Gynecology & Obstetrics
ISSN: 2161-0932

Vijaya Krishna, Gynecol Obstetric 2011, 1:1
DOI: 10.4172/2161-0932.1000e101

Editorial Open Access

Prenatal Screening for Down Syndrome

Vijaya Krishna Varanasi

Post doctoral research, Washington State University, USA

Editorial

Open access (OA) refers to unrestricted access via the Internet to articles published in international scholarly journals such as "Gynecology and Obstetrics". OMICS Publishing Group strongly supports this open access initiative and all articles published by OMICS Publishing Group are freely accessible to everyone immediately after publication. Some of the special features of OMICS group journals include digital formatting, audio listening, language translation and ability to share views on articles via social networking. There are many benefits of open access model where the end users including researchers, patients, students, clinicians and policy makers can have immediate access to latest research findings throughout the world. Open Access articles are cited much more than the non open access articles [1] and have greater visibility in the scientific community and public. Due to this reason impact factor of open access journals is on the rise for the last couple of years [2]. Researchers in developing nations are more likely to access information published by OA journals. In recent years the question that was asked repeatedly was- "if the public is paying for this work, why cannot the public see the results?" The advocates of open access model have now persuaded governments across the world that if the public pays for the research then the public has a right to see the results.

A new DNA-based prenatal blood test developed recently at Brown University, United States was a major breakthrough in the ongoing battle against Down syndrome. The blood test, ready to be introduced into clinical practice can reduce the number of risky diagnostic procedures needed to identify a pregnancy with Down syndrome. The results of this worldwide study led by Dr.Palomaki and Dr.Canick were recently published [3]. It is a major landmark in the sense that prenatal screening for Down syndrome has improved, but the number of resulting invasive diagnostic procedures such as amniocentesis or chorionic villus sampling (CVS) remains problematic. If this new test is introduced into clinical practice, nearly all women with a normal pregnancy could avoid an invasive diagnostic procedure and its associated anxiety, cost, and potential for fetal loss.

It all started in the year 1997 when it was found that about 3–6% of cell-free DNA in maternal blood was of fetal origin suggesting noninvasive ways to detect Down syndrome [4]. Down syndrome or trisomy 21, is a chromosomal condition caused by the presence of all or part (translocations) of an extra 21st chromosome. It is named after John Langdon Down, the British physician who described the syndrome in 1866 [5]. Individuals with Down syndrome tend to have a lower cognitive ability and severe to high degree of mental retardation and stunted growth. The different physical characteristics of individuals suffering from Down syndrome are microgenia (an abnormally small chin) [6], 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 instead of a double crease across one or both palms), poor muscle tone, and a larger than normal space between the big and second toes. Trisomy 21 is caused by a meiotic nondisjunction event and is the cause of approximately 95% of observed Down syndrome cases, with 88% coming from nondisjunction in the maternal gamete and 8% coming from nondisjunction in the paternal gamete.

The new DNA based technique for screening Down syndrome is based on massively parallel shotgun sequencing (MPSS) on Illumina platform [7]. Using relatively small sample sizes, two groups have earlier identified fetal Down syndrome in the year 2008 [8,9]. A larger sample size involving 4,500 women enrolled at 27 prenatal diagnostic centers worldwide was used in the study done by Palomaki et al. [3]. This technique is based on sequencing the first 36 bases of millions of DNA fragments to determine their specific chromosomal origin. If the fetus has a third chromosome 21, the percentage of chromosome 21 fragments is slightly higher than expected.

The chances of having a baby with Down syndrome increase with the age of the mother. Due to this reason many health care providers recommend that women over age 35 have prenatal testing for the condition. Testing the baby before it is born to see if he or she is likely to have Down syndrome allows parents and families to prepare for the baby's special needs. Current prenatal screening tests for Down syndrome combine maternal age with information from the measurement of maternal serum markers and ultrasound markers in the first and second trimesters of pregnancy. While these tests can detect up to 90% of Down syndrome cases, they also incorrectly identify 2% to 5% of normal pregnancies as positive. The new DNA-based test will reduce this "false positive" rate while maintaining the detection rate for Down syndrome. Similar noninvasive prenatal tests should be made available for screening other chromosomal abnormalities and neural tube defects. More research efforts are needed and I am sure the journal "Gynecology and Obstetrics" from OMICS Group will make a positive impact in this direction.

References

1. Eysenbach G (2006) The open access advantage. J Med Internet Res 8: e8.
2. www.biomedcentral.com/info/presscenter/pressreleases?pr=20050623
3. Palomaki GE, Kloza EM, Lambert-Messerlian GM, Haddow JE, Neveux LM, et al. (2011) DNA sequencing of maternal plasma to detect Down syndrome: An international clinical validation study. Genet Med 20: 1-8.
4. Lo YM, Corbetta N, Chamberlain PF, Rai V, Sargent IL, et al. (1997) Presence of fetal DNA in maternal plasma and serum. Lancet 350: 485-487.
5. Down JL (1995) Observations on an ethnic classification of idiots 1886. Ment Retard 33: 54-56.
6. Weiss M (1994) Conditional love: parents' attitudes toward handicapped children. Bergin and Garvey.
7. Ehrich M, Deciu C, Zwiefelhofer T, Tynan JA, Cagasan L, et al. (2011) Noninvasive detection of fetal trisomy 21 by sequencing of DNA in maternal blood: a study in a clinical setting. Am J Obstet Gynecol 204: 205.
8. Chiu RW, Chan KC, Gao Y, Lau VY, Zheng W, et al. (2008) Noninvasive

***Corresponding author:** Vijaya Krishna Varanasi, Post doctoral research, Washington State University, USA, E-mail: vijaya.varanasi@gmail.com

Received December 23, 2011; **Accepted** December 25, 2011; **Published** December 28, 2011

Citation: Vijaya Krishna V (2011) Prenatal Screening for Down Syndrome. Gynecol Obstetric 1:e101. doi:10.4172/2161-0932.1000e101

prenatal diagnosis of fetal chromosomal aneuploidy by massively parallel genomic sequencing of DNA in maternal plasma. Proc Natl Acad Sci USA 105: 20458-20463.

9. Fan HC, Blumenfeld YJ, Chitkara U, Hudgins L, Quake SR (2008) Noninvasive diagnosis of fetal aneuploidy by shotgun sequencing DNA from maternal blood. Proc Natl Acad Sci USA 105: 16266-16271.