GastroIntestinal Endoscopy

newsletter the insider

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Celebrating 30 years

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Our Mission at GastroIntestinal Endoscopy is to deliver experienced and accessible endoscopy services with the highest quality of healthcare standards to improve the health outcomes of patients and the communities we serve.

GIE operates an Open Access Endoscopy Service from four locations:

- **SUNNYBANK** – Brisbane Endoscopy Services
- **CHERMSIDE** – Chermside Day Hospital
- **EVERTON PARK** – North West Private Hospital
- **AUCHENFLOWER** – The Wesley Hospital

Dr Neville Sandford

Lynch Syndrome

The cause of Colorectal cancer (CRC) is multifactorial with both environmental and genetic influences, and although sporadic disease occurs in 70–80% cases, in the remaining 20–30% cases, heritable factors are important.

Lynch Syndrome (LS) is the most common cause of inherited CRC accounting for 3% of newly diagnosed cases, and was named after Dr Henry Lynch, the first author of the original 1966 publication that described the familial clustering of CRC with stomach and endometrial tumours and termed this condition the "cancer family syndrome". This was later changed to "hereditary nonpolyposis colorectal cancer" (HNPCC) to differentiate it from familial adenomatous polyposis (FAP). In the early 1990's mutation of the DNA mismatch repair (MMR) genes was found to be the cause of this autosomally dominant condition, and in many cases genetic testing can confirm the diagnosis at a molecular level and identify at-risk individuals who require screening. The current term Lynch Syndrome is used in preference to HNPCC as most patients will develop one or several adenomatous polyps which makes the term "nonpolyposis" misleading.

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www.gastros.com.au

Colorectal Cancer

The lifetime risk of CRC in LS is dependent on sex and also the MMR gene mutated and varies from 30–74% in MLH1 and MSH2 gene mutation carriers, to 10–22% with MSH6 mutations and 15–20% with PMS2 mutations with generally higher risk in males than females. The mean age of diagnosis is 44–61 years compared with 69 years for sporadic CRC and tumours occur more commonly in the right hemicolon. There is also a higher risk of metachronous tumours. The precursor lesion is often a flat high-risk adenoma (with villous histology or high-grade dysplasia), and the adenoma-carcinoma sequence is more rapid than in sporadic cancer (3 years compared to 10–15 years). This is the rationale for a shorter surveillance interval between colonoscopies (1–2 years) than for patients with colonic polyps without LS (3–5 years). Although LS CRC is more frequently poorly differentiated, LS patients have an improved survival, stage for stage, compared to sporadic cancer.

Extracolonic neoplasms

LS patients also have a significantly increased risk for a wide range of extracolonic malignancies. This is highest for endometrial cancer (54% in MLH1 and MSH2, 15% in PMS2 and 71% in MSH6), but also cancer occurs in the urinary tract, ovary, stomach, hepatobiliary tree, small bowel, brain and skin (cutaneous sebaceous neoplasms). A small increased risk for pancreatic, breast and prostate cancer has been reported in some LS kindred.

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Changes to access for Capsule Endoscopy

In 2014 amendments were made to the Medicare Benefits Schedule for Item 11820 Capsule Endoscopy removing age restrictions and the requirement that the qualifying upper gastrointestinal endoscopy and colonoscopy be performed in the previous six month period.

The updated guidelines for Capsule Endoscopy are as follows:

Capsule Endoscopy to investigate an episode of obscure gastrointestinal bleeding, using a capsule endoscopy device if:

(a) The patient to whom the service is provided:

i. Has recurrent or persistent bleeding; and

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Clinical Diagnosis

In 1990, the Amsterdam Criteria were established to diagnose HNPCC. This required the presence of three confirmed cases of CRC in non-FAP families, one of which was a 1st degree relative of the other two, in at least two generations and with one case aged less than 50 years. This was widened in 1999 to include the extracolonic cancers: endometrium, small bowel, ureter or renal pelvis. Now other tumours in the ovary, stomach, hepatobiliary tract and brain are included. The Bethesda Guidelines have been developed to identify at-risk individuals who deserve investigation for LS. (Namely: CRC <50years, synchronous or metachronous CRC or LS-tumours, CRC with LS-type histology <60years, patient with CRC with 1st degree relative with CRC or LS-tumour < 50yrs, patient with CRC with CRC or LS-type tumour in two 1st or 2nd degree relatives).

