

# GORLIN SYNDROME

SUPPORT GROUP

*News and Views - February 2003*

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Dear Group Members and Associates



## **Jim Costello 2<sup>nd</sup> June 1951 – 9<sup>th</sup> December 2003**

Words cannot really express my sadness since hearing of Jim's death.

Jim died very peacefully in St Catherine's Hospice with Margaret, Jane, Helen and Richard at his side. I am sure I speak for all of us in extending our condolences to them during this most difficult time.

Before Jim and Margaret established the group in 1992, we were all enduring the isolation, lack of information and inappropriate treatments experienced with rare disorders. Recognising this, Jim set up the support group with a handful of members that have now extended to 250 families within the UK, links with support groups in

America, the Netherlands and Norway and the running of very successful annual conferences. Jim strove to establish medical advisor links to ensure we had regular and timely access to information on the best treatments, support services and advice. These links led to the development of national partnerships with the government to have a voice in the health care planning and with links to the media, the most notable the BBC documentary, Bitter Inheritance.

Despite the unconceivable difficulties and pain he suffered, Jim inspired us in his ability to make the most of his life circumstances. Jim was our counsellor and friend, whether on the phone or in his company. Jim rarely spoke of his condition, always wanting to hear of others, listening, consoling and lifting our spirits, along with his natural warmth and sincerity Jim had an incredible sense of humour and a wonderful presence of being.

Jim's gift to us is incalculable and irreplaceable. He has left us an invaluable network that will ensure our children and future generations receive the best information to make choices and treatment options available.

I am sure I also speak for all of us in sincere gratitude to Margaret for her continued support with the group. Over the past 12 months Margaret has virtually ran the group with the help of Helen, Jane and Richard despite caring for Jim. With the help of the Committee, group members, our patrons and medical advisers we aim to continue the group the best way we can.

We will always have a permanent vacancy for our counsellor and friend, but we can continue building on the legacy Jim left for us and to make the most of life as Jim did.

*Sally Webster - Vice Chair*

## **A Message from the Costello Family**

In December it was with a great deal of sadness that we informed everyone of Jim's death. Jim died peacefully in St. Catherine's Hospice on 9<sup>th</sup> December 2003 – aged 51 years, following a

long illness. Deterioration over the last two years lead to Jim spending a total of 30 weeks in the hospice since summer of 2001. The dedicated care and support of hospice staff was of great value to us. Despite the illness and associated problems, Jim retained his dignity and wonderful sense of humour. In the face of adversity Jim coped admirably. Our loss is a dedicated, compassionate, loving and caring husband and father. Wonderful memories remain. We are extremely proud of Jim's achievements and consider ourselves privileged to have shared his life.

Jane, Helen, Richard and I would like to express sincere thanks for kind expressions of sympathy, support, cards of condolence and flowers received following Jim's death. Thank you for all donations to St. Catherine's Hospice and the Genetics Trust Fund – to date £4000.00 has been donated. Each charity has received almost £2000.00 in Jim's memory. We extend our thanks to everyone who cared for Jim and provided support to us all. Especially pleasing was to see so many at Jim's funeral. Many took time out of very busy working schedules and travelled long distances to be with us, and for this we are grateful. Jim would have been proud and at the same time humbled by the experience.

Jim worked tirelessly to raise awareness of the condition and campaigned for improved treatments. Many advances have been made since Jim formed the group but he often confided in us and advised there was still much to do. With the valued and continued support of our Medical Advisory team the work will continue in Jim's memory. Jim would have expected nothing less and we wouldn't dare do otherwise!

*Margaret, Jane, Helen and Richard Costello*

### **Group Patrons**

We are pleased to announce that Professor Gareth Evans and Professor Peter Farndon have accepted our request to become patrons of the group. Many thanks for your continued support and loyalty to the group.

### **Charity Status**

The accounts have now been audited by Robert Webster and charity status applied for. Many thanks to Robert for his help with the accounts and Margaret for endurance with completing multiple application forms.

### **Grant from the London Law Trust**

Pending charity status endorsement the London Law Trust have awarded the group £2,500 per year for 3 years to support the running and development of the group.

### **Useful Website Address**

[www.bad.org.uk/doctors/guidelines/disease/20management/basal/20cell/20carcinoma](http://www.bad.org.uk/doctors/guidelines/disease/20management/basal/20cell/20carcinoma)

### **Annual Conference – Saturday, 9<sup>th</sup> November 2002**

The conference took place at Park Hall Hotel in Chorley. The event was well attended by patients and their families. It was good to meet up with old friends and we were particularly pleased to welcome a number of new families to the event. We are grateful to the Medical Advisory Board members who attended, giving precious time at the weekend to share experiences and their expertise in the care/treatment of the condition.

