

Images in clinical medicine



Clinical image of a 3-month-old male infant with Bohring Opitz syndrome demonstrating right eye anophthalmia with staphyloma

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Clinical image of a 3-month-old male infant with Bohring Opitz syndrome demonstrating right eye anophthalmia with staphyloma

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Image in medicine

A 3-month-old male infant presented to the outpatient pediatric department with multiple congenital anomalies and respiratory distress. The infant had complaints of cough, difficulty in breathing, and poor feeding since birth. On examination, dysmorphic features suggestive of Bohring-Opitz syndrome, a rare multisystem genetic disorder, were noted. Respiratory assessment revealed tachypnea with increased work of breathing. A sacral pit was also observed on physical examination. Ophthalmologic evaluation showed right eye anophthalmia with associated staphyloma. Echocardiography revealed a 2.5 mm ventricular septal defect with patent foramen ovale. Chest radiography findings were suggestive of bronchopneumonia, correlating with the presenting symptoms.

Laboratory investigations demonstrated hypothyroidism, indicating endocrine involvement. Based on the clinical presentation and supportive investigations, a diagnosis of Bohring-Opitz syndrome with associated congenital cardiac defects, ocular malformation, sacral anomaly, bronchopneumonia, and hypothyroidism was established. The patient was managed with supportive care including oxygen therapy, intravenous antibiotics, and thyroid hormone replacement therapy. The child showed clinical improvement with stabilization of respiratory distress during hospitalization. This case highlights the importance of early recognition and multidisciplinary management of rare congenital syndromes with multisystem involvement, ensuring timely diagnosis, appropriate intervention, improved clinical outcomes, and long-term follow-up for such complex pediatric cases.



Figure 1: clinical image of a 3-month-old male infant with Bohring Opitz syndrome demonstrating right eye anophthalmia with staphyloma