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Editorial

Down Syndrome Prenatal Testing

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EDITORIAL

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Researchers in developing countries are more likely to use open-access publications to find knowledge. "If the public pays for this work, why can't the public see the results?" has been a recurring question in recent years. The proponents of the open-access approach have persuaded governments all around the world that if the public pays for research, the public has a right to see the results.

In the ongoing struggle against Down syndrome, a novel DNA-based prenatal blood test developed recently at Brown University in the United States was a huge breakthrough. The blood test, which is ready for clinical use, could reduce the number of dangerous diagnostic procedures required to detect a Down syndrome pregnancy. Dr. Palomaki and Dr. Canick just published the findings of their global study.

Although prenatal screening for Down syndrome has improved, the frequency of invasive diagnostic procedures such as amniocentesis or Chorionic Villus Sampling (CVS) that result from it remains a problem. Nearly all women with a normal pregnancy could avoid an invasive diagnostic procedure and its accompanying anxiety, cost, and risk of fetal loss if this new test is integrated into clinical practice.

It all began in 1997, when researchers discovered that 3%-6% of cell-free DNA in maternal blood was of fetal origin, implying noninvasive methods for detecting Down syndrome. Down syndrome, also known as trisomy 21, is a chromosomal disorder characterized by the presence of an additional 21st chromosome in whole or in part (translocations). It was named after British surgeon John Langdon Down, who first characterized the illness in 1866.

Down syndrome patients have a poorer cognitive ability, as well as severe to severe mental retardation and limited growth. Microgenia (abnormally small chin), an unusually round face, macroglossia (protruding or oversized tongue), an almond shape to the eyes caused by an epicanthic fold of the eyelid, upslanting palpebral fissures (the separation between the upper and lower eyelids), shorter limbs, a single transverse palmar crease (a single rather than a double crease across one or both palms), poor.

Trisomy 21 is created by a meiotic nondisjunction event, which accounts for about 95% of Down syndrome instances, with 88 percent resulting from nondisjunction in the maternal gamete and 8% from nondisjunction in the paternal gamete.

The new DNA-based screening method for Down syndrome is based on Illumina's Massively Parallel Shotgun Sequencing (MPSS) platform Two studies had previously found fetal Down syndrome in 2008 using relatively small sample numbers Palomaki et al. employed a larger sample size of 4,500 women who were enrolled at 27 prenatal diagnostic clinics around the world. This method involves determining the chromosomal origin of millions of DNA fragments by sequencing the first 36 nucleotides of each fragment. The fraction of chromosome 21 fragments is slightly higher than expected if the fetus possesses a third chromosome 21.

The likelihood of having a baby with Down syndrome rises with the mother's age. As a result, many health care practitioners advise women over the age of 35 to get prenatal testing for the illness. Parents and families can prepare for the baby's unique needs by having the infant tested before birth to see whether he or she is likely to have Down syndrome.

Current Down syndrome prenatal screening methods combine maternal age with data from maternal blood markers and ultrasound markers measured in the first and second trimesters of pregnancy. While these tests can detect up to 90% of Down syndrome cases, they also mistakenly classify 2% to 5% of healthy pregnancies as positive. The new DNA-based test will lower the percentage of "false positives" while still detecting Down syndrome. Noninvasive prenatal testing for various chromosomal disorders and neural tube defects should be made available. More research is

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needed, and I am confident that the Longdom Group's magazine "Gynecology and Obstetrics" will help in this regard.

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