QUESTION TIME

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Can you be an organ donor with Polyposis syndromes or leave your body to medical science?

This will not stop you doing so but they will look very specifically at you. As the conditions do not affect the kidney, heart, lungs corneas etc. then there would not be any reason to be stopped donating.
Leaving your body to medical science is not an option today as research tends to be carried out on live bodily fluids – like blood, bile, poo samples from live people.

Inheritance - What is the risk factor to a child of a parent with FAP?

The FAP gene is autosomal dominant and this implies there is a 50% chance for each child. This does not mean that if you have 4 children then 2 will definitely have Polyposis but this is the risk.

Genetic Testing – what level of information should be given?

This gives the position in the gene of the mutation and St Marks discusses this with patients as there is a relationship between the genetic spelling mistake and the condition diagnosed. You should be referred somewhere to get the level of information you require.
An individual's condition might not be categorized easily because it depends on other information as well.

What research is being done on the faulty gene?

The problem is that the faulty gene is in every cell in the body. This gene mainly affects the bowel and is a code for a protein that is broken and that leads to steps in forming a cancer, but it is in every cell in the body. The need is to replace the gene at conception. IVF creates embryos which are tested to ensure there is no polyposis and then the embryo is implanted knowing it will stop the disease being passed on.

CRISPR is gene editing on sperm, eggs and embryos – removing the faulty gene and replacing it, but as this changes the genetic make-up of the person - this is unethical and illegal at present, but this might change.

Genetic Testing - POLE gene

The POLE gene has recently been associated with multiple adenoma. The POLE gene mutation is extremely rare and therefore the data is pretty poor.
Only 70-80 people worldwide have this mutation and the numbers are not there to know much about the association. The APC gene is a more common mutation.

How long does the process take to do family genetic testing?

A family goes through cascade gene testing. This begins with taking the full name and their date of birth and using this information on family members to build up the database. GPs are then written to about the family members risk and to invite members for testing.

Genetic testing takes about 3-6 months (diagnostic testing looking at the whole genome – at about £500), but predictive testing where they know which gene is at fault then it might only take 4-6 weeks for a confirmation because of the recorded familial connections, (£250). These prices are changing rapidly as it becomes more mainstream.

How soon should colonoscopy be done in Peutz-Jeghens Syndrome?

Colonoscopy should be done at age 8 if the child is well (earlier if there are symptoms). The surveillance is pan-enteric. If there are no symptoms then it does not have to be repeated until 18. If there are symptoms then the scoping should be repeated every three years.

Does Chemo prevention work in PJS?

Chemotherapy (oral selective mTOR inhibitors) is being researched because there have been some successes. Research has been done to slow down the growth rate of PJS polyps. Work needs to be done to show a reduction in childhood polyps and intussusception.

What is the inheritance risk for PJS?

PJS is a dominant inherited condition so the risk to inherit is 50%.

How often does endoscopy have to be carried out in Juvenile Polyposis Syndrome?

If only a few polyps a year this can be managed endoscopically. If there are a lot more polyps then more frequent scoping and if controlled then the scoping can be at more extended intervals. In some patients there is a "carpeting of polyps" and then surgery may be required but this will be the minority of JPS.

Can you opt out of elective surgery?

It is a choice whether you take the specialist advice. You have to know that if a FAP patient progresses to cancer there is a 50% chance that they will not survive.
If a patient decides to not have the recommended surgery, then we have to ask do they have capacity – are they capable of making a rational decision or have they impairment to make a decision. If this is true, then they will be assessed by a Health Professional for their own interest.

If a J-Pouch failed after many years -can another one be made?

Questions: why has the pouch failed and is there enough room for another join? You can create another one further up the small intestines provided there is enough material to join to. One consideration is why has the pouch failed – if it due to polyps forming then they will form on the new pouch as well. Remember that it takes half metre of bowel to make a pouch – and we have about 3 metres so two pouches will use a third of the small intestine.
Duodenum removal uses a third of the bowel so after several operations a lot of bowel could have been used up. If the surgery has to be repeated, then further replumbing uses up more of the bowel each time. The question has to be asked - if it has failed once will a pouch not fail again?

Do a high percentage of J-pouch patients have complications?

Less than 10% - nearer 8% of pouches are removed. This is a difficult operation because of where the surgery takes place; there can be damage to the bowel, ureters, bladder, pelvic nerves. Normally 1-2% will have erection/ejaculation problems; with removal of a pouch this rises to 25%. The anus is removed with the pouch and this can lead to scarring in an area difficult to heal. Every removal of any structure means that there is less small intestine to work with. Try and only do when you have to. Removal is not often done as 90% of pouches will remain in situ.

Can the pouch be improved with diet, exercise etc?

Not really.
Pouches fail due to leaks and infections after surgery and having a normal weight and healthy lifestyle will only help with healing. (Pouches are mainly seen in ulcerative colitis).
Work needs to be done to discover why polyps develop in pouches as we don't know why but it may be due to contact with substances that are not normal for the tissue to be exposed to, especially the bile content which can be different in Polyposis syndromes. This is being researched.

What is life without a colon like?

Can you have alcohol? YES
If you don't have a large bowel – you don't need the fibrous food used for movement through the bowel. With an IRA there is frequent pooing, with a pouch even more frequent and with an ileostomy is even more frequent and liquid. These foodstuffs can speed up transit which can be a problem with an ileostomy and can create the equivalent of gastroenteritis all of the time. Meals should probably be little and often, working out ways to stop the bag filling up. Dehydration can become a problem and therefore there may be a need for fluids and electrolyte supplements. The colon moves water and salts back into the blood and therefore there has to be a careful watch on dehydration.

