My Genetic Journey by Mick Mason From the *ia Journal*

It is nearly four years since I had an operation, which resulted in my permanent ileostomy. This happened after around 30 years of mild but persistent ulcerative colitis. The operation was necessary because of multiple polyps and I was a bit astounded to hear the consultant say at my final colonoscopy "I've never seen anything like it in my life". I think it was then that my wife Ann and myself realised the full extent and were not totally surprised to hear after the operation that they had found a tumour and I had cancer of the colon.

Whilst in hospital the words Familial Adenomatous Polyposis were mentioned for the first time. Luckily, although I hate abbreviations, it is commonly called F.A.P. (sounds like it's straight from Thunderbirds).

This is where at the age of 56 my Genetic Journey began. My father and an Aunty also had colon cancer and it was suggested I saw a Professor Trembath in the Department of Clinical Genetics at the Leicester Royal Infirmary. It was mentioned that it could take quite a while. If I had the gene it was then a 50/50 chance that each of our children could be affected and if so virtually certain that polyps would eventually appear and at least one turn cancerous.

In November 1999 we had a home visit from Valerie of the Genetic Clinic. I had seen a television programme about a couple who had to decide whether to have a genetic test or not and was instantly reminded of it. Going through a family history was quite a test of memory but only concerned those on my father's side of the family.

We remembered a lot but afterwards had a chat with my brother and sister to complete the tree. It soon became apparent that if the test was positive it could affect around 50 relatives, right down to grandchildren of second cousins.

The next step was a chat with Professor Trembath and the mention of an eye test. This wasn't to look for anything dangerous but for freckles at the back of the eye, another guide to a final decision.

Things were then quiet until January 2002 when I had the eye test but no freckles were found. I thought this was a good sign and although not the final answer was starting to think all was OK. It was a surprise then when within a week I was asked to see our Professor and was told I did have the faulty gene. The strange point was it usually starts to cause problems in the early teens. Another bit to our strange family.

Now I know it wasn't a fault that would cause the certain death of a relative like some genetic disorders but it was a blow all the same. However if relatives decided to have a blood test the results would be known in about six weeks.

I was asked if I would contact 13 relatives on my father's side and was glad I had been on the *ia's* Visitors Course for what I had learnt certainly helped me to talk to my relatives on rather a sensitive issue. They were invited to ring the clinic if they wanted.

A harder task was telling our three children. The thought now is will I feel guilty if I have passed the gene onto my children? They say I shouldn't and pointed out it could have started anywhere in the family history. I'm not sure and only time will tell. Even if they do not have a test details of a possible genetic condition will be on their medical records.

I had seen on the Internet about cysts and boney bits on the jaw being related to this gene and from the age of 11 had about 120 cysts removed from all over the place.

This led to being asked to see another friend of the Prof. who would take an x-ray of my jaw. It was a relief when none were found and I was given some free advice that a back tooth was rather large and my sinus cavities also large. This would mean if the tooth were ever extracted I would have to have an operation. Also a visit to a dermatologist, again about my cysts was referred to as Gardner's Syndrome. This has led to another two consultants examining this strange person and a bit of minor surgery to remove a few this year.

So I have ended up with my ileostomy, which so far for a three year old has behaved quite well and been on a journey that has been fascinating yet worrying. Obviously at times it has been eased by the knowledge that a positive result would not affect my own health and there are far more serious genetic faults, which others have to cope with. Also when my wife Ann is not around to give a cuddle there is always my 'Therapy dog' called Friday who is my daughter's seven-year-old Newfoundland.

It is now November 2002 and one of my immediate family has the gene and multiple polyps in the colon. Soon a chat to the surgeon will take place about an operation to remove the entire colon, which will prevent colon cancer. It wasn't until a few days after receiving the news that I realised how it had affected me. For a few days I had become a bit 'broody' and I suppose withdrawn, which is most uncommon for me. With the help of my wife Ann I have realised that these things happen and now look to the future and the support other members of our family might need.

Like so many things in life everyone at some time has to make decisions and whilst advice and help can be offered only one person can make the final decision. The feeling of guilt has gone but in some ways I still feel responsible.