

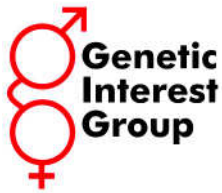
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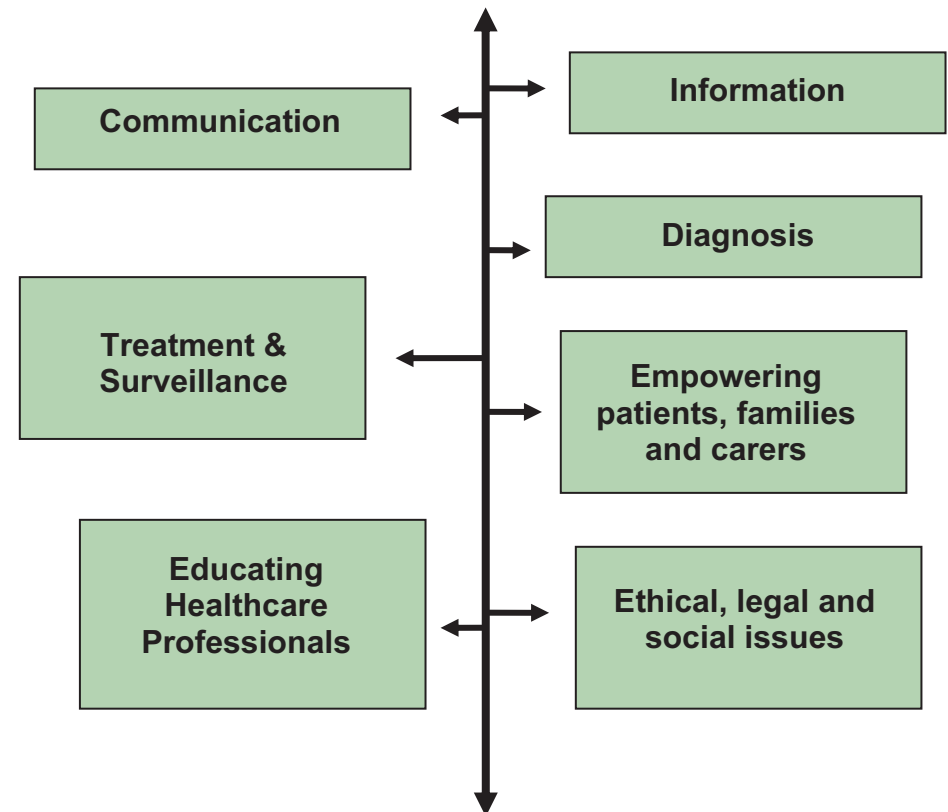
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This leaflet has been prepared in good faith to provide patients with a guide to current services and information. Neither GIG nor the Gorlin Syndrome Group can be held responsible for the accuracy of the information it contains. Links to other organisations are included for information purposes only and are not recommendations from GIG or Gorlin Syndrome Group .

Gorlin Syndrome

Family Route Map

This Family Route Map is a guide to current services and information.



INTRODUCTION

This leaflet is designed to provide signposts to sources of current information and appropriate services for patients, their families and carers, together with healthcare professionals. The issues and concerns raised during a series of patient focus groups in 2006 organised by the Genetic Interest Group (GIG) identified seven themes (see front cover) which have been used as the basis for developing this resource with the help of patients, families and carers, and clinical staff with experience and expertise of this condition.

The Gorlin Syndrome Group was formed in the UK in 1992 by Mr Jim Costello (now deceased), with assistance from the Clinical Genetics Departments at St. Mary's Hospital, Manchester and Birmingham Women's Hospital, Professor G R Evans and Professor P Farndon respectively and their staff. Official charity status was granted in March 2003.

The Group is organised by patients affected by Gorlin Syndrome and their families, with support from a medical advisory board.

The main aims and objectives are to:

- offer guidance, advice and information to those affected by Gorlin Syndrome
- the advancement of the education of the medical profession into Gorlin Syndrome and its implications for the family.
- promote research into the causes, effects, treatment and management of Gorlin Syndrome.

Contact details:

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Web: www.gorlinggroup.co.uk

Information about the many aspects of Gorlin Syndrome is available from the Gorlin Syndrome Group, details above. They can also provide detailed information about the genetics relating to the condition.

INFORMATION FOR HEALTHCARE PROFESSIONALS

Information and articles about Gorlin Syndrome are available from the Gorlin Syndrome website: www.gorlinggroup.co.uk

Use of radiotherapy for the treatment of BCCs in Gorlin syndrome can lead to the development of thousands of BCCs in the radiation field [Strong 1977; Evans, Birch et al 1991] and is not recommended.

