

# GORLIN SYNDROME

SUPPORT GROUP

*News and Views - March 2005*

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## **Dear Group Members and Associates**

A very belated Happy New Year and a warm spring welcome to group members old and new, including all the spring chickens. Apologies for the delay in this newsletter and the cancellation of the autumn and spring conferences. Leaving unforeseen circumstances in 2004 we are now planning the 2005 conference on Saturday 22<sup>nd</sup> of October at Park Hall in Chorley. This year the conference will include information on the range of current treatments and developments in Gorlin syndrome from our medical advisors, personal history presentation from individual group members/families, alternative therapies and stress management. Whilst all this is going on the children can join in a separate arts workshop if they wish to.

Far more important than the agenda is the opportunity to meet old and new friends, have fun, support and help each other taking home memories lasting a lifetime. This is the foundation Jim left for us. Please put the dates in your diaries and iron your best party outfit. Margaret has included details of the conference and accommodation information in this newsletter. As in previous years we will be able to support group members and families to attend. Best wishes and take good care of yourselves.

*Sally Webster - Vice Chair*

## **Annual Conference**

Many of our regular readers were disappointed that no meeting took place last year. We are pleased to advise that this year's conference will take place on **Saturday, 22<sup>nd</sup> October** at Park Hall Hotel and Conference Centre in Chorley, Lancashire. The event will open at 9.30 am with tea and coffee on arrival for a 10.00 am start and the day will run through until 4.00 pm, lunch will be provided. The cost of attendance is subsidised at £10.00 per adult and £5.00 for a child. Funding has been set aside to enable patients on low income or benefits to attend. Further information regarding financial assistance can be obtained by contacting Margaret Costello on 01772 517624 between 6.00 pm – 9.00 pm daily.

A full and interesting programme is planned with excellent medical speakers who will update us on current research and treatments for the condition. You will have the opportunity to meet old friends, and for new comers to meet people and families to discuss common concerns. For those staying over at the hotel the evening social session extends the warmth of the day. For families attending there will be an art and craft workshop for the children, alternatively the theme park will be open for children young and old as will the swimming pool.

Anyone wishing to attend should complete and forward the attached form to Margaret along with a cheque made payable to 'Gorlin Syndrome Group'. **Closing date** for applications **no later than Friday, 7<sup>th</sup> October 2005**. If there is anything you would like included in the event, please let us know.

## **Welcome**

A warm welcome to all new newsletter readers. If anyone has suggestions or would like to submit an article for publication please do so. We would also like to extend a very warm welcome to Carolyn Deveney who has recently joined the committee.

## **NOWGEN – North West Genetics Knowledge Park**

Nowgen, the North West Genetics Knowledge Park, is one of six UK Genetics Knowledge Parks. Our work is split into broad categories: Genetics & Healthcare, Genetics & Society, Genetics & Knowledge and Genetics & Enterprise.

One of our major achievements for 2004 was the completion of our £3.5 million building: the Nowgen Centre. We are keen for support groups to benefit from using the facilities – the Centre provides a venue for public and professional events, including workshops, debates and regular support group evenings.

The Gorlin Syndrome Group is one of a number of genetic support groups that we have worked with over the last year and we will continue to do so in 2005. Recent and forthcoming events include a support group evening on Wednesday, 2 March and a series of workshops run by Sayward Morley, GIG's Artist-in-Residence at Nowgen ([sayward.morley@cmmc.nhs.uk](mailto:sayward.morley@cmmc.nhs.uk)). The Nowgen photo exhibition *Human Genetics and Society* will also be on show in the Centre from March.

As well as our work with support groups, we have a range of other activities. Our Education Officer, Kate Mathieson, has run regular Communications Workshops for genetics professionals, as well as working with the North West Science Learning Centre to run courses for teachers. We worked in conjunction with the Life Genetics Knowledge Park to run the *Disability and Reproductive Choice* workshop. The academic research programme evaluating current genetics services has progressed well, with support groups participating keenly in the relevant focus groups.

Nowgen has presented posters at the British Society of Human Genetics and the American Society of Human Genetics conferences. In addition, two reviews of Public Engagement in Human Genetics in the UK and of Ethical Frameworks and Ethnicity have been completed.

Successful bids to the Department of Health have led to funding for a pharmacogenetics research project (the TARGET study), as well as three service development bids.