**SESSION 1** Mr John Corner, Oral & Maxillo-Facial Surgeon, Royal Preston Hospital

**Topic: Odontogenic Keratocysts**

Odontogenic Keratocysts - a condition with many different names, which involve the dental lamina, which is formed in the foetus. The condition is one of the symptoms of Gorlin syndrome.

Patients develop fluid filled cavities in the jawbone with Keratin in the fluid (yellow), usually with daughter cysts in the epithelium (the 'skin' of the cyst). The cyst enlarges by osmotic pressure. Bone withdraws under pressure of the fluid. If allowed to progress they can cause fracture of the jaw. Oral x-ray is the only method of diagnosis.

Standard removal technique is by surgery with scarification (scrapping) or cryogenic (freezing) treatment of the cavity. In some places cytotoxic (cancer killing) treatment has been used in the cavity. No clinical evidence that the cytotoxic treatment has any effect good or bad. When the contents of the cyst have been removed the drain needs to be kept open to allow bone re-growth. Full re-growth should take less than 6 months.

Although x-rays are highly cautioned for Gorlin, the risk of low-level radiation from the oral x-ray needs to be balanced against potential for harm of unchecked cyst growth. There may be some advantage in the removal of overlying soft tissue to reduce the growth of the dental lamina, which may be implicated in the formation of cysts.

**SESSION 2** Dr E Allen, Dermatologist, Christie Hospital, Manchester  
Dr J. Moore, Laboratories, Christie Hospital, Manchester  
Anne Haylet, Senior Scientific Officer, Christie Hospital, Manchester

**Topic Photodynamic Therapy**

Photodynamic Therapy comprises of topical local treatment with ALA cream, the passage of a period of time and then illumination with intense pure red light (Patterson lamp)

Over several years the clinic has developed the use of pulsed ultrasound scanning to measure the thickness of the cancer. For very thin cancers (0.5 to 0.7 mm) "Efudix" (Fluorouracil) cytotoxic cream can be used as an alternative and has been very successful at the Christie Hospital for many years. PDT is used for medium thick cancers. PDT has been in use at Buffalo in U.S.A. since 1960's

Evaluation of treatments undertaken at the Christie Hospital has shown significant success following the use of gentle abrasion and massage of the site when applying the photo-sensitising (ALA) cream. Constant research into improving sensitises (creams) and red light sources are part of the work of this team. Dr Moore demonstrated the latest red light source, which is capable of irradiating (illuminating) much larger areas so reducing the necessity for repeated sessions.

Anne Haylet studies variations in the healing process. Scars are mostly collagen. There are many different types of collagen. Collagen production is linked to a protein called Hsp47. Hsp47 can be used as an indicator for potential scarring.

In order to establish an appropriate correlation between Hsp47 levels and scarring, tests are carried out on laboratory samples and on the skin of patients using ballistometer (A small lightweight plexiglas probe is dropped onto the skin surface via a free falling pendulum. The probe rebounds and again drops to the skin surface, causing less indentation, etc. The movement

of the probe is converted into a graphics display through the use of an image capturing software program. The resultant display is essentially a dampened sinusoidal wave pattern). This work is at an early stage. Watch this space for further developments. The availability of PDT is very limited; the committee is considering the best way to raise awareness of this method of treatment.

**SESSION 3** Dr Neil Walker, Dermatologist, Oxford

**Topic: The Role of the Dermatologist in Gorlin Syndrome**

Dermatologists are Physicians as opposed to Surgeons. When a patient first attends their GP(General Practitioner) they may be referred to an Oncologist, a Plastic Surgeon or a Dermatologist. “Diagnosis and treatment by GPs is generally poor”

The first task of the Dermatologist is diagnosis, then screening of the family and then management of the individual lesions. Dermatologists who have not specialised knowledge of skin cancer are likely to pass treatment to an Oncologist or Plastic Surgeon.

Management of Skin Cancer – Considerations

|                     |                           |
|---------------------|---------------------------|
| Clinical Appearance | Size and Depth            |
| Duration            | Presence of other lesions |
| Solar Damage        | Cell Type (?)             |
| Anatomic Site       | Primary or Recurrent      |
| Previous Treatment  | General Health            |

Treatment is influenced directly by the size and depth of the lesion. The best (only) way of accurately determining the size is by Moh’s surgery. [Moh's surgery is a technique where the specimen is excised and the surgical margins are immediately inspected. Any residual tumour is resected and the process is completed until all margins are free of tumour. Most surgeons advocate the Moh's surgery only for large skin cancers, skin cancer in anatomically difficult locations, recurrent skin cancers, or very poorly defined skin cancers.]

