GORLIN SYNDROME.

There is room for EVERYONE.

HOW TO MONITOR AND MANAGE GORLIN SYNDROME.

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 organise 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.

GORLIN SYNDROME

www.gorlinsyndromesupportaustralia.weebly.com

ALSO KNOWN AS:

NEVOID BASAL CELL CARCINOMA SYNDROME

BASAL CELL NEVUS SYNDROME

MULTIPLE BASAL CELL CARCINOMA

MORE THAN SKIN DEEP.

...most marathons end at about 40 kms, but the race is still on for people who live with a rare cancer, such as Gorlin Syndrome. This is a beast that requires your attention and perseverance, it is a race to just keep up with it let alone get ahead of it...

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, such as other organ systems and central nervous system.
- **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.

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
- Hedgehog inhibitor therapy

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

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.