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Short Communication

Bulgarian Down Syndrome Prenatal Testing

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ABSTRACT

With the emergence of non-invasive prenatal testing, Down syndrome screening and diagnosis technologies have advanced quickly in recent years. Not all of the women, however, are aware of the additional test alternatives. As a result, the purpose of our study is to look at women's awareness of prenatal Down syndrome tests in Bulgaria. The survey's findings reveal a lack of understanding of the available screening and diagnostic tests for Down syndrome, particularly NIPT. Women should be better educated and counselled throughout their pregnancy appointments. According to the most recent EUROCAT study, Down syndrome, also known as Trisomy 21, is the most prevalent chromosomal anomaly, with a frequency of 23.02 per 10000. Intellectual handicap, characteristic facial traits, heart abnormalities, and other clinical indications are well-known.

KEYWORDS: Down syndrome, trisomy 21, chromosome abnormality.

INTRODUCTION

Since 1968, when Nadler reported one of the earliest diagnoses of Trisomy 21 from cultured amniocytes, amniocentesis has been the first procedure for prenatal detection of Down syndrome. Merkatz and colleagues discovered a link between low levels of betha-horiongonadotropin and a high incidence of Down syndrome in 1984. That was the beginning of maternal Down syndrome screening, which used biochemical markers in the blood of pregnant women to assess the chance of Down syndrome. Since then, prenatal testing for Down syndrome has advanced significantly, with the launch of Non-invasive Prenatal Tests (NIPT) in August 2011 in Hong Kong and the United States. Prenatal screening and diagnostic testing for Down syndrome are now widely utilised and integrated into standard prenatal treatment for expecting women. NIPTs, which are aggressively provided by private firms, are meant to replace commonly done biochemical first and second trimester screening tests because to their great sensitivity [1]. Nonetheless, the issue remains whether women of reproductive age are aware of the benefits and limits of Down syndrome testing accessible from health care practitioners or direct-to-consumer firms. When compared to biochemical screening, NIPT has a better sensitivity and specificity. It is still used as a screening procedure, with a Chorionic Villus Sampling (CVS) or Amniocentesis performed following a positive result (AC) should be made available to verify the outcome. NIPT, on the other hand, might assist identify high-risk pregnancies and avoid more intrusive treatments with a higher risk of problems [2]. The advantages and disadvantages of each test are discussed during prenatal genetic counselling sessions, but not every woman in Bulgaria has access to such consultations with a medical genetics professional. Medical societies are aware of the many limits of various tests, but are women of reproductive age similarly knowledgeable and able to make an informed decision? As a result, the purpose of our study is to look at women's awareness of prenatal Down syndrome tests in Bulgaria [3].

CONCLUSION

The most crucial aspect of today's prenatal treatment is preventative medicine. Down syndrome screening tests are available in all nations with well-developed healthcare systems. However, the results of the study suggest that pregnant women in Bulgaria are unaware of the capabilities and limits of existing prenatal Down syndrome testing. They also point out that, in comparison to commonly done tests, there is a dearth of understanding regarding NIPT; nevertheless, as technology advances and costs fall, NIPT is projected to become the first choice test. Women should be better educated and counselled throughout their pregnancy appointments. Qualified obstetricians and medical geneticists should collaborate to develop a patient education programme in order to achieve this.

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