

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:

- Electrodesiccation 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 \(US Group\)](http://www.bccns.org)

www.bccns.org

www.gorlingroup.org (UK Group)

info@gorlingroup.org

Patient Fellowship is available from the Australian Gorlin Syndrome Mutual Support Group: <http://home.vicnet.net.au/~gorlinsyndromesupport/>

Contacts:

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<http://gorlinsyndromeaustralia.blogspot.com>

"Gorlin Syndrome In And Beyond Australia" on Facebook

GORLIN SYNDROME

also known as

Nevoid Basal Cell Carcinoma Syndrome

Basal Cell Nevus Syndrome

Multiple Basal Cell Carcinoma

Victoria	NSW
Monash Medical Centre 246 Clayton Road CLAYTON VIC 3168 Phone: (03) 9594 2026	Royal Prince Alfred Hospital CAMPERDOWN NSW 2050 Phone: (02) 9515 5080
Peter MacCallum Cancer Centre St Andrew's Place EAST MELBOURNE VIC 3002 Phone: (03) 9656 1199	St George Hospital Gray St KOGARAH NSW Phone: (02) 9350 3815
Royal Melbourne Hospital Grattan St PARKVILLE VIC 3050 Phone: (03) 9342 7151	Nepean Hospital PO Box 63 PENRITH NSW 2750 Phone: (02) 4734 3362
Austin Repatriation Hospital Banksia Street WEST HEIDELBERG VIC 3081 Phone: (03) 9496 5000	Hunter Genetics PO Box 84 WARATAH NSW 2298 Phone: (02) 4985 3132
The Canberra Hospital GARRIN ACT 2606 Phone: (02) 6244 4042	Prince of Wales Hospital High Street RANDWICK NSW 2031 Phone: (02) 9382 2551
Northern Territory Women's and Children's Hospital NORTH ADELAIDE SA 5006 Phone: (08) 8204 7375	St Vincent's Hospital Victoria Rd DARLINGHURST NSW 2011 Phone: (02) 8382 3395
Queensland Herston Hospital Complex HERSTON QLD 4029 Phone: (07) 3636 1686	Westmead Hospital WESTMEAD NSW 2145 Phone: (02) 9845 6947
Brisbane North Breast Cancer Family Clinic 534 Hamilton Road CHERNSIDE QLD 4032 Phone: (07) 3350 7411	Royal North Shore Hospital Level 2 Vindin House ST LEONARDS NSW 2065 Phone: (02) 9926 5665
Western Australia Genetic Services of WA 374 Bagot Road SUBIACO WA 6006 Phone: (08) 9340 1603	Wollongong Hospital South Australia Women's and Children's Hospital NORTH ADELAIDE SA 5006 Phone: (08) 8161 6995
Telephone: (08) 9483 2824 Mount Hospital 140 Mounts Bay Road PERTH WA 6000	Tasmania Royal Hobart Hospital PO Box 1061L HOBART TAS 7000 Phone: (03) 6222 8296