Recent changes to genetic testing

Samantha Crockett
Familial Cancer Specialist

February 2016

Background to the idea of familial breast/ovarian cancer

- Approx 5-10% of ovarian and breast cancers thought to occur within the context of family history/suspected genetic pre-disposition

- 1994 Identification of BrCa1 gene
- 1995 Identification of BrCa2 gene

- Genetic testing available to high risk families with multiple cases of breast and/or ovarian cancer

- Blood sample required from a relative with breast/ovarian cancer

Moving forward….

- 2004 NICE guidelines for Familial Breast Cancer
- Mostly with regard to risk classification and screening for in relation to breast cancer families

- 2013 update to NICE guidelines
- Recommend offering genetic testing to families with at least a 10% chance of finding a faulty BrCa1/2 gene
- Lowering the threshold for offering genetic testing meant more families were eligible
- Also consideration of offering genetic testing to unaffected individuals if no affected relative is available to give a blood sample.

Moving forward….

- 2014 Marsden study
- 200 women with a new diagnosis of ovarian cancer were offered BrCa testing irrespective of their family history
- 17% tested positive for a BrCa1/2 mutation

- ? More ovarian cancers are due to BrCa genes than previously thought

Moving forward….

- Genetic testing can now be offered to any woman with a diagnosis of (non-mucinous) ovarian cancer.

- Implications of a positive BrCa gene test result

    - Increased risk of breast cancer
        - Additional screening/ risk reducing surgery

    - Potential candidate for treatment with parp inhibitors – approved by NICE in Jan 2016 under specific circumstances

    - Implications for family – possibility of predictive testing for relatives

Moving forward….

Genetic testing is becoming part of the routine pathway in oncology in order to make treatment decisions.

Mainstreaming Cancer Genetics

- Making genetic investigations part of routine treatment
- Pilot studies done at several locations, gradually to be rolled out nationally
  - Genetic testing to be discussed by oncology team treating patient, not as a tertiary referral to a specialist genetic centre.
  - In Derby – Familial Cancer Service (FCS)