Fundamentally there is nothing that a Polyposis patient cannot do – although for some, it might take a little more planning. So, by enlarge there is nothing that they cannot do.

With an IRA, the general advice is not to have too much leafy vegetables. Some will need Fybogel soluble fibre (grainy substance) to make bulk, to make the consistency gloopier and less liquid.
A call for all patients undergoing endoscopic procedures – no fibre beforehand.

Can lifestyle and diet reduce polyps?

Smoking is banned in polyposis because of its detrimental effects on the body and links to cancer.
FAP is genetically determined so generally lifestyle and diet will not have any effect, but diet may have subtle effects. Turmeric is the one mentioned at the moment but there have been no studies linking it to cancer prevention.
There is no study on any drug that has had an advantageous effect on polyps.
In serrated polyposis, vitamin D has been shown to have some effect. Some fish oil and celecoxib (COX-2 selective NSAID) can reduce polyps and make them disappear, but this causes a problem, including heart attack side effects. Polyps cause problems when they become quite large, with the bigger ones becoming higher risk. Patients rectum needs to be removed in high risk patients (elderly), drugs can make polyps go away but then they come back with cancer.

But reducing the size of a polyp with a drug does not reduce the risk altogether. In the US drugs are especially given to young patients to reduce polyps. If there are no polyps there is no sign – there is no enlarging polyp to say there is a problem. More polyps, more risk, more cancer and this is the problem with drugs – they might reduce polyps but they do not reduce the cancer risk. The polyps are a warning sign and they give a warning brief which can be watched.

How successful is endoscopy/colonoscopy?

The miss rate is variable. Especially poor results if the bowel prep is poor and then things can be missed. There is different success with different types of polyps and their removal: lumps on a stalk (easy), flat lump (difficult), serrated polyposis (much more difficult).
In JPS and PJS it is more obvious but FAP can be variable. The danger points on scoping are when the polyps are behind the folds or just going around the corners.

Historical data shows that there is a 30% missed rate but this is based on old data and knowledge of what to look for is constantly improving and the cameras are so much better.
One approach is to bring back earlier if the scoping is not giving clear results. Please no sweet corn! Bowel pre is the most important factor in giving clear results even with meticulous endoscopic techniques, nothing will be seen if the bowel has not been prepared properly.

What is the rate of small bowel cancer?

Why isn't Pillcam (is innovative capsule endoscopy solution provides clear images of the colon to support detection of polyps with a non-invasive, patient-friendly device) given to all patients specially to look at the duodenum? There is no advantage in doing this, as there are not many polyps seen here in FAP. Mainly occurs at site of stoma and in the duodenum or jejunum. Don't follow the US very closely – as we treat differently.
Thyroid cancer is more common in FAP but this can be treated and is curable. US they treat very aggressively but we believe there is no advantage in doing this. This can do more harm than good – and this is the same for the small bowel. Identifying thyroid cancer early does not affect the results, as it is treatable with surgery.

How would you treat a 13year old girl with MAP?

MAP is recessively inherited and therefore both copies of the gene will be faulty.
How do polyps behave? It takes quite a while to become cancer. Supercharged ones would grow more quickly. The procedure interval used depends on this. Treatment is the same as with FAP with small polyps removed every three years, but the treatment and investigative procedures have to be personalised due to the size and number of polyps.

Can silver nitrate be used for polyps on a stoma?

Most patients with a stoma do not have FAP (but ulcerative colitis). Inflammatory (granular tissue) polyps form where the delicate bowel meets the sticky appliance. Then stoma nurses use silver nitrate to burn these off. In FAP you can get polyps on the small bowel because of constant contact with poo and bacteria, which is not normally in contact with this tissue.

Polyps can be cancerous so remove and collect tissue and send to pathology, can be an inflammatory polyp or real polyp. These are removed in theatre as they bleed a little. Removal by snaring rather than cauterising.

Are piles linked to polyposis?

Piles are not linked to polyposis but to being human. Two groups of people – people with piles and people who haven't got them yet! Tends to be because we are upright.
Colonoscopy needed to check the remaining large bowel. Bleeding will not suggest anything sinister if up to date with colonoscopy.

Are there plans for extending the registry?

Can become anal about the Registry!
Research only as good as the information put in, and therefore we are fastidious about the way we keep the information. The patients get classified as FAP only if it is a known mutation but there are other different categories including unclassified polyposis that are recorded. The Registry is very accurate about the diagnosis and if it is not clear that is how it is recorded. In NZ and Scandinavia there is compulsory national registering but this is not what we are recommending.

There are also really accurate and verified records of polyps and desmoids.

We are careful to only include patients who have had some of their care here at St Marks. We work with a network of specialists, where surgery can be done in most places. But in problem cases there might be a greater need for an in-depth diagnosis. We are aware that criteria can be different at different centres.

This is not a research data base but one of really good clinical information to enable the best day to day management of Polyposis Syndrome Patients.

We need to ensure that all these specialists are up to speed with the latest approaches. By joining together, we have made an App which gives both the European and International Guidelines and we need your help in taking this out there to GPs and Hospital Clinicians that you come across. The App contains diagnostic and clinical information with guidelines on the recommended treatments.

We need to ensure others are using these guidelines. Ideally we need to have 10 centres up to speed using the guidelines and we need to join together and this should be a political priority and this is where the Registry is hoping that PolyPeople can help.