MedicalXpress

Health of entire families at risk through under-use of genetic testing

June 7 2013

A new study of the use of genetic testing for cancer-causing mutations in affected families in France has found that its take-up is very low. Professor Pascal Pujol, Head of the Cancer Genetics Department, Montpellier University Hospital, Montpellier, France will tell the annual conference of the European Society of Human Genetics tomorrow (Sunday) that analysis of data from the French National Cancer Institute covering the years 2003 to 2011 showed that, although there had been a steady increase in tests performed for the breast and ovarian cancer-causing mutations BRCA1 and BRCA2, this was not the case with the MMR mutation, implicated in Lynch syndrome (a form of colorectal cancer). Only a third of relatives of individuals with either mutation underwent genetic testing themselves.

"Given that such testing can provide many options to enable individuals to manage their cancer risk, it is vital to encourage awareness and acceptance among both the public and medical professionals", he will say. "For example, removal of the ovaries in women over 40 years old who carry a BRCA mutation decreases their overall cancer mortality by 20% and prophylactic mastectomy can reduce the chances of breast cancer in women carrying such a mutation by around 90%. Those who are unwilling to undergo prophylactic surgery can benefit from increased surveillance, with regular MRI (magnetic resonance imaging) scans. For familial colon cancer, screening by colonoscopy has been shown to decrease mortality. It is therefore regrettable that so few people seem to be aware of the benefits of genetic testing in families with a history of breast, ovarian, or colorectal cancer."

Professor Pujol and colleagues from cancer centres across France analysed 240134 consultations and 134652 genetic tests from patients referred for a predisposition to breast or colorectal cancer. They found a substantial increase in tests for BRCA1/2 – from 2095 a year in 2003 to 7393 in 2011 – but for MMR mutations the increase was tiny – from 1144 to 1635 a year over the same period.

Mutations in BRCA1/2 genes are thought to be responsible for about 5% of all cases of breast and ovarian cancer. A woman with such a mutation has a risk of up to 87% of having breast cancer before she reaches the age of 80, as opposed to a risk of 8% in the general population. Such cancers are diagnosed at an average age of 43, as opposed to 60 in the general population, and are often more aggressive. In the case of ovarian cancer, a woman carrying a BRCA1 mutation has a risk of ovarian cancer of up to 63%.

Individuals with Lynch syndrome, or hereditary nonpolyposis colorectal cancer, have a 45% risk of developing colorectal cancer by the age of 70, and women with the syndrome are at increased risk of endometrial and ovarian cancers.

"While the increase in BRCA testing is encouraging, it is far from optimal. And the uptake of MMR testing for Lynch syndrome - responsible for 5% of all colorectal cancers - is frankly disappointing", says Professor Pujol. "And of course, positive test results may have implications for other family members.

"While we have only studied the situation in France, we believe that our findings would be likely to be replicated in many other countries across the world. It is extremely worrying that such a simple test, which has the potential to spare whole families from devastating illness, is being so under-used. We urgently need a major programme of awareness among all those concerned, involving medical education and training,

information programmes for patients and their families, public health campaigning, and improved genetic counselling," he will conclude.

Provided by European Society of Human Genetics