acceptance of NBGS. NBGS also has the potential to advance carrier screening practices. After learning that a baby is a carrier, many parents are willing to undergo carrier screening before having another child, especially for diseases with a relatively high incidence, thereby reducing the risk for the next child.

More information: Xin Wang et al, Utility, benefits, and risks of newborn genetic screening carrier reports for families, *Journal of Global Health* (2024). DOI: 10.7189/jogh.14.04044

Provided by BGI Genomics