

News & Views

August 2007

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Over half way through the year and here I am only just getting around to producing the first newsletter. I hope you are all well

Maureen has attended a number of events on behalf of the group, LTCA and GIG, just to name two. Maureen and I also attended the British Association of Dermatologists Conference in Birmingham in July. This is an important event in terms of further raising awareness of the GS to health care professionals.

New bank accounts are in place. The accounts were submitted to the Charities Commission for audit in April. Thanks to Maureen and to Robert Webster for their time on this task.

At our meeting in April the committee began planning for the patient conference to be held in October. This year we will be back in the North – apologies to all of you in other regions. It will be great to see those of you who can make the trip.

Read on for all the latest news.

Best wishes to all.

Margaret

Warm greetings to all.

We continue to challenge the government to deliver the promises of the White Paper to improve the quality and access to services for skin cancer patients. Spreading the word about Gorlin Syndrome is also important and we will attend two national conferences this year.

To ensure that fair and equal decisions in the use of group funds for travel expenses we are to draft travel guidelines. In addition are to draft research guidelines which will ensure when approached to take part in research, the best interests of the group are maintained.

All committee members are volunteers, many of us work full or part time, have family commitments or are involved in a variety of environments that assist us to support the development of work supporting the function of the committee and group. Often we have to seek outside help. Sometimes there are just not enough of us to meet all the commitments. If anyone has experience or skills that would assist our work, for example helping with the conference, this could be registration, the raffle, signposting new members and welcoming them, or telling us your personal story for inclusion in newsletters, we would love to hear from you.

We get invited to attend meetings to share the needs and represent the voice of Gorlin Syndrome, or attend information days spreading the word to health professionals and the public. These meetings are across the UK. Attendance is usually in pairs, mostly for moral support. If you would like to be involved or have any ideas, please do get in touch. We work mainly by e-mail and extra help would be truly appreciated. If you feel you can assist please contact Margaret on 01772 517624.

Take the very best care.

Sally

The newsletter is available on-line at www.gorlingroup.co.uk. In an endeavour to reduce administration costs, we would like to hear from you if you no longer wish to receive paper copies. Let us know by contacting Gorlin Syndrome Group.

A Matter of Choice!

This article outlines the experience of one couple in choosing to have children without Gorlin Syndrome. Other people may decide to make different choices and the couple respect that. However, they felt it was important to share their story to go some way to helping others make an informed choice.

I was diagnosed with Gorlin Syndrome (GS) at the age of 14 and in the intervening 23 years I've had numerous operations for skin cancers and jaw cysts. I married in 2002 and my wife and I knew we wanted children but we didn't want children with GS. So what were the options? Obviously, a decision like this is complicated and emotional with many factors to be considered. We arranged to have genetic counselling at the Regional Genetics Service in Birmingham, which was extremely helpful. I strongly recommend using the service. The location of Services around the country can be found on the Gorlin Syndrome website www.gorlingroup.co.uk or the Genetics Interest Group (GIG) website at www.gig.org.uk/services

Our options were:

1. A diagnostic test in the early stages of pregnancy with a view to termination if the test identified the presence of GS.
2. Pre-implantation diagnosis, involving eggs that have been fertilised in-vitro (in a lab, outside of the womb), with only non affected embryos being placed back in the womb to establish pregnancy.
3. Using donor sperm (or eggs when the woman has GS).
4. Adoption.

We ruled out option 2 due to cost, and options 3 and 4, choosing option 1 CVS Chorionic Villus Sampling as we wanted the baby to be our biological offspring. CVS was preferable to the other diagnostic test, amniocentesis, as it is carried out at 11 – 13 wks, rather than at 15 – 20 wks. At 15+ wks termination is carried out by inducing labour, whereas at 11–13 wks it is carried out by Dilation and Evacuation (D&E) / Dilation and Curettage (D&C. It is worth noting the tests can only be carried out to diagnose GS if the 'genetic fault' has been identified in the affected parent. My 'genetic fault' was identified in 2000.

We started trying for a baby in Nov 2003 and my wife became pregnant in June 2004. Fantastic news, but the next few weeks were very difficult whilst we waited for testing to see if the foetus had GS (a 50% chance), our lives were on hold. CVS can be performed in one of two ways, either by withdrawing placental tissue via a hollow needle inserted through the abdomen (trans-abdominal CVS) or through a flexible catheter inserted through the cervix. Trans-abdominal CVS testing was performed in September 2004. We were saddened and bitterly disappointed that the foetus did have GS. Termination was carried out the next day.