Further information can be accessed as follows:

Surveillance guidelines are as published in Journal of Medical Genetics, Volume 30, 1993, pages 460-464

Farndon PA., Gorlin syndrome in Management of Genetic Syndromes Eds Cassidy SB and Allanson JE, 2005: 265-278 Wiley, New Jersey (2nd edition)

Review of Patients with Basal Cell Nevus Syndrome. Taylor et al. *Ophthalmic Plastic and Reconstructive Surgery*. Vol. 22, No. 4, pp 259-265 (recommends gold standards)

Basal cell carcinomas of the inner canthus: incidence of incomplete excision according to topographical localization of tumours. F. Boriani, F. Marconi. *British Journal of Dermatology*. Vol. 157, Issue 6 Page 1301 Dec. 2007

NICE Guidelines 'Improving outcomes for people with skin tumours including melanoma: The manual.' www.nice.org.uk

Orphanet (European database) www.orpha.net Free-access website providing information on rare diseases

General Practitioner Notebook– a UK medical encyclopaedia on the web <http://dev.gpnotebook.co.uk/simplepage.cfm?ID=x20041218200929159860>

British Oculoplastic Surgery Society www.bopss.org

Education for Healthcare Professionals:

NHS National Genetics Education and Development Centre www.geneticseducation.nhs.uk

PATIENTS, FAMILIES AND CARERS

Psychological help

Psychological counselling is not commonly offered on diagnosis or even later on. If you feel that you need extra help in coming to terms with your condition or any other aspect of your life, don't be afraid to ask for this via your GP or Consultant. Practical, medical, emotional and financial support is also available from:

Macmillan Cancer Support. www.macmillan.org.uk Tel: 0808 808 2020
Cancerbackup www.cancerbackup.org.uk Tel: 0800 800 1234

Education

It may be helpful for parents to talk to teachers and other staff at school about the educational and other needs of their children, particularly as Sunscreen is required to be used. 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. You can ask to speak to your school's SENCO.

Financial Assistance

Living with a long-term condition requiring frequent hospital visits or admissions for check-ups or treatment can put a strain on finances. Patients in receipt of welfare benefits may be able to claim back travel expenses so it is worth checking this out with the hospital. The Citizens Advice Bureau (CAB) can help with advice on benefits. Visit their website www.citizensadvice.org.uk CAB also has on-line advice and information on benefits, taxes and debt: www.adviceguide.org.uk

Other useful organisations:

Carers UK	www.carersuk.org	Tel: 0808 808 7777
Contact-A-Family:	www.cafamily.org.uk	Tel: 020 7608 8700
Disability Benefits Office		Tel: 08457 123456
The Family Fund	www.familyfund.org.uk	Tel: 0845 1304542

OTHER INFORMATION

The Department of Health has produced a set of 'Questions to ask' to take with you to your appointment with a specialist. Available in several different languages, pick up a leaflet at your GP Surgery or use the 'Search' option on the website www.dh.gov.uk to view it.

NICE Guidelines on skin tumours outlines how healthcare services for people with skin tumours should be organised. NICE has produced a leaflet for the patient, their family and carers, 'Improving the outcomes for people with skin tumours including melanoma: Information for the public.' Available from their website www.nice.org.uk or by phoning the NHS Response Line Tel: 0870 1555 455 and quoting Ref: N0958.

INFORMATION

Gorlin Syndrome is a medical condition caused by a fault in just one of the thousands of genes which humans possess. A study in the North West of England indicated approximately 1 in 56,000 are affected by the condition (Evans, D.G.R. et al. *Journal of Medical Genetics* 1993)

The condition is also known as Nevoid Basal Cell Carcinoma Syndrome (NBCCS), or Basal Cell Nevus Syndrome (BCNS) and is characterised by the development of multiple jaw cysts (keratocysts) and/or basal cell carcinomas (BCCs). Most individuals have pits (small depressions) on the palms of the hands and the soles of the feet and skeletal anomalies such as bifid ribs (ribs that are divided at the end) or wedge-shaped vertebrae. Other features occur in the syndrome, although most are rare but these can include ophthalmic problems, cleft lip and palate, ovarian fibromas (a fibromas is a benign tumour), cardiac fibromas which can affect heart function and medulloblastoma (type of brain tumour). The Gorlin Syndrome website lists many of the symptoms. Your geneticist will be able to confirm if your symptoms are related to Gorlin Syndrome.

