Scientific Essay about

'Ethnically relevant consensus Korean reference genome towards personal reference genomes (Nat commun. YS Cho, 2016)'

[Supplementary Essay 1]

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Abstract

This paper is a scientific essay of a paper; 'Ethnically relevant consensus Korean reference genome towards personal reference genomes (Nat commun. YS Cho, 2016)'. Therefore, it includes not only the summary of experiments in the paper but also problems and defects based on scientific reasons.

Summary

In this paper, authors took issue with an ethnic variant of the standard human reference (currently GRCh38). It was not suitable for reference to research about Korean genome analysis. This paper challenged new sequencing method for Korean genome mapping. They recruited representative genome donors and compared with other human genomes (CHM1_1.1, NA12878_single, Mongolian, African and so on). Also, they investigated structural variants and found ethnicity-specific functional marker.

Discussion

1. insufficient scientific reasons of ethnicity of Korean people or genome in the donors.

   This paper show 2 supplementary figures (Supplementary Figure 1,2) which were reasons that volunteers' genomes were suitable for using sample of genome analysis. First one show the genetic distance of the sample did not fall outside the common Korean population range using MDS plot. The other show there were no abnormalities in the chromosomes using G-banded karyotype analysis.

   However, there did not include reasonable data which donors had ethnicity-specific genomes. The possibility which they were mix-blood people remained. At least, I think, authors must be check genealogy of donors and show that. All of data and analysis in this paper assumed 16 donors' genome totally represented Korean genome but there were no scientific reasons.

2. No ethnicity-specific genomic information of chronic diseases.

   Authors reported the first consensus Korean genome reference and found that ethnically-relevant

reference help to check variants. However, they cannot investigate ethnically variants of the gene related to chronic disease (ex, BRCA1,2). One of the purpose of the finding and analysis variants was to cure chronic disease especially cancer. It was hard to see all of gene related to disease but I think it is necessary to show data or clues which variants of one or two genes in the Korean genome were related to disease incidence.

3. Weakness of confirming data for accuracy of the Korean reference

(It related to first discussion subject) At the end of experiments, they did not show comparing step for common Korean people. They just present a statistic data which KOREF was useful to found variants and how many difference to compare other genome reference. I think an experiment which show accuracy of KOREF to apply analysis for other common Korean genomes should necessary.

Reference

1. Ethnically relevant consensus Korean reference genome towards personal reference genomes (Nat commun, YS Cho et al, 2016)
2. https://en.wikipedia.org/wiki/Posttranslational_modification
3. Epigenome Maintenance in Resopnse to DNA Damage (Mol cell. J. Dabin et al, 2016)