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A 3-month-old male infant with Goldenhar syndrome: A clinical case report from Woldia, Northeast Ethiopia

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Abstract

Goldenhar syndrome is a multifactorial congenital anomaly that involves structures that develop from the first and second pharyngeal arches. In this report, we present a clinical case of a 3-month-old male infant diagnosed with Goldenhar syndrome, born to a known retro-viral infected mother who was receiving antiretroviral therapy. The baby was brought to the hospital with complaints related to upper respiratory system. On examination, he had typical signs and symptoms of Goldenhar syndrome: an asymmetrical face with small left facial bones, a low-set ear, left anophthalmia, an atretic left ear with only small ear appendages, and a complete cleft lip and palate. His family had no history of birth defects or exposure to the known causes of birth defects. The baby was treated for severe community-acquired pneumonia, the diagnosis for his current presentation to our hospital, and he is now on multidisciplinary follow-up for possible medical and surgical management of the Goldenhar syndrome.

Keywords

Goldenhar syndrome, congenital anomaly, pharyngeal arches, retro-viral infection

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Introduction

Goldenhar syndrome (GS) is a developmental anomaly that involves structures derived from the first and second pharyngeal arches.[1] It is also called Franceschetti Goldenhar syndrome, facio auriculo vertebral spectrum, first and second pharyngeal arches syndrome, or oculo-auriculo-vertebral syndrome.[2] Carl Ferdinand Von Arlt was the first physician from Germany to record this anomaly in 1845.[2] However, Maurice Goldenhar best described the clinical features of GS in 1952, due to this reason, the credit for GS discovery was given to him.[3] In 1963, Gorlin named this syndrome oculi auriculo-vertebral.[4] Smith in 1978 used the term facio-auriculo-vertebral sequence to describe both GS and hemifacial microsomia.[5] The incidence of this syndrome is 1:3500 to 1:5600 with a male-to-female ratio of 3:2.[2] Most of the cases are sporadic in nature; however, autosomal dominant inheritance has also been reported. There is no suggested geographic or racial predilection.[6]

The clinical presentations of GS include epibulbar desmoids, anophthalmia, microtia, mandibular hypoplasia, and vertebral anomalies.[7–9] This syndrome may also present with heart disease (commonly tetralogy of Fallot, septal defects, and situs inversus)[10] and hypoplasia of the zygotic, mandibular, and maxillary bones.[11] Some authors have also pointed out facial muscle hypoplasia, anatomical and morphological abnormalities of the tongue, vertebral abnormalities, anomalies of the eyes,[12] cleft lip and cleft palate,[11] disturbances of the central nervous system, and other visceral anomalies.[12]