Available Treatments

|   |                              |
|---|------------------------------|
| Curettage and cautery/ electrodesiccation | Cryosurgery                  |
| Radiation (Not for Gorlin!)               | Laser Vaporisation           |
| Photo-Dynamic Therapy                     | Excision & Repair            |
| Cytotoxic Treatment e.g. Efudix           | Moh’s Microsurgery           |
| Retinoids                                 | Interferon (v. experimental) |

**SESSION 4** Bud Caruso of NBCCS Support Network of U.S.A.

**Topic: The Work and Progress of NBCCS Support Network**

Bud Caruso gave us an update on the recent activities of the organization including attending a variety of medical associations conferences. Hosting booths in the exhibitor’s halls to distribute information on Gorlin Syndrome and the existence of support groups. They have been very well received by the medical community and have had numerous responses as a result of their efforts. Bud added a description and make up of BCCNS and their Medical Advisory Board. The group has also enhanced their efforts by becoming a member of the Coalition of Patient Advocates for Skin Disease Research (CPASDR) and being listed with the Alliance of Genetic Support Groups and the National Organization of Rare Diseases (NORD).

A tour of the website [www.bccns.org](http://www.bccns.org) was given with emphasis on the Patient Forum <http://forums.delphiforums.com/n/nav/start.asp?webtag=Gorlinsyndrome&fpi=yes> and the chat

sessions on a Sunday Evening <http://www.bccns.org> in an effort to open up communication with our European friends. It has been discussed that we set an alternative time for the chat sessions once a month, providing the opportunity for everybody to participate.

BCCNS are also working with the medical community distributing information on current research regarding Gorlin Syndrome. Current are studies: Dr David Bickers and Dr Ervin Epstein who are conducting clinical trials of chemoprevention using Celecoxib (Celebrex), COXII inhibitor for preventing growth of basal cell carcinomas. <http://bccns.org/events.htm#Celecoxib> Dr. Lo Muzio of Italy, is soliciting x-ray of the jaws of patients to assist in proving what he describes as a new sign for Gorlin Syndrome, Bilateral Hyperplasia of the Coronoid Process. [http://wwwcsi.unian.it/medicina/docenti\\_lomuzio.html](http://wwwcsi.unian.it/medicina/docenti_lomuzio.html). QLT Inc has developed a new sensitizer for PDT. With their new product the residual photosensitive period is shortened from 6-8 weeks down to one week. <http://www.mbccstudy.com/>

Bud impressed on members the importance of members participating in the trials. The example cited was Dr Lo Muzio's study for the new sign of Gorlin Syndrome. Dropping a set of x-rays in the mail could lead to describing the one feature of the syndrome that makes possible the diagnosis of a young person who has not yet manifested some of the more common features of the disorder, enabling their family and health care providers to be proactive in monitoring closely for the other features and treating them as soon as they arise. Any newly described sign could also turn out to be the one missing "piece of the puzzle" of information that helps unravel the remaining mysteries of Gorlin Syndrome.

Lastly Bud encouraged group members to attend the BCCNS International Retreat Orlando Florida, 26th - 29th April 2003. The event is being held just ahead of the Society of Investigative Dermatology International Meetings in Miami, Florida. The European Society and Japanese society will be in attendance, so the group hopes to capture the attention of the international medical community at their event. Details [info@bccns.org](mailto:info@bccns.org)

*Bud Caruso – BCCNS Life Support Network - USA*

**SESSION 5** Professor P Farndon, Consultant Clinical Geneticist, West Midlands

**Topic: Clinical Genetic Service Birmingham**

By way of visual aids Professor Peter Farndon introduced us to the various members of his team in the Genetics Unit and described the advantages of new equipment capable of mapping and finding the precise location of the faulty gene in individual families. Professor Farndon described in detail the genetics responsible for the syndrome.

**SESSION 6** Margaret Costello and Sally Webster

**Topic: Gorlin Group Progress and Administration**

The committee members held their 1<sup>st</sup> AGM on 13<sup>th</sup> September 2002. The agenda covered the following points: -

- Documents to apply for charity status. Once the accounts have been audited, the documents will be forwarded to the charity commission.
- Each committee member reviewed their role/responsibility in respect of applying for charity status
- The accounts are currently being audited and then Helen Mulligan will formally act as Treasurer

- The London Law Trust have approached us to apply for a grant of up to £2,500 per year, application submitted
- The web page is updated by an external IT specialist
- Review of patient information leaflets highlighted the need to develop information for children and raise awareness amongst health professionals of the needs of children with Gorlin Syndrome. An external specialist will be approached to develop this area of work.

