

Welcome to all newsletter readers

Since our last newsletter excitement continues in the ongoing trials and new treatment possibilities for individuals with Gorlin Syndrome.

We have been approached to ensure group members are aware of the phase 2 drug trial for LDE225. The details of this study are included in the newsletter explaining who may be eligible and where to obtain further information. LDE225 blocks the overgrowth of cells in the genetic pathway. This trial involves oral treatment and is open to patients worldwide with basal cell carcinoma not just those with Gorlin Syndrome.

All of a sudden we are using words used in the research world and I have attempted to explain what they mean in an article within the newsletter, which I hope you find useful.

Following the success of last year's patient conference where we heard so much about these new developments, as a committee we had hoped to provide a patient conference this year however sadly this has not been possible.

The committee is made up from a small group of volunteers, most of us managing life, work, families and Gorlin Syndrome. The main bulk of the operational organisation, the phone line and the website maintenance are undertaken mainly by Margaret. With this in mind and other pressures, with regret we are unable to commit to providing annual patient conferences. As a committee, we are meeting soon to look at this further, to ensure contact with new patients, the newsletter, website and ongoing developments with national bodies continue to further improve information, access to treatments and research into Gorlin Syndrome. We have decided more realistically to aim for two yearly patient conferences.

Again if you have any spare time to help us out we would really welcome any extra support. Few of the committee members have much experience of previous committee work, just a passion for nothing but the best for Gorlin Syndrome patients and families. So if you are skilled at organising events, or obtaining quotes for the best and most cost effective option or keen to express views on behalf of others, happy to talk to others about your experiences, or provide a sympathetic ear please do contact Margaret on 01772 496849 if you could spare time to help us keep this group going. We communicate mainly by computer, phone and meet usually together at least once a year.

Finally to wish all readers well and hope you find this newsletter informative and helpful.

Sally Webster
Chair

LDE225 Trial open in UK

A new worldwide study of an oral drug for patients with BCC that is either extensive or spreading (metastatic) is now open in the UK.

It will assess the effectiveness and safety of a new oral drug called LDE225, in these patients.

To be eligible patients need to either have a basal cell carcinoma (BCC) that has spread within their bodies or have a BCC greater than 1.5 cm, which is felt to be untreatable by other therapies such as surgery or radiotherapy.

(NB – the use of radiotherapy for the treatment of BCCs in Gorlin Syndrome can lead to the development of multiple BCCs in the radiation field and is not recommended)

The study is open for patients with or without Gorlin Syndrome. The study hopes to recruit 120 patients worldwide and patients selected will be allocated to receive one of two possible drug strengths of LDE225.

Patients are/are to be recruited in Glasgow, Guildford, Manchester, Leicester, Newcastle upon Tyne, London, Yeovil.

You can access further information about the study and see details of the location of investigative sites and contact details by visiting the following website.

http://www.clinicaltrials.gov/ct2/show/study/NCT01327053?term=LDE+BCC&rank=3&show_locs=Y#locn

Clinical Trials

How new drugs or treatments are tested to make sure they are safe and work properly.

There are different sorts of clinical trials, the following explains the process for ensuring drug treatments can be licensed for use by a doctor.

When a laboratory study finds a new drug that has not been tested on humans but shows the promise of treating disease, researchers develop a plan called a clinical trial protocol to start testing the new drug. This is normally led by a doctor who has to state the reason for the study, how many people are needed, who can take part, which drug will be used, any tests that are required and information needed. The process for the trial is standardised, very strictly monitored and has to be agreed by a research ethics committee.

The trial is normally sponsored by an organisation seeking better treatments for the disease and all those taking part volunteer to do so after giving their full informed consent. The first part of the trial is called phase 1, where the main focus is on the safety of the drug. This usually involves a small group of patients (20-80). When phase 1 trials show evidence the drug is safe, phase 2 trials commence. Phase 2 trials focus on whether or not the drug is effective and works. This usually involves a larger group of patients (100-300).

After phase 1 and 2 trials are completed, phase 3 is where the drug is given to a much larger group including other countries (1,000 to 3,000) to confirm its effectiveness,

Phase 4 trials are carried out after a drug has been shown to be safe and effective and has been given a licence so doctors can prescribe it. Phase 4 monitors how well the new drug works, the long term benefits and any long term or rare side effects.

