

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.

The concerns expressed in the open letter highlight the writer's perspective on the decline of attention and support for (molecular) cytogenetics in the field of Human Genetics.

While these concerns may be valid from the writer's viewpoint, it's essential to note that perspectives can vary, and different stakeholders may have different views on the priorities and directions of scientific fields. Here are some points to consider revision:

- The letter suggests that the exclusion of (molecular) cytogenetic sessions from meetings is a sign of a severe issue in the field. However, it does not provide a comprehensive analysis of the reasons behind this exclusion. There might be reasonable reasons, such as evolving scientific priorities or a shift in research trends, which are not adequately addressed in the letter.
- The letter expresses disappointment that the cytogenetics field is declared not to be of interest anymore. However, it does not delve into the reasons behind this shift in focus, and there may be scientific justifications for emphasizing other aspects of genetics. Understanding the rationale behind such decisions is crucial for a more nuanced evaluation.
- While the open letter suggests the exclusion of cytogenetic sessions at human genetics conferences, it lacks a comprehensive analysis of the reasons behind this exclusion. A stronger argument could be made by providing data, such as an examination of abstracts from conferences, to determine the extent of the issue. The author could have explored the trend in the use of cytogenetics-related words in published conference abstracts, such as "karyotype" or "copy number variant".
- The letter mentions cases where cytogenetic results were allegedly ignored in favor of sequencing. Without concrete examples and data, it may be challenging to assess the prevalence and significance of such instances.
- The concerns raised about cytogenetics training should be framed within existing opportunities, such as courses supported by the European Society of Human Genetics (*Basics in Human Genetic Diagnostics – A course for CLGs in Education and the Goldrain Cytogenetics Course*) and the European Cytogenetics Association (*European Advanced Postgraduate Course in Classical and Molecular Cytogenetics*).
- The claim of total neglect in cytogenetics training should be revised in light of existing guidelines from the European Union of Medical Specialists for medical genetics physicians. Evaluating whether additional training opportunities are needed is a valuable consideration and whether more guidelines should exist for cytogenetics training in other professions.
- The critique of pathologists' involvement in practice requires a more nuanced framework, considering historical

practices and the advantages and disadvantages of including other professionals.

- The open letter expresses alarm over the absence of patient support groups at recent conferences. While this highlights a potential issue, the letter does not provide concrete evidence or specific examples to substantiate this claim. It would strengthen the argument to include data demonstrating the decline in patient representation, fostering a more thorough understanding of the perceived disconnect between the scientific community and patient advocacy.

In addressing these concerns, a more in-depth analysis, inclusion of concrete examples, and engagement with relevant stakeholders would enhance the open letter's credibility and contribute to a constructive dialogue about the future direction of Human Genetics as a field.