

GORLIN SYNDROME

SUPPORT GROUP

News and Views - March 2004

Dear Group Members and Associates

Sending a sincere welcome to all newsletter readers old and new.

Many thanks to members here in the UK and across the seas who attended the conference in October helping to make another memorable event. In addition, all members who were unable to attend due to health or personal problems we were grateful for your wishes and support.

Again we were very privileged to have our visiting guest speakers and representatives from our medical advisory panel, which proved an invaluable part of the group question and answer session. It was lovely to meet so many new members and their families and to be able to share concerns and worries but also to have some fun and relaxation together. In particular we had many young people attending with their parents, who were able to meet and enjoy some time together and make new friends. Jim's absence was noticeably missed, but we did our best to continue the legacy he has left for us. We had our low points and I kept expecting Jim to turn up and be there for us as he always was. I think we did him proud and thanks for all those who helped and supported me, especially Margaret, Jane and Helen.

Sally Webster - Vice Chair

Annual Conference – Saturday, 25th October 2003

The event took place at Park Hall Hotel and Conference Centre in Chorley, Lancashire. We are extremely grateful to Professor Peter Farndon, Dr E Allan, Dr Kumar, Mrs Dot Wilkinson and Chris Baker for their input, and to committee members for their help with the various workshops on the day.

Feedback from delegates has been extremely positive. Patients and their families welcomed the opportunity to meet others with similar issues and to share experiences in relaxed and comfortable surroundings. The focus groups were also welcomed, as was the opportunity to join in the question and answer session with the medical advisors in attendance.

Some concerns were raised about the facilitation of one of the focus groups, with delegates pointing out that it is wholly inappropriate for facilitators to target individuals and expect those individuals to take on board the facilitator's personal beliefs. The Gorlin Syndrome Group Committee would fully endorse this principle and ensure that all facilitators are fully briefed in relation to this issue.

The children and their families were able to take advantage of Park Hall's generous donation of Camelot Theme Park tickets, and despite poor weather conditions enjoyed the 'thrills' of all the rides and attractions. Many of the children also took part in the hotel's colouring competition. One young member Jacob Wareing was rewarded for his efforts by winning first prize consisting of a boxed set of cars. Well-done Jacob!

Annual General Meeting

The Annual General Meeting was held on the afternoon of the Annual Conference. The minutes of the last AGM were agreed and accepted as a true record. Two new members were co-opted

onto the committee Mrs Maureen Gregory and Mrs Carolyn Deveney. We would like to take this opportunity to welcome them. Accounts for year ended 2003. Treasurer Helen Mulligan was unable to attend because of health problems. Summary of accounts below:

Gorlin Syndrome Group

For Year ended 31st March 2003

Balance brought forward		£ 7,000.52
Income		
- Donations, grants and interest		£ 15,487.50
Outgoings		
- Administration costs - telephone, internet access, postage, stationery etc	£ 970.08	
- Annual Conference costs	£ 1,695.20	
- Donation to Genetic Research Fund	£ 5,000.00	
- Conference fees/subscriptions	£ 110.45	
		£7775.77
	Total	£ 7775.77
		Balance £ 7711.73

Priorities / Plans for 2004 – 2005

At the February meeting the committee reviewed objectives outlined in Constitution. Objectives are as follows:

1. The relief of sickness and the protection and preservation of the health of persons affected by Gorlin Syndrome and any related condition and their families and carers.
Existence of telephone help line, newsletters, email/web site facilities, medical advisory team, production of adult/children's leaflets, involvement in national initiatives and supporting families to attend meetings demonstrate a commitment to achieving this objective.
2. The advancement of the education of the medical profession and the general public into Gorlin Syndrome and its implication for the family.
The web site, input to medical conferences, along with appropriate mail shots to regional genetics units demonstrate a commitment to this area of work.
3. The promotion of research into the causes, effects, treatment and management of Gorlin Syndrome and to disseminate the useful results thereof.
Involvement in studies, i.e. Royal Eye Hospital, Clinical Genetics Unit, on-going support and donation to research unit demonstrate a commitment to the objective.

Discussion followed in relation to the support of patients and the appropriate use of funds. The following expenditure was deemed acceptable for the coming financial year:

- Continued use of funds to support the telephone help line, broadband for email and web site facilities
- Production of updated adult and children's leaflets
- Mail shot of updated leaflets to regional genetics units
- Continued use of funds to support families to attend the annual meeting
- Attendance of committee members at appropriate regional and national conferences to support the continued work of the group.

We are happy to hear from anyone with suggestions as to how we can best meet your needs. We are also pleased to receive articles or questions for inclusion in the newsletter. Let us have your comments, articles or suggestions. Please feel free to email, write or telephone with your ideas.

Paul Holroyd is renowned for his poetic skills. Paul's poem is dedicated to his good friend Jim Costello who passed away Dec 2002.

