Familial Adenomatous Polyposis (FAP)

ConvaTec

What is it?

Familial Adenomatous Polyposis (FAP) is a hereditary bowel condition. FAP was previously known as Gardner's Syndrome or Familial Polyposis Coli.

Incidence

FAP is an uncommon gene dominant disease affecting about 1 person in 8,000.

Signs and Symptoms

1. **Bowel polyps**
   In FAP large numbers of polyps are usually found on the lining of the colon (large bowel) and rectum. Polyps are usually benign (non-cancerous) lumps that can occur in other parts of the body. People without FAP can also have polyps in the colon and rectum but the important feature of FAP is the large number of polyps (hundreds and thousands) that are present. In FAP these polyps are adenomatous polyps, which means that if they are left untreated they will become malignant (cancerous).
2. **Skin cysts and lumps on bones**
   Both of these are harmless in themselves.
3. **Congenital Hypertrophy of the Retinal Pigment Epithelium (CHRPEs)**
   Most people with FAP have this eye condition which does not affect their vision. On close eye examination small clots are seen on the back of the eye.
4. **Desmoid Tumour**
   These are soft tissue growths that can occur inside or outside of the abdomen and can invade surrounding tissues. Most occur inside the abdomen. About 5-10% of people with FAP are affected. It is more common in females than males (3:1 ratio) and treatments may include surgery, radiotherapy, chemotherapy, hormone treatment and non steriodal anti-inflammatory drugs.

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Gene Dominant

Genes determine such things as hair and eye colour and they come in matching pairs – one set from each parent. FAP occurs because of a fault on a gene; you only need to have the gene from one parent to have FAP because it is a dominant gene.

It also means that someone with FAP has a 50/50 chance of passing it on to their children.

Diagnosis

There are several ways that a diagnosis can be made:

1. **Family History**
   This is checked to identify any other family members who may be at risk of having FAP.
2. **Genetic Testing**
   Copies of genes are carried in different cells including blood cells. A blood test can be used to show whether someone carries the FAP gene and which part of the gene is faulty. Most Health Authorities have a team of people working in genetics. This team will include a genetics counsellor who you could talk to, to obtain help, advice and information about FAP that will be specific to your child and your family.
3. **Bowel Examinations**
   To look at the colon and rectum to check for polyps.

Methods

Colonoscopy – Insertion of a longer flexible tube into the bottom, allowing examination of the inside of all the rectum and colon, under general anaesthetic/sedation. A biopsy will be taken.

Screening in FAP

Screening involves regular check-ups which will include physical examinations and colonoscopies. This is so that the status of the disease can be checked, and any treatment advised can be given as soon as possible.

Treatment

If polyps are found, the surgeon will take a biopsy to be examined under the microscope. If they are benign they can be left and regular screening continued. If they are malignant, the surgeon will recommend an operation. Some families choose to have an operation as soon as polyps are found whether they prove to be benign or malignant.

Surgical Treatment

The aim of surgery is to remove the part of the bowel that is or may be affected by FAP. There are several operations that may be considered and the surgeon will discuss the advantages and disadvantages of each one and which one will be best for your child.

1. **Ileo-Rectal Anastomosis (IRA)**
   This can be done if polyps occur in the colon but not in the rectum. The colon is removed and the ileum (the last part of the small intestine) is joined to the rectum. This means that the child will open his/her bowels in the usual way but the movements maybe more frequent. It also means regular screening of the rectum for polyps will still be required.
2. **Pan-Procto Colectomy**
   This is done when the rectum needs to be removed. The whole of the colon, rectum and anus is removed. The end of the ileum is brought out onto the surface of the abdomen. This is called an ileostomy. Bowel motion passes through the illeostomy into a bag worn on the abdomen.

3. Ileo-Anal Pouch (IAP)

The colon and rectum are removed. A pouch is made from the last part of the small intestine. The pouch is joined to the anus. The pouch gradually takes over the function of the colon and rectum. The Surgeon may advise a temporary ileostomy while the pouch heals.

Future Possibilities of Treatment

Gene Therapy – researchers are investigating ways of replacing or repairing faulty genes.

Drug Treatment – research continues to try to identify drugs that can stop polyps occurring.

At present the only option for treatment is surgery to remove the parts of the bowel that are affected.

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PSNG
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