Clinics in Surgery

Case Report
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Goldenhar Syndrome: Case Report & Review of the Disorder

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Abstract

Goldenhar syndrome is an autosomal dominant genetic disorder in which there is abnormal prenatal development of the head and face leading to subsequent asymmetry of craniofacial structures. Most often, those affected will have very small and asymmetric ears and mouth with hypoplasia of the mandible and can also have missing ears and malformations of the eyes, vertebrae and palate. A case report of an adolescent with Goldenhar Syndrome that was seen is presented along with a brief review of the disorder in order to enlighten Podiatric physicians of this rare syndrome.

Introduction

Goldenhar syndrome is an autosomal dominant genetic disorder in which there is abnormal prenatal development of the head and face leading to subsequent asymmetry of craniofacial structures [1]. Most commonly, individuals affected will have very small and asymmetric ears and mouth with hypoplasia of the mandible [2,3]. Moreover, children affected can also have missing ears and malformations of the eyes, vertebrae and palate [4]. The syndrome is also known as Hemifacial Microsomia [5] and Oculo Auriculo Vertebral Spectrum [6]. A literature search revealed orthopedic [1], dermatological [4] and cardiac [7] manifestations; however the only mention regarding the foot was that of clubfoot which can occur in 20% of cases [8]. This article will serve as an introduction to this rare malady so that podiatric physicians are more acquainted with it should they have a patient with the disorder.

Case Presentation

KP, a twelve year old female presented to the office for evaluation of a growth on the bottom of her right foot, present since she was four years of age. The child's mother stated that when the lesion appeared, her child was worked up with what she recalls were X-rays, blood tests and a PET scan and was eventually told it was nothing that needed treatment. Since that time it had slowly grown and more recently it began to become painful and at times bleed spontaneously. No prior treatment had been sought since she was four years old. She was born full term to non consanguineous parents after an uneventful pregnancy. Her past medical history was significant for Hemifacial Microsomia, Asthma and Depression. She underwent facial reconstructive surgery to repair her mandibular and ocular deformities at the age of three and also had a tonsillectomy when she was eight. She had been taking fluticasone propionate, montelukast and sertraline as per her PCP and had no known allergies. She is an otherwise active seventh grader with two older unaffected siblings. Physical exam revealed a well nourished and pleasant twelve year old female, ambulating well without limp or

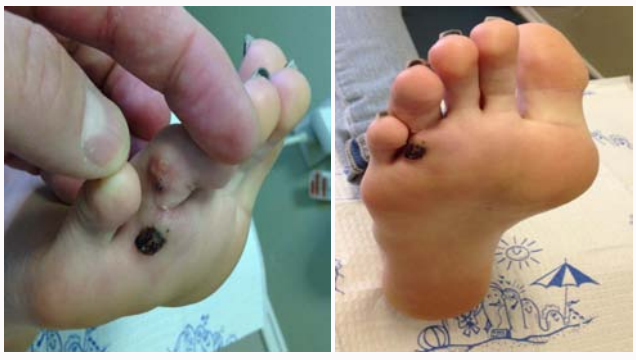

Figure 1 and 2: Hemmorhagic lesion upon presentation.

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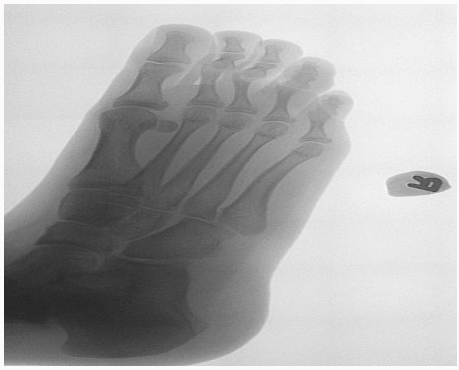

Figure 3: Radiograph displaying mild soft tissue swelling with no osseous abnormalities.

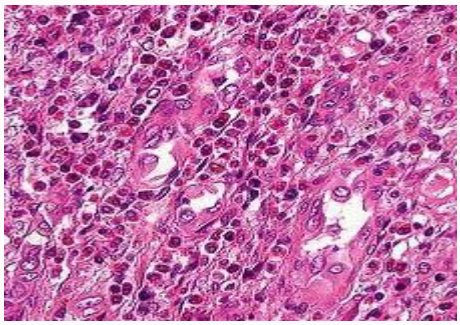

Figure 4: Microscopic analysis revealing hemangiomatous lesion with no cellular atypia.

assist. Lower extremity neurovascular exam was within normal limits with no focal deficits. A 12 mm hemorrhagic and tender lesion was present at the plantar aspect of the right 5th metatarsophalangeal joint with a similar 4mm lesion seen at the lateral aspect of the right 4th proximal interphalangeal joint (Figure 1 and 2). X-rays of the foot revealed mild soft tissue swelling with no osseous abnormalities or soft tissue calcification and open growth plates consistent with her age (Figure 3). Pathology revealed the lesion to be a hemangiomatous lesion with no malignant features (Figure 4).

Discussion

Goldenhar Syndrome affects approximately 1 in 3000-5000 births; the cause is unknown and can occur in families with no history of genetic disorders. In addition to visible irregularities such as cleft lip and palate, scoliosis and or kyphosis along with the aforementioned deformities, children can have respiratory, renal, cardiac and central nervous system defects [9]. Diminished pulmonary functions are often a result of improper spinal curvature with or without missing or fused ribs [10]. Moreover, unilateral hearing loss can occur and can be partial or full on the affected side [4].

Conclusion

A case report and description of Goldenhar Syndrome has been presented. Although the syndrome affects multiple body systems, children afflicted can have an excellent prognosis: The disorder carries no cognitive disabilities and those afflicted can live a normal life with a normal lifespan. As adults, they can reproduce without any greater chance of having children with the disorder than the general public.

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