The next few months were very emotional, knowing that we could have had a baby boy (the foetus was sexed at our request) the following March. We were keen to try for another baby and were delighted when my wife became pregnant again in March 2005. Again, we felt a strong sense of our lives being on hold whilst waiting for the CVS test. Trans-abdominal CVS testing in June could not be carried out as the placenta was 'low posterior' and could not be accessed by the needle as it was on the other side of the foetus from the abdomen. The risk to foetus was too high. The test was repeated a week later, again it was unsuccessful extending our anxiety and stress. The placenta could still potentially be accessed by a trans-cervical CVS, but we had to go to City Hospital, Nottingham, a week after the second unsuccessful test. We received the results a week later, on my birthday and whilst on holiday – the foetus did not have GS. Without doubt, this is the best birthday present that I will ever receive! Whilst on holiday my wife had some light bleeding so we returned home. A scan revealed there were no problems and a week later we were thrilled to find out we were to have a girl. Our daughter was born in November 1995 and has been the light of our lives ever since.

Despite all the difficulties, we still wanted more children so we tried for a baby as soon as we could. My wife became pregnant again in Feb 2006. Once again, there was the long, emotional wait for testing. Again the CVS test at Birmingham could not be carried out, but this time we went to Nottingham the following week for the trans-cervical test rather than wait for a second trans-abdominal test. We were overjoyed when we received the news that the foetus did not have GS and that we were going to have another baby girl. It is now mid-November as I write this article and our beautiful second daughter was born just last week!

We have been very lucky, the results of the CVS tests that did not show GS could just have easily gone the other way and it would have been devastating if they had. It's an issue we will have to face should we decide to try for more children but for now we look forward to our two lovely daughters growing up together without them having to face the consequences of having GS. For us, this makes the difficulties of the last two and a half years a price worth paying.

Thank you for sharing your story with us

The full SunSense range of sun creams are now available on prescription, as is Uvistat SPF 50. All dermatology approved and worth a try. Ask your Doctor for details.

Research Projects

Over the last 12 months or so the Gorlin Syndrome Group have been contacted by a number of organisations wishing to carry out research to draw upon the experience of people living with long term medical conditions. The number of requests has risen dramatically and obviously as a voluntary organisation it has been necessary to embark upon those which relate to GS.

GIG offered their services, along with funding, to produce a Family Route Map. Patients and health professionals have worked with Anna Lane to build up a picture of services families receive. The project will develop ways to gather and disseminate information and will assist people to access information and services. We were pleased to welcome Anna and Melissa to the annual patient event last October where they ran a patient focus group.

NHS National Genetics Education and Development Centre made contact in October in relation to a project 'Experiences and preferences of people receiving genetic information'. Results will assist development of future services. We participated in a similar project with ROCC at the University of Edinburgh. The findings will also feed into future care.

Not strictly research but the GS group were represented at the Skin Care Campaign Scotland in Glasgow on 17th March 2007. The event raised awareness of condition North of the border.

Annual Patient Conference

The event this year will take place at Park Hall Hotel and Conference Centre in Chorley, Lancashire on 20th October 2007 between 9.30 am and 4.00 pm and as is usually the case we look forward to seeing as many of you who are able to make it. Feedback from last year's event was extremely positive with all commenting on the benefits of attending. Again we were delighted to welcome a number of new families. For many of the 'new faces' it was the first time they had met people in a similar situation as themselves and they reported how good it was to share experiences. It is also an opportunity to speak to Doctors who really know about the condition and can advise on the most appropriate treatment.

A booking form is attached giving full details. Kindly ensure they are returned by Friday, 28th September 2007 so that final arrangements can be made with Park Hall conference staff.

It isn't all heavy stuff! The social aspect is also important. We look forward to seeing you.

Christmas Cracker!

Apologies for mentioning Christmas at this time of year but I need to thank Jason & Sandra Knuckey of Cornwall. They were busy in the run up to Christmas with Jason decorating their home with Christmas lights. Family, neighbours and friends gathered for the switch on and for a second year the press were there to mark the occasion. There was a raffle on the night and a collection box at the end of the drive giving people the chance to donate.

Jason and Sandra are to be admired as they also organised a Christmas Karaoke and Disco in late November at the Sandy View Hotel in Newquay. Special thanks to staff at the hotel their welcome on the day. Thank you too to all who supported the event and to the local businesses who donated raffle prizes. The children were thrilled that Father Christmas took time out to be there.

£793.76 raised and greatly appreciated. A big thank you to all!

**Chatroom now 2 nights
a week. Sunday and
Wednesday evenings
8.00 – 10.00 pm**

Following the resignation of Sonya Dean we have a vacancy on the committee and we will be looking to elect at the AGM. Our thanks go to Sonya for her contribution over the years. On behalf of all, our very best wishes to Sonya and her family.

