https://www.cell.com/ajhg/fulltext/S0002-9297(21)00096-3
Hi,
Thank you very much for putting together this paper!
I originally had a longer comment, but I thought that it was hard to read. So, in a sense, I am glad that it got flagged so that I could focus on posting a shorter comment in this system.
For context and details, you can see something similar to that earlier version here .
However, I think these are the most important questions to ask:
1) I have concerns about the consumer IcWGS that I have received, such that I would be concerned about use for medical applications. However, I thought results like Figure 4 and Table S4/S5 might match my experiences, while also giving a reason not to unfairly discount a result labeled as "IcWGS".
In other words, do you think some of the problems that I encountered were an issue of how "low" the coverage sequencing is (such as 0.1x-0.5x, versus 4-6x)?
Also, do you think the ~0.5x sequencing is a fair representation of what you see currently available to consumers, across various companies?
2) For your comparisons, do you always include imputations (both IcWGS and SNP chip)?
In other words, for medically actionable results, I might group imputed SNP chip <i>or</i> lcWGS in one category, and directly measured SNP chip genotypes (perhaps for tested populations) in another category.
So, I think there are at least some situations where I would prefer a SNP chip with measured genotypes (without any imputations) over ~0.5x IcWGS. Do you think this is fair?
Thank you again for your contributions to the field!
Sincerely,
Charles