

Cancer genetic testing and counselling on the move

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The field of familial cancer genetics is continuously on the move. In this presentation I will highlight three topics that are of growing interest. The first topic is the use of gene panels as a means not to only test for the genes that are related to the cancer that triggered referral, i.e. diagnostic testing, but also to screen all other genes on that panel for possible mutations. A recent development in the Netherlands is to perform whole genome sequencing of both tumour and lymphocytes in cancer patients; the latter with the goal to help selecting the somatic variants. However this germline WGS would offer the possibility to screen for a wide range of genetic conditions, relevant for patients and their families. There are many pros and cons to be considered.

The second topic is the mainstreaming of genetic testing, i.e. DNA testing being ordered by non-geneticist clinicians, e.g. medical oncologists and surgeons. Several models are now being tested in the field in various countries, typically for breast cancer. The third topic is the use of polygenic risk scores for cancer, as opposed to the traditional testing of genes for Mendelian, single gene, disorders. Something that was for many years regarded as being a predominantly genetic-epidemiological scientific exercise, is now being adopted for clinical application.