Review of: "[Perspective] Is There Any Reason to Stay in Human Genetic Societies as Cytogeneticists?"

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Potential competing interests: No potential competing interests to declare.

Title of the reviewed manuscript: [Perspective] Is There Any Reason to Stay in Human Genetic Societies as Cytogeneticists?

Comment:

There is a great reason to stay in the Genetic Societies as cytogeneticists.

Cytogenetics now became the cytogenomics (Chromosmics) science with the advent of many new applications.

The conventional cytogenetics (CC) is very important in diagnosis of many genetic disorders, by simple test we can diagnose numerical and structural abnormalities in autosomes and sex chromosomes. Conventional cytogenetics can diagnose carriers of balance translocation which is sometimes missed even by whole genome sequencing (WES), (Hochstenbach et al 2019). The molecular technology involved WGS and whole exome sequencing (WES) are very expensive and not available in many laboratories and cannot be applied to all patients especially in the developing countries.

Fluorescence in situ hybridization (FISH) is a very promising technique and have many applications and in conjunctions with CC can diagnose balanced translocation. Balanced translocation is a great problem and if diagnosed probably can prevent the occurrence of multiple congenital anomalies (MCA) and intellectual disability (ID) in the next generations. In study of Mohamed et al (2015), they reported on balanced translocation transmitted through four generations and gave 19 affected patients with MCA and ID. FISH technique has many available commercial probes, FISH probes also can be generated in house using PCR amplification and labeling or nick translation and labeling of bacterial artificial clones (BAC).

There are many FISH technology that combined the FISH with the molecular technology. Oligonucleotide (oligo)-based FISH has developed as an important tool for the study of chromosome organization and gene expression. A CRISPR/Cas9-Based In Situ Labeling Method and CRISPR/Cas Genome Editing now have vast applications. The SNP microarray analysis can diagnose small structure copy number variance and loss of heterozygosity and can detect uniparental dizomy.

The science of cytogenomics is a live science with rapid progression and updating. The human genetics societies and conferences must involve this great science in all its activities. Also genetic societies must incorporate the patients and

their families in their meeting and explain to them in a simplified approach their problem and the methods of management.

Hochstenbach, R., van Binsbergen, E., Schuring-Blom, H., Buijs, A., and Ploos van Amstel, H. K. (2019). A Survey of Undetected, Clinically Relevant Chromosome Abnormalities when Replacing Postnatal Karyotyping by Whole Genome Sequencing. Eur. J. Med. Genet. 62, 103543.

Mohamed AM, Kamel A, Mahmoud W, Abdelraouf E, Meguid N. 2015. Intellectual disability secondary to a 16p13 duplication in a 1;16 translocation. Extended phenotype in a four-generation family. Am J Med Genet Part A. 167A:128–136.