The group is a member of the following organisations

- Long Term Medical Alliance
- Genetics Interest Group
- Contact A Family/Rare Disorders Alliance

These organisations work at a national level assisting and supporting the work of support groups, influencing government policy, standards of care and advice for patients.

Recently the group has been approached by Central Government and the Department of Health to contribute to the development of new guidelines in the treatment and care of skin cancers. Work has been undertaken with the: -

- Commission for Health Improvement
- National Institute for Clinical Excellence
- Parliamentary Group on Skin Cancer
- Human Genetics Commission

*Summary of Medical Speakers kindly produced by John Doohar*

### **Fund raising activity and donations**

**Samantha and Super Gran!** We congratulate and thank Samantha Mulligan and her Gran, Beryl for raising an amount of £239.00 by taking part in a sponsored walk. This time Beryl went for a shorter distance than her previous marathon 'West Highland Way'. The cheque was presented to Jim Costello at the Annual Conference.

**Disco Charity Night Following** a year of fund raising activity culminating with a disco in June Mick and Lauretta Chester of Kettering presented Jim Costello with a cheque for £2400.00.

**Other donations received include:** J Robertson £250.00, Margaret M £200.00, Flora – Sponsor of Marathon £10.00 in respect of two participants, and an anonymous cheque received for £2000.00.

### **First Meeting of Netherlands Group**

Willem Breurken attended the Gorlin Syndrome meeting three/four years ago and since they have provided support in Holland, Belgium and Germany. Their first meeting was in November. <http://members1.chello.nl/~wf.breurken/Persoonlijke%20webpagina.htm>

### **Radiotherapy**

The newsletter in September included an item on radiotherapy. Medical guidelines are '*Radiotherapy should be avoided because of clinical evidence that new lesions can appear in the radiated field... It may be that some families are not as radiosensitive as others, until laboratory tests can detect these, radiotherapy should be avoided in all families.*'

Following the information a letter was received from Ms L reporting she was tempted to entitle her letter ***“I had radiotherapy and survived!”*** Ms L has become increasingly concerned about the negative publicity attributed to this form of treatment, and goes on to say, ***“Over the last ten years I have had numerous BCC’s, all on my face and all previously treated with surgery, removed in this way, with no ill effects, and certainly with no return of the lesions in the radiated sites. As is usual with the NHS the type of treatment offered may depend on where you live but doctors here in the South favour the ‘long, slow burn’, i.e. 1 – 1½ minutes of treatment per day for 10 – 15 days. I may just be lucky – but I have a wonderful oncologist and over the years we have learned from one another about Gorlin’s and the effects of certain treatments and I trust him completely, which is very important to me. So please, on my behalf, advise those people that have enquired that medical guidelines are just that – they are not cast in stone, and they should not be afraid to give it a try.”***

We are happy to include all views. Ms L points out that patients have options but geographical area may dictate the availability of treatments. Medical evidence shows that some patients treated with radiotherapy in the UK and abroad have suffered adverse effects resulting in the acceleration of BCCs to the site. It is with this in mind medical advisors suggest ***“It may be that that some families are not as radiosensitive as others, but until laboratory tests can detect these, radiotherapy should be avoided in all families.”*** Being in receipt of this information from medical advisors there is a duty on us to share with patients. At the end of the day the choice lies with the patient.

**If members would like to comment email, write or telephone for inclusion in the next newsletter.**

### **Royal Eye Hospital Study**

Awaiting details of the report from the Royal Eye Hospital, Manchester following the recent study. Once the information is to hand will include in a future newsletter

### **Midlands Regional Group**

Howard and Alison are hosting the first meeting of the Midlands Regional Group on 1<sup>st</sup> March 2003 at Yew Tree Cottage Restaurant, Stoney Lane, Yardley, Birmingham B25. Interested in attending then contact Howard or Alison on 0121 783 5009 or via email [gorlin@tyers.fsnet.co.uk](mailto:gorlin@tyers.fsnet.co.uk)

### **Funding to benefit research**

Ms J Foster of Brighton contacted the group following the screening of Bitter Inheritance. She was so moved by the programme she offered financial support. We are extremely grateful to Ms Foster for her generous donation of £5000.00. The funding has been passed to Peter Farndon’s department to aid further research.

### **Recycling Initiative and Yorkshire Water ‘Waterwheelers’**

John Pismeny’s fund raising efforts over the last year have really paid off. John set himself a target of £500.00 by end of 2002. As the time approached John realised he was short of the target. Not to be deterred he approached his employers Yorkshire Water Charity Trustees, The Waterwheelers who donated £200.00, bringing the total to £614.50. Well done John and special thanks to The Waterwheelers.

