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

Pavel V. Nikitin[1]

1 University of Houston

Potential competing interests: No potential competing interests to declare.

The article "Is There Any Reason to Stay in Human Genetic Societies as Cytogeneticists?" presents several critical observations and concerns about the current state of human genetic societies, particularly focusing on the European Society of Human Genetics (ESHG) and the German Society of Human Genetics (GfH). Here's an analysis of the core arguments and implications:

Shift in Focus of Human Genetics Societies: The author notes that these societies, which once covered a broad range of topics in human genetics, have now predominantly shifted their focus towards DNA-oriented approaches, mainly sequencing. This observation raises questions about the inclusivity and diversity of research topics in these societies. The lack of representation of certain subfields, like cytogenetics, could lead to a narrow scope of research and discussion, potentially overlooking crucial aspects of human genetics.

Absence of Patient Support Groups in Meetings: The article highlights the concern that patient support groups no longer attend these meetings. This absence suggests a potential disconnect between the societies' activities and the needs or interests of patients. The core purpose of human genetic research, which is to aid in patient care and provide insights into genetic disorders, may be undermined if patient perspectives are not integrated into these discussions.

Underestimation of Cytogenetics: The author argues that there is a growing tendency in the field to believe that sequencing alone can solve every genetic case. This perspective undervalues the role of cytogenetics, which, as the author points out, is crucial in understanding complex genetic diseases and genomic imbalances. Ignoring cytogenetic data can lead to misdiagnosis or incomplete understanding of genetic disorders, which is detrimental to patient care.

Educational and Professional Concerns: The dwindling focus on educating cytogeneticists in universities, particularly in Germany, is another significant concern raised. This trend could lead to a shortage of skilled professionals in this important subfield, affecting both research and clinical practice. Furthermore, the absorption of human genetics by other fields like pathology, as mentioned in relation to the USA, Australia, and Canada, suggests a potential dilution of specialized expertise in human genetics.

Call for Action: The author's suggestion to draft letters of concern to these societies is a call for action. It underscores the need for a re-evaluation of the priorities and focus areas of human genetic societies to ensure a balanced and comprehensive approach to genetics research and application.

Overall, the article effectively highlights critical issues within human genetic societies concerning the representation and prioritization of various subfields, especially cytogenetics. The implications of these issues are far-reaching, affecting research diversity, patient care, professional training, and the future direction of the field. The concerns raised warrant serious consideration and action from the involved societies to ensure that the field of human genetics remains inclusive, comprehensive, and patient-oriented.

I strongly agree with the argument against underestimating cytogenetics. In the current landscape of genetic research, there is increasing evidence that the geometrical and spatial features of chromosomes play a crucial role in the regulation of gene expression. This insight is particularly relevant in the burgeoning field of epigenetics, which explores how genes are expressed and regulated beyond just their sequence.

One significant example of this is the studies of chromatin architecture. Researches have shown that the way DNA is packaged and structured within the cell nucleus – aspects that are a focus of cytogenetics – can significantly impact gene expression. For instance, the positioning of genes within topologically associating domains (TADs) has been found to influence their activity. Disruptions in TADs can lead to misregulation of gene expression, contributing to various diseases, including cancer.

Another area where cytogenetics plays a vital role is in the study of chromosomal abnormalities. Techniques such as karyotyping and fluorescence in situ hybridization (FISH) have been instrumental in identifying chromosomal rearrangements and aneuploidies that are associated with genetic disorders. For example, the identification of chromosomal translocations in certain cancers has led to a better understanding of the disease mechanisms and the development of targeted therapies.

Furthermore, the integration of cytogenetic data with sequencing information offers a more comprehensive view of the genome. This approach can be particularly important in cases where sequencing alone may not provide a complete picture, such as in the detection of large genomic imbalances or complex structural variations.

In summary, while sequencing data is invaluable, a holistic approach that includes cytogenetic insights is essential for a deeper understanding of genomic function and regulation. This broader perspective is crucial for advancing our knowledge in genetics and improving patient care.