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INTRODUCTION

Focal dermal hypoplasia (FDH) is a rare genetic mesoectodermal disorder, described by Goltz et al. in the year 1962.[1] The terminology, FDH was used to describe linear or reticulate atrophic macules with fat herniation and a wide variety of defects in the facial skeletal, urinary, gastrointestinal, cardiovascular and central nervous system. We describe a case of FDH presenting with a labial lipomatous mass and atrophic skin in sacrococcygeal area at birth followed by lipomyelomeningocele (LMMC) at 2 years of age.

CASE REPORT

A full-term female baby weighing 3200 g was born to non-consanguineous parents by emergency cesarean section for fetal distress. Antenatal period was uneventful with normal serology and antenatal scan. At birth, the baby was noted to have a swollen left labia majora and atrophic lesions affecting the sacrum and left buttocks. The left labial mass was soft to firm on palpation and was non-tender. At birth, the skin over the left labial mass was pearly white area with an erythematous border. The posterior end of the left labium was nodular and extends over to the anus. No active ulcers or bleeding noted. The right labium was normal. The sacral skin showed punched out and puckered lesion with an erythematous margin and extends from the posterior end of the left labial lesion to the spine behind the anus [Figure 1a and b]. Anus was patent and the baby was passing meconium. Urethral meatus and vaginal orifice appeared normal. There was no discrepancy in length of the lower limbs or muscle mass. No facial dysmorphism was noted. Scalp hair was sparse. There was no family history of similar genital or skin lesions.

On day one of life, her blood counts and renal function tests were within the normal limits. Varicella immunoglobulin M and TORCH screen were negative. Chromosome study showed a normal 46, XX karyotype. X-ray of the long bone did not show osteopathia striata. No cardiac lesions were detected in echocardiogram. An ultrasound scan of the spine, pelvis and the renal system were reported as normal. A detailed ophthalmological examination was unremarkable. Histopathology of the biopsied left labial lesion revealed normal epidermis, atrophy of the dermis with decreased connective tissue and the presence of large subcutaneous fat mass just beneath the epidermis confirming the diagnosis of FDH.

As the baby was thriving well, symptomatic treatment was offered by the dermatologist. At 2 months of age, the left labial mass was noted to be bulky with a more pearly white appearance. The borders were irregular and had a lipomatous feel [Figure 2a and b]. The old puckered lesion was healing, but new similar lesions were appearing over the right sacral and gluteal areas.

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She developed urinary incontinence and constipation at the age of 2 years. Magnetic resonance imaging of the lumbar spine showed a transitional type of LMMC with tethered cord [Figure 3a and b]. The urinary bladder was distended and both kidneys showed bilateral hydronephrosis secondary to neurogenic bladder [Figure 4]. She underwent L5-S1 laminectomy. Per-operative findings included a large subcutaneous lipoma extending through the dura and adherent to the dorsal surface and the tip of conus medullaris. The cord was found to be low lying. The distal dysplastic cord along with the adherent lipoma was divided from the subcutaneous lipoma, untethering the cord. She is currently on regular enema to empty the bowel and has a bladder catheter to maintain the urine output. Her mental and motor milestones are appropriate for the age.

DISCUSSION

FDH, more commonly known as Goltz syndrome, is a multisystem disorder primarily affecting the two germ layers, ectoderm (skin, eye) and mesoderm (bones, teeth) and less often the endoderm (pharyngeal mucosa). FDH is an extremely rare condition and the exact prevalence is unknown. FDH is an X-linked dominant disorder, 90% of affected individuals being females. Mosaicism for mutations in the PORCN gene on chromosome Xp11.23 has recently been implicated in the genesis of FDH. FDH is a developmental defect due to a block of Wnt signal transmission from cells carrying a detrimental PORCN mutation on an active X-chromosome with focal distribution of affected tissues. The variability in the severity of expression is due to lyonization. Surviving males with FDH have milder signs and symptoms than females, suggesting somatic mosaicism.\(^1\)