### **News from Contact a Family (CAF) – Rare Disorders Alliance**

A rare disorder, as defined by the European Union, is a condition that affects 5 or less people in every 10,000. Individually rare disorders affect relatively few. Collectively they affect the lives

of between 5-8 percent of the European population of 25-30 million. An estimated 4 million people are affected by rare disorders in the UK.

The Alliance believes rare disorders should be a public health priority. The large numbers of people affected, as well as the severity of some of the disorders, means that those with rare disorders have to compete for equal access to health resources and social services. In the UK there is little recognition of rare disorders at a national level. The Government has yet to take on board the recommendations of the European Union to consider rare disorders within the context of public health programmes (Article 6(2) of Decision 2951991EC. As rare conditions have little recognition at national level the Alliance is working to ensure that they are included in the current government National Service Frameworks (NSF) for children and long-term medical conditions.

**Central London congestion charging – Hospital appointments (CAF)** On February 17th 2003 Congestion Charging becomes part of life for those travelling within central London. This means that if you are driving into central London for a hospital appointment you need to pay the charge on the same day or you could find that you are charged a penalty fine. However, those who have a Blue/Orange Badge are exempt from the charging, subject to registration and a fee of £10. Registration is required on an annual basis although there is no charge after the initial payment. Log on to <https://www.cclondon.com> for information or telephone 0845 9001234 or visit [http://www.mayorwatch.org.uk/info\\_congestion.html](http://www.mayorwatch.org.uk/info_congestion.html).

**Meeting medical needs in mainstream education – (CAF)** The Research Department at NCB is beginning a new study looking at how the medical needs of children who attend mainstream secondary schools are met. Recent education policy has shown a definite shift towards the inclusion of all children in mainstream schools, and local education authorities are to a greater or lesser extent in the process of facilitating greater inclusion. However, new policies have focused in the main on the needs of pupils with physical and learning disabilities and less attention has been paid to the issue of medical needs and how these may interfere with optimal educational progress and social adjustment. The new study will include a large-scale survey of secondary school pupils in two local authorities to ascertain the level of medical needs and how well young people feel they are met in the school setting. Individual pupils' experience of school life will be explored in depth with interviews with pupil, peers, parents, teachers and health care staff. If you would like further information about the study or could contribute insights or experiences of either good or poor practice in supporting pupils with medical needs, please contact Jessica Datta 020 78436338 <mailto:jdatta@ncb.org.uk> or Nicola Ryder (020 7843 1161, <mailto:nryder@ncb.org.uk>

**Disabled – education and disability – a parent's guide to rights from nursery to university (CAF)** Contact a Family has produced the above publication, which will be of assistance to parent's with children who have special educational needs. It is a free publication funded by the Department for Education and Skills in England. For a copy telephone 0808 808 3555 between 10.00 am and 4.00 pm Monday to Friday. Alternatively visit [helpline@cafamily.org.uk](mailto:helpline@cafamily.org.uk) or minicom: 020 7608 8702.

## **News from LMCA (Long Term Medical Conditions Alliance)**

### **Written Evidence Request - Psychological & Social Consequences of Skin Disease**

The All Party Parliamentary Group on Skin is conducting an Enquiry into the Psychological & Social Consequences of Skin Disease. Their principal aim is to evaluate the ways in which skin diseases affect people's lives. LMCA has been asked to provide a written submission and we

would be keen to hear your views on this subject. Suggested questions are “How does having a skin disease affect a person’s day to day life i.e. work, leisure, relationships etc”; “What are the social & psychological effects of having a skin disease, particularly in relation to how society views skin diseases?” “Are there adequate support services available?” These questions are intended to act as a guide – if you have any relevant information or references to studies or statistical data that you would like us to include then please email [alliance@lmca.demon.co.uk](mailto:alliance@lmca.demon.co.uk).

### **New Regional Post for Genetic Interest Group (GIG)**

Anna Lane, Development Officer - Clinical Genetics, is working on behalf of the Genetics Interest Group (GIG) and the West Midlands Regional Clinical Genetics Service raising awareness of Genetic Services and helping to educate healthcare professionals, students and the public. Ultimately the aim is to strengthen and build capacity within the community to enable people to be involved in decisions regarding planning and commissioning services. Anna would welcome the opportunity to develop links with local Support Groups and would be delighted to hear from you, please contact her on 0121 623 6905 or email [anna.lane@bham-womens.thenhs.com](mailto:anna.lane@bham-womens.thenhs.com)

### **The All Party Parliamentary Group on Skin**

The above group is *conducting an enquiry* into the *Psychological and Social Consequences of Skin Disease*. The *principal aim* of the enquiry is to *evaluate the ways in which skin diseases affect people’s lives*. The group is *keen to obtain a wide range of evidence from people with skin diseases, clinicians, parents, carers and others who wish to contribute*.

