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Gynaecological Oncology News

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One in six ovarian cancers is positive for BRCA1 or BRCA2

Genetic testing justifiable for all patients with ovarian cancer

New Australian research suggests that 16% of all ovarian cancers test positive for BRCA1 or BRCA2. BRCA genes are hereditary and patients carry a 50% risk of ovarian and an 80% risk of breast cancer.

As part of the Australian Ovarian Cancer Study the samples of 1000 patients with ovarian cancer were analysed through gene sequencing.

Especially, ...

- 44.4% of women with a personal history of breast cancer were BRCA1/2 mutation positive
- 100% of women who reported that their mother had both breast and ovarian cancer were mutation positive
- 31% of women whose mother had breast cancer and 52% of women whose mother had ovarian cancer were mutation positive.

57.3% of mutation positive women did not have a family history suggestive of a carrier

The implications of these findings are:

Patients with BRCA mutations should undergo intensified efforts for surgical debulking. These patients may be ideal candidates for aggressive and high-tech surgery to remove all macroscopic cancer.

Patients with BRCA mutations respond better to platinum-based chemotherapy. This means that patients who developed recurrence of ovarian cancer can be re-treated with Carboplatin chemotherapy, which is normally well tolerated. Taxanes are less effective.

The survival chances of patients with BRCA mutations are better compared to patients who are BRCA negative.

Patients with BRCA mutations may become eligible for novel treatments. PARP inhibitors target the BRCA gene and miraculous responses have been observed in even previous “hopeless” cases.

Patients with BRCA mutations have also an increased risk of breast cancer and will benefit from intensified breast screening.

Newly diagnosed patients with ovarian cancer should be offered genetic testing for BRCA1/2.

Novel tumour marker HE4

HE4 offered by Sullivan & Nicolaides Pathology

HE4 is now routinely available from S&N pathology as an ovarian cancer tumour marker at a cost of $40. No Medicare rebate is available as yet.

The use of HE4 should be limited to:

1. Follow up after surgery: HE4 is only worthwhile in patients with stage 1 ovarian cancer whose CA125 was negative before primary diagnosis. In all other patients CA125 will be elevated should recurrence develop and therefore HE4 will not provide any additional value.
2. Triage of patients with pelvic masses: HE4 is superior to CA125 in triaging patients with an incidental finding of a pelvic mass who are asymptomatic and medically compromised. Some of those elderly and medically unfit patients can be spared surgery. Patients at high risk of malignancy still require surgical exploration.
3. Diagnosis of endometriosis: Endometriosis is positive for CA125 but negative for HE4. Patients with high CA125 and negative HE4 should be referred to a gynaecologist with an interest in endometriosis. Patients with CA125 and HE4 positive should be referred to a gynaecological oncologist.

HE4 should not be used for ovarian cancer screening as erroneously suggested in a recent article in The Australian. While screening for ovarian cancer would be fantastic if effective, this is unfortunately not justifiable at present.

New Website www.Obermair.info.

The website has been re-designed to become a resource not only for medical professionals but also for patients, family and friends who seek advice on uterine, ovarian, cervical, vulva and vaginal cancers.

It provides information on medical imaging (e.g., PET/CT, the use and limitations of tumour markers as well as familial cancer syndromes (BRCA1/2, Lynch). I also explain safety and quality procedures we routinely run in my practice, such as audit and the research program that I participate in.

My practice offers patients the opportunity to pre-register prior to their appointment to save time.

Please do not hesitate to give me a call if you wish to discuss an aspect of the above or a specific patient with me.

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