Published on *My46* (https://www.my46.org) 01/23/2022

My46 Trait Profile

Goltz syndrome

Other Names: Focal Dermal Hypoplasia

Goltz syndrome is a rare genetic condition characterized by abnormalities of the skin, hair and nails, and skeletal system. It is caused by mutations in the PORCN gene which is located on the X chromosome.

Characteristics of Goltz syndrome

Goltz syndrome is characterized by abnormalities in multiple body systems. Children with Goltz syndrome are typically born with streaky or patchy areas of thin or absent skin or skin with abnormal pigmentation. There may be fat nodules in the skin that appear as soft, yellow-pink bumps. The hair may be thin and sparse and nails may be ridged, small, and thin.

Goltz syndrome typically also affects the skeletal system. Individuals may have limb malformations such as a split hand or foot (i.e., a birth defect where it looks as though the hand or foot has been split down the middle) and fused or missing fingers or toes. The long bones may have a lined appearance on x-ray and can also be abnormally developed in severe cases. Though not always apparent on physical exam, there may be abnormalities of the vertebra or ribs which are visible by x-ray. These abnormalities may lead to complications such as scoliosis in later childhood.

Some individuals with Goltz syndrome also have small or absent eyes. Other eye problems include colobomas (i.e., a key-hole shaped lesion of the iris (the colored part of the eye), the retina (light detecting layer at the back of the eye) or the optic nerve (the nerve that sends visual information from the eye to the brain), and problems with the tear ducts. Depending on the severity of the eye findings, the individual may be blind. Facial features common to this condition include a pointed chin, asymmetry, simple prominent ears and notches in the cartilage in the nasal area. Some individuals with Goltz syndrome may have a cleft lip and cleft palate. The size, shape and number of teeth may also be abnormal. The teeth enamel can be ridged and thin causing an increased risk for dental caries.

Less commonly, there are developmental problems of the kidneys, abdominal wall defects (an opening in the abdominal wall exposing the intestines), and diaphragmatic hernias (an opening in the muscle that separates the abdominal and chest cavities). Some individuals with Goltz syndrome also have intellectual disability.

Diagnosis/Testing

Most individuals with Goltz syndrome have a change or mutation in a gene called PORCN that makes a protein called protein-cysteine N-palmitoyltransferase porcupine. It is not entirely known why or how mutations in the PORCN gene cause the features seen in Goltz syndrome.

Management/Surveillance

Management of Goltz syndrome often involves regular dermatology visits to treat the skin lesions which may be painful and prone to infection. Some individuals with Goltz syndrome develop papillomas (wart-like skin growths) around the mouth and genital area. Rarely, these papillomas involve the larynx (voice box) affecting swallowing. An Ear Nose and Throat doctor should be consulted if swallowing problems are noted and surgery or laser therapy may be needed.

Depending on the type of skeletal abnormality present, some individuals benefit from surgery. Others increase their

function through physical and occupational therapy and with assistive devices. Individuals with rib or vertebral abnormalities should be followed by an orthopedist to screen for and manage complications such as scoliosis.

Given the high risk for dental caries routine visits to a dentist are essential.

Mode of inheritance

Goltz syndrome is inherited in an X-linked dominant pattern. This means only one copy of the PORCN gene whether in a female with two X chromosomes or males with one X chromosome must have a change or mutation for an individual to be affected with Goltz syndrome. It is thought that in males, Goltz syndrome is fatal before birth because a second normal X chromosome is needed for survival to birth. However, some males with Goltz syndrome have been reported. It is thought that these surviving males have mutations in the PORCN gene in only some of their cells, which is called mosaicism. In most individuals, the mutation in the PORCN gene occurs brand new (de novo). However in about 5% of cases, the mutation is inherited from a parent.

Risk to family members

The risk to family members depends on whether or not the individual with Goltz syndrome has a parent affected with Goltz syndrome. If a mother has Goltz syndrome, the risk of passing on her PORCN gene mutation is 50% with each pregnancy. Male fetuses that inherit the mutation do not typically survive, but female fetuses that inherit the mutation can survive and have Goltz syndrome. Thus, at delivery, there is a 33% chance to have an unaffected male, a 33% chance to have an unaffected female, and a 33% chance to have an affected female. If a father has Goltz syndrome then depending on his level of mosaicism (i.e., meaning how many of his sperm carry the mutation), up to 100% of his daughters may be affected with Goltz syndrome. None of his sons would have Goltz syndrome. If neither parent has Goltz syndrome, the risk of other siblings being affected is very low.

Special considerations

None

Resources

National Foundation for Ectodermal Dysplasias (NFED)
http://www.nfed.org
Genetics Home Reference: Focal Dermal Hypoplasia
http://ghr.nlm.nih.gov/condition/focal-dermal-hypoplasia

References

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