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Editorial

Challenges and Prospects for the Future: Down syndrome

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INTRODUCTION

Down syndrome (DS) is the most common chromosomal abnormality in humans, affecting 1 in 400-1500 babies born based on maternal age and prenatal screening schedules in various populations. DS is the most prevalent neurological cause of intellectual disability in the world, though a significant percentage of people suffer from a variety of other health problems. The syndrome is caused by trisomy of all or part of chromosome 21 in all or certain cells of the body, followed by an increase in expression due to the trisomic genes' gene dosage [1]. Mental retardation, congenital cardiac abnormalities, gastrointestinal disturbances, weak neuromuscular speech, dysmorphic characteristics of the head, spine, and airways, audiovestibular and visual dysfunction, distinctive facial and physical features, hematopoietic conditions, and a higher prevalence of other medical problems are all associated with this condition. The risk of raising a child with Down syndrome grows with the mother's age. However, since younger mothers have higher fertility rates, the risk of raising a child with DS grows with the mother's age, and more than 80% of children with DS are born to women under the age of 35. Maternal age was the first method for detecting aneuploidy. Nondisjunction predisposes to DS and other foetal chromosomal defects in advanced maternal age [2]. In reality, advanced maternal age was described as being 35 years old or older at the time of childbirth because her risk of developing an aneuploid foetus was equal to or greater than the expected risk of pregnancy loss due to an amniocentesis. Trisomy 21 is linked to brachycephaly, duodenal atresia, heart abnormalities, mild ventriculomegaly, nasal hypoplasia, echogenic bowel, mild hydronephrosis, femur shortening, sandal void, and clinodactyly (middle phalanx hypoplasia) of the fifth digit. In conclusion, Down syndrome is a birth condition with significant physical and social consequences, and there is currently no medical treatment for it. As a result, all pregnant women should be screened for DS. Since false positive and false negative test findings are still produced, NIPS for foetal aneuploidy, which has been in clinical practise since November 2011, is not yet considered a diagnostic test. Covering chromosome 21 with Xist RNA tends to be the mechanism of transcriptional silencing caused by the Xist transgene, resulting in stable heterochromatin alteration. Induction of the newly implanted transgene in the iPS cells resulted in the expression of XIST noncoding RNA, which coated chromosome 21 and caused it to be inactivated. As a result, invasive dialysis. After a positive cfDNA foetal aneuploidy screening procedure, invasive medical treatment such as CVS or amniocentesis is recommended. The cffDNA test's success in screening for trisomy 21 is superior to other screening approaches, with a diagnosis score of more than 99 percent and a false positive rate of less than 0.1 percent. Despite the test's universal adoption, its high cost prohibits it from being used on all patients that have been identified as such by another conventional first-line screening process. The nuchal scan is considered the most effective first-line form of screening by using cffDNA scanning [3].

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