

The path to individualised breast cancer screening

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Stellingen behorende bij het proefschrift

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- 1. While common low risk variants are clearly associated with breast cancer in families, the polygenic risk score of the proband in such a family is a poor predictor of the polygenic risk score of the proband's relatives (this thesis).
- 2. Addition of the polygenic risk score to family history-based risk prediction has a large impact on screening recommendations for women with a negative genetic test-result as well as carriers of *ATM* and *CHEK2* pathogenic variants (this thesis).
- 3. Although women with an *in situ* breast carcinoma have on average a lower polygenic risk score than women with an invasive breast carcinoma, a polygenic risk score is predictive for in situ carcinoma as well (this thesis).
- 4. Only collaborative research in large consortia results in cancer risk estimates for rare genetic variants being precise enough for use in the clinic (this thesis).
- 5. Proper clinical interpretation of relative disease risks is not possible without information on absolute risks (this thesis).
- 6. Researchers should at least present the effect size of polygenic risk scores per unit standard deviation to enable meaningful comparison with other studies.
- 7. The lack of ethnic diversity in most published genetic studies to date hampers clinical implementation of polygenic risk scores in present-day societies.
- 8. We must standardise data acquisition in electronic health records of patients to enable proper use in clinical research and improve health care.
- 9. Because every individual is unique, early cancer detection programs will not be of equal value to everyone (Adapted from D. Crosby et al., Science 2022).
- 10. De eisen voor proefpersoneninformatie voor WMO-plichtig wetenschappelijk onderzoek leiden tot een verhoogde inclusie van hoog opgeleide personen.
- 11. Conclusies in de wetenschap worden gekenmerkt door zijn vele nuances, keuzes in de kliniek zijn echter voornamelijk zwart-wit.
- 12. De PhD student van nu kan niet zonder informaticus aan haar zijde.