Towards solving the missing heritability in pharmacogenomics

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behorende bij het proefschrift

Towards solving the missing heritability in pharmacogenomics

1. The vast increase of available genetic data forced a shift in the field of pharmacogenomics towards approaches that are better capable at processing and interpreting these large amounts of data. *This thesis (introduction).*

2. Ultimately, selecting the right technology is not a matter of fact but a matter of choosing the most appropriate technique for the problem at hand. *This thesis (chapter 2).*

3. Compared to other genes, long-read sequencing results for pharmacogenes are superior with regard to variant calling accuracy and resolution of larger phased haploblocks. *This thesis (chapter 4).*

4. A continuous scale instead of a categorical approach improves predictions for CYP2D6 enzyme activity. *This thesis (chapter 5).*

5. The explosion in SNP discovery in pharmacogenetics emphasizes the need for databases that are frequently updated with respect to the nature and functional consequences of the different allelic variants […] *Ingelman-Sundberg M, Daly AK, Oscarson M, Nebert DW. Pharmacogenetics. 2000 Feb;10(1):91-3.*

6. Do not let the perfect be the enemy of the good. *Krebs K, Milani L. Hum Genomics. 2019 Aug 27;13(1):39.*

7. In order to fully understand and appreciate *CYP2D6* genetic tests, test options, their interpretation and limitations, and ultimately, integration of this knowledge into clinical action, a basic understanding of this highly polymorphic gene locus is invaluable. *Gaedigk A. Int Rev Psychiatry. 2013 Oct;25(5):534-53.*

8. Without careful consideration of the methods and biases embedded in a trained artificial intelligence system, the practical utility of these systems in clinical diagnostics is limited. *Dias R, Torkamani A. Genome Med. 2019 Nov 19;11(1):70.*

9. In science nothing is perfect. *Reviewer 3 (2021).*

10. When you think another language is strange, remember that yours is just as strange, you've just got used to it. *Anonymous.*