It is an autosomal dominant genetic condition (affecting either sex) which means each child of a person with Gorlin Syndrome has a 1 in 2 (50:50) chance of also inheriting the faulty gene, and so developing signs of the condition. The severity of the condition is extremely variable between individuals and within families. There is also the possibility that the fault in the gene causing the syndrome happened for the first time in that family in the egg or sperm which went to make the person. This is called the result of a 'new mutation' and we know that this happens fairly frequently to the Gorlin Syndrome gene.

Understanding Inherited Conditions

The Genetic Interest Group (GIG) website has a series of leaflets explaining about inherited conditions and has one about 'Dominant Inheritance' and a glossary of genetic terms together with a leaflet about possible questions to ask when considering genetic testing:
www.gig.org.uk/eurogenest_patientleaflets.htm

Genetic Testing:

It is possible for family members to be tested to see if they carry the faulty gene. Genetic testing and counselling is available at 23 regional NHS genetic centres throughout the UK. A list of these centres can be found at www.bshg.org.uk See the section 'For Patients' which includes details of what genetic counselling is. The UK Genetic Testing Network (UKGTN) has produced a patient leaflet:
www.ukgt.nhs.uk/gtn/UKGTN-information/Patient-leaflet.html

DIAGNOSIS

Patients can present to different specialists, depending on the first sign of the syndrome. It is very common for families to remain undiagnosed for several generations despite having been seen by doctors from a variety of disciplines. Information about Diagnostic Criteria is available at www.gorlingroup.co.uk

Measurement of head circumference and examination of the skin plus X-rays are necessary to verify a clinical diagnosis of Gorlin Syndrome.

Diagnosis is vital for subsequent surveillance for complications such as basal cell carcinomas (BCCs) and jaw cysts, and for giving genetic information. Families should be offered regular screening, ideally with one clinician or a genetic department monitoring and co-ordinating the care.

A referral to a genetic centre is usually made through your GP or specialist. However, if you or a family member were seen several years ago in a Clinical Genetics Unit it may be possible to contact them directly as a self referral if further questions or concerns arise.

SURVEILLANCE

During pregnancy-most babies with Gorlin Syndrome have large heads and so may need assistance in delivery either by forceps or by Caesarean section. An ultrasound scan may help in predicting this.

Neonatal physical examination-when the baby is born, it may be apparent that he or she has a larger than average head circumference which would raise the suspicion that the baby had inherited the condition. In these cases X-rays may confirm bifid ribs or vertebral anomalies.

Childhood-annual dental screening should commence from about 8 years for the detection and early treatment of jaw cysts. There is also a need for at least annual surveillance from the specialist taking responsibility for treatment of the skin.

Adults-should inspect their skin regularly. Annual surveillance of the skin by a Plastic Surgeon or Dermatologist is recommended and dental screening should continue into adult life, its frequency depending on the findings of each X-ray. New jaw cysts seem to slow after the middle thirties. It is unusual (but not impossible) for cysts to appear after this age.

TREATMENT

The treatment options for BCCs will depend upon the age of the patient and the size, location and extent of the lesion. The following treatments are available:

- *Electrodessication and curettage*
- *Cryosurgery*
- *Laser Vaporisation*
- *Surgical Excision*
- *Micrographic (Moh's) Surgery*
- *Topical 5-Fluorouracil*
- *Oral Retinoids*
- *Photodynamic Therapy*

Patients should discuss treatment options with their specialist to ascertain which would suit their individual situation and give the best outcome. Information about the above treatments can be accessed at www.gorlingroup.co.uk

Jaw cysts require surgical removal and it is important that patients seek prompt treatment if complications and extensive surgery is to be avoided.

Treatment Priorities

To ensure effective management of Gorlin Syndrome it is vital that patients are treated in the early stages and for this reason regular surveillance is recommended to ensure:

- ⇒ *Prompt treatment for best cosmetic effects*
- ⇒ *Eradication of aggressive BCCs*
- ⇒ *Particular attention to individual lesions occurring around the eyes, nose, mouth and ears*
- ⇒ *Preservation of normal tissue to prevent disfigurement*
- ⇒ *Early detection and surgical excision of jaw cysts*

It is highly recommended that patients with Gorlin Syndrome are seen on a regular basis by an experienced Dermatologist.

Primary Care Trusts hold lists of clinicians who are specialised in particular areas, for example, they can usually advise on a local or regional Dermatologist who provides **MOHs** surgery to remove BCCs (a technique for removal of skin tumours with a minimum of normal tissue).

Care in the Sun

As sunlight may be one of the environmental agents promoting the appearance of BCCs it is sensible to use a good quality, high factor sun cream offering UVA/UVB protection. When the sun is at its highest it is advisable to wear a tee-shirt, a wide brimmed hat and sunglasses to offer protection to the area around the eyes.