PAEDIATRIC DERMATOLOGY

GOLTZ SYDROME: REPORT OF TWO CASES

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Background: Goltz syndrome or Focal dermal hypoplasia (FDH) is a rare congenital dysplasia of the mesoectodermal derived tissues. It Is an X-linked Inheritance syndrome caused by mutations in the PORCN Gene mapped on Xp11.23. We report two cases of GOLTZ syndrome.

Case reports: Case n ° 1: A 4 months old female infant, born of a non-consanguineous marriage, a mother presenting amniotic band syndrome, has cutaneous atrophy and some pigmented lesions, with an omphalocele and cleft lip and palate. A skin biopsy was made but remained non specific. At the age of 6 years, she presented diffuse polymorphic skin lesions : dyschromia, cutaneous atrophy along lines blaschko and perianal papillomatous lesions. A second cutaneous biopsy was performed showing focal dermal hypoplasia. Systemic examination was normal but chest x-ray revealed right clavicle hypoplasia. The little girl was then referred to surgery department for the omphalocele surgery and reconstructive surgery of cleft lip and palate.
Case n ° 2: A 4 years old girl who had linear lipomatous hamartomas associated with cutaneous atrophy, thumb hypoplasia and bilateral nystagmus of both eyes. In addition, the patient had a IV grade stunting and microcytic hypochromic anemia. X-rays examination reveals clavicular hypoplasia, P2 of right thumb hypoplasia with spinal stiffness.

Discussion / Conclusion: FDH or Goltz syndrome is characterized by multiple abnormalities of the mesodermal and ectodermal tissue. As a result, patients suffer from cutaneous, bone, oral, dental and ocular disorders. Characteristic cutaneous features include asymmetric linear streaks of atrophy, linear hypo- or hyperpigmentation along the lines of Blasckho; fat herniation and telangiectasia usually present at birth. When a first affected offspring is observed, genetic counselling is a must.