The UK clinical research collaboration has produced two really helpful publications: - 'Clinical trials: what they are and what they're not' and 'Understanding clinical trials', both can be downloaded at:

www.ukcrc.org/publications/informationbooklets

Sincere Condolences to the Dean Family

We were shocked and deeply saddened to hear of the tragic and sudden death of Ashley Dean. Ashley aged 40 years died on 17th September 2011 and his funeral took place at Crewe Crematorium on Thursday, 20th September 2011.

Ashley, a loving husband to Sonya and dad to Jasmine, Oliver, Ryan and Alfie will be sadly missed by family and friends.



On behalf of all associated to the Gorlin Syndrome Group we offer sincere condolences to Sonya and the children.

Sincere Condolences go to the Crampton-Barden family

Victoria, aged 25 years of Southampton, lost her long battle with Gorlin Syndrome and the cancers associated with it in the early hours of Saturday, 12th November 2011.



Despite the many 'mountains' Victoria had to climb during her short life she remained optimistic for her future. She welcomed and drew strength from the many friends she made as a result of being associated to the Gorlin Syndrome Group.

Victoria was a warm, kind and caring young woman. She coped admirably with her condition and the many treatments she endured and will remain an inspiration to all who had the good fortune to know her. Victoria will be sadly missed but remembered fondly.

Our sincere condolences, thoughts and prayers are with Victoria's parents Sue and David, and her sister Sarah.

Yorkshire Water

Once again we thank the Yorkshire Water Waterwheelers for their recent donation. This time around we received a cheque for £50.00 following their Jeans for Genes Day collection. We are extremely grateful for the continuing support and thank all concerned for their generosity.

Thank you Marjorie

Thank you Marjorie of Tonbridge for your generous donation of £100.00. Donations of this nature ensure our continuing work.

Join us on Facebook

A recent update to the Facebook page has seen an increase in 'new faces' and posts. Topics of conversation are varied and allow people the chance to network with others. Log onto Facebook, click on Groups and search for Gorlin Syndrome Support Group.

Service Issues - Lincolnshire

The British Association of Dermatologists has advised of service issues in North Lincolnshire. Late last year Commissioners decided to tender for a community Dermatology service which will include all outpatient services. Assura was awarded the contract after the local trust decided not to bid. There are issues around Assura being able to provide 2 week wait skin cancer services compliant with NICE guidelines and it is unclear how patients with complex skin conditions requiring access to multidisciplinary care will continue to receive care locally. Concerns were raised about the lack of public and patient consultation in the commissioning process. The Trust has stipulated that the Dermatology unit will still provide a service. However, it's important patients are aware of their rights and choice to continue using the service at North Lincolnshire and Goole NHS rather than being automatically referred to the new service by GPs. Discussions are ongoing with the commissioners, the National Cancer Action Team and the Health and Overview Scrutiny Board. Patients in the area can also voice their concerns directly with the Trust.

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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 Doohar – 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.

Loz's London Marathon



On behalf of all sincere thanks are expressed to Loz for her energy, effort and the funds raised when she ran the London Marathon in April this year in a time of 4 hours 30 minutes. Loz, a friend of the Farmery family in Bristol raised £3078.00 and £2280.00 came to the Gorlin Syndrome Group, the rest went to fund a much needed break and Karate Club trip for Charlotte (Lotte) Farmery. Thanks to all those who sponsored Loz.

Margaret Costello (left) travelled to Bristol to be presented with the cheque and is pictured with Loz (centre front), Lotte Farmery and her Dad. A huge thank you to Loz!

Children and Education

If your child has Gorlin Syndrome it may be useful for you as a parent to advise teachers and other staff at school about your child's educational and medical needs.

UK schools have a Special Education Needs Coordinator (SENCO) whose role is to ensure every effort is made to tailor the individual needs, physical, emotional or educational, of a child within the school.

Does your child have Special Educational Needs?

IPSEA is a registered charity offering free and independent advice to parents of children with special educational needs in England and Wales on:

- local authorities' legal duties to assess and provide for children with special educational needs;
- exclusions of children with special needs/disabilities;
- actions or inaction by local authorities and/or schools which discriminate against children with disabilities.

The website is a great resource and gives advice on how to deal with any issues children may be experiencing at school. IPSEA has produced 40 new on-line resources to help parents with common Special Educational Needs issues. They can be downloaded on the website at <http://www.ipsea.org.uk/What-you-need-to-know>

Finally, as we approach the end of 2011 we take this opportunity to wish all our readers Seasonal Greetings and a Happy New Year.