No screening for 'silent killer'

Despite being the leading cause of death from gynaecological malignancy in the western world, there is still no national risk assessment or genetic screening programme for ovarian cancer in Ireland. **Michelle McDonagh** reports

Emer Casey died of ovarian cancer in 2006, aged 28

> "There are approximately 334 new cases of ovarian cancer diagnosed in Ireland every year and 226 deaths from the disease

WHEN EMER Casey died of ovarian cancer in June 2006 at the age of 28, her family vowed to do all they could to help progress research into early detection of the disease known as "the silent killer".

Her mother, Juliette, believes that the youngest of her five daughters might still be alive today if there was an ovarian cancer screening programme in Ireland.

"My big concern is the complete lack of standardised screening in this country. I feel that ovarian cancer is a disease that's poorly understood and while great progress has been made in breast cancer, we are just not equipped to deal with ovarian cancer."

There are approximately 334 new cases of ovarian cancer diagnosed in Ireland every year and 226 deaths from the disease.

The symptoms are vague and non-specific meaning that often, by the time ovarian cancer is diagnosed, the tumour has spread beyond the ovaries.

A Women's Health Council (WHC) report published earlier this year highlighted the fact that the incidence rate of ovarian cancer in Ireland is 22 per cent above the western European average, while the death rate from the disease is 37 per cent higher.

According to the WHC, those diagnosed with the disease have only a 15 per cent chance of surviving beyond the first five years if the cancer is detected at an advanced stage. However, if it is detected at an early stage, the five-year survival rate jumps to 80 per cent.

A native of Youghal, Co Cork, Emer Casey had been working with the Dublin-based law firm, Matheson, Ormbsy, Prentice Solicitors when she was diagnosed with ovarian/uterine cancer on February 28th, 2006. She died less than four months later on June 10th, 2006, only four days after her 28th birthday.

Her mother explains that Emer was leading a very active life, busy at work and in her social life, when after complaining about a pain, she went for a scan which showed up an ovarian cyst.

"She was told to go back in six weeks but the pain got worse so we brought her to the doctor and she was operated on for an ovarian cyst. The operation appeared to go well but Emer went downhill rapidly. In the meantime, the cyst went to the lab and the results came back positive for cancer."

Juliette, her husband, Brendan, and their four daughters were totally devastated by the loss of the vibrant, lively Emer but her untimely death also had a huge impact on her friends, colleagues and everybody who knew her. During her lifetime, Emer frequently ran marathons for charities such as Goal and the street children of Calcutta and shortly after her death, her former boss, Padraig Madigan, announced the establishment of the Emer Casey Foundation.

The charity, which is largely run by Emer's family, is currently funding three PhD fellows to conduct research at the Discovary consortium, which is composed of researchers from several Irish institutions dedicated to carrying out world-class research in the area of ovarian cancer. Details of upcoming fundraising events or on how to donate can be found on www.emercaseyfoundation.com.

Director of pathology at the Coombe and chairman of Pathology at Trinity College, Prof John O'Leary explains that the Emer Casey PhD fellowship students have begun working on identifying novel serum markers to detect early ovarian cancer and are developing novel therapeutics for patients who no longer respond to chemotherapy.

One student is looking at the effect of low oxygen in patients with ovarian cancer and its impact on the development of chemo-resistant cancer. A second is working with O'Leary and Prof Dolores Cahill from UCD on an investigation into a unique signature protein which has been discovered in women with early and late ovarian cancer. "Our hope is to develop a blood test which could be used to detect early ovarian cancer," he says.

A third PhD student is looking at markers in ovarian cancer cells which appear to drive chemo-resistance and to significantly reduce overall survival and disease-free survival.

Master of the Rotunda Hospital, Dr Sam Coulter-Smith, consultant obstetrician and gynaecologist, says risk assessment services and genetic screening should be available to all women on a national and regional basis who are in a high-risk category. He points out that there are very few formal ovarian cancer screening services in place in Ireland where women could have their risk of developing the disease identified so that mechanisms could be put in place to reduce this risk and promote early detection.

Coulter-Smith has been running a risk assessment service at the Rotunda since 1997 for women with a strong family history of breast and ovarian cancer which are strongly linked.

"If a woman is unfortunate enough to carry one of the genetic mutations that we know causes ovarian cancer, their risk of developing the disease is very high, between 20-80 per cent depending on the type of mutation so obviously anything we can do to reduce the risk is very important.

"The most effective way to prevent ovarian cancer is to remove the ovaries while they are still healthy, which reduces the risk to about 1 per cent, a serious risk reduction. The downside is that if a close relation developed the disease in their 50s or 60s, you would need to have your ovaries removed in your 40s and go straight into menopause at this age."

Coulter-Smith says there is now a greater awareness about the fact that this "faulty gene" can be inherited and the number of people presenting for genetic screening has increased.

"The problem is that BRCA-1 and BRCA-2, the two primary genetic mutations linked to ovarian cancer that we know about, only account for about 25 per cent of mutations that cause genetic ovarian and breast cancer so even with screening, we are only identifying 25 per cent of faulty genes."

His advice to people with a strong family history, ie two or more close relations with breast or ovarian cancer, is to seek advice from their GP and see if they need to be referred to an ovarian screening service.