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

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Potential competing interests: I am the founder of ClearSKY Genomics. We are developing a genome browser design to simplify "nucleotide-focused" genomics, by adapting certain cytogenetic perspectives.

This letter highlights how the clinical genomics space is changing and evolving. The author expresses concern about the place of cytogenetics within modern genetic societies - describing this letter as an urgent distress signal. While I am worried that this signal might be ignored by those who see the shifting focus to DNA sequencers and single nucleotide variants as part of the inevitable march towards the future, as cytogenetics plays a crucial role in modern clinical genetics, this perspective is important to highlight.

Here, the author highlights that a large proportion of genetic diseases can be attributed to large, megabase-scale aberrations, and that the value of techniques that can effectively (and economically) detect these changes is often overlooked to discuss newer technologies and methodologies within genetics societies. As someone who has noticed a similar pattern, I can empathise with the author's frustrations.

This shifting focus can, to some extent, be attributed to the rapidly evolving world of clinical genomics, with (potentially) clinically significant advancements routinely being made in areas as diverse as sequencing technologies, bioinformatic methods, and curation strategies. While, at times, this nucleotide-centric focus can seem a bit reductive, the discussion of the advancements in this space is important to an informed workforce and critical to ensuring these new approaches are implemented safely and clinically effectively.

I must admit that I am new to Qeios, so please forgive me if I've misunderstood the scope of the review process, and I'm providing unwarranted feedback - however, if there is an option to provide advice for the next iteration of this letter, I would like to make two suggestions.

Firstly, it would be beneficial to precisely quantify the role cytogenetics plays in molecular diagnosis in a modern health service. Knowing the proportion of molecular diagnoses that can be attributed to cytogenetic approaches in an NGS-enabled healthcare system will strengthen the argument and (hopefully) prevent people from dismissing this perspective as support for an "outdated approach."

Secondly, I would like to see the author propose a model (or even an outline) that allows cytogeneticists to use their expertise and experience to help guide the future iteration of clinical genetics. While future technologies MAY replace SOME common cytogenetic techniques, the practical knowledge provided by cytogeneticists is invaluable, and it would be a tremendous mistake not to utilize this experience.