

GORLIN SYNDROME

SUPPORT GROUP

News and Views - January 2006

Dear Group Members and Associates

Warm winters greetings to members new and old, from the armchair at the side of the fire. Hoping this newsletter finds you well and able to make the most of the shorter colder days.

It was fantastic to see so many people at the conference. As always we had many opportunities to share and discuss our fears, concerns and experiences with each other. Meeting members from all ends of the country and making new friends with members from as far away as Sweden. Keeping Jim's vision, it's so good to talk, especially to someone who has shared the same journey and knows and understands how you feel.

Many thanks to our medical speakers who gave us their time, energy, knowledge and patience to keep us updated with the current issues and treatments.

Sorry to go on so for those who could not make it, but need to say special thanks to Sonia for relaying Oliver's story and to Jane and Helen for so bravely telling the story of their Dad, their hero. Not a dry eye in the house and a well deserved standing ovation. Thankfully Jasmin dried our tears and cheered us up with a very talented guitar accompanied song.

During the AGM the following committee changes took place Secretary, Margaret Costello (re-elected). Maureen Gregory takes over from Helen Mulligan, many thanks to Helen for her support as Treasurer over the past three years. The post of chair currently held by Jim Costello is now held by me, Sally Webster (previous vice chair). I accepted the role of chair with mixed emotions of sadness, inadequacy and honour; I just hope I can do Jim proud. Jim was voted unanimously to be known as founder member of the Gorlin Syndrome Group, I hope you all support and agree with this most fitting tribute to Jim.

Further to the conference, we have been asked to look at organising an update meeting south of Manchester to enable easier access for members. We aim to explore this at the next committee meeting.

I hope that you all had a peaceful and relaxing Christmas. On behalf of the committee all the very best wishes for 2006. Take good care.

Sally Webster - Vice Chair

Hello from Australia

My name is Nicole and I have Gorlin Syndrome as well as cleft pallet. I'm a bit shy but would like to chat to anyone who is interested. I am 28 yrs of age live in Adelaide, been married 7 yrs now and have 2 beautiful girls who can be pain in the necks at times but make me smile everyday.

I would like to speak to anyone who has Gorlin Syndrome or anyone who would like to chat as I'm a friendly down to earth kinda person and enjoy having a good chat time to time.

Nicole dolphin013@bigpond.com

Message from Prof R Gorlin

A 60 year old with Gorlin in Seattle with documented cysts and abundant BCCs was talked into \$60,000 of dental work. She was given her diagnosis and at the time had 2 obvious cysts. Of course, the bridgework had to come out within 2 years. Do we not tell patients that only temporary stuff should be inserted? Does your literature address this?

Love Bob

It does now Bob. Thank you for the advice and lovely to hear from you.

Calling all teenagers!



Calling all teenagers! Take a look at the website. This is your chance to chat and share experiences with people of your own age. <http://groups.msn.com/BccnsGorlinsTeens>

Skin Information Day – Sat, 24th Sept 2005 at Queens Park Hospital in Blackburn was attended by Sally and Margaret who were on hand to pass on information about Gorlin Syndrome to people who attended. Many nurses from Dermatology Units in Lancashire attended and were pleased to receive information about the condition and existence of the support group.

The **All Party Parliamentary Group** on Primary Care and Public Health is undertaking an inquiry into prescription charge scheme. LMCA has written to the All Party Group stating that the current system of charges and exemptions is over due for reform, highlighting the anomalies that exist. LMCA has urged the MPs on the Group to consider the risks people with long term health conditions may face if they discontinue treatment on cost grounds.

Annual Conference

The conference proved to be a popular event with about eighty five people in attendance. It was good to see so many there.

Professor Peter Farndon and Mr John Cornah unfortunately were unable to attend. Peter was unwell on the day and John was called to theatre to carry out an emergency procedure. We were delighted to welcome Professor Gareth Evans, Dr Ernest Allan and Dr Anne Cook who all gave excellent presentations. Prof Evans talked about Gorlin Syndrome and its many characteristics and admirably filled Prof Farndon's 'slot' to speak about the genetics associated with the condition, whilst Dr Allan updated on PDT and Dr Cook reported on the findings of the Royal Manchester Eye Hospital study undertaken a couple of years ago.

Photo Dynamic Therapy

Dr Ernest Allan, Christie Hospital, Manchester

Results of first 338 patients – Sporadic BCCs and Gorlin BCCs. We know that Gorlin BCCs respond to PDT in exactly the same way as sporadically occurring BCCs. A number of patients required several treatments to eradicate their lesions, but for 20 there appeared to be little benefit from PDT and 13 of the sporadic BCCs were ultimately treated by radiotherapy and 7 treated by surgery.

The success rate was therefore 94%. This is at least as good as most other forms of treatment but we are looking at ways of improving the effectiveness still further and reducing the numbers of treatments sometimes required in order to produce complete tumour resolution.

Another problem is that it is difficult to treat many lesions at one time. This is because it is difficult to keep the sensitising cream in place and also to achieve pain control during treatment. We have therefore undertaken a pilot study using an intravenous sensitizer called Photofrin. Of 10 patients treated, 4 have had complete resolution of their lesions and in 6 there has been considerable improvement. The treatment is painless and it is possible to treat thicker lesions by means of insertion of a laser fibre into the lesion. The initial results are therefore encouraging. A few patients have had brisk painful reactions following treatment but there have been no long term side effects. We are treating more patients with Photofrin but I think that it will take a little time before we achieve the excellent results reported by the Americans.

