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Focal Dermal Hypoplasia: A Case Report and Literature Review

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Case Report

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Abstract: Focal dermal hypoplasia (FDH) or Goltz syndrome is a rare genetic skin disorder; characterized by abnormalities of both ectodermal and mesodermal structures. We report the case of a girl of 8 years of Moroccan origin, second of a fratery of two, resulting from a non-consanguineous marriage and without any particular family history. Examination at admission notes that the patient presents with an overall hypotrophy, right facial asymmetry with low-set protruding ear, narrow nasal bridge. The examination of the oral cavity notes: Arborescent papillomas of the oral mucosa, hypertrophy gingival, abnormous tooth form, ectopic extensive dental caries eruption. Skeletal abnormalities: Ectrodactyly ("lobsterclaw" hand deformity) of the right hand, Syndactyly bilateral second and third toes. Hypopigmentation of the abdomen skin, microphthalmia of the right eye. The rest of the clinical examination is without particularity. The genetic study confirmed the diagnosis of FDH by highlighting a mutation of the PORCN gene. This mutation is from Novo. The patient is scheduled for dental care and orthopedic surgery on the right hand. a review of the literature was made

Keywords: Focal dermal hypoplasia, Goltz syndrome.

INTRODUCTION

Focal dermal hypoplasia (FDH) or Goltz syndrome is a rare genetic skin disorder [1, 2]. Characterized by anomalies of both ectodermal and mesodermal structures [3]. It was first described by Liebermann in 1935 as “atrophodermia linearis maculosa et papillo-matosis congenitalis”. Goltz, in 1962, mentioned the term “FDH” [1]. The disease is associated with a PORCN gene mutation.

Its protein plays a key role in the Wnt pathway, which has an impact on em-bryonic development. The inheritance is X-linked dominant, therefore 90% of affected peo-ple are female [2].

We report the case of a Goltz syndrome in an 8-year-old girl attending the otolaryngology department of the military training hospital Mohamed V of Rabat, with a review of the literature.

CASE DESCRIPTION

This is a girl of 8 years of Moroccan origin, second of a fratery of two, resulting from a non-consanguineous marriage and without any particular family history. She was sent to us for ENT examination.

Examination at admission notes that the patient presents with an overall hypotrophy, right facial asymmetry with low-set protruding earnarrow nasal bridge (Fig 1 & 2). The examination of the oral cavity notes: Arborescent papillomas of the oral mucosa, gingival hypertrophy, abnormous tooth form ectopic extensive eruption dental caries (Fig 3 & 4). Skeletal abnormalities: Ectrodactyly ("lobsterclaw" hand deformity) of the right hand (Fig-5), Syndactyly bilateral second and third toes (Fig-6); hypopigmentation of the abdomen skin, Dystrophic nails, microphthalmia of the right eye, good psychomotor development. By all echocardiography, abdominal and renal ultrasounds are normal.

This table evokes a dermal hypoplasia in areas or Goltz syndrome. The genetic study confirmed the diagnosis by highlighting a mutation of the PORCN gene. This mutation is from Novo because found in the girl and absent in the parents the family was reassured when the recurrence of the disease in siblings except gonadal mosaicism. The patient is scheduled for dental care and for orthopedic surgery on her right hand.

Fig-1: Right facial asymmetry

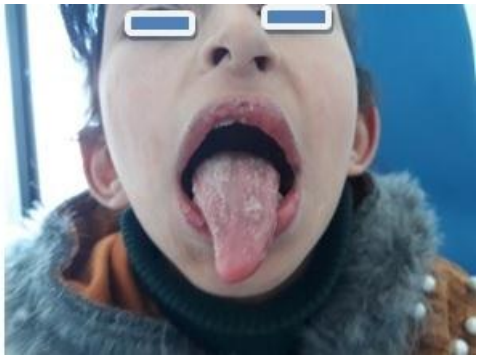

Fig-2: Asymmetry of the right hemilangue

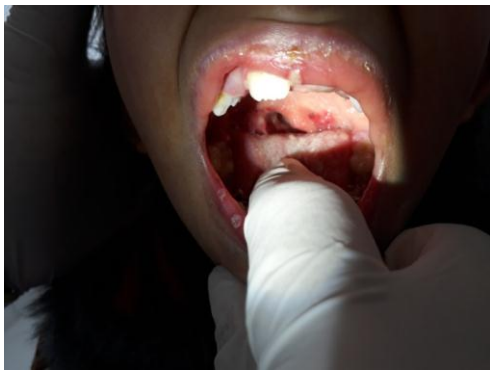

Fig-3: Arborescent papillomas of the oral mucosa

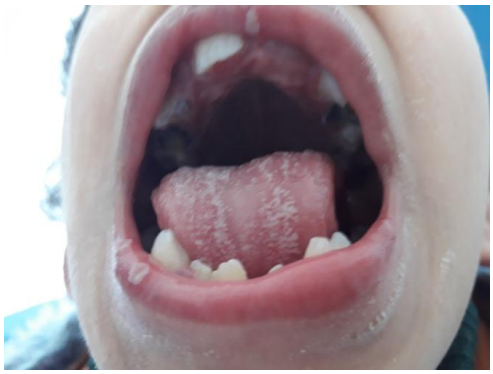

Fig-4: Gingival hypertrophy, abnormous tooth form, ectopic extensive eruption and dental caries