LS is applied to patients or families in which the genetic basis can be linked to a germline mutation in one of the DNA MMR genes which maintain the fidelity of the DNA during replication by correction of nucleotide base mis-pairs. The LS cancers develop only after a second hit occurs within somatic tissue which causes total loss of DNA MMR activity in the cell and subsequent microsatellite instability (MSI) and cancer development. Patients with MSI high tumours can have immunohistochemistry testing of their tumours, checking for loss of expression of the DNA MMR genes. Testing of tumour tissue can be done on archival formalin-fixed tissue from affected family members to see if LS is likely. Family members can then be referred for genetic counselling for consideration of germline testing. If a genetic mutation is then found, other family members can be checked for the mutation to determine if they require surveillance. If no mutation can be found, LS cannot be excluded so all family members would require ongoing surveillance.

Management of Lynch Syndrome patients

Screening for CRC by colonoscopy is recommended in LS patients or 1st degree relatives of LS patients every 1–2 years from age 20–25 years or 5 years before the youngest age of diagnosis of CRC in the family. Patients with CRC or unresectable polyps should undergo colectomy with ileorectal anastomosis.

Although the level of evidence for benefit is low, screening for endometrial and ovarian cancer should be offered to female patients with annual pelvic examination and endometrial sampling and transvaginal ultrasound from age 30–35 years. Hysterectomy with bilateral oophorectomy is recommended for women who have finished childbearing or from age 40.

Screening for gastric cancer should be considered with upper endoscopy and gastric biopsy every 2–3 years and for cancer of the urinary tract with annual urinalysis from age 30–35 years, although the strength of evidence for these guidelines is low.

Routine screening for small intestinal, pancreatic, breast or prostate cancer is not recommended.

There is some evidence that chemoprevention with aspirin may be of benefit in LS patients, and this should be considered after discussing the potential risks with the patient.

Image courtesy of Lynch Syndrome Australia www.lynchsyndrome.org.au

Reference: Gastroenterology 2014; 147:502-526

Checklist for Open Access Endoscopy Referrals

Including sufficient detail in the referral letter will assist in the bookings process to provide an efficient open access service to you and your patients.

- ☑ **Patient is aged between 16 and 85 years**
- ☑ **Procedure/s requested**
- ☑ **Indication for procedure/s**
- ☑ **Current medications**
- ☑ **Current medical history/ comorbidities**
- ☑ **Allergies**
- ☑ **BMI <40**

For referral templates go to: www.gastros.com.au or call us for more information.

ii. Is anaemic or has active bleeding; and

(b) An upper gastrointestinal endoscopy and a colonoscopy have been performed on the patient and have not identified the cause of the bleeding; and

(c) The service has not been provided to the same patient on more than two occasions in the preceding 12 months; and

(d) The service is performed by a specialist or consultant physician with endoscopic training that is recognized by The Conjoint Committee for Recognition of Training in Gastrointestinal Endoscopy; and

(e) The service is not associated with balloon endoscopy.

GIE offers Capsule Endoscopy services at our four sites as well as St Andrew's War Memorial Hospital. Capsule Endoscopy is not available on an open access basis but can be arranged directly through Dr Michael Miros (Ph: 07 3801 2233) or Dr Roderick Roberts (Ph: 07 3831 2704).

The Incidence and Mortality of Colorectal Cancer is Increasing in People Younger than 50

Dr Andrew Bryant

Colorectal cancer (CRC) is a disease with an incidence that increases with age but over the last few decades its rate is falling in the highest risk groups (i.e. those aged >50). From 1975 the incidence of CRC fell by 22% and mortality fell by 26%. About half of this is due to screening while the other half is due to better and earlier investigation of symptoms and lifestyle improvements. From 2000 to 2009 the incidence fell in those >50 by 2–3% year but over the same period the incidence rose by 2% per year in those aged <50. At the end of the decade the incidence in the older group fell from 180 to 150 per 100 000 (down 17%) of the population but increased from 5.9 to 6.6 per 100 000 in the younger group (up 12%).

