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

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The topic of this perspective is very interesting and other Authors also discussed challenges and opportunities for clinical cytogenetics in the 21st Century (for instance see Heng E, Thanedar S, Heng HH. Challenges and Opportunities for Clinical Cytogenetics in the 21st Century. Genes, 2023 Feb 15;14:493) since genome sequencing, combined with the analysis of cell free DNA, is greatly reducing the request of karyotype analysis. Indeed, even if it is true that both the genome and chromosome architectures are relevant of the gene expression, diagnostic labs and patients seem to be more interested in sequencing data than in other approaches. As suggested by the Author in a previous paper, we must remind that “all chromosome-related studies should be summarized to introduce novel ideas and concepts in biology and medicine, thus having an integrative effect on the field” so that cytogenetics may be extremely useful also in the genome sequencing era. Cytogenetics is indeed relevant to monitor genome instability, to identify and classify new types of chromosome/nuclei variations and for the study of chromosomal polymorphisms.

The present proposal advocating for a reassessment of the significance of clinical cytogenetics in the 21st century is therefore very important and timely, since it emphasizes the importance of cytogenetics alongside sequencing technologies rather than replacing them. This recommendation is based not only on the crucial genomic topological insights provided by cytogenetics for the understanding of gene organization within biological systems, but also in the fact that altered karyotypes serve as end products reflecting a range of individual molecular mechanisms. This, in turn, can enhance clinical diagnosis with more accurate predictions.

As a whole, even if it is true that conventional cytogenetics is a time consuming and labor-intensive process (automation and digitalization driven by the rise of artificial intelligence of cytogenetic analyses are still ongoing), cytogenetics has substantially enhanced the detection and quantification of chromosomal aberrations/changes and facilitated clinical and non-clinical investigations making cytogenetic methods essential for the tool kit of human geneticists.