FDH most often presents with skin manifestations as blisters or erosions at birth. Erythematous, hypopigmented atrophic macules, arranged in a linear or blashkoid pattern or in a reticulate grouping, may be seen anywhere on the body. Lipomatous lesions, papillomatous lesions and telangiectasia are more often seen in older patients where as subtle skin manifestations are invariably noted at birth. Raspberry papillomas are usually not present at birth but develop with age and are rarely found around the nostrils, lips, anus and female genitalia. The skin lesions might be pruritic and photosensitive. Skin manifestations include atrophic and hypoplastic areas, fat nodules presenting as soft, yellow-pink cutaneous nodules and pigmentary changes. The nails can be ridged, dysplastic or hypoplastic. Hair can be sparse or absent. A wide range of ocular features including microphthalmia, anophthalmia, coloboma of the iris, choroid, retina or optic disc, corneal defects, blue sclera...
and ptosis have been described. A variety of oral and dental defects such as prognathism, malocclusion, enamel defects, cleft lip and cleft palate have been reported in patients with FDH. Anomalous pulmonary venous drainage and mediastinal dextroposition are the cardiac lesions seen in FDH patients. Hypoplastic external genitalia and human tail has been reported in patients with FDH. Less frequently, abnormalities of the kidneys and gastrointestinal system are present.[1-5] In the reported case, lipomatous and atrophic skin manifestations were present at birth. Her bladder and bowel dysfunction due to LMMC and tethered cord are not described as a manifestation of FDH even though meningocele with hydrocephalus was reported.[6] LMMC is a closed neural tube defect.

Some of the cases of FDH have characteristic facies with a triangular face and pointed chin. Face may be asymmetric with hemi-hypertrophy. Hand and foot abnormalities such as oligodactyly, syndactyly and ectrodactyly are common in FDH. X-ray of the long bones may show streaks of altered bone density in the metaphyses, known as osteopathia striata. It is seen in 20% of the cases of FDH; however, it is not a diagnostic feature of FDH.[1,5,7]

Diagnosis is based on clinical findings, histopathology of the lesion and molecular genetic testing. Histopathology of the skin reveals marked reduction in the thickness of the dermis and the deposition of fat cells up to the epidermis as seen in this reported case. A study by Bornholdt et al. identified 23 different PORCN mutations in 24 unrelated FDH patients from different ethnic backgrounds which included 3 microdeletions, 12 nonsense mutations resulting in loss of function, 1 splice site mutation and 8 missense mutations. Extreme skewing of X-chromosome inactivation in female FDH patients overcomes the consequences of potentially lethal X-chromosomal mutations. Molecular characterization of PORCN is relevant in patients with FDH for genetic counseling of families since carriers of the mutation might otherwise be overlooked due to significant phenotypic variability due to mosaic status. Prenatal testing/karyotype analysis, sequence analysis and deletion analysis are available in very few centers world-wide.[4,8-10]

The differential diagnoses for FDH are incontinentia pigmenti, Rothmund Thomson syndrome and microphthalmia, dermal aplasia, sclerocornea (MIDAS). Presence of atrophic lesions at birth rules out both incontinentia pigmenti and Rothmund Thomson syndrome, latter usually presents after 3 months of age. Generally, MIDAS syndrome shows dermal atrophy limited to the upper half of the body without fat herniation.[10]

A multidisciplinary follow-up facilitates early detection and appropriate preventive and/or corrective treatment of patients who were diagnosed to have FDH. Treatment with flash lamp-pumped pulse dye laser may ameliorate the pruritic symptoms. Papillomas may need repeated surgical intervention.[10]

CONCLUSION

This is the first reported case of FDH presenting at birth with isolated unilateral labial mass and sacral atrophic lesions with scalp hypotrichosis. FDH should be suspected whenever a child, especially female, presents with generalized, or at times localized, areas of atrophic plaques in a linear and blaschkoid pattern along with soft boggy swellings suggesting fat herniation. Unless an early accurate diagnosis is made, future plans targeting early detection of anomalies, timely corrective treatment planning with the goal of achieving optimal functional and esthetic results cannot be made.

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