We will continue to provide an environment for discussion on human genetic issues. We welcome comments and newsletter articles. For more information visit [www.nowgen.org.uk](http://www.nowgen.org.uk) or email [info@nowgen.org.uk](mailto:info@nowgen.org.uk). Alternatively, you can call Jo Ayres on 0161 276 8943 or Stuart Nicholls on 0161 276 8947.

### **Himalayan Bike Ride**



Congratulations and well done to Annie Macrorie who took part in a sponsored 350 kilometer, off-road mountain bike ride adventure from Pokhara to Kathmandu in Nepal. Annie was delighted to complete the course and in Annie's words 'chuffed to bits' to raise £680 for Gorlin Syndrome funds. Thank you Annie your efforts are greatly appreciated.

### **Sponsored Climb**

The committee would also like to extend sincere thanks to Sharon Hobson of Cumbria. Despite many recent health problems Sharon showed real determination undertaking a gruelling climb in the Lake District. Thank you to Sharon whose effort raised £103.00 which she donated to group funds. It is such donations which ensure the continued work of the group in supporting patients and their families affected by the condition.



**Charity Sunday Lunch** - Clare Bean sent a cheque for £85.00 which had been raised by a friend of hers Pat. Pat organised a Charity Sunday Lunch. Thank you to Claire and Pat for your donation. .

## **Funding – London Law Trust**

We are again thankful to the London Law Trust for their continued support in the form of a three year grant. The last of the three cheques to the value of £2500.00 was received in March meaning a healthier bank balance.

## **Web Site**

The web site continues to be well used with an average of 25,000 hits per month. Don't forget to check out for all the latest information. The site can be accessed at [www.gorlingroup.co.uk](http://www.gorlingroup.co.uk)

## **Jeans for Genes**

The Genetic Interest Group (GIG) has been chosen as one of the four guest charities for the Jeans for Genes Appeal. One of the main aims of the appeal is to raise awareness of genetic conditions in order to raise £4,000,000 this year for research and to fund family support services. As a guest charity GIG will receive 2% of the net profits made on the actual day. GIG will use the funding to carry out Project QUICK, the patient charter project. A Project Worker will be employed for two years and work with five smaller support groups (Gorlin Syndrome Group being one of them) with a view to producing a patient charter for each. These are guidelines drawn up in consultation with affected families and expert professionals which aim to lay out clearly what is currently thought to be good practice which they ought to expect to receive or provide.

In return GIG will be looking for volunteers willing to raise awareness of their condition and the Jean for Genes appeal. This could be through speaking to local school children in school assembly or telling their story to magazines, newspapers, or television. If you feel able to assist with the media appeal we would be delighted to hear from you either by email or telephone (address and telephone no at end of newsletter). You may wish to encourage local schools or businesses to register in which case we can supply a supporting letter for you to send out. Any help will be greatly appreciated. GIG has committed to donate £1000.00 a year for two years to group funds and to work with us to produce a patient charter, something which will be of benefit to all of us affected by Gorlin Syndrome.

## **National Institute of Clinical Excellence**

The committee has been involved in the consultation process for Service Guidance for improving outcomes in children and young people with cancer. A number of suggested amendments were forwarded as follows:

- Local and national voluntary support groups need to be included in partnership in local service delivery planning.
- In planning each child's care the multi development team must be aware and signpost parents to any relevant support group.
- The scope needs to map and consider the lack of resources for rarer cancers, e.g. PDT.
- The scope needs to consider the provision for travel and accommodation support to families who have to travel to access services.

The fourth point was taken on board and the text of the scope has been amended to cover the issues. Although the panel agreed on the first two points, on the first they commented that the guidance can only advise on the commissioning of NHS services and the second they felt unable to make such detailed recommendations. The third point they suggested that the mapping exercise is outside the remit of the guidance.

## **Department of Health - Section 64 Funding**

A recent application for funding from the Department of Health was unfortunately unsuccessful. The Department responded saying they had received a very high level of applications for the Section 64 General Scheme of Grants and with cash limited funds many organisations including ours were disappointed.

## **I stumbled across Gorlin Syndrome almost 3 years ago**

Hi everyone, I am Carolyn, the newest member of the committee. I stumbled across Gorlin Syndrome almost 3 years ago whilst looking for an explanation for some of my daughter's health anomalies.

