A CASE REPORT OF GOLTZ SYNDROME: A NEWBORN WITH DIGITAL ANOMALIES AND SKIN LESIONS

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Introduction

Goltz syndrome or Focal Dermal Hypoplasia (FDH) is an uncommon genetically inherited disorder characterized by distinctive skin abnormalities and a wide variety of multisystem defects which was first described by Goltz (an American dermatologist) in 1962. About 200- 300 cases have been reported worldwide. FDH can be inherited in an X-linked dominant manner with in-utero lethality in males. Majority of the cases are sporadic with new mutations arising in the embryo and not inherited from a parent. Approximately 10% of cases occur in males; postzygotic somatic mosaicism accounts for the findings in these affected males. FDH is caused by abnormalities or mutations at the PORCN gene in the X chromosome. We report a case of FDH with characteristic skin lesions as well as multiple digital anomalies - oligodactyly, syndactyly and ectrodactyly.

Case Report

Baby NHI, a term baby girl of birth weight 1.95kg, was born with multiple congenital anomalies:-

- Cutaneous lesions - atrophic erythematous skin lesions over bilateral pinnae, bilateral inguinal region, left elbow and left shin (Figures 1,3,5); wart-like skin lesion over left shin (Figure 2) and hyperpigmented skin over inguinal area (Figure 5).
- Skeletal anomalies - left hand oligodactyly forming "lobster claw" deformity (ectrodactyly) and left foot syndactyly with abnormally placed little toe (Figures 4,8,9).
- Other dysmorphic features - bilateral low set ears, flat nasal bridge with a pointy nose, small chin, simian crease left palm and irregularly sparse hair (Figure 1).
- Ophthalmologic review - no coloboma noted.

Bedside ultrasound cranium, kidneys and ECHO were normal. Parents were not agreeable to skin biopsy proposed by dermatology team.

Follow-up at 2 months old revealed progression of the skin lesions - atrophic skin lesion became hypopigmented and had further extended to involve a larger area of the trunk with a Blaschkoid distribution (Figures 6,7). New papillomatous lesions appeared on several fingers and toes bilaterally (Figures 8,9).