While most CRC still occurs in the older population, 11% of colon cancer and 18% of rectal cancer occurs in the younger group. Unfortunately CRC tends to present later in young people leading to more advanced disease at diagnosis and lower five year survival rates. The cancers in young people tend to have more mucin and also may have signet ring cells. They are less differentiated. They tend to be more left sided. About 20% of cancers in young people are due to known family cancer syndromes such as Familial Adenomatous Polyposis (FAP) or Lynch Syndrome but the rest are due sporadic cancer or unknown risks which may be genetic or relate to acquired risks such as inflammatory bowel disease.

It is important to enquire about family history of polyps and CRC. This is especially important if multiple relatives are involved or a close relative was also young at presentation. It is also vital to investigate symptoms such as rectal bleeding, pain and change of bowel habit. It is also important to investigate anaemia appropriately, especially iron deficiency.

The youngest person I diagnosed with CRC was a 23 year old otherwise well man who reported rectal bleeding to his astute GP. Other family members were clear so his tumour was probably sporadic. More recently I diagnosed a woman in her late 20s with multiple liver metastases from a sigmoid primary. In her case the delay in diagnosis occurred because she was in South East Asia when she started having bowel symptoms and it was thought to be an infective process by the doctors she consulted then.

The take home message is that while CRC in the young is rare it still happens, the incidence is rising and due to late diagnosis, they don't do well. Investigate those with concerning symptoms or test results and start screening early in those with strong family histories.

For more information:
www.mayoclinicproceedings.org/article/S0025-6196(13)00822-7/fulltext

Frequently Asked Questions Dr Neville Sandford

Q With patients investigated via open access endoscopy, who is responsible for pathology review and recommendations?

A In all cases the results of the pathology from patients having Open Access procedures is reviewed by the gastroenterologist performing the procedure (or another Partner of GIE if the proceduralist is a locum or on leave), and in addition a copy of the pathology is forwarded to the referring doctor. Once the pathology has been reviewed, if there is any change to the follow-up recommendations as printed on the procedure report, an amended report with new follow-up recommendations will be sent to the referring doctor. If significant pathology or unexpected results are received, the referring doctor will also be contacted by phone. All patients at the time of their scheduled follow-up will be sent a reminder, and if this is not acted upon, the patient will be phoned. A recent medico-legal case where a malignant polyp was not followed up appropriately by either the proceduralist (at a Public Hospital) or by the referring GP and where both were found culpable, highlights the fact that the responsibility of review of the pathology and the appropriate follow-up rests with both parties.

Q What do you recommend for screening family members of bowel cancer patients <50 years of age?

A Once genetic syndromes such as Familial Adenomatous Polyposis, MUTYH Polyposis, or Lynch Syndrome are excluded (see attached article on Lynch Syndrome), family members should commence screening colonoscopies from age of 10 years younger than the affected family member's age at diagnosis. Subsequent colonoscopies should be done five-yearly unless polyps are found which may necessitate earlier review.

If you require A5 referral pads, please contact one of our four locations below. Electronic referral templates can be downloaded from our website www.gastros.com.au

GIE practice locations and contact details For all appointments, call 1300 4 GASTRO (1300 4 427876)

Brisbane Endoscopy Services

Suites 16–18
McCullough Centre
259 McCullough Street
Sunnybank QLD 4109

Phone: 07 3344 1844
Fax: 07 3344 2739

Chermside Day Hospital

Level 1
Chermside Medical Complex
956 Gympie Road
Chermside QLD 4032

Phone: 07 3120 3407
Fax: 07 3120 3443

The Wesley Hospital

3rd Floor, East Wing
451 Coronation Drive
Auchenflower
QLD 4066

Phone: 07 3870 3799
Fax: 07 3870 5069

North West Private Hospital

Endoscopy Unit
137 Flockton Street
Everton Park
QLD 4053

Phone: 07 3353 3322
Fax: 07 3353 9325

Private practice locations and contact details

DR RODERICK ROBERTS MB BS FRACP AGAF
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