FOCAL DERMAL HYPOPLASIA

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Focal dermal hypoplasia (FDH), also known as Goltz syndrome, is a multisystem genodermatosis. Genodermatoses are rare genetic diseases have cutaneous findings. FDH is an X-linked condition that is characterized by mucocutaneous, musculoskeletal, and central nervous system abnormalities. FDH is a rare condition of which 90% of affected individuals are female. With only 200-300 cases reported in the literature, the exact incidence and prevalence are unknown.

The porcupine O-acyltransferase gene (PORCN), located on the X chromosome, is responsible for the clinical manifestations of FDH. The PORCN gene typically plays a key role in several developmental pathways in utero. When one abnormal copy of this gene is present, the standard developmental process of several organ systems is compromised. While an abnormal copy of this gene can be inherited from a parent, most cases arise from spontaneous mutations.

Clinically, FDH is characterized by a collection of commonly occurring defects. In the skin, areas of hypoplasia (thinning) with underlying fat bulging are seen in approximately 75% of patients at birth. These areas of hypoplasia tend to be hypopigmented (lighter in color) compared to surrounding skin and may display freckling or telangiectatic vessels. Thin areas are also prone to recurring crusted ulcerations. These skin abnormalities are generally accompanied by papillomatosis of the oropharyngeal and respiratory mucosa, patchy or diffusely absent scalp hair, and nail dystrophy. In addition to the mucocutaneous findings, musculoskeletal and orofacial abnormalities are also characteristic of FDH. Short stature, syndactyly, ectrodactyly, and osteopathia striata are commonly observed. Microcephaly, cleft lip/palate, and dental anomalies are a few of the noted orofacial malformations. Furthermore, ophthalmic, urogenital, and central nervous system defects are also regularly present.

As the baby is developing in the womb, the presence of intrauterine growth restriction, limb malformations, or thoracoabdominal wall defects can suggest the presence of FDH. A series of clinical diagnostic criteria have been proposed and necessitate the presences of cutaneous and musculoskeletal findings. Specifically, three characteristic skin findings and one major limb malformation are required for diagnosis. FDH is confirmed by the identification of an abnormal or missing copy of the PORCN gene via blood sample.

Treatment of FDH requires a multidisciplinary approach that is directed by individual clinical findings. Cutaneous manifestations should be followed closely by a dermatologist as regular evaluation of erosive lesions and papillomas is critical. Within the first year of life, assessment of dental and oral manifestations by a pediatric dentist is recommended. Co-management between orthodontic and prosthodontic specialists may be necessary. With a propensity for feeding and growth issues, standard pediatric and nutritional evaluations should take place. Referral to endocrinology to asses for growth hormone deficiency may be required. Limb and musculoskeletal abnormalities should receive radiographic evaluation and orthopedic referral when appropriate. Finally, evidence of developmental delay and behavioral issues should prompt management with physical, speech, or behavioral therapy.

This information has been provided to you compliments of the American Osteopathic College of Dermatology and your physician.