Figure 1-5. At birth

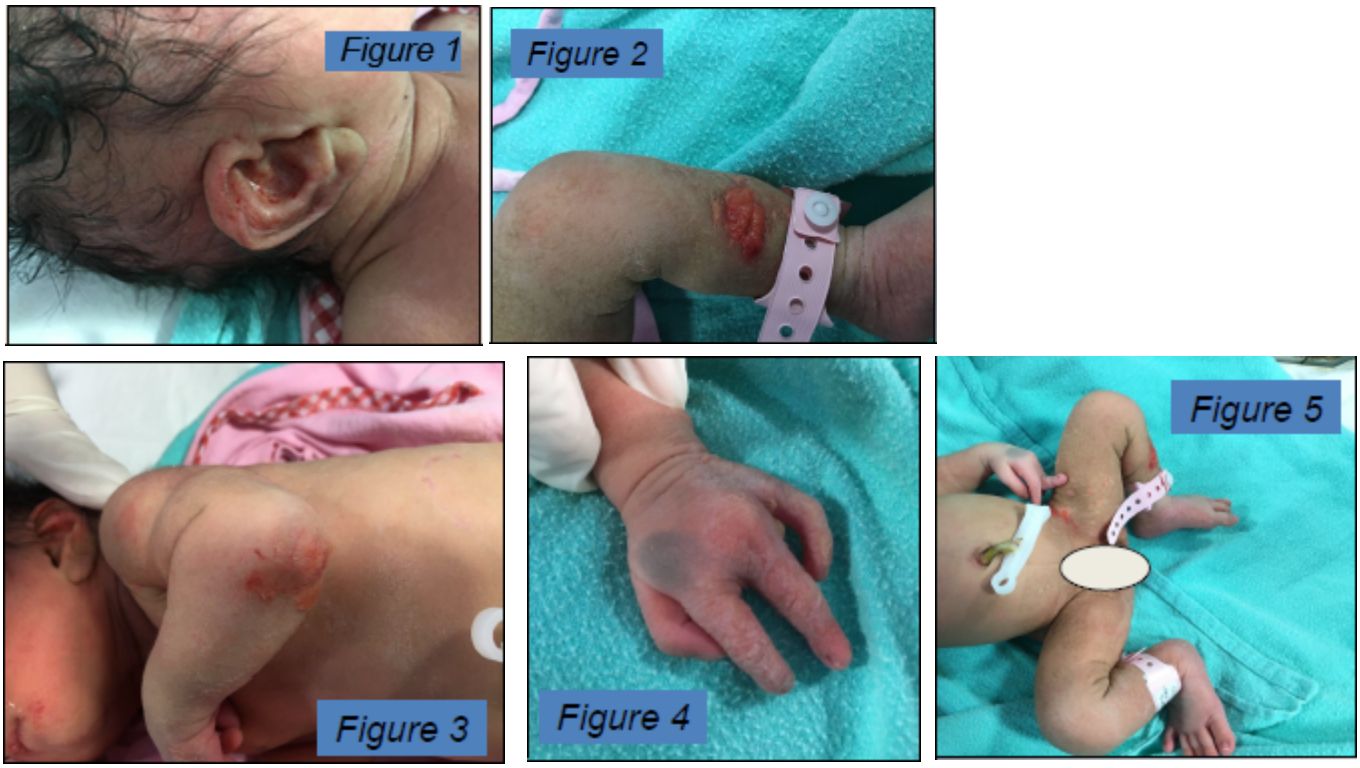
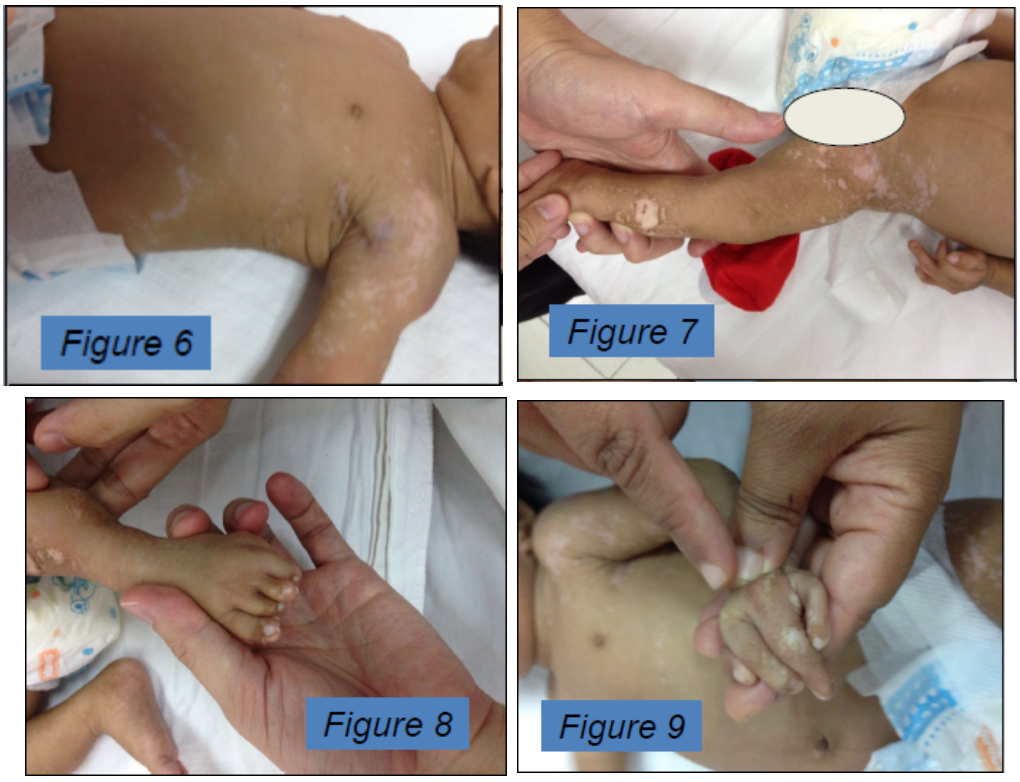