Interested parties are invited to provide *written submissions in response to the questions below*. Submissions of *no more than three sides of A4 should be sent by 1<sup>st</sup> March 2003* to the Administrative Secretary at The All Party Parliamentary Group on Skin, 26 Cadogan Square, London SW1X. The group would prefer electronic submissions to email address: [natalie.delima@portcullisresearch.com](mailto:natalie.delima@portcullisresearch.com). *Please respond only to the areas that you feel best able to address*. The questions are quite broad and are intended to act as a guide, but the group will be *pleased to receive any information you think relevant to the Enquiry*, as well as reference to studies or statistical data.

#### **Questions**

- How does having a skin disease affect a person’s day-to-day life, i.e. work, leisure, schooling, relationships, life plans etc.? What factors are important in maintaining a good quality of life?
- How do skin diseases affect parents/and or carers? What are the time implications, social implications and cost implications of supporting a person with a skin disease?
- What are the social and psychological effects of having a skin disease, particularly in relation to how society views skin diseases?
- Do consultant dermatologists and/or general practitioners and nurses understand the psychological and social effects of having a skin disease? What kind of support should be expected from clinicians?
- Are there adequate support services available for people with skin diseases? Are carers/parents supported in any way?

- If not, what services do you think should be available, or how could current support services be improved? Are there any fast-tracking systems in place for patients access specialist services?
- What are the main blocks to developing services in this area – resources, other priorities, lack of awareness?
- Are you aware of any reports of peer-reviewed research into the social, psychological or ‘quality of life’ issues associated with skin diseases? If so, please will you provide copies (ideally electronically) or references?’
- Are there sufficient resources to conduct research into the psychological and social effects of skin disease?
- What are your key concerns about the priority that is given to the quality of life implications of having a skin disease by clinicians and by those who develop Government health policy?
- What more could the Government and other agencies do to support people with skin disease?

This is an opportunity to make a real difference. Only by forwarding your response will views be considered. Kindly respond to: **The All Party Parliamentary Group on Skin.**

The content of this newsletter will give you an indication of how busy things have been since the last newsletter. We hope you find the information both interesting and informative. Feedback on any of the articles or issues is always welcome.

On behalf of the committee we wish you well and trust that all up and coming treatments are successful and recovery speedy. Until the next time!

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**Gorlin Syndrome Group**  
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 Registered Charity No 1096361

# GORLIN SYNDROME

SUPPORT GROUP

*News and Views - January 2006*

## Dear Group Members and Associates

Warm winters greetings to members new and old, from the armchair at the side of the fire. Hoping this newsletter finds you well and able to make the most of the shorter colder days.

It was fantastic to see so many people at the conference. As always we had many opportunities to share and discuss our fears, concerns and experiences with each other. Meeting members from all ends of the country and making new friends with members from as far away as Sweden. Keeping Jim's vision, it's so good to talk, especially to someone who has shared the same journey and knows and understands how you feel.

Many thanks to our medical speakers who gave us their time, energy, knowledge and patience to keep us updated with the current issues and treatments.

Sorry to go on so for those who could not make it, but need to say special thanks to Sonia for relaying Oliver's story and to Jane and Helen for so bravely telling the story of their Dad, their hero. Not a dry eye in the house and a well deserved standing ovation. Thankfully Jasmin dried our tears and cheered us up with a very talented guitar accompanied song.

During the AGM the following committee changes took place Secretary, Margaret Costello (re-elected). Maureen Gregory takes over from Helen Mulligan, many thanks to Helen for her support as Treasurer over the past three years. The post of chair currently held by Jim Costello is now held by me, Sally Webster (previous vice chair). I accepted the role of chair with mixed emotions of sadness, inadequacy and honour; I just hope I can do Jim proud. Jim was voted unanimously to be known as founder member of the Gorlin Syndrome Group, I hope you all support and agree with this most fitting tribute to Jim.

Further to the conference, we have been asked to look at organising an update meeting south of Manchester to enable easier access for members. We aim to explore this at the next committee meeting.

I hope that you all had a peaceful and relaxing Christmas. On behalf of the committee all the very best wishes for 2006. Take good care.

*Sally Webster - Vice Chair*

### **Hello from Australia**

My name is Nicole and I have Gorlin Syndrome as well as cleft pallet. I'm a bit shy but would like to chat to anyone who is interested. I am 28 yrs of age live in Adelaide, been married 7 yrs now and have 2 beautiful girls who can be pain in the necks at times but make me smile everyday.

