

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

Ivan Y. Iourov¹

¹ Mental Health Research Center of Russian Academy of Medical Sciences

Potential competing interests: No potential competing interests to declare.

Firstly, I would like to express my support for Prof. Liehr's initiative to highlight the problem of being a cytogeneticist in Human Genetic Societies. It might raise wider concerns about the place of cytogenetics in current human genetics or even biomedicine. Since cytogenetics spans far beyond the diagnosis of chromosome abnormalities, genetic societies have to pay attention to the artificial diminishing of researchers'/scholars' interest in cytogenetics, an important part of genetics and bioscience. Certainly, problems surrounding cytogenetics' role in current human genetics are not limited to technological aspects (i.e., suggested outdateding of cytogenetic methods, which indicates a misunderstanding of basic cytogenetic technologies because of sophistication than true outdateding). Thus, Prof. Liehr's perspective paper is a perfect opportunity to start the discussion about modern cytogenetics.

For brevity reasons, I would like to consider current problems of cytogenetics and cytogeneticists in four dimensions: theoretical (i), empirical (ii), diagnostic (iii), and educational (iv). (i) Unfortunately, theoretical aspects of chromosome biology and cytogenetics are currently addressed by a variety of biomedical disciplines (e.g., cell biology, cancer biology, etc.), among which (human) genetics/genomics is absent. So, chromosomal aspects of the human genome are lost to human genetics and genomics. (ii) Empirically, human cytogenetics, or better to say, molecular cytogenetics and cytogenomics (cytopostgenomics), is actively developing and combining achievements in visualization (microscopic) and whole-genome technologies. Thus, the lack of knowledge about these developments can only cause diminished interest among genetic scholars in studying human chromosomes. (iii) Prof. Liehr properly indicated the impossibility of diagnosing chromosomal abnormalities (including cryptic and mosaic ones) without molecular cytogenetic or visualization/microscopic techniques. Here, it is not a matter of outdated technologies; it is a matter of logics. For instance, no molecular techniques are able to give comprehensive info about the localization of chromosomal abnormalities. (iv) As noted by Prof. Liehr, educative efforts in cytogenetics are unfairly rare. The problem is worldwide. The main reason is referred to the complexity of performing visual analysis of chromosomes by banding techniques (the majority of scholars/researchers prefer the easiest ways to succeed). On the other hand, the lack of cytogenetic knowledge stops further developments in human genetics and genomics, i.e., to propose efficient therapeutic interventions in genetic diseases. Indeed, the unawareness about gene localization and gene behavior at the chromosomal level leads to the impossibility to interfere correctly with mutated nuclear DNA. If you miss knowledge in human cytogenetics, you consider genes as loose substances floating in a mysterious space. It's funny, but useless. For more details, one may see https://doi.org/10.1007/978-3-030-62532-0_9. In total, it seems that there is an urgent need to solve the problems surrounding human cytogenetics in all the four aforementioned dimensions. Fortunately, Prof. Liehr has made an important step towards the solution (if it actually

exists!).

Finally, it is undoubted that every cytogeneticist has to answer the title question according to their own experience and future sightedness. It is to note that one may expect a rise in interest in human cytogenetics, since interests in specific science areas generally obey oscillatory laws. Researchers who studied chromosomal imbalances in the mid-2000s and beginning of 2010s should remember an increased interest in studying chromosomes in the human genetic context due to the introduction of array methods. Visualization (cytogenetic and molecular cytogenetic) techniques also gained some attention during that époque.