Women's preference after a high risk screening test result for fetal Down syndrome: Chromosomal microarray vs. NIPT

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Objective

To determine women's preference for choice of follow-up test after a high risk (term risk 1: 250 or higher) Down syndrome screening test result.

Methods

Women with a singleton pregnancy attending for their Hospital Authority Down syndrome Screening test between July 2015 and March 2016 were provided with written information prior to undergoing screening describing their available options if their post screening test result indicated that they were at increased risk of having a Downs syndrome affected pregnancy. Women with a post screening term risk of 1: 250 or higher with no additional fetal ultrasound abnormalities were offered individual counselling and the option of either 1) undergoing a blood withdraw and NIPT test [SafeT21Express, Xcelom, HK] for common aneuploidies; 2) undergo a chorionic villus sampling (CVS) or amniocentesis followed by chromosomal microarray analysis using the FetalDNA Chip [version 2, Dept O&G, CUHK] to detect 100 recognized micro-deletions, micro-duplications and uniparental disomies; 3) Decline any further follow-up. During counselling women were informed that i) amniocentesis and CVS carried an additional 0. 1-0. 2% miscarriage risk of miscarriage; ii) a confirmatory diagnostic procedure would be needed to confirm a 'positive' NIPT test result; iii) all options were free of charge and iv) test results would be available in 5-7 days.

Results

Of the 300 women with a term risk of 1: 250 or higher, 2 (0. 7%) declined further testing, 3 had a spontaneous miscarriage before their intended diagnostic procedure could be performed, 17 (5. 7%) had a concurrent ultrasound abnormalities, 59 (19. 7%) opted for a private self-financed NIPT from other providers before counselling. Post counselling 96 (44. 2%) of the remaining 217 women opted for diagnostic procedure plus microarray versus 121 (55. 8%) who opted for NIPT. Twenty four (25%) women had abnormal result microarray findings (9 Trisomy 21; 2 Trisomy 18; 4 chromosomal mosaic; 2 pathogenic CNVs; 6 CNV of unknown significance; 1 loss of heterozygosity). Five (4. 1%) women who elected for NIPT had either Trisomy 21 (n=4) or Trisomy 18(n=1). Two (1. 6%) NIPT were non-reportable results due to low fetal fraction. Supplementary NIPT reports indicated increased chromosomal DNA counts in 3 (2. 5%) women, all elected to undergo a further diagnostic test. Overall, 60% of women opted for NIPT.

Conclusion

Only 60% of women opted for NIPT in preference to diagnostic procedure with offered a definitive and comprehensive fetal genetic test result using chromosomal microarray after adjusting for cost and time to reporting differences and informing them that the procedure related loss rate was 0. 1 to 0. 2%.