Gorlin Syndrome Group

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The Gorlin Syndrome Group
was formed in 1992 by
Jim Costello (deceased)

Patrons

Professor R J Gorlin
(deceased)

Professor P A Farndon

Professor G R Evans

Trustees / Committee

Sally Webster – Chair

Maureen Gregory – Treasurer

Margaret Costello – Secretary

Carolyn Deveney

John Doohar

Vicky Gill

*Backed by a full Medical
Advisory Board consisting of
health care professionals
covering the many aspects of
Gorlin Syndrome.*

*We are grateful for their
continuing support.*

Living with a Long-term Illness: The Facts

Oxford University Press. Published in January 2006

0-19-852882-5 978-0-19-852882-1 Price £12.99

Frankie Campling, a writer who has a long term illness and Michael Sharpe, Professor of Psychological Medicine and Symptoms Research, School of Molecular and Clinical Medicine, University of Edinburgh.

The book identifies the challenges posed by illness and suggests a wide variety of ways in which you might meet them. Key to this is the idea of becoming an expert in managing your illness and learning how best to deal with it. The authors accept that you know more than them about how you experience it, so that rather than telling you what to do, they offer a tool box from which you may pick the strategies that best suit you. The two authors combine their expertise and experience to offer a practical and comprehensive guide along your own unique journey.

Snippets

To all who sent donations and supported us through fund raising efforts a big thank you. It is through your efforts we are able to continue our work.

Thanks to the Community & Environmental Committee at Johnson & Johnson, High Wycombe for their support and donation following the sponsored walk by one of their employees.

Family, friends and work colleagues of Paul Drury raised £450.00 following his sudden death in the summer of 2006. This was obviously a testimony to Paul's popularity. Thank you so much.

Website continues to receive 60,000 + hits per month.

Congratulations to Samantha Mulligan and her partner Michael on the arrival Leigh. She arrived on 11th June weighing 6lb 5oz.

Very best wishes to Matthew Jones and Gemma Gregory of Kent. The couple were married in Faversham on Saturday, 18th June 2007.

Congratulations to Jane and Paul on the birth of Aimee who arrived on 25th April 2007. For those of you who aren't aware this is a first grandchild for Margaret Costello. Here she is at 3 weeks of age.



One of our younger members, Gemma Fox is off to Cyprus to work teaching after success in applying for a job. Gemma joins her Mum who lives out there. Our very best wishes to you Gemma, enjoy! Don't forget the suncream and keep in touch.

Those who receive treatment in Manchester will be pleased to hear that plans are well on to implement NICE guidelines for the treatment of GS patients. A multi disciplinary approach/care plans are on the way. Would be good to hear how this is progressing in other parts of the country.

Keep in touch. Till the next time!

Gorlin Syndrome Group Annual Conference and AGM

Saturday, 20th October 2007

9.30 am – 4.00 pm

Park Hall Hotel and Conference Centre
Charnock Richard, Chorley, Lancashire PR2 5LP

This form can be printed off and returned to the Gorlin Syndrome Group with a cheque for the appropriate amount at address below. Cheques payable to Gorlin Syndrome Group. Thank you.

Park Hall hotel is situated between Preston and Wigan and is well sign-posted from the M6 (junction 27 Northbound and junction 28 Southbound) or the M61 (junction 8), following brown tourist signs for Park Hall and Camelot Theme Park. If arriving by train the nearest railway station is Chorley. Taxis and local buses are available directly opposite the station.

Name:

Address:

Name/s of people attending	Age (if under 16 yrs)

The cost of the conference for the day is £20.00 per adult and £7.50 per child (subsidised) to cover refreshments and lunch.

Single, twin, double and family rooms are available on Friday, 19th and Saturday, 20th October. Prices quoted are for dinner, bed and breakfast.

**Twin or double room x 2 guests @ £62.50 per person (normally £79.50)
Family rooms x 2 children and 2 adults @ £62.50 per adult and £20.00 per child
Single rooms @ £82.50 per person (normally £99.50)**

For those people wishing to stay at the hotel, bookings should be made direct with Park Hall Hotel Tel No 01257 455000 quoting 'Gorlin' special rates.

Guests resident at the hotel have free membership to the hotels Health and Fitness Club with its 2 swimming pools, sauna and gym during their stay. For anyone wishing to take advantage of Camelot Theme Park, tickets can be purchased at £9.00 per person (normally £18.00)

Limited funding is available to those patients/families who would otherwise find it difficult to attend. Contact Margaret on the number below for further details.

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