Briefly, Charlotte now 6 had been diagnosed with hydrocephalus as a baby and was shunted at 7 months old. Though this improved her condition overnight there were still so many issues that were not explained by hydrocephalus alone. After a routine visit to a new neurosurgeon, where, for the first time when I voiced my concern that there was 'something else' I was listened to and respected, I was spurred on to seek the answers.

After inputting key words into a search engine on the internet, I discovered Gorlin Syndrome. After reading information from several websites including the Gorlin Group the pieces of the jigsaw started locking together. I contacted Jim Costello who pointed me in the direction of St Mary's Hospital. After bombarding our GP with information on GS Charlotte was referred to St Mary's. Although I was told it was highly unlikely that Charlotte actually had GS we were seen by a genetic counsellor and her life story was taken. I explained all of the features of the syndrome that I believed Charlotte had. As she was just 3 years old at the time and there is no apparent family history of Gorlin's, I think they thought I had gone mad. A skeletal survey was performed more to appease me than anything else.

Concurrently Charlotte was being seen by another genetics department who dismissed her completely, and by a maxillofacial surgeon who told me to bring her back as a teenager if her 'looks' bothered her. This infuriated me, but I was determined to get to the root of Charlotte's condition and was not to be thwarted by this insensitive consultant.

After several months we were recalled to St Mary's and after the negative result from the other genetics department I was prepared for us to be sent on our way. Then came the words 'Well you must be a doctor...' and Gorlin Syndrome officially entered our lives.

So over two years on, we play the waiting game, not quite knowing what to expect or when. I know that I have a beautiful daughter who lives life to the full enjoying every minute and I know that she will deal with everything that comes her way with courage and determination. There will be difficult times ahead of that I have no doubt, but with the right support, I am sure we will get through them.

Personally, I have flourished as the result of being an advocate to have Charlotte's condition recognised, I have gained in confidence, met many new people and have become involved in many voluntary activities, charities, projects and committees in St Helens, most of which support families of children with disabilities. I am also studying for a Psychology degree....there just aren't enough hours in the day!

## **With Sympathy**

The Committee would like to extend sincere condolences to Brian Cooper and family of Leyland in Lancashire on the passing of his daughter. Gina aged 37 died in hospital on 1<sup>st</sup> December 2004. Gina had lived with Gorlin Syndrome for many years and endured many treatments in her short life. Gina will be sadly missed.

It is also with sadness that I report the death of Margaret Young from Cardiff, South Wales. Margaret was also a young woman with Gorlin Syndrome and she died suddenly on 17<sup>th</sup> September 2004. We extend our sympathy to her family and friends.

**Until the next time**, look after yourself and take care.

# Gorlin Syndrome Group Annual Conference and AGM

**Saturday, 22<sup>nd</sup> October 2005**

9.30 am – 4.00 pm

Park Hall Hotel & Conference Centre

Charnock Richard, Chorley, Lancashire PR7 5LP

Tel: 01257 452090

Web site: [www.parkhall-hotel.co.uk](http://www.parkhall-hotel.co.uk)

The venue is accessed via the M6 or M61 motorways (well signed posted from both- Park Hall and Camelot), and is a ten-minute taxi ride from Chorley Railway Station.

Name:

Address:

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

**The cost of the conference is £10.00 per adult / £5.00 per child (under 16 years of age) and includes lunch. Please make cheque payable to Gorlin Syndrome Group and return to address below**

Adult places at £	
Child places at £	
Total amount payable (cheque attached)	£

**Twin or Double room x 2 guests** £60.00 per person per night–dinner/bed/breakfast  
(Children under 16 years £10.00 when sharing room with adults)

**Single rooms** £90.00 per person per night–dinner/bed/breakfast

Guests resident at the hotel have free membership to the Health and Fitness Club during their stay, which includes: indoor pools, gymnasium with weight and fitness area, sauna, spa whirlpool bath and steam room.

**For those members wishing to take advantage of accommodation at the hotel, bookings should be made direct with** Park Hall Hotel and Conference Centre, Chorley their website, or with Sue Lester via e-mail [sue@parkhall-hotel.co.uk](mailto:sue@parkhall-hotel.co.uk) or on Tel 01257 452090 – quoting 'Gorlin' for preferential rates.

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