GORLIN SYNDROME

WHAT IS GORLIN SYNDROME?

Gorlin syndrome is an inherited "autosomal dominant" condition, which means there is a 50% chance that children can inherit it from a parent with the condition. In some people it can start as a brand new gene mutation but it is important to remember that they can still pass the gene mutation onto their own children. Typically, symptoms start to show in young adulthood but sometimes young children can show some signs.

SYMPTOMS INCLUDE:

Common:

Basal Cell Carcinoma (BCC): Multiple BCCs over the skin and not just in sun-exposed areas.

Jaw Cysts: These are also known as "odontogenic keratocysts" and develop in the mandible and maxilla bones of the jaw. They often cause no symptoms but can cause swelling or pain. Often found in teenage/young adult years.

Rare:

Brain Tumours: Known as Medulloblastomas and can develop in childhood.

Fibromas: Benign tumours of the heart or in a woman's ovaries.

APPEARANCE

Palmar/Plantar Pits: Very small pits or depressions on the palms of the hands and soles of the feet.

Facial Differences: Larger head than usual "I can't find a hat to fit", a prominent forehead or jaw line, wide-set eyes and a variety of eye problems including cataracts. Children can have a cleft lip or palate.

Skeletal Changes: Prominent curve of the spine, a sunken or protruding chest and rib changes such as "double", "extra" or "missing" ribs

Head: Calcification of the lining of the brain seen on x-ray "falx calcification". This does not cause symptoms but is a useful diagnostic sign.

BASAL CELL NEVUS SYNDROME

WHAT ARE THE TREATMENT OPTIONS?

These include:

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

Radiation therapy is generally not suitable for people with Gorlin Syndrome as it can stimulate even more BCCs to form.

CART-WHEEL
Center for Analysis of Rare Tumors

Research is required to make the progress needed to find better treatments and provide more information about Gorlin syndrome. Due to the rarity of Gorlin Syndrome, clinical trials of new drug treatments for symptoms such as BCCs are not readily available in Australia. Part of the problem is that researchers need to be able to recruit enough people with Gorlin syndrome so that clinical trials can be opened in Australia.

You can be involved in research and help to attract clinical trials to Australia by registering your interest with the Centre for Analysis of Rare Tumours (CART-WHEEL) as well as keeping in touch with your doctors and regularly asking about the opportunity to be involved in research in this field.

www.cart-wheel.org

NEVOID BASAL CELL CARCINOMA SYNDROME

WHAT SURVEILLANCE DO I NEED?

During Pregnancy: An ultrasound during pregnancy can help predict if a baby has a large head, so extra help with the delivery can be made available.

Newborn: X-rays can confirm the inheritance of Gorlin syndrome by detecting bone abnormalities, especially of the ribs. No action is usually needed.

Childhood: Yearly dental screening should start from about 8 years of age to look for jaw cysts and start treatment early. Regular (at least yearly) skin checks by a dermatologist from puberty to look for BCCs and easy access to dermatologists in-between scheduled appointments.

Adulthood: Adults should inspect their skin regularly. At least yearly skin checks by a dermatologist is recommended. Dental screening should continue into adult life . Appearance of new jaw cysts often slows from the mid-thirties.

Prevention: Use of sunscreen, hats and sunglasses to limit UV exposure to sun-exposed skin is very important.

Get help: Find a doctor or medical service you can develop an ongoing relationship with to organize your regular checks and put you in touch with other specialists if any problems arise. Join a support program/group of people in a similar position.

GENETIC TESTING AND COUNSELLING

Gorlin Syndrome results from a problem in the "Sonic Hedgehog Signalling Pathway" in the body's cells, usually caused by an alteration ("mutation") of the PTCH-1 (Patched) tumour suppressor gene. Familial Cancer Centres and general genetics services can offer genetic testing to look for a gene alteration but also provide support for families and can help manage their screening. Contact details of Familial Cancer Centres within Australia are listed over the page.

CONTACTS AND SUPPORT

The information in this leaflet is intended as a brief summary about Gorlin syndrome only.

If you would like more detailed information and advice please refer to the following websites and support groups or contact one of the genetic services listed on the back page.

More information and support is available from:

BCCNS Life Support Network
www.bccns.org (US Group)

www.gorlingroup.org
info@gorlingroup.org (UK Group)

Patient Fellowship is available from the Google Group:

Australian Gorlin Syndrome Mutual Support Group

Contacts:

Anna Hickey (Melbourne) -
annahickey43@gmail.com
Margaret Emery (Adelaide) -
pmemery@bigpond.com
Julie Hagedorn BSc (Adelaide) -
augusta222@hotmail.com
http://gorlinsyndromeaustralia.blogspot.com

"Gorlin Syndrome In And Beyond Australia" on **Facebook**

FAMILY CANCER CLINICS

Victoria

Monash Medical Centre
246 Clayton Road
CLAYTON VIC 3168
Phone: (03) 9594 2026

Peter MacCallum Cancer Centre
St Andrew's Place
EAST MELBOURNE VIC 3002
Phone: (03) 9656 1199

Royal Melbourne Hospital
Grattan St
PARKVILLE VIC 3050
Phone: (03) 9342 7151

Austin Repatriation Hospital
Banksia Street
WEST HEIDELBERG VIC 3081
Phone: (03) 9496 5000

Australian Capital Territory

The Canberra Hospital
GARREN ACT 2606
Phone: (02) 6244 4042

Northern Territory

Women's and Children's Hospital
NORTH ADELAIDE SA 5006
Phone: (08) 8204 7375

Queensland

Herston Hospital Complex
HERSTON QLD 4029
Phone: (07) 3636 1686

Brisbane North Breast Cancer Family Clinic
534 Hamilton Road
CHERMSIDE QLD 4032
Phone: (07) 3350 7411

Western Australia

Genetic Services of WA
374 Bagot Road
SUBIACO WA 6006
Phone: (08) 9340 1603
Telephone: (08) 9483 2824

Mount Hospital
140 Mounts Bay Road
PERTH WA 6000

NSW

Royal Prince Alfred Hospital
CAMPERDOWN NSW 2050
Phone: (02) 9515 5080

St George Hospital
Gray St
KOGARAH NSW
Phone: (02) 9350 3815

Nepean Hospital
PO Box 63
PENRITH NSW 2750
Phone: (02) 4734 3362

Hunter Genetics
PO Box 84
WARATAH NSW 2298
Phone: (02) 4985 3132

Prince of Wales Hospital
High Street
RANDWICK NSW 2031
Phone: (02) 9382 2551

St Vincent's Hospital
Victoria Rd
DARLINGHURST NSW 2011
Phone: (02) 8382 3395

Westmead Hospital
WESTMEAD NSW 2145
Phone: (02) 9845 6947

Royal North Shore Hospital
Level 2 Vindin House
ST LEONARDS NSW 2065
Phone: (02) 9926 5665

Wollongong Hospital
Phone: (02) 4222 5576

South Australia

Women's and Children's Hospital
NORTH ADELAIDE SA 5006
Phone: (08) 8161 6995

Tasmania

Royal Hobart Hospital
PO Box 1061L
HOBART TAS 7000
Phone: (03) 6222 8296

GORLIN SYNDROME

also known as
Nevoid Basal Cell Carcinoma Syndrome

Basal Cell Nevus Syndrome

and
Multiple Basal Cell Carcinoma

Website:
gorlinsyndromesupportaustralia.weebly.com