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Case Report

Unravelling the Mystery: A Case Report of a Baby with Suspected Goldenhar Syndrome

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ABSTRACT

We present a case of a preterm male infant who was admitted to the neonatal intensive care unit at our affiliated hospital. The infant displayed multiple congenital anomalies involving the CNS, eyes, ears, heart and the skeletal system along with subtle dysmorphic features. The anomalies were not detected on antenatal investigations. Despite a comprehensive workup postnatally, including whole exome sequencing and mitochondrial gene sequencing, all genetic tests returned normal results. Given the clinical presentation and known limitations of genetic testing, Goldenhar syndrome is suspected but remains unconfirmed. After receiving supportive care in the neonatal intensive care unit for a month, the infant was discharged in stable condition. This case emphasizes the challenges of diagnosing rare congenital syndromes in the absence of conclusive genetic findings.

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INTRODUCTION

Congenital anomalies are the structural or functional anomalies or metabolic disorders which are present at the time of birth.[1] Congenital anomalies are a leading cause of neonatal morbidity and mortality globally. The prevalence of congenital anomalies varies globally and is influenced by factors such as genetic predisposition, environmental exposures and maternal health.[1,2]

Some congenital anomalies are the manifestation of a syndrome. A syndrome is a collection of symptoms and signs that occur together and are characteristic of a particular condition or disease but may not necessarily have a single identifiable cause.[3]

Early detection through prenatal screening and diagnostic imaging plays a crucial role in the identification of congenital anomalies, enabling timely interventions and better outcomes. We present this case to highlight the challenge faced in diagnosing a baby with multiple congenital anomalies with an emphasis on the importance of early recognition and comprehensive care. The consent of the parents was taken prior to writing this manuscript.

CASE REPORT

A male preterm infant was admitted to the neonatal intensive care unit (NICU) shortly after birth. The baby was born at our affiliated Burjeel Speciality Hospital, Sharjah in August 2024. The Cesarean section was performed at 31+5 weeks of gestation due to severe oligohydramnios and suspected placental abruption secondary to preterm prelabour rupture of membranes (PPROM) for one month. The baby weighed 1900 grams and had a poor APGAR score. As a result, resuscitation was performed by the neonatal team and the new-born was intubated. Later the baby was transferred to the NICU and was given respiratory support, IV antibiotics, IV fluids and other supportive therapies including surfactant, which successfully stabilized the patient's condition.

On detailed examination after stabilisation, the baby was mildly hypotonic, had subtle dysmorphic features and multiple congenital anomalies (Figure 1A and 1B). The anterior fontanelle was small in size and there was cloudiness in the right cornea and microphthalmia, bilateral nystagmus, rudimentary right external ear, wide-spaced nipples and grade 3/6 pan-systolic murmur at left lower sternal border. In addition, left-sided deviation of angle of the mouth was noted.

The patient was the fifth child of the parents, who are second-degree cousins. The pregnant mother was initially booked at another medical facility. Initially it was a spontaneously-conceived low-risk pregnancy and the first trimester was unremarkable with no significant findings. All the initial booking tests and NT scan were normal. However, the combined test or serum screening were not performed.

A 2-D anomaly scan performed at 21 weeks of pregnancy detected fetal bilateral pyelectasis with a

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suspected small right orbit. However, no major anomaly was detected on the anomaly scan. A fetal medicine was suggested and sought. She was screened for gestational diabetes at 24 weeks which was not detected. At 27 weeks of gestation, the mother had PPROM. She was given antibiotics and a single course of antenatal corticosteroids with outpatient follow-up visits later. She was monitored twice-weekly until labor with no apparent signs of chorioamnionitis or sepsis.



Figure-1A: Subtle dysmorphic features were noted including cloudiness of the right cornea and unilateral deviation of the angle of the mouth. Figure-1B: The external ear was rudimentary

At 31+5 weeks, the mother presented to the ER with heavy vaginal bleeding and suspected placental abruption. An ultrasound showed fetal breech presentation and severe oligohydramnios. Magnesium sulphate was administered and the patient was delivered by an uncomplicated emergency Cesarean section.

The baby remained at the NICU for 31 days. Meanwhile, investigations showed butterfly vertebrae with fusion of vertebrae at the D8-D9 level, right atretic external auditory canal and hypoplastic middle ear, staphyloma in the right eye, asymmetrical right lobe of the thyroid and hypoplastic superficial lobe of the right parotid. Fundoscopy confirmed congenital cataract, microphthalmia and microcornea while ventricular septal defect with a left-to-right shunt, patent ductus arteriosus and atrial septal aneurysm was detected on echocardiography. Abdominal ultrasound and X-ray of the urinary tract were normal. TORCH screening including PCR for cytomegalovirus, whole exome sequencing (WES) and mitochondrial gene sequencing were negative. The baby was later discharged from the hospital in a stable condition with a weight of 2.2 Kg.

