

In this issue

<i>CMOs Report</i>	2
<i>Clinical Trials</i>	2
<i>GIG Conference</i>	2
<i>BAD Conference</i>	3
<i>Walk for Skin</i>	3
<i>Claire's Great North Run</i>	4

Spring has arrived! I sit here on a bright sunny April morning pulling together information for this newsletter reflecting on how time passes by so quickly. It's hard to believe the last edition was produced last summer. It's been a busy time both on a personal front and with Gorlin related activities.

In late October we welcomed regulars and first-timers to the patient conference and Annual General Meeting. Feedback has been excellent and very encouraging, with many people confirming the benefits of attendance. It was good to see you all.

We continue to work closely with many external bodies, the Genetics Interest Group, Rare Disease UK, the British Association of Dermatologists and the European Medicines Agency to name a few, to ensure that diagnosis and treatments for those with rare diseases are firmly on the national agenda.

Read on for all the latest news!

Margaret Costello

Welcome to the spring edition of our newsletter.

Thanks to your continuing support. We hope you find all the updates and information within this newsletter helpful and of interest.

Behind the scenes Margaret has been extremely busy representing us at national and European initiatives to further increase awareness of Gorlin Syndrome across a wide range of health care professionals, civil servants, politicians and other patient support forums, to ensure the provision of an equitable, accessible, timely and appropriate service for patients and family members affected by Gorlin Syndrome. Not stopping there, Margaret has also been involved in the further understanding of treatment options via various research opportunities and developments, also exploring developments in patient partnership and the important role patient support groups provide for rare conditions.

John has been instrumental in ensuring the proper operation of our IT function and governance, also in securing funding from BAD for further IT equipment to support the operational management of the group.

As always we run on a small number of volunteers who offer what little time, mixed skills and experience that we have with sincere commitment to support and help the group. We would always welcome any group or family member who could offer any time or support with supporting the group function. Margaret would be more than happy to discuss further if anyone feels they could lend us some time.

Wishing you all the very best and hope you all enjoy what so far seems to be a good promise for a good old British sunny summer.

Sally Webster
Chair

Visit the Gorlin Syndrome Group website and help raise funds with Every Click.
See the 'Support our Work' section for full details.
We value your support.

Giving Patients a Voice – CMO's Report

Each year the Chief Medical Officer (CMO) produces a report with a focus on the major health challenges needing to be addressed in the UK. The CMO is the Government's principal medical adviser and the professional head of all medical staff in England. In this year's report CMO, Professor Sir Liam Donaldson includes a chapter entitled "**Rare is Common**" relating to the needs of people with rare conditions.

Recommendations included in the report are:

- Strengthen the network of reference centres for rare diseases to enable better coordination of specialist services, including the transition from paediatric to adult services.
- Ensure that adequate numbers of specialists are trained so that future service needs can be met.
- Appoint a National Clinical Director for rare diseases to oversee the development of clear standards and pathways for the treatment and surveillance of rare diseases, with national registers to support service planning and delivery as well as research.
- Strengthen research, including translational research with economic incentives, to develop and market medicines for the 'orphan diseases'.
- Raise public and professional awareness of this neglected group of diseases.
- Support international collaborative efforts to share information and resources for rare diseases.

The inclusion of these recommendations offers hope for the future of health care for people with rare conditions. Equally it is also a boost for the work being undertaken by the Genetic Interest Group (GIG) and Rare Disease UK (RDUK) on behalf of patient organisations. Both get a mention in the report and much of the data within it has been supplied by them. Here at the Gorlin Syndrome Group we are committed to continuing our relationship with GIG and RDUK to take forward this area of work. The report is available on the Department of Health website http://www.dh.gov.uk/en/Publicationsandstatistics/Publications/AnnualReports/DH_113912

Genetic Interest Group Conference 2010

The 2010 GIG Conference takes place on the 8th June in London. The event will focus on important developments in patient partnership in healthcare and attendance is free for patients and patient groups. Full details about the event and how to book a place can be found on the GIG website at www.gig.org.uk. John Doohar and Margaret Costello will be representing the group at this event.

Clinical Trials Extension

Clinical trials are currently underway in Austria to evaluate the safety and local tolerability of LDE225, a topical formulation, for the treatment of basal cell carcinomas in Gorlin Syndrome patients. The pharmaceutical company Novartis are looking to extend the trials to the USA and UK. Rigorous procedures are in place in the UK to safeguard patients and before any clinical trial is given approval it must go before the European Medicines Agency (EMA). Margaret was invited to attend a Scientific Advice Discussion Meeting at offices of the EMA in March in the capacity of patient group representative. She listened to proposals for the trial and was able to raise issues or concerns about aspects of the proposed trial with the panel, which consisted of Doctors from various EU countries and Novartis staff. We await the outcome and hope to update all as soon as we hear anything further.

Annual BAD Conference

Sally and Margaret will attend the British Association of Dermatologists Conference in July at the Manchester Central Convention Centre. The event is an important one and further promotes the continuing affiliation between the BAD, its members and the associated patient support groups.

We were saddened to hear of the death of long time GS member Robert Thorndale who passed away on Wednesday, 21/04/10. Sincere condolences go to his wife Val and daughter Rebecca. Our thoughts are with each of you.

