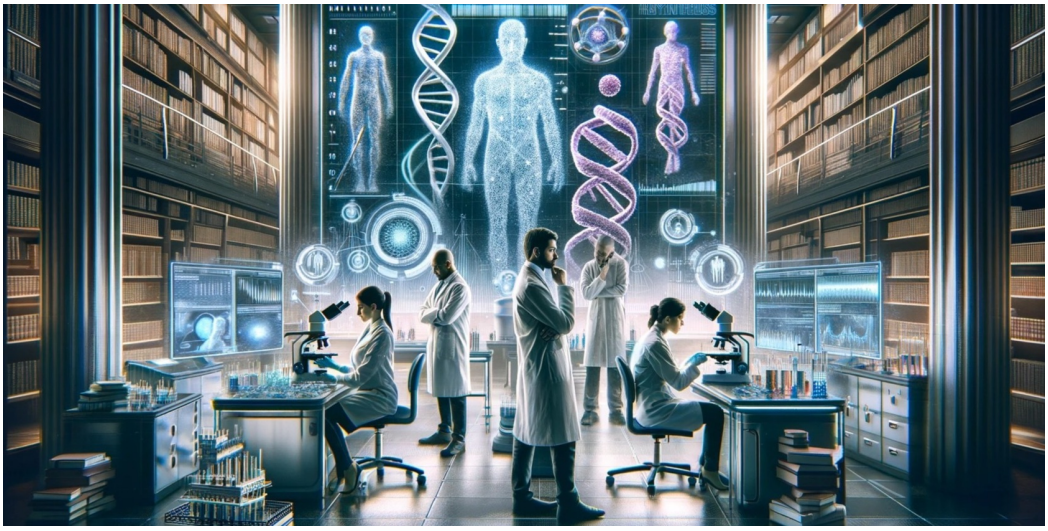

# [Perspective] Is There Any Reason to Stay in Human Genetic Societies as Cytogeneticists?

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## Abstract

In this paper, I express my concerns that for about a decade, Human Genetic societies have ceased to represent the full field. Specifically, clinical and scientific data provided by (molecular) cytogenetics are practically ignored and not considered as topics that need attention at congresses, in professional politics, or in the education of young scientists or MDs. Simultaneously, Human Genetic society meetings have lost family support groups as regular participants. This trend suggests that the field, by focusing solely on high-quality research and diagnostics in the narrow field of gene mutations, may have chosen the wrong direction. I provide a draft letter in English, which may serve as an urgent distress signal to national and European societies of Human Genetics. They are urgently requested to reconsider their politics and to again consider chromosomes, banding cytogenetics, and molecular cytogenetics as topics worth presenting at meetings and educating young scientists about. Otherwise, Human Genetic societies may fade and disappear, as has already happened, for example, in Australia, where the field is now represented by Pathology.

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When I joined the European (ESHG -https://www.eshg.org/home) and our national Society of Human Genetics in Germany (Deutsche Gesellschaft für Humangenetik – GfH - https://www.gfhev.de/) about 25 years ago, they both represented and covered the entire field of Human Genetics. This included clinical genetics, syndromology, genetic counseling, chromosomes (cytogenetics and molecular cytogenetics), nuclear architecture, DNA (cloning, sequencing, imprinting, etc.), and new methodological developments. Meetings organized by these societies were not only oriented towards diagnostics, research, and industrial exhibitions, but also towards patients. This was nicely highlighted by the fact that many patient support groups were present at these meetings.

For the past 10 years (or even more), such meetings have not included any (molecular) cytogenetic sessions. At the same time, no patient support groups are attending these meetings! The latter point is particularly alarming to me: For whom do we do our work? – For patients! Moreover, if the representatives of patients with genetic disorders see no reason to attend our conferences anymore, something must be going severely wrong in our field.

If we analyze the content of these meetings in terms of research nowadays, they primarily focus on all possible variants of DNA-oriented approaches, mainly sequencing. They also emphasize high-quality research on rare diseases and single gene mutations. More specifically, the technical progress of sequencing machines seems to be the main topic. Chromosomes are declared not to be of interest anymore, even though the recognition of topologically associating domains (TADs) and the introduction of optical genomic mapping were small signs of hope that:

- It might be coming back to the attention of specialists that the human genome is more than (not one but) 46 long DNA strands with possibly disease-causing genes on them.
- That there may be more than just assigning a variant in one of 5 (arbitrary) classes;
- And that in 70% of cases, a genetic disease is multifactorially caused and/or by a genomic imbalance of many megabase pairs.

Unfortunately, there is an increasing attitude in the field that every genetic case can be solved by sequencing alone. I am aware (I believe we all are) of cases where cytogenetic results were ignored and a single gene SNP was incorrectly blamed as the potential cause of the patient's problems. However, such issues are practically never discussed at Human Genetic meetings, as there are no cytogenetic sessions anymore.

Overall, there have been no attempts by GfH or ESHG to strengthen (molecular) cytogenetics. In Germany, I know that all private labs conducting postnatal/fertility diagnostics need cytogeneticists, just like tumor genetic labs do. Almost all University-based Human Genetic institutes in Germany have stopped educating cytogeneticists. As a result, German private labs now have to train new coworkers in banding cytogenetics, as these skills are no longer provided by universities. This type of education cannot be considered equivalent to what was previously offered at universities. Even worse, we know from countries like Australia, the USA, and Canada that Human Genetics has been absorbed by Pathology. Therefore, the fact that new tumor genetic labs have been and continue to be opened by Pathologists in

Germany over the past few years, and that they are looking for cytogeneticists, should alarm national societies and prompt them to re-strengthen this field.

Perhaps more ‘chromosome people’ reading this share the same concerns about the development of our national and international Human Genetic societies and may write corresponding letters of concern to them. A draft letter in English is provided as a supplement to this paper.

## Supplementary File

Draft letter to Human Genetic societies, which can/may/should be adapted and sent out by email or mail and distributed to colleagues to generate some response. Additionally, it can be translated into your national language using Google Translate or similar platforms.