

9th Yazd International Congress and Student Award on Reproductive Medicine with 4th Congress of Reproductive Genetics

Key Lectures

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Non-invasive prenatal testing (NIPT) challenges in future

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Non-invasive prenatal testing (NIPT) for select fetal trisomies became clinically available in 2011 and transformed the landscape of prenatal screening in many countries. With very high sensitivity and very low false-positive rates, this advance represented a tremendous step forward in Down syndrome detection. However in the case of the following conditions the interpretation of NIPT results might be challenging and even error-prone: mosaicism, co-twin demise/vanishing twin, maternal genetic abnormalities, maternal medical conditions (e.g. organ transplant, malignancy), copy number variation of the portion of a fetal chromosome

too small to be detected by the standard cytogenetic testing.

Since it is noninvasive, safe and allows the early detection of abnormalities, NIPT expanded rapidly and the test is currently commercially available in most of the world. As NIPT is being introduced globally, its clinical implementation should consider various challenges, including financial-economic, social, and organizational/ educational challenges. Subsequently, with a higher depth of sequencing and improved bioinformatics analyses, NIPT expanded to include detection of a number of microdeletions. Genome-wide screening for copy number variants has also been reported and is now offered clinically. Noninvasive identification of fetal single-gene disorders, and ultimately analysis of the fetal genome, has become the “next frontier” in prenatal diagnosis. By increasing the portfolio of what can be offered by NIPT it will be necessary to think ahead the challenges of interpreting the incidental findings that will come up and how to share these findings with the families in future.