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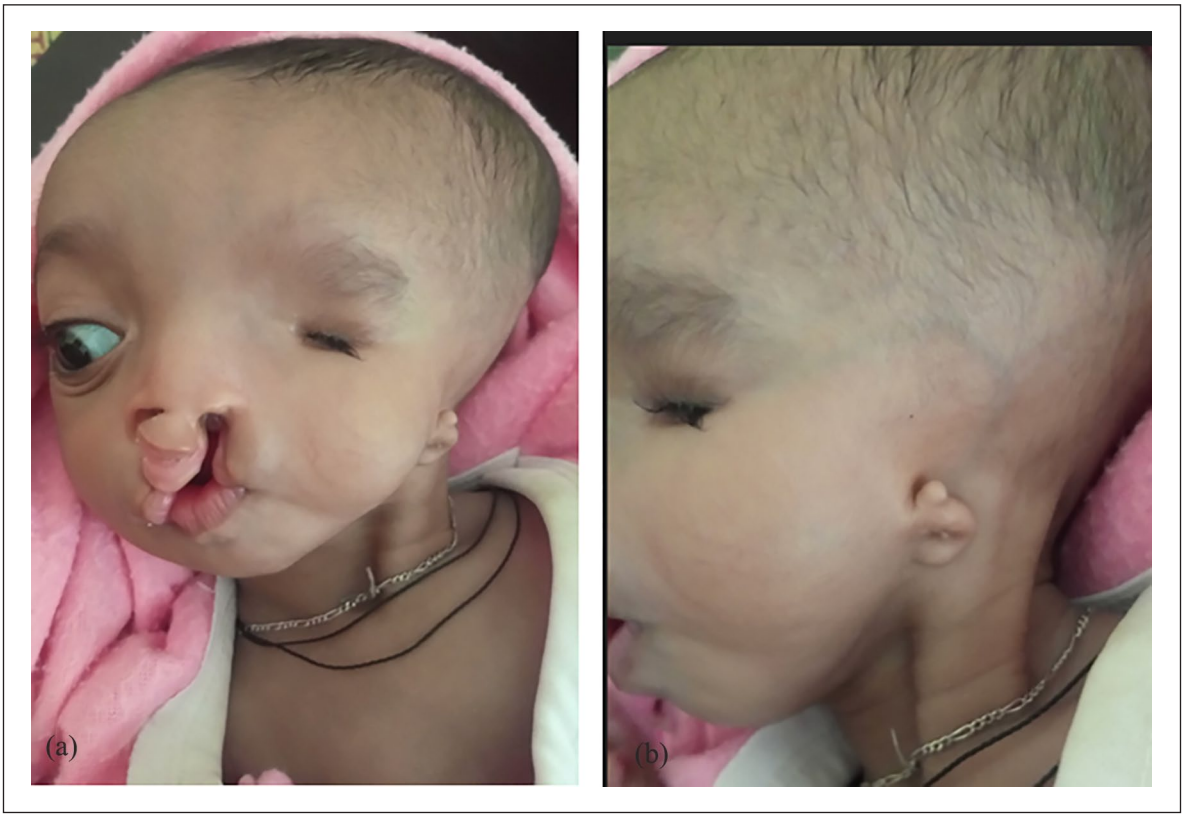

Figure 1. Photograph of a 3-month-old baby boy diagnosed with Goldenhar syndrome. (a) Anterior view showing cleft lip, cleft palate, anophthalmia, and asymmetrical face with mandibular hypoplasia. (b) Lateral view showing atretic left ear with only small ear appendages.

To the best of our knowledge, no published articles reported the incidence of this anomaly in Ethiopia. Thus, our case report presents a case of GS in a 3-month-old male patient from Woldia Comprehensive Specialized Hospital, Northeast Ethiopia.

Case presentation

This is a 3-month-old male infant born full-term to a para-one mother who had regular antenatal care follow-up at a nearby health center where there is no ultrasound service. This 32-year-old mother is a known retroviral infected (RVI), by human immunodeficiency virus (HIV), patient for 8 years, but discontinued antiretroviral therapy (ART) for the past 6 years. Her follow-up was at a nearby health center but no recent viral load determination. Otherwise, there was no history of similar illnesses in the family and no history of drug intake other than those mentioned above. The mother had no history of hospital admission or other known medical illness. She has no blood relationship (consanguinity) with her husband.

She gave birth at a nearby health facility via cesarean section due to failure of induction after prolonged labor and a ruptured membrane. At birth, the infant had no birth-related complications. However, it was noticed that he had a dysmorphic face with a small left eye, absent left ear opening with only simple appendages, and defects in the lips and palates (Figure 1(a) and (b)).

Since exposure to HIV, the baby received the recommended antiretroviral prophylaxis (nevirapine, zidovudine, and cotrimoxazole). He was on exclusive formula feeding and to check the serostatus, DNA PCR done at 6 weeks of age tested negative. Currently, the baby is presented to Woldia Comprehensive Specialized Hospital with complaints of cough, fever, and fast breathing 3 days duration after preceding upper respiratory tract symptoms and contact with his caregiver having the same complaint. Since birth, no abnormal body movement or changes in mentation were observed. On physical examination, asymmetric facial features with small left facial bones, low set ear with left anophthalmia, an atretic left ear with only small ear appendages, a complete cleft lip, and a cleft palate were observed (Figure 1). However, no other morphological and or congenital abnormalities, in any other system, were observed. In addition, chest X-ray, echocardiography, transfontanel as well as abdominal ultrasound did not show visceral abnormalities. Finally, the baby was admitted to the hospital for 7 days and treated for severe community-acquired pneumonia (ampicillin and gentamycin for 7 days) and is currently undergoing multidisciplinary follow-up for possible medical and surgical management of the GS. The diagnosis of this case was solely made based on clinical presentation. As per many literatures, the characteristic combination of external ear

anomalies and facial underdevelopment is the hallmark of this GS. Due to a lack of facility, no genetic analysis was done.

