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Commentry

Whole-Genome Sequencing: The future of Down Syndrome Diagnosis?

Olagunju Abdul Rahman A*

Student, Federal University of Technology, Akure, Nigeria

***Corresponding Author:** Olagunju Abdul Rahman A, Student, Federal University of Technology, Akure, Nigeria.

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The whole journey started in 1866 when John Langdon Down initially identified Down syndrome (DS) approximately 153 years ago [3]. DS is the genetic manifestation of trisomy of chromosome 21 [8] [Delabaretal 1993; Korenberg., et al. 1994; Constestabile., *et al*. 2010].

Down syndrome continues to be the most common chromosomal disorder. Each year, about 6,000 babies are born with Down syndrome, which is about 1 in every 700 babies born. Before, the diagnosis of DS is typically made through genetic karyotype testing, with post-natal confirmation via the identification of characteristic syndrome-based physical and/or medical features. Today, an expansive menu of prenatal tests for Down syndrome (DS) is already available to pregnant women around the globe, but new tests are likely to become the most popular entrees. Presently, pregnant women can choose among the many prenatal screening tests–triple screen, quadruple screen, first- trimester combined screen, stepwise sequential screens, and fully integrative screens–to receive statistical chances that their fetuses have DS, to varying degrees of detection [1]. However, an open question still remains; with the availability of these DS diagnosis tests, will the birth incidence of DS even decrease further? The main problem is that women has the opportunity to make a decision about the continuation of their pregnancies in private after DS diagnosis. If desired, a woman could decide to terminate without anyone ever knowing that she was pregnant which has blighted the right of life of the child. Does this actually solve the problem? Why can't we correct this genetic mutation as soon as its detected?

Recently, new sequencing methods capable of rapidly analyzing the genome at increasing resolution have transformed diagnosis of single- gene or oligogenic genetic disorders in pediatric and adult medicine.

Whole genome sequencing, also known as WGS, is a laboratory technique in which the entire coding (exon) and non-coding regions of the genome are obtained. It provides a complete, comprehensive map of a person's genetic makeup and allows extensive analysis of all genes to be performed. It's a single lab test that obtains all of the data on all of your genes.

An obvious advantage of the next-generation sequencing techniques is the greater potential to identify the genetic component of health problems, and probably, in the near future, at a lower cost than that of the current techniques. The sheer mass of data generated can reveal diseases causing alleles that could noted otherwise. Moreover, affordable technology that generates more genomic information may aid the translation of applications to further improve healthcare.

During whole genome sequencing, researchers collect a DNA sample and then determine the identity of the 3 billion nucleotides that compose the human genome. The very first human genome was completed in 2003 as part of the Human Genome Project, which was formally started in 1990. Today, sequencing technology is much more efficient, and a human genome can be sequenced in a matter of days for under $10,000. The first human genome cost $2.7 billion [5].

To assess for fetal genetic risks and defects, pregnant women now have access to very sensitive and specific noninvasive screening methods for the common autosomal-trisomies. Today, nonin-

vasive screening for a few selected deletion syndromes is being investigated [4] and has been added by some providers [2], but the performance of noninvasive detection of such microdeletions is still being evaluated. Although, for a definitive prenatal diagnosis of DS, there are currently just two options, both of which are invasive: chorionic villus sampling (CVS), generally performed between 9 and 12 weeks of gestation, and amniocentesis, traditionally offered between 15 and 20 weeks of gestation which was recently done by some researchers in which they demonstrated the feasibility of generating an accurate whole-genome sequence of fetus from either the cellular or cell-free DNA (cfDNA) of an amniotic sample [7]. By nature of being invasive, both of these diagnostic tests carry small, albeit real, risks of spontaneous abortions.

Despite hundreds and thousands of articles published and billions of dollars spent on genetic diseases especially DS research worldwide, the correction of the mutated gene remains obscure and controversial. Developments are just done around its diagnosis.

Furthermore, none of these techniques can detect other types of mutations, such as point mutations and small insertion–deletion (indels) mutations, that cause the now more than 4600 known single- gene disorders and others yet to be characterized [9]. We implore researchers to focus more on ways to correct the mutation.

The author of this article still feels and believes as stated by [5] that the role of most of the genes in the human genome is still unknown or incompletely understood. Therefore, a lot of the "information" found in a human genome sequence is unusable at present. Till the time we can address and find an easy, non-invasive and effective method and technology to accurately correct the genetic errors in DS, we will still be circling back to the same issue.

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Volume 3 Issue 9 September 2019