DISCUSSION

The baby was born with multiple anomalies involving the CNS, eyes, ears, heart and the skeletal system. This case is important as it involves an infant born with a range of congenital anomalies despite normal infection screen and genetic testing. This case highlights the possibility of non-genetic factors influencing fetal development, offering educational value to healthcare providers in managing similar cases.

The differential diagnosis included Goldenhar syndrome and CHARGE syndrome. CHARGE syndrome is characterized by coloboma, heart defects, atresia of the choanae, retarded growth and genital and ear abnormalities.[4] Goldenhar syndrome, also called oculo-auricular vertebral (OAV) spectrum, is commonly found in male babies, is unilateral and on the right side. It classically involves abnormalities in the development of ears, eyes and vertebrae and mandibular hypoplasia resulting in facial asymmetry and may include underdeveloped ears and spinal deformities.[5,6] The baby in our case had all the features of Goldenhar syndrome.

While the exact cause of Goldenhar syndrome is unknown, it is believed to result from sporadic genetic mutations or disruptions rather than from an inherited genetic pattern. The condition has been associated with mutations in the EDNRB gene and Tbx1 gene, both of which play important roles in the development of facial and cranial structures.[6,7] In many cases the condition appears to occur sporadically where 5p deletions, 14q23.1 duplications or abnormalities of chromosomes 18 and 22 may be observed.[6,7]

In this case, whole exome and mitochondrial gene sequencing were normal. However, despite being powerful diagnostic tools, these sequencing techniques have some limitations.[8] WES focuses on sequencing the protein-coding regions of the genome which constitute about 1-2% of the entire genome.[8] Additionally, WES may not detect structural variants, such as large deletions or duplications and certain types of mutations or epigenetic modifications. Therefore, a negative result does not completely exclude the risk/carrier status for these disorders tested.[8]

We believe documenting this rare case will encourage and prompt further research into unexplained anomalies in infants to improve diagnosis, treatment and long-term outcomes for similar patients.

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Authors' Contribution

Following authors have made substantial contributions to the manuscript as under:

AR & AMHA: Data acquisition, data analysis, critical review, approval of the final version to be published.

HASA & YFE: Study design, data interpretation, drafting the manuscript, critical review, approval of the final version to be published.

Authors agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

REFERENCES

1. Malherbe H, Modell B, Blencowe H, Strong K, Aldous C. A review of key terminology and definitions used for birth defects globally. J Community Genet 2023; 14: 241–262. https://doi.org/10.1007/s12687-023-00642-2
2. Türkbay D, Canpolat FE, Derme T, Altuğ N, Yılmaz Y. The birth prevalence of selected major congenital anomalies: Six-years' experience in a tertiary care maternity hospital. Turk Pediatri Ars 2020; 55(4): 393-400. https://doi.org/10.14744/turkpediatriars.2020.36097
3. Verma RP. Evaluation and Risk Assessment of Congenital Anomalies in Neonates. Children 2021; 8: 862. https://doi.org/10.3390/children8100862
4. Pearce JMS. Disease, diagnosis or syndrome? Pract Neurol 2011; 11: 91–97. https://doi.org/10.1136/jnnp.2011.241802
5. Goswami M, Bhushan U, Jangra B. Goldenhar Syndrome: A Case Report with Review. International Journal of Clinical Pediatric Dentistry 2016; 9(3): 278–280. http://doi.org/10.5005/jp-journals-10005-1377
6. Digilio MC, McDonald-McGinn DM, Heike C, Catania C, Dallapiccola B, Marino B, et al. Three patients with oculo-auriculo-vertebral spectrum and microdeletion 22q11.2. American J of Med Genetics Pt A 2009;149A:2860–2864. https://doi.org/10.1002/ajmg.a.33034
7. Strömland K, Miller M, Sjögreen L, Johansson M, Joelsson BE, Billstedt E, et al. Oculo-auriculo-vertebral spectrum: Associated anomalies, functional deficits and possible developmental risk factors. American J of Med Genetics Pt A 2007; 143A: 1317–13125. https://doi.org/10.1002/ajmg.a.31769
8. Wortmann SB, Oud MM, Alders M, Coene KLM, van der Crabben SN, Feichtinger RG, et al. How to proceed after "negative" exome: A review on genetic diagnostics, limitations, challenges, and emerging new multiomics techniques. J of Inher Metab Disea 2022; 45: 663–81. https://doi.org/10.1002/jimd.12507