Discussion

GS is a clinically heterogeneous anomaly that makes it difficult to clearly agree on its diagnostic criteria. The symptoms vary in type and severity from case to case. However, it is well known that this syndrome includes anomalies of the eyes, ears, and face.[4] Our report presented a case of baby with GS born to a known HIV-infected mother who took ART. This glares the light for further investigating the association between GS, HIV infection, and ART.

Auricular appendages and microtia are present in almost all GS cases.[11] Similarly, the current case had a unilateral atretic left ear with only a small auricular appendage. In addition, as many studies have reported, the current patient had unilateral mandibular hypoplasia and so an asymmetric face. In patients with GS, multiple ocular features, such as coloboma of the eyelid, iris, and retina, epibulbar choristoma, dermoid, and microphthalmia have been reported.[13,14] However, in our case, unilateral absence of the eye (anophthalmia) was observed.

Even though some studies have reported cardiovascular anomalies associated with GS,[15,16] our case did not reveal this and no cardiac anomalies were detected. The study done by Tasse et al.[8] reported that in GS patients, the presence of ocular anomalies and or orofacial clefts is indicative of brain anomalies.[10] Although our patient presented with ear and eye abnormalities and cleft lip and palate, no such associated anomalies were detected.

With regard to the cause and pathogenesis of GS, which is still not well understood, it is believed to be multifactorial, both genetic and environmental. Some authors associated the development of GS with disturbance of blood supply to the first and second pharyngeal arches and abnormal formation of neural crest cells.[17] On the other hand, during pregnancy, maternal exposure to viral infections such as rubella and influenza, as well as drug intake (such as thalidomide and tamoxifen) can also cause GS.[10] In the present case, there was no documented family history of congenital anomaly. However, the mother has been a known RVI patient under ART for 8 years, which has been discontinued in the last 6 years. Therefore, the association between RVI/ART and GS requires further investigation.

In case when the pregnant mother has a known infection, especially one that affects the development of the fetus, an early ultrasound scan with fetal echocardiography can help to detect the anomalies. In sonographic evaluation, a dysmorphic face with asymmetry, ear abnormalities, and cleft lip/palate can help the prenatal diagnosis of GS. Thus, comprehensive evaluation of the fetus, counseling and supporting the mother, regular monitoring of the pregnancy, and collaborative treatment are needed.[18,19] The treatment of GS varies based on the type and severity of the anomalies and the extent of vital organ involvement. Collaborative management among experts, pediatricians, ophthalmologists, neurosurgeons, orthodontists, maxillofacial surgeons, and ear–nose–throat specialists is required.[6,20,21] As a complication, GS may result in difficulties with eating, breathing, hearing, and speech. Aspiration pneumonia may also occur. In rare cases, if there is experience of immunodeficiency, the individual may be predisposed to recurrent infections. Thus, timely, proper, and holistic management is required.[22]

In conclusion, our report presented a rare congenital anomaly in the Ethiopian population, the first of its kind to the level of our literature search, particularly, born to an RVI patient on ART for 8 years needing further investigation to elucidate possible cause–effect relationship. Health professionals should encourage early ultrasound scans for better detection of anomalies. Comprehensive and multidisciplinary management should be employed for an individual with GS.

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Data availability

All data are incorporated in the manuscript.

Declaration of conflicting interests

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Ethical approval

Written informed consent was obtained from the parents of the baby for taking the picture and publishing the case. We have also obtained ethical approval from Woldia Comprehensive Specialized Hospital.

Informed consent

Written informed consent was obtained from the patient(s) for their anonymized information to be published in this article.

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