

Management Protocol

Pediatric Patients

Baseline MRI with contrast and epilepsy protocol of the brain, repeat yearly (if asymptomatic) until age 8, then d/c

Baseline cardiac ultrasound

Baseline dermatologic exam, repeat yearly until first BCC develops, then every 6 months (or more frequently as needed) throughout life

Digital 6" film panorex of jaw at age 3 or 4 years (or as soon as tolerated), then yearly until first jaw cyst develops. Repeat annually (or more frequently based on patient occurrence) or until age 21.

Pelvic ultrasound in girls at age 18 (or earlier if symptomatic)

Molecular diagnosis

Baseline spine film at age 1; if abnormal, follow for scoliosis risk

Adult Patients

Baseline MRI of brain (for comparison if symptoms develop later)

Skin exam by a dermatologist every 4-6 months (or more frequently if new lesions present at each exam)

Digital Panorex of jaw yearly

Genetic Counseling (and molecular diagnosis)

Medical Genetics appointment annually

Appropriate Psychological referral and support

Pregnancy Management for at risk Couples

Prenatal/Preconception Genetic Counseling

OB recommendations

Cardiac ultrasound for fibroma

Follow OFC



Basal Cell Carcinoma Nevus Syndrome

Life Support Network

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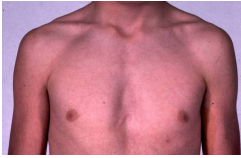
Basal Cell Carcinoma Nevus Syndrome
Life Support Network



Advancing Awareness . . .
. . . Finding A Cure

Designed for
Families,
Caregivers,
Healthcare Providers

What is Basal Cell Carcinoma Nevus Syndrome (BCCNS)?



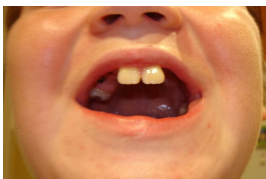
Pectus Excavatum



Scoliosis



Palmar Pits



Jaw Cysts - KCOTs



Multiple BCCs

BCCNS, aka Gorlin Syndrome, is a genetic disorder affecting 1:55,600 persons. This disorder was described in 1950 by Hale et al.; further identified by Dr. Robert Gorlin as a syndrome in 1955; and to be a result of genetic alterations in the *patched* (PTCH1) gene, 9q22.3 in 1996.

Inheritance is passed on by one parent (autosomal dominant) or occurs as a spontaneous mutation at conception. Approx. 70-80% of people have one affected parent and multiple affected relatives.

Manifestations: Basal cell carcinomas (BCCs); invasive oral tumors (KCOTs keratocystic odontogenic tumors) occur in 78% of all cases; hand (palmar) or feet (plantar) skin pits; fusion of the cranial bones of the skull (premature calcification of the falx cerebri); curvature & tumors of the spine; skeletal abnormalities; cardiac & ovarian fibromas; brain tumors, i.e. medulloblastoma, meningioma, among others.

The BCC's typically begin appearing in the first three decades of life. Although there may be medical con-

sequences to all of the phenotypic characteristics, the frequency with which BCCs occur and their treatment may arguably result in the greatest morbidity from this disorder. BCCs can occur on all areas of the body, but are most prevalent in the head & neck, shoulder, back, arms and trunk region.

Individual variability in the frequency of BCC's, results in persons having only a few lesions to thousands over a lifetime, requiring repeated doctor visits for monitoring, treatment and counseling. Treatment most often requires surgical excision, leading to disfigurement over time. Persons with BCCNS have significant risk factors for depression and their underlying genetic condition significantly impacts their quality of life.

The new frontier of gene therapeutic agents holds great promise for all the manifestations of this complex, lifelong condition.



Syndactyly



Developmental & Cognitive Delays



Plantar Pits



Strabismus

Current Possible Treatments

Oral Cyst Treatment:

Pantomographic evaluation every 6 months
Enucleation; Curettage; Cryotherapy;
Ostectomy; Excision; Marsupialization
Decompression; non chloroform Carnoy's solution.

These are often used in combination w/ the goal of complete lesion removal. Save as many teeth as possible. Increased incidence of dental Caries warrants frequent dental exams.

Skin Cancers (Nodular, Morpheaform, Ulcerative)

Curettage; E, D & C;
Topical 5 Fluorouracil (5FU)
Imiquimod
Photodynamic Therapy with ALA
Surgical Removal with layer evaluation to achieve clear margins: MOHS Surgery w/ Layer Evacuation

Brain Tumor

Excision ** Radiation treatment may trigger hundreds of skin cancers further exacerbating the proliferation. Cyber knife may reduce the area of exposure, when necessary.

Macrocephaly ~ Hydrocephalus Shunt placement to reduce pressure and drain excessive cranial fluid build-up

Vision, Hearing & Speech screenings, regularly
Cognitive & Learning evaluations, regularly.

Nutrition -- Vitamin A, B Complex, C and especially D should be regularly evaluated.

**Frequent CAT scans are cumulative and may be contraindicative. Please use Magnetic Resonance Imaging (MRI) whenever possible.