James

*9th December was the terrible day
When our good friend Jim went away
It was an awful way for us to part
As Jim had touched all our hearts
Nothing was too much trouble for Jim
As anyone would know who knew him
A patient man full of wisdom and kind
Had a problem, Jim wouldn't mind
He would help you with any worries
Though he had plenty of his own
You could always ring him for a chat
Or be made welcome at his home
You see they say time is a healer
And this thing time goes on and on
But this makes things no easier
Now our good friend Jim has gone
Though Jim lives on in our memories*

Helen Costello wrote this (aged 16) some nine years ago following a hospital visit for the removal of a jaw cyst.

Ode to Gorlin Syndrome

*Another x-ray taken, another cyst on my jaw
Wouldn't be as bad if it wasn't so sore.
So it's another hospital visit and operation for me.
It's all over; I just wonder when the next will be.
The nervousness of waiting on the ward to go down
Feeling stupid in that awful hospital gown.
Its another tooth lost and more missing bone,
The blood and the sickness and more stitches sewn.
Now I'm recovering from my ordeal,
Oh how lousy, sick and tired I feel
From the anaesthetic and tablets they shove down
Is it any wonder my face wears a frown?
But after all the suffering, worry and pain
I realise it's important to go through it again.
Better to grin and bear and put up with the strife
I **will not let Gorlin** rule my life!*

£20.00 well earned!

Committee member Maureen Gregory recently spent a Sunday morning being a patient for Doctor's exams. The arrangement was that doctors were told to look at Maureen's skull x-ray, the scarring on her face and back, and the palmar pits on her hands. Between 10 and 20 extremely nervous doctors took part as she read the Sunday papers and feasted on tea and muffins over a 4 hour period- a very easy £20 and a way of spreading the word in the South – It astounded her that more than a handful actually made the correct diagnosis!!

Royal Society of Medicine Skin Cancer Conference – Birmingham Sept 2003

Attended by Dermatologists and Plastic Surgeons the group was represented by Margaret Costello. A question and answer session followed Margaret's presentation 'Diagnosis and self awareness'. Margaret gave patient feedback and outlined treatment priorities, inequality of access to treatments, and concerns relating to the use of radiotherapy in Gorlin Syndrome. Feedback has been positive. Other invitations have been extended to Margaret, the first of which will be at Queens Medical Centre, Nottingham in March 2004. We have also been approached to organise a National Study Day and are currently looking into the feasibility of doing so.

National Service Framework for long-term conditions

Results of the consultation to inform the NSF for long-term conditions, led by LMCA and the Neurological Alliance, have been published. Key concerns focus on the very basics of services for people with long-term conditions. People are let down by basic failings, for example:

- access to health and social services are poorly coordinated,
- people should be at the centre of planning the support they need, they should be provided with full information about their condition and the services available, and
- the attitudes and awareness of service staff will need to change to see things from the perspective of the service user.

The report, along with a summary, is available on the LMCA website at www.lmca.org.uk.

An article by Gemma Fox

email gfox2@student.uscm.ac.uk

“You look like a guinea pig!” My brother’s reaction after my first operation, I was thirteen he was ten. I remember thinking what will my friends say when they see me like this. I didn’t think about my health and how it was for the better. I’d had an operation to remove two cysts from my jaw and a hole in my gum to allow a tooth to be pulled through which had fallen to the bottom of my gum. Can you imagine my friends’ faces when I told them why I was going into hospital?

Five years, four operations, twenty cysts and around fifteen teeth later one of the removed cysts were tested and it was found that I had ‘Gorlin Syndrome’. I’d no idea what ‘Golden Syndrome’ was (this was what I told my mum it was called when my doctor told me what I had). Being a fan of the Internet I decided to search for information. I was amazed to discover it is a genetic disease. To think that my mum or dad carried the gene and had passed it on to me not knowing they were carriers. It is even scarier to think that I risk giving it to any children that I may have in the future. It was a shock to see how rare the disease is. Why had I to be the 1 in 50,000 people to have the disease? I guess that is what anyone would think.

I was so pleased (and quite relieved) to discover a support group for Gorlin syndrome sufferers. To be able to talk to others knowing they are going through similar difficulties to you helps you deal with things more easily and reassures you that you are not alone. After visiting the annual conference for the first time and hearing people talk about being a family (from all over the world) made me realise that something good comes out of anything bad. It’s nice to be in contact with people who know exactly how you feel because they are going through similar things.

Sometimes I wish it would all go away. I get sick of having to go for regular six-month checks and the consultant telling me how many cysts have grown since my last visit. It is made easier by having a friendly sympathetic doctor. Just before I go under anaesthetic, he always comes to talk to me, holds my hand and reassures me. Usually at that time I am crying and shaking with fear.

