4th Annual FAPGene Family/Information Day Report

Swarkestone Sailing Club Saturday March 28th 2009

We are pleased to report that the event was another very successful day. Numbers beforehand indicated around 46 would be attending but we expected less allowing for the unpredictable nature of FAP. So in the end 38 found it a most interesting and rewarding place to be given the unpredictable weather forecast.

Mick welcomed both old and new visitors and also gave a brief over view of the day. Several had asked how Bonnie a young lady who many had met last year at Swarkestone was. Unfortunately Bonnie passed away shortly afterwards through secondary liver cancer but she will always be remembered for her vitality and hopes for raising awareness of FAP. Parts of a letter from Bonnie's mum were read out including her wish that everyone would enjoy a day which they had found so rewarding the year before.

Apologies were sent by Krystle Konteh of The Genetic Interest Group due to family circumstances and Dr Alison Meltcalfe who was away but sent a report on The Family Talk Project. Mick then introduced John Roberts FAP Gene's Chairman for his Introduction to FAP talk.

For some reason we have always been unable to attract a speaker locally for this talk. However it is understood that FAP is a rare condition and whereas several local Health Professionals deal with patients and Families with FAP it is usually only for the immediate problem and they do not need to know all the intricacies of the human plumbing world as affected by FAP.

John formed his talk around what we now call The Verona Article (A 4,000 word article written by John and Mick on The Role of an FAP Support Group for The Rosa Gallo Foundation in Italy and printed in a Springer Publication in December 2008). The two main operations were shown with diagrams explaining what was and wasn't removed in each case. Also included was a bit of Family History and how rare hepatoblastoma, a form of childhood liver cancer was in those with FAP. A warm response was given to Johns talk and questions seemed to reflect mostly on the frequency of bowel motions with the different operations. More of that later.

With a 30 minute space Mick then gave an update on The Family Talk Project from Dr Alison Metcalfe at Birmingham University. All the results are in but until they are accepted and published through the appropriate authorities little could be said about them. However Alison thanked those who had contacted her to take part following her talk in 2008. A total of 33 families including seven with FAP were included in the study. Alison has also expressed an interest in seeing everyone at the proposed Chat Day in September 2009.

Details of the Facilitating Network Project from Krystle Konteh of The Genetic Interest Group were given and Mick impressed that this was a three year project and FAP Gene were delighted to be one of the seven groups chosen. Krystle is also interested in the Chat Day. Mick mentioned a previous project from GIG which looked at Insurance for Rare Diseases and hoped that this would eventually benefit all 130 member groups of GIG.

Then it was the turn of Jo to give a really interesting talk on her family and how FAP had involved them. 20 years ago Jo's mom had died at the age of 34 leaving seven children aged between 5 and 15. Since then several of the family have been diagnosed with FAP and had surgery. One has had problems with a Desmoid Tumour and the lack of knowledge of FAP is shown in the following taken from her talk

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"What really does stick in my head is taking her to A&E a few weeks after removal of her Desmoid Tumour complete with stomach pains.
The Triage Nurse asked for PMH. The inevitable…'Whats FAP?' Another Nurse…the same. The Dr…the same….all never heard of it…yet the Dr decided these stomach pains were unrelated and she was sent home with the advice 'take painkillers'. Luckily it was noting to do with the FAP…but how was this diagnosed???

I have more respect for the two consultants that removed the Desmoid that didn't mind telling us that they had to do some research prior to the op and it was completely new to them".

We have always supported projects around the UK involving patients with FAP. This year was the turn of 'Sculpting a Voice for Genetic Disorders'. Julia Thomas is an artist with a research background in bioscience. After having gone back to college to further her studies in Art & Design, she now wants to combine the skills and knowledge of both her scientific and artistic backgrounds. In particular, her mission is to work with health related issues and health/science organisations to inspire artwork that will help to raise awareness for those affected.

She told of the talks she had with those with genetic disorders and how she then expressed those thoughts in sculpture. Several images were shown and those present were invited to chat with Julia afterwards about helping with this very intriguing project.

Lunchtime was when we all enjoyed the buffet supplied by Claire at the Sailing Club. Jacket potatoes and the famed steamed pudding with custard. When in Verona giving a talk Mick mentioned that the Family Day was worth attending if only for these puddings. Even translated into Italian it raised quite a peal of laughter from the audience.

On the more serious side this was the time for CHAT and many views exchanged about the many variances in how FAP can affect individuals. That it lasted almost 25 minutes longer than planned didn't really matter for our planning is always quite loose to say the least.

During the afternoon we had two speakers from St. Mark's FAP Registry. Kay Neale the registry manager gave a talk about frequently asked FAP questions and Dr. Ash Sinha a research Fellow, who is conducting research on Desmoid Tumours in patients with FAP.

This was Kay's third time at Swarkestone and she still makes the genetics of FAP sound interesting enough top keep everyone's attention. There were quite a few smiles when she explained how gene are like a recipe and how a fault in the gene of a single letter can make such a change. The case in question was a recipe for a Ham Sandwich. The letter changed was the H in Ham to a J. Hence a very surprised customer with a Jam Sandwich.

Dr Ash Sinha gave a history of the development of the FAP registry from its beginning in 1924 and how the knowledge of FAP has increased. The positive effect of genetic screening, so that surgery can be used on members of a family with FAP at the appropriate time has greatly reduced the number proceeding to develop bowel cancer. He also spoke about research into the treatment of desmoid tumours which are non-cancerous but at the moment the most difficult of the extra colonic lesions to deal with. They only affect around 20% of those with FAP and in only 2% of those do they cause serious problems. For those in this 2% with the more aggressive form it could be comforting to know the problem is recognised and a solution being looked for.

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So after five hours it was time for those thus inclined to sample a ride on the rescue boats and others to say their farewells hopefully loaded with information and a better knowledge of this rare condition.

Mick had mentioned about the lack of a national registry and how it seems the Regional Genetic Centres always seem to quote the number of families with FAP, not individual numbers. He explained this was possibly because family members are treated by their nearest Genetic Centre often in other parts of the UK and numbers diagnosed not passed back to the original source.

John was asked about the childhood liver cancer, hepatoblastoma, which his son George was found to have. He explained it was very rare and might affect a child up to the age of five. It can be found by ultrasound scanning but although this has been asked about for all under 5's it is not seen as a viable approach considering the numbers it would find.

One of the main discussions during the day was the frequency of toilet visits. It was amazing that people who had never met before were willing to chat openly about what is to others an almost taboo subject. It actually varied from four to twenty four times a day in those present. This was despite trying different diets and medicines. Most had keys for disabled toilets 'RADAR KEYS' and cards stating their need to use toilets in an emergency due to bowel problems.

Another point of interest was not absorbing coated tablets due to the removal of the colon. This seemed too often to be not understood by some GP's but hopefully will improve. Finally a mention of tiredness and the vitamin B12. Several had regular injections of B12 and found a great improvement in concentration and energy afterwards. They are usually given every 12 weeks and a simple blood test shows if there is any deficiency.

All the speakers were presented with book tokens as a thank you for giving their time and all agreed that Swarkestone 2009 was an outstanding success whether a patient, relative, friend or health professional.

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