Summary of Review of Patients with Gorlin Syndrome

Dr Anne Cook - Manchester Royal Eye Hospital.

Purpose: Review patients with Gorlin Syndrome (GS), documenting presentation, referrals, treatment patterns, and associated morbidity.

Methods:

Cross-sectional review and retrospective data collection of 40 patients with Gorlin Syndrome. Patients from the GS support group were invited to be examined. Those that were unable to attend were questioned via telephone or post. Demographics, presenting features, associated pathologies, and treatment modalities were recorded. Demographic data, age at presentation and diagnosis, spectrum of ophthalmic and peri-ocular disease, treatment modalities used, and peri-ocular deformities developed.

Results:

40 patients were included.

Age range = 8-72 years.

A variety of medical practitioners were involved in the care of these patients. In 16 patients, jaw cysts were the presenting features. In 12 patients, BCC was the presenting pathology. A wide range of other presentations were recorded. 29 had developed BCC's, 16 did so before the age of 30, 8 before 20 years.

Conclusion:

Multi-disciplinary care is essential in the care of the patient with GS. Early diagnosis of GS may allow for skin protection and surveillance at an earlier age. Early aggressive treatment may reduce peri-ocular morbidity. Co-ordination of a referral 'network' may improve the efficiency of referrals and management planning.

The Features and Genetics of Gorlin Syndrome

Professor Gareth Evans – St Mary's Hospital, Manchester

It has been a number of years since Prof Evans last attended the event but it was a pleasure to welcome him back and listen to his very informative and clear presentation on the features and genetics of the condition. Both are well documented but patient feedback was positive, reporting a better understanding in these areas.

Following this session patients were allowed the opportunity to raise issues and ask questions of the three Doctors in attendance.

Whilst parents were busy in the main conference room the children were fully occupied and enjoying activities in the Art and Craft session kindly provided by Michelle Hughes. All were very proud of their Christmas card creations when they were able to show them off to all in the main conference room.

For those people resident at the hotel there was the opportunity to get to know each other whilst relaxing in the restaurant at dinner or over drinks in the hotel lounge.

Thank you to all who contacted us with your appreciation and feedback. Here's to the next event!

National Blood Service

Committee member John Mills:

I have been informed by the National Blood Service (NBS) that I will not be able to give blood because I have 'ongoing' treatment for recurrent/multiple basal cell carcinomas (BCCs). They said that they gave the same response to another Gorlin patient last week. They informed me that this rule would apply to anyone who has ongoing treatment for BCCs. The rule applies for all donors with no exceptions for individuals. [A one-off BCC for which the patient had been treated and discharged would not be a problem.] The rule is in place to reduce the risk of infections being passed on through the blood (e.g. infection from the BCC /broken skin/rodent ulcer). Not clear on the risk to me as the blood donor.

GS website indicates that people with the Syndrome CAN give blood. I have explained this to NBS and they said that it will probably have to be changed. I have asked NBS to send me a letter clarifying this issue so that we can use appropriate wording on the website.

Contact has been made with Profs Farndon and Evans. Peter's response 'This is news to me, and I agree with John it seems over the top. I think the best thing is to wait for the letter John has requested and then for Gareth and/or myself to talk to the blood people.'

NHS Reforms and health equality

The Socialist Health Association has organised six meetings around England (the first in Ipswich in late January) to consider the effects of proposed health service reform on health inequality. For more details of the events and a booking form, see the association's website. www.sochealth.co.uk

Worth noting!

NICE Guidance for the treatment of patients with skin cancer will be launched in January 2006 meaning Gorlin syndrome patients are designated a special group. Commissioners have a duty to make sure that all the necessary treatments are available to treat the skin lesions in patients with Gorlin Syndrome and that the necessary expertise is available to administer these treatments.

Do Once and Share

This is a six month project funded by NHS Connecting for Health (Programme for IT in the NHS) and is aiming to create an Integrated Care Pathway for Genetics. The project is in Southampton but the idea is that it can be shared once completed with other clinical genetic units to help harmonise the information and treatment offered. It forms one of many project teams based around the UK who are developing pathways for all types of services that the NHS provides. Once the project is completed the information will be used by NHS Connecting for Health to help with their ongoing IT programme to integrate services at both a local and national level.

A care pathway helps guide patients through the "patient journey", assisting them to access information and care about their condition at appropriate times.

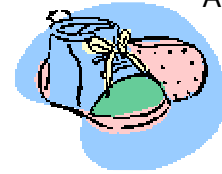
Jeans for Genes

Special thanks go to all who contributed to the success of the 2005 appeal. Many gave up time raising awareness in schools whilst others contacted businesses for support.

And finally! Thank you for the many Christmas Cards, best wishes and the generous donations to group funds. Best wishes to all for 2006.

Until the next time!

Congratulations go to



Anne and John Mills on the arrival of their first child a girl.

Yolanda arrived early and weighed in at 7lb 13 oz.

Christmas lights

News from Cornwall came in the form of Christmas lights and a request from Jason and Sandra Knuckey of Newquay. Each year Jason decorates their home with festive lights and this year was no exception. In March Shania their 4 year old daughter was diagnosed with Gorlin Syndrome and they wanted to make this years display special and raise funds for the Gorlin Syndrome Group.

Family and friends attended the switch on with David White from Radio Cornwall flicking the switch. Local businesses supported the event offering raffle prizes and a collection box was placed to enable passers by the opportunity to donate. The event featured in the Cornish Guardian. Thank you Jason and Sandra, your efforts are appreciated.

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