Walk for Skin 2010

In keeping with previous years the British Skin Foundation walks will take place during May and June. The Gorlin Syndrome Group is pleased to announce its affiliation again with the British Skin Foundation 'Walk for Skin 2010'. The Walk for Skin is the biggest multi charity fundraiser for skin disease and will take place at various venues across the UK.

There will be seven fabulous walks all themed around gorges, country parks, coasts and cities. Walks are generally 3 to 8 miles long, with shorter and longer options available at some venues. This year the walks will include added attractions and a Walk for Skin t-shirt when registering for any of the events. Joining the walks couldn't be easier – simply choose a venue and register either on-line at Walk for Skin or by phone on 0207 391 6341. Once registered, walkers will receive a sponsor form, walkers' pack and information about the venue of their choice.

This year 75% of participant sponsorship monies will go to their nominated affiliated skin disease charity. Thinking of walking?

We would value and appreciate your support.

Experiencing Problems Accessing Dermatology?

Andrew Langford of the Skin Care Campaign over recent months has brought to our attention the problems patients are experiencing in accessing Dermatology services in various locations across the UK because of a lack of resources and Dermatologists. Additionally, a number of Dermatology Departments are threatened with closure. This situation has led to the cancellation of appointments and in some cases leaves patients with no option other than to travel further for treatment. Andrew is continuing to address this matter and is working closely with the Department of Health with the intention of ensuring patients have access to a full range of services.

If you are experiencing problems accessing services in your area then do let us know so that we can feedback information to Andrew. Call Margaret on 01772 496849 or email at info@gorlingroup.org. Alternatively you can contact Andrew on 07810 564913 - Email alangford@skincarecampaign.org giving brief details.

We look forward to your hearing from you if you are encountering problems.

It's a boy!

Belated congratulations to the Dean family of Crewe on the arrival of Alfie Jack who weighed in at 8lb. Sonya, Ashley and children Jasmin, Ryan and Oliver welcomed their new son and brother on Tuesday, 19th January 2010. Best wishes to you all!

Thanks to Sonya for offering to be interviewed by Contact a Family (CAF) for a podcast about Oliver, her elder son's diagnosis of Gorlin Syndrome. Visit the CAF website to listen – access under Podcast Rare Diseases - Gorlin Syndrome // April 2010.

80th Birthday Celebration

Maureen and Nigel of Burnley made contact early in the New Year to advise that Maureen's mum Monica celebrated her 80th birthday in mid December. Rather than accept birthday gifts Monica asked family and friends to make donations to the Gorlin Syndrome Group. We were pleased to receive a donation of £265.00. Thank you to Monica and guests for their generosity, we are extremely grateful.

LTCAS

We recently became members of the Long Term Conditions Alliance Scotland. For UK groups membership offers the opportunity to link to the long term conditions agenda and policy developments thus ensuring the needs of patients with long term conditions are not overlooked. LTCAS are key partners in the Scottish Government and manage the £2m a year Self Management Fund. They drive forward the long term conditions agenda with the support of its membership organisations.

Gorlin Syndrome Group

11 Blackberry Way
Penwortham
Preston PR1 9IQ

Phone: 01772 496849

Email
info@gorlinggroup.org

Web site
www.gorlinggroup.org

Registered Charity No: 1096361

The Gorlin Syndrome Group was formed in 1992 by
Founder and First Chair
Jim Costello (deceased)

Patrons

Professor R J Gorlin (deceased)

Professor P A Farndon

Professor G R Evans

Trustees / Committee

Sally Webster – Chair

John Dooher – Treasurer

Margaret Costello – Secretary

Vicky Gill

Robert Webster

Helen Costello

Backed by a Medical Advisory Board consisting of healthcare professionals covering the many aspects of Gorlin Syndrome

We are grateful for their continuing support.

Gorlin Syndrome Group Awarded BAD Funding

We were delighted to be awarded British Association of Dermatologist funding for the purchase of laptop computers and advertising banners for use at conferences and for presentations. We are grateful to all involved at BAD for the continuing support of our work.

Care in the sun

Spring has arrived and with it some glorious weather. The sun has been out. Important to remember is the need for sun protection and staying safe in the sun, particularly babies and children with their delicate skin.

Recommendations

- Stay in the shade whilst the sun is at its highest, usually between 11.00 am and 3.00 pm.
- Wear wide brimmed hats and protective clothing.
- Wear 100% UV sunglasses to protect the area surrounding the eyes. Be sure that sunglasses carry quality marks such as BS Kite and CE marks. These will guarantee that they are giving you a high level of protection against UVB and UVA rays of the sun.
- Use sun-block or high factor sunscreen, the higher the number the better the protection. Look at the label and ensure that the product protects from UVA and UVB and that it carries a 4 star rating.

Claire Runs in Support of Gorlin Syndrome

Claire Edge, niece of Margaret Costello has contacted Margaret with the news she has successfully registered for the Great North Run, an event which takes place in the North East annually. With 54,000 runners, the BUPA Great North Run is the world's biggest half marathon. The event this year takes place on Sunday 19th September. Claire has indicated that sponsorship raised will be coming our way.



We are sincerely grateful for Claire's kind offer and take this opportunity to wish her well with training and the very best of luck over what is known to be a quite a gruelling course.

Thank you Claire, we really do appreciate your gesture.

We welcome your stories, articles and items for inclusion in newsletters. All contributions are gratefully accepted and can be submitted by email or post.