Last year’s operation was one of the worst. I had four cysts removed. It was the 12th of June (three

days after my twentieth birthday). I’m always nervous the night before any operation. I did not get much sleep and was already awake when my mum came to my room at 5:00 am, just as I had to get up for breakfast because from 6:00 am it’s nothing to eat. This is big downfall with having operations on your mouth; **YOU CANNOT EAT PROPERLY FOR AGES!!!**

The time leading up to the operation hunger kicks in but it’s after the operation when starvation hits. You can’t eat much as not only is your mouth swollen but also your gums are still extremely delicate and tender from the operation and the stitches. The only solution is to suck blended food through a straw. This is a long process and not very attractive for your family and friends to witness. You would not believe the number of straws that I have used over the years. My mum always buys a box of thirty before my operation. Being like this makes you feel incredibly helpless. I could not move around very much as I was still incredibly weak. I had a self medicate machine attached to my left hand. This machine was my best friend. It removed all my pain in short bursts. However one of the side effects of morphine is that it makes you scratch your nose constantly. What with my swollen mouth and always scratching my nose I really did look like a guinea pig!

Being summer it was warm in the wards so I used a rolled up magazine as a fan to cool me down. Big mistake! I was talking to one of my friends, lost concentration for a moment and hit my mouth with the magazine. The pain I felt for the next few hours was unbearable; I had constant tears running down my eyes for ages. I learnt never to do that again.

I wish that there were some sort of therapy or medication to take to reduce the growth of cysts. It is so frustrating having to go into hospital at least once a year for operations.

During the recovery period I get sick of being uncomfortable, unable to eat, sleep or go out because of the way I look. People also look at me with either pity or disgust. It is these reactions that make me feel so ugly. This is not my fault so why should I be treated any differently to any one else? I am still a human being.

I am sure many of you will relate to this. Thank you Gemma. We are always happy to receive items for the newsletter.

Apologies to everyone who has struggled to locate the Gorlin Syndrome web site. This has been due to on-going technical problems and a move to a new service provider.

**Visit the updated and newly launched Gorlin Syndrome Web Site www.gorlingroup.co.uk
Your feedback and comments are welcome.**

Also note new email address gorlin.group@btconnect.com

Greetings from Kent and grateful thanks to Margaret for delaying the newsletter until I got around to reporting my recent (and less recent!) attendances!!

As a 'burnt-out nurse' it is a great honour to have the time (and energy) to be a patient representative of the Gorlin Syndrome Group at healthcare events down here in the South. Two years ago I was still having panic attacks when I ventured out and last March I was somewhat over-awed by my surroundings at Westminster's Church House. One year on and I have spent 9 days in London representing the group although I feel sure that dear Jim would be amazed by the length of time I can keep quiet, without managing a 'dig' about the 'postcode lottery', which persists in many areas of healthcare.

I feel sure my NHS burnout was the result of the frustration of trying to nurse from the patient's perspective. Consequently it is good to hear attempts to re-focus the process of health care provision back to the patient. It shouldn't have left the patient in the first place and it's lovely to hear many delegates question the need for so many costly management tiers added to the process. Politics aside, whilst there are many speakers still prone to waffle and jargon, there are many special people trying to put across the message. I would like to see more nurses and doctors at the events but the age-old time and funding issues remain so it is left to each of us to remember that we are the experts of our own body.

Last year's King's Fund workshops introduced me to the concept that life is a journey of discovery and the emphasis on improving the patient journey by redesigning processes. As we all travel a variety of different methods and routes, not to mention the different tools (maps, signposts, etc.), we should expect to have different journeys and cope with them in different ways. When I attended the GIG (Genetic Interest Group) AGM last October, a group of patients spoke of being both helped and hindered by the drug company which manufactured enzymes vital to their existence. In other words, not every discovery is a positive journey but we all deserve high quality information to allow us to make informed choices. Hence the current emphasis is to challenge professional boundaries and work in new ways to develop patient led change. This was debated at the most recent event I attended, the AGM of CHIQ (The Centre for Health Information Quality), which I thought was a government organisation but it is a charity based in Winchester!! Nevertheless, it billed the guest speaker as Rosie Winterton, MP, Minister of State for Health whilst I would've given that title to Clare Rayner, Head of Patients Association and fellow nurse. Her contribution was apparently much more deep and meaningful but I missed it, courtesy of delays from the train service. The conference was titled "Better Information, Better Communication in Healthcare" and there was much emphasis on PALS, the Patient Advisory Liaison Service – If you have a problem, they are apparently the people to approach.... Personally I'd ask Margaret Costello.

On that note I should stop otherwise I shall need a newsletter of my own!! I shall bring all the leaflets to our AGM but in the meantime, enjoy your journeys!!

Maureen Gregory

One of our younger members Zak will be 13 years old on 19th March and would love to communicate via email with members of a similar age to share experiences. Anyone interested? Then email Zak at zakim@cutlers209.freeserve.co.uk

Keep well and enjoy a well earned rest at Easter. Until the next time!

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