Figure 6-9. At 2 months old



Discussion

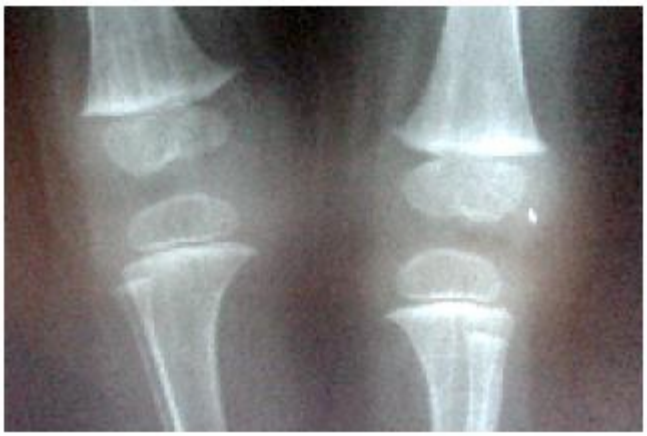
Goltz syndrome or Focal Dermal Hypoplasia (FDH) is an uncommon multisystem disorder. It involves all the three tissue layers i.e. ectoderm (skin, eyes), mesoderm (bones, teeth) and endoderm with variable expression. In our patient, genetic study to pinpoint the exact nature of the defect could not be done due to lack of the facility and parents were not agreeable for skin biopsy. Therefore the diagnosis was clinical as evident by the typical cutaneous lesions and skeletal malformations.

Cutaneous lesions are regarded as an essential component of this condition. At birth, the lesions may present as erosions or the typically red, hypopigmented atrophic macules arranged in a linear or Blaschkoid pattern which usually heal soon after birth leaving behind typical atrophic scar. Fat herniation and raspberry papillomas are the other 2 characteristic cutaneous features observed. Fat herniation typically appears as soft, pink- brown nodules overlying the thin atrophic skin. Raspberry-like papilloma occur most frequently at junctions between mucosa and skin, on or around lips, in the vulval and perianal areas. Papillomas can also be seen at other sites - on the fingers and toes as in our patient (noted during her 2-month old review).

Dental anomalies are the commonest extracutaneous manifestation of FDH - such as enamel defect, slow eruption of defectively formed teeth and absence of teeth.

Skeletal malformations are seen in almost 80% of the patients. Typical skeletal system findings are also noted in our patient, namely, oligodactyly, syndactyly and ectrodactyly (Figure 4,9). A "lobster claw" type of deformity is particularly characteristic. Radioimaging may show striated bones (osteopathia striata - Figure 10).

Figure 10. Linear radiodense parallel striations in a juxta-articular location suggestive of osteopathiastriata



The hair of these patients are usually sparse and brittle. Localized areas of alopecia are sometimes seen. (Figure 1)

Ophthalmic manifestations of FDH occur in 40% of cases. Ocular colobomas and microophthalmia are the most frequent manifestations.

Patients with FDH may have distinctive facial features - pointed chin, small ears, notched nostril and facial asymmetry. These features are typically subtle.

FDH generally involve both sides of the body. However, there are few reported cases of FDH with all the manifestations occurring unilaterally.

Management of FDH is mainly supportive. As it is a multisystem disorder, a multidisciplinary approach is needed. Early recognition of Goltz syndrome is important as this allows early intervention by ophthalmologist, occupational therapist, dentist, dermatologist, orthopedician and paediatrician.

Summary

FDH is a rare multisystem discorder. The mnemonic ***FOCAL*** can be used to remember the key features of this syndrome: **F**emale sex; **O**steopathia striata; **C**oloboma; **A**bsent ectodermis-, mesodermis- and neurodermis-derived element and **L**obster claw deformity. The evaluation, treatment and further management of a child with Goltz syndrome involve a multidisciplinary approach with the goal of achieving optimal functional and aesthetic results. The overall prognosis in females is good and life expectancy is normal. Proper genetic counselling, emphasizing on X-linked dominant inheritance pattern should be provided to parents with affected offspring.

Acknowledgement

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References

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