I would like to speak to anyone who has Gorlin Syndrome or anyone who would like to chat as I'm a friendly down to earth kinda person and enjoy having a good chat time to time.

Nicole [dolphin013@bigpond.com](mailto:dolphin013@bigpond.com)

### **Message from Prof R Gorlin**

A 60 year old with Gorlin in Seattle with documented cysts and abundant BCCs was talked into \$60,000 of dental work. She was given her diagnosis and at the time had 2 obvious cysts. Of course, the bridgework had to come out within 2 years. Do we not tell patients that only temporary stuff should be inserted? Does your literature address this?

Love Bob

*It does now Bob. Thank you for the advice and lovely to hear from you.*

## Calling all teenagers!



Calling all teenagers! Take a look at the website. This is your chance to chat and share experiences with people of your own age. <http://groups.msn.com/BccnsGorlinsTeens>

**Skin Information Day** – Sat, 24<sup>th</sup> Sept 2005 at Queens Park Hospital in Blackburn was attended by Sally and Margaret who were on hand to pass on information about Gorlin Syndrome to people who attended. Many nurses from Dermatology Units in Lancashire attended and were pleased to receive information about the condition and existence of the support group.

The **All Party Parliamentary Group** on Primary Care and Public Health is undertaking an inquiry into prescription charge scheme. LMCA has written to the All Party Group stating that the current system of charges and exemptions is over due for reform, highlighting the anomalies that exist. LMCA has urged the MPs on the Group to consider the risks people with long term health conditions may face if they discontinue treatment on cost grounds.

## Annual Conference

The conference proved to be a popular event with about eighty five people in attendance. It was good to see so many there.

Professor Peter Farndon and Mr John Cornah unfortunately were unable to attend. Peter was unwell on the day and John was called to theatre to carry out an emergency procedure. We were delighted to welcome Professor Gareth Evans, Dr Ernest Allan and Dr Anne Cook who all gave excellent presentations. Prof Evans talked about Gorlin Syndrome and its many characteristics and admirably filled Prof Farndon's 'slot' to speak about the genetics associated with the condition, whilst Dr Allan updated on PDT and Dr Cook reported on the findings of the Royal Manchester Eye Hospital study undertaken a couple of years ago.

## Photo Dynamic Therapy

Dr Ernest Allan, Christie Hospital, Manchester

Results of first 338 patients – Sporadic BCCs and Gorlin BCCs. We know that Gorlin BCCs respond to PDT in exactly the same way as sporadically occurring BCCs. A number of patients required several treatments to eradicate their lesions, but for 20 there appeared to be little benefit from PDT and 13 of the sporadic BCCs were ultimately treated by radiotherapy and 7 treated by surgery.

The success rate was therefore 94%. This is at least as good as most other forms of treatment but we are looking at ways of improving the effectiveness still further and reducing the numbers of treatments sometimes required in order to produce complete tumour resolution.

Another problem is that it is difficult to treat many lesions at one time. This is because it is difficult to keep the sensitising cream in place and also to achieve pain control during treatment. We have therefore undertaken a pilot study using an intravenous sensitizer called Photofrin. Of 10 patients treated, 4 have had complete resolution of their lesions and in 6 there has been considerable improvement. The treatment is painless and it is possible to treat thicker lesions by means of insertion of a laser fibre into the lesion. The initial results are therefore encouraging. A few patients have had brisk painful reactions following treatment but there have been no long term side effects. We are treating more patients with Photofrin but I think that it will take a little time before we achieve the excellent results reported by the Americans.

## Summary of Review of Patients with Gorlin Syndrome

Dr Anne Cook - Manchester Royal Eye Hospital.

**Purpose:** Review patients with Gorlin Syndrome (GS), documenting presentation, referrals, treatment patterns, and associated morbidity.

**Methods:**

Cross-sectional review and retrospective data collection of 40 patients with Gorlin Syndrome. Patients from the GS support group were invited to be examined. Those that were unable to attend were questioned via telephone or post. Demographics, presenting features, associated pathologies, and treatment modalities were recorded. Demographic data, age at presentation and diagnosis, spectrum of ophthalmic and peri-ocular disease, treatment modalities used, and peri-ocular deformities developed.

**Results:**

40 patients were included.

Age range = 8-72 years.

A variety of medical practitioners were involved in the care of these patients. In 16 patients, jaw cysts were the presenting features. In 12 patients, BCC was the presenting pathology. A wide range of other presentations were recorded. 29 had developed BCC's, 16 did so before the age of 30, 8 before 20 years.

