PREVENTION IS BETTER THAN CURE

Tena Walters, Consultant Breast Surgeon MBBS, FRCS, MS

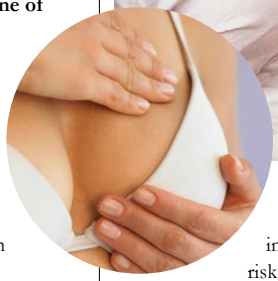
A study at the Royal Marsden Hospital in 1940 first suggested that breast cancer could run in families, but it wasn't until 1994 that the first genetic abnormality, which caused breast cancer was identified.

Approximately 5% to 10% of the 4 million breast cancers diagnosed in the world, per year are due to genetic mutations. Approximately 1:400 people in the population carry the abnormal gene.

More than half of all women with breast cancer, who are found to have an abnormal gene do not have a family history of breast cancer, but currently only patients with breast cancer who are considered to have a greater than 10% risk of carrying one of the abnormal genes is offered testing. This is based on their age, type of cancer and family history.

Patients are offer testing if they have one of the following criteria:

- One first degree relative with breast cancer younger than 40 years of age
- One first degree male relative at any age
- One first degree relative with bilateral breast cancer, when the first was diagnosed younger than 50 years old
- Two first degree relatives or one first degree and one second degree relative with breast cancer at any age



However, a recent study has suggested that it would be more cost effective to offer testing to all women with breast cancer, which would identify more gene carriers and their relatives, who would benefit from preventative therapy.

Testing patients with a family history only means that 50% of gene carriers are missed. This is because there may be a lack of awareness of the significance of family history by patients and clinicians leading to restricted access, and underuse of genetic testing services with only 20-30% of eligible patients tested. This means that 97% of carriers of an abnormal gene in the population remain unidentified.

If patients are known to have an abnormal gene then they may, like Angelina Jolie opt to have a risk reducing mastectomy with reconstruction. Patients who have breast cancer may opt to have a mastectomy instead of lump removal and a risk reducing mastectomy on the other side with breast reconstruction. This may also reduce the need for radiotherapy following lumpectomy. They may be also eligible for different, new preventative drugs. Most importantly, it is now possible to test embryos for the genetic mutation and this could prevent the gene from being passed on the future generations.

Identification of gene carrying relatives could allow those individuals to have selective and advanced screening techniques with MRI and tomo synthesis mammography or risk reducing surgery.

Prevention is better than cure and patients with breast cancer should discuss genetic testing with their clinicians. ■

For appointments please call 01622 873058. For more details call Tena's PA Helen on 07982 249612.Or email tenakerrywalters@gmail.com

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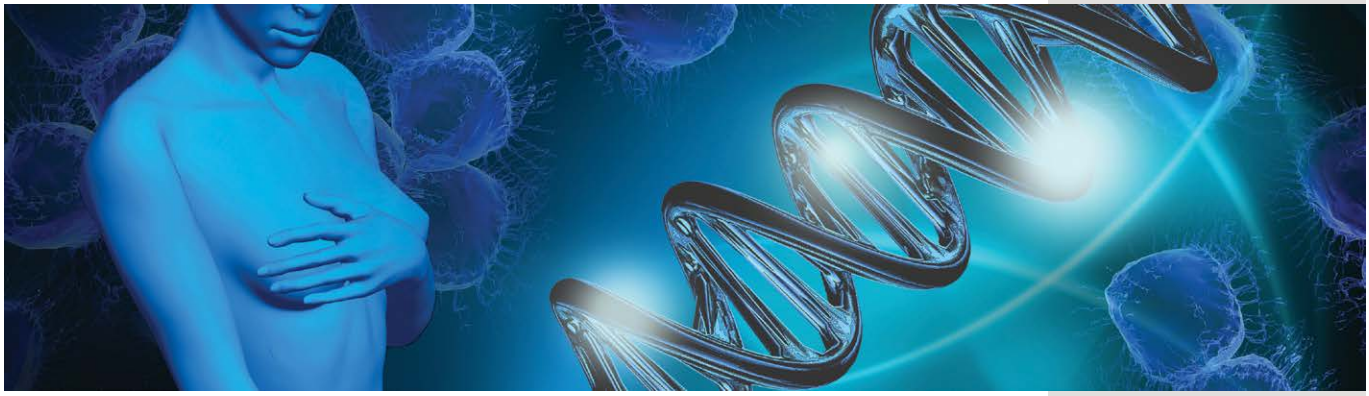


COLORECTAL CANCER MAKING IT'S UNPOPULAR DEBUT AMONGST THE YOUTH

As a consultant colorectal surgeon, I have come across a fair share of haemorrhoids (piles), fissures, and other benign anorectal conditions which are extremely common affecting over 30% of the population. Patients who are diagnosed with these conditions present symptoms such as bleeding, pain, itching, irritation, a swelling or lump or discharge from the anus. These symptoms however, can also be the presenting symptoms of anal, rectal and even colon (large intestine) cancer as well as polyps (benign pre-cancerous growths) and inflammatory conditions such as colitis or Crohn's disease.

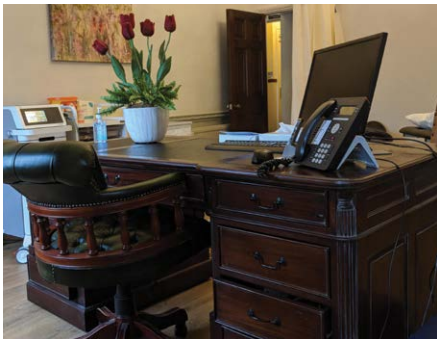
With 40,000 new cases of colorectal cancer (CRC) diagnosed every year in the UK, equivalent to a lifetime risk of 1 in 17, the NHS states that, 'Bowel cancer is one of the most common types of cancer diagnosed in the UK.'

CRC is commonest in the elderly, majority being over the age of 60 years, and the numbers are low in the younger age groups. However, in recent years there is increasing evidence that shows the incidence is rising more steeply in the young, especially in 20-29 and 30-40 age groups in Europe and USA. The cause of this is not known but obesity may be a factor. More than 50% of CRC diagnosed in under 50 year olds are symptomatic suggesting symptoms are being ignored resulting in late diagnosis.

In a majority of cases a clinical examination is conducted which would include a rigid sigmoidoscopy (small endoscope) by a specialist colo-proctologist. This would help the consultant to diagnose the benign condition, reassure and start treatment immediately. However in some cases further investigation becomes necessary post the initial consultation. This would involve a colonoscopy or CT virtual colonography to examine the entire large bowel if the diagnosis was not clear at first consultation.

As most consultants would suggest, early diagnosis is essential and leads to a cure. With survival rate of CRC at 57% and mortality at 43% of which over 50% are considered preventable and were diagnosed late, it is imperative that patients keep an eye out for any early warning signs and symptoms no matter what age they are at.

Mr Emin Carapeti is a Consultant Colorectal Surgeon with a special interest in management of anorectal conditions and endoscopy. He has published widely on anorectal disorders and pilonidal sinus disease and has pioneered new surgical techniques in complex recurrent pilonidal disease and day case surgery for haemorrhoids. ■



Mr Emin Carapeti,
Consultant Colorectal Surgeon at
The London Rectal Clinic,
108 Harley Street, London
W1G 7ET.

THE DETAILS

The London Rectal Clinic
108 Harley Street
London
W1G 7ET
To book an appointment contact our team on
0207 563 1234
info@108harleystreet.co.uk

For more information, visit us at
www.108harleystreet.co.uk