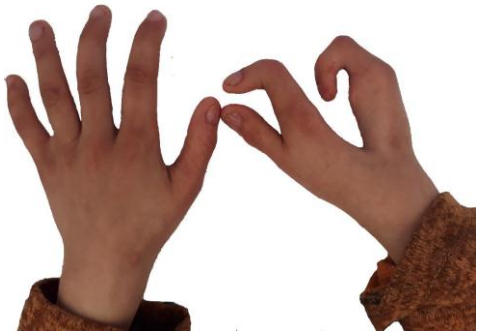

Fig-5: Ectrodactyly ("lobsterclaw" hand deformity) of the right hand

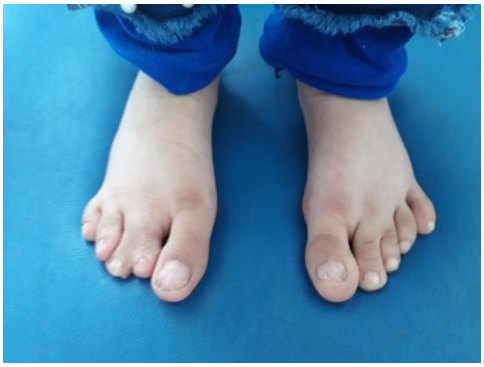

Fig-6: Syndactyly bilateral second and third toes

DISCUSSION

The Goltz syndrome is described as focal dysplasia of the dermis that is associated with multiple skin defects caused by disorders of the ecto- and the mesoderm, manifesting as improper formation of collagen IV and abnormal look of its bundles [4].

Focal dermal hypoplasia (FDH) is a rare genetic skin disorder [2]. With about 200 cases described worldwide; it was reported for the first time by Liberman [5] in 1935. In the 1960s Goltz *et al*., [6] and Gorlin *et al*., [7] Outlined characteristic traits of the disease, which was later attributed to Goltz in the nomenclature. Other commonly used names include: Goltz-Gorlin syndrome and focal dermal hypoplasia (FDH) [4].

The inheritance of FDH or Goltz-Gorlin syndrome is X-linked dominant [2] and the disease is associated with a mutation of the PORCN (porcupine homologue) gene located on Xp11.23 chromosome, which has variable expression [8]. PORCN codes for a transmembrane endoplasmic reticulum protein, which allows signalling and secretion of Wnt proteins involved in embryonic tissue development [13]. The majority of mutations are single nucleotide substitutions or small deletions/insertions that can be identified through sequence analysis [1]. About 95% of the cases appear de novo, and 90% of them are females [1, 2].

FDH affects primarily females with no ethnic or racial predilection. Most males affected by FDH die in utero and reports of living males represent cases of sporadic new mutations or mosaic, as non mosaic hemizygous males are not viable [9].

Theoretically every tissue of meso- and ectoderm origin can be altered. Intelligence is mostly unaffected [2]

It is characterized by vermiculate dermal atrophy, outpouchings of fat, telangiectasias, and hypopigmentation. Additional features include periorificial raspberry-like papillomas, dystrophic nails, sparse hair, abnormal teeth, split hand/foot (“lobster claw”) malformations, ocular abnormalities (e.g., microphthalmia), and the radiographic finding of osteopathia striata in long bones [1]. The skin and dental alterations manifest along the Blaschko lines [2].

Because all tissues of ectodermal origin are affected in FDH, a variety of orofacial and dental manifestations can be observed in this syndrome, as Facial abnormalities Asymmetry Low-set protruding ears Midfacial hypoplasia Narrow nasal bridge Skeletal abnormalities Cleft lip or palate High-arched palate Micrognathia Pointed chin

By far the most prevalent oral alteration found in FDH is generalized enamel hypoplasia, with malformed teeth that are more susceptible to dental caries [10]. Papillomas in the oral mucosa are also frequently cited as a characteristic of FDH [11].

Skeletal anomalies found in FDH include syndactyly, polydactyly, "lobster-clawlike" oligodactyly, short stature, and osteopathia striata [11. Diagnosis is usually made on the basis of clinical examination. Additionally, molecular genetic tests may be conducted [9].

Prenatal diagnosis is advised for pregnancies at increased risk, such as when the disease causing mutation has been identified in the family [12].

Surgical interventions are often needed during childhood for the correction of skin, skeletal, intestinal, andophthalmologic alterations. The removal of recurring multiple papillomas may require repeated surgeries. Early correction of skeletal alterations in upper and lower limbs, such as syndactyly, may be beneficial to the development of motor coordination, including oral hygiene skills [9, 12]. Another type of intervention that is common in children with FDH is surgical correction of omphalocele. Painfuland pruritic erosive skin lesions are prone to infection and demand regular visits to the dermatologist. Photodynamic therapy with flash lamp–pumped pulse dye laser may relieve itching symptoms in the skin and improve the esthetic appearance of telangiectatic and erythematous lesions [12].

Severely affected individuals often do not survive past infancy and family pedigree analysis shows a high prevalence of miscarriages and stillbirths. Individuals with minor expression may have a normal life span, depending on the associated presence of systemic alterations [9].

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