Germline variants in the mismatch repair genes: Detection and phenotype

Suerink, M.

Stellingen behorende bij het proefschrift getiteld:

Germline variants in the mismatch repair genes: detection and phenotype

1. Testing for constitutional mismatch repair deficiency (CMMRD) should be considered in children with a phenotype reminiscent of NF1 but without an NF1 germline mutation, if at least one other diagnostic/clinical feature is present. (this thesis)

2. Small bowel adenocarcinomas are frequently mismatch repair deficient and newly diagnosed cases should always be analyzed for mismatch repair deficiency for the benefit of the patient and his family. (this thesis)

3. Well documented high-grade serous ovarian cancers should not be screened for mismatch repair deficiency. (this thesis)

4. Colonoscopic surveillance of PMS2- and MSH6- associated Lynch syndrome can be delayed until age 30-35 years. (this thesis)

5. Increased detection of cancer predisposition syndromes comes with an increased need for improved risk estimations and personalized surveillance protocols.

6. Interpretation of germline variants of unknown significance (VUS) in genes associated with increased cancer risk can be aided by molecular tumor analysis.

7. Mainstreaming of (onco)genetic testing requires improved knowledge of genetics from the specialists involved.

8. Dealing with bias is the largest challenge in (oncogenetic) research.

9. Teaching equals learning and should be part of any PhD trajectory.

10. Current publication pressure in the medical sciences has a negative impact on research quality.

11. It is right that we should stand by and act on our principles; but not right to hold them in obstinate blindness, or retain them when proved to be erroneous; a healthy sense of skepticism is therefore essential to practice science. — adapted from Michael Faraday, 'Observations on Mental Education' (1854)