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Gorlin syndrome

Gorlin syndrome is also known as Gorlin–Goltz syndrome, Gorlin's syndrome, nevoid basal cell carcinoma syndrome, and basal cell naevus syndrome. The so-called basal cell naevi are actually tiny basal cell carcinomas.

What is Gorlin's syndrome

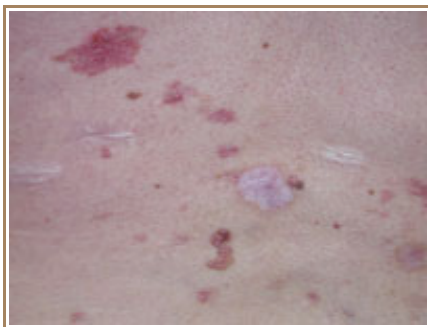
Gorlin's syndrome is a rare inherited condition characterised by:

- Multiple and early onset basal cell carcinomas
- Other tumours including melanoma, medulloblastoma, meningioma, breast carcinoma, non-Hodgkin's lymphoma, and ovarian fibroma

Developmental abnormalities in affected individuals may include:

- Odontogenic cysts in the jaw
- Broad forehead
- Pits on palms and soles
- Rib anomalies
- Calcified falx cerebri (noted on skull X-ray)

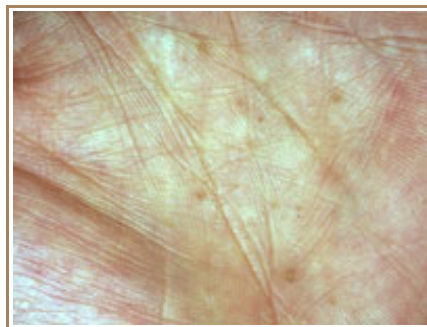
Gorlin's syndrome



Numerous basal cell carcinomas



Multiple scars from cryotherapy



Palmar pits

What is the cause of Gorlin's syndrome?

Gorlin's syndrome is due to an abnormal *PTCH* (patched) gene on chromosome 9q22.3–q31. This gene normally functions as a tumour suppressor so when it is not working properly it may allow cancers to grow.

Gorlin's syndrome is an autosomal dominant condition meaning half of an affected person's children also have the syndrome. It affects one in every 50 to 100,000 people.

How is the diagnosis made?

A diagnosis of Gorlin's syndrome can be made if there are 2 major or 1 major and 1 minor criteria.

Major Criteria

- Multiple (>2) basal cell carcinomas at any age or one basal cell carcinoma less than 30 years or >10 basal cell naevi
- Histologically proven odontogenic keratocyst or a polyostotic bone cyst
- Palmar or plantar pits (3 or more)
- Ectopic calcification: lamellar or early (<20 years) calcification of the falx cerebri
- Family history of Gorlin's syndrome

Minor Criteria

- Congenital skeletal defects: bifid, fused, splayed, or missing rib, or bifid, wedged, or fused vertebra
- Large head with occipitofrontal circumference >97th percentile, with frontal bossing
- Cardiac or ovarian fibroma (benign tumour in heart or ovary)
- Medulloblastoma (a malignant brain tumour that usually arises in young children)
- Lymphomesenteric cysts (abdominal cysts full of lymph fluid)
- Congenital malformation: cleft lip and/or palate, polydactyly (extra fingers or toes), congenital eye defect such as cataract, microphthalmos (small eye) or coloboma (iris tumour)

What is the treatment for Gorlin's syndrome?

The first sign of the disease may be the development of a medulloblastoma in a child aged 2 to 5 years, but luckily this is uncommon. Only a few children with medulloblastoma also have Gorlin's syndrome. If detected early enough, the tumour may be treated by surgery and chemotherapy.

Patients with Gorlin syndrome often require surgery to remove jaw cysts in their 20s. Often, it is not until they are in their 30s or 40s that the basal cell carcinomas begin to appear so the diagnosis of the syndrome is often delayed.

All patients with Gorlin's syndrome should see a dermatologist for regular skin examinations so that basal cell carcinomas can be treated when they are small. This may require surgery or one of the many other treatments available for these tumours including [cryotherapy](#), [photodynamic therapy](#), [fluorouracil cream](#) and [imiquimod cream](#). They should not receive treatment with [irradiation](#) as this is liable to provoke the development of more tumours.

Some patients may require long term treatment with oral retinoids such as [isotretinoin](#) or [acitretin](#).

Sun protection is vital to reduce the number of skin cancers developing but even complete protection will not prevent all basal cell carcinomas in patients with Gorlin's syndrome.

Related information

References:

On DermNet NZ:

- [Basal cell carcinoma](#)

Other websites:

- [Gorlin syndrome](#) - emedicine dermatology
- [Gorlin syndrome group](#)

Books about skin diseases:

See the [DermNet NZ bookstore](#)

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DermNet does not provide an on-line consultation service.

If you have any concerns with your skin or its treatment, see a [dermatologist](#) for advice.

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