**Conclusion:**

Multi-disciplinary care is essential in the care of the patient with GS. Early diagnosis of GS may allow for skin protection and surveillance at an earlier age. Early aggressive treatment may reduce peri-ocular morbidity. Co-ordination of a referral 'network' may improve the efficiency of referrals and management planning.

**The Features and Genetics of Gorlin Syndrome**

Professor Gareth Evans – St Mary's Hospital, Manchester

It has been a number of years since Prof Evans last attended the event but it was a pleasure to welcome him back and listen to his very informative and clear presentation on the features and genetics of the condition. Both are well documented but patient feedback was positive, reporting a better understanding in these areas.

Following this session patients were allowed the opportunity to raise issues and ask questions of the three Doctors in attendance.

Whilst parents were busy in the main conference room the children were fully occupied and enjoying activities in the Art and Craft session kindly provided by Michelle Hughes. All were very proud of their Christmas card creations when they were able to show them off to all in the main conference room.

For those people resident at the hotel there was the opportunity to get to know each other whilst relaxing in the restaurant at dinner or over drinks in the hotel lounge.

Thank you to all who contacted us with your appreciation and feedback. Here's to the next event!

**National Blood Service**

Committee member John Mills:

I have been informed by the National Blood Service (NBS) that I will not be able to give blood because I have 'ongoing' treatment for recurrent/multiple basal cell carcinomas (BCCs). They said that they gave the same response to another Gorlin patient last week. They informed me that this rule would apply to anyone who has ongoing treatment for BCCs. The rule applies for all donors with no exceptions for individuals. [A one-off BCC for which the patient had been treated and discharged would not be a problem.] The rule is in place to reduce the risk of infections being passed on through the blood (e.g. infection from the BCC /broken skin/rodent ulcer). Not clear on the risk to me as the blood donor.

GS website indicates that people with the Syndrome CAN give blood. I have explained this to NBS and they said that it will probably have to be changed. I have asked NBS to send me a letter clarifying this issue so that we can use appropriate wording on the website.

**Contact** has been made with Profs Farndon and Evans. Peter's response 'This is news to me, and I agree with John it seems over the top. I think the best thing is to wait for the letter John has requested and then for Gareth and/or myself to talk to the blood people.'

## **NHS Reforms and health equality**

The Socialist Health Association has organised six meetings around England (the first in Ipswich in late January) to consider the effects of proposed health service reform on health inequality. For more details of the events and a booking form, see the association's website. [www.sochealth.co.uk](http://www.sochealth.co.uk)

## **Worth noting!**

NICE Guidance for the treatment of patients with skin cancer will be launched in January 2006 meaning Gorlin syndrome patients are designated a special group. Commissioners have a duty to make sure that all the necessary treatments are available to treat the skin lesions in patients with Gorlin Syndrome and that the necessary expertise is available to administer these treatments.

## **Do Once and Share**

This is a six month project funded by NHS Connecting for Health (Programme for IT in the NHS) and is aiming to create an Integrated Care Pathway for Genetics. The project is in Southampton but the idea is that it can be shared once completed with other clinical genetic units to help harmonise the information and treatment offered. It forms one of many project teams based around the UK who are developing pathways for all types of services that the NHS provides. Once the project is completed the information will be used by NHS Connecting for Health to help with their ongoing IT programme to integrate services at both a local and national level.

A care pathway helps guide patients through the "patient journey", assisting them to access information and care about their condition at appropriate times.

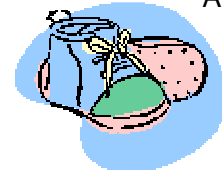
## **Jeans for Genes**

Special thanks go to all who contributed to the success of the 2005 appeal. Many gave up time raising awareness in schools whilst others contacted businesses for support.

**And finally!** Thank you for the many Christmas Cards, best wishes and the generous donations to group funds. Best wishes to all for 2006.

Until the next time!

## **Congratulations** go to



Anne and John Mills on the arrival of their first child a girl.

Yolanda arrived early and weighed in at 7lb 13 oz.

## **Christmas lights**

News from Cornwall came in the form of Christmas lights and a request from Jason and Sandra Knuckey of Newquay. Each year Jason decorates their home with festive lights and this year was no exception. In March Shania their 4 year old daughter was diagnosed with Gorlin Syndrome and they wanted to make this years display special and raise funds for the Gorlin Syndrome Group.

Family and friends attended the switch on with David White from Radio Cornwall flicking the switch. Local businesses supported the event offering raffle prizes and a collection box was placed to enable passers by the opportunity to donate. The event featured in the Cornish Guardian. Thank you Jason and Sandra, your efforts are appreciated.

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