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Case Report

Variant of bohring-opitz syndrome: A rare case report

Ramesh Choudhary[1,*], Romesh Gauttam[1], Vishnu Pansari[1], Anand Kumawat

[1] *Dept. of Pediatric Medicine, Swai Man Singh Medical College, Jaipur, Rajasthan, India*

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ABSTRACT

Bohring–Opitz syndrome also known as Opitz C syndrome or Oberklaid–Danks syndrome is a rare syndrome. We are reporting a 2 months old male child with Bohring-Opitz like syndrome with all classical features and eventration of diaphragm (left side) which has not been reported yet with this syndrome. To our knowledge, a total of 23 cases with this syndrome have been reported in the medical literature to date and this is probably the first case report from India. Although there is overlap, a clinical distinction from the Bohring-Opitz syndrome and other syndromes seems possible, and thus a specific causal entity may be postulated.

1. Introduction

Bohring–Opitz syndrome was first described in 1999 by Bohring et al.,[1] who described four new patients and identified similarities with two patients who had previously been reported as having Opitz C syndrome.[2,3] As one of the patients was initially described by Oberklaid and Danks,[2] this syndrome is also known as Oberklaid–Danks syndrome. To our knowledge, a total of 23 cases have been reported in the medical literature till date and this is probably the first case report from India.

2. Case Report

This male child was the fourth child born to healthy, consanguineous parents. The pregnancy was complicated by polyhydramnios. Family history was uneventful. A 2-month-old boy presented with complaints of respiratory and feeding difficulty with poor weight gain. The infant was born at term. Birth weight was 1.7kg (<3rd centile), length 42 cm (<3rd centile), and Occipital Frontal Circumference 31.5 cm (50th centile). Now at 2 months weight was 2.3 kg (<3rd centile), length 50 cm(<3rd centile) and Occipital Frontal Circumference 33.5cm (50th centile).

On examination patient has highly characteristic combination of facial anomalies including facial anomalies including typical trigonocephaly/prominent metopic ridge, retrognathia, prominent eyes with hypoplastic supraorbital ridges, upslanting palpebral fissures, depressed nasal bridge, low-set ears, cleft palate and broad alveolar ridges, microcephaly, IUGR and short stature, joint abnormalities, abnormal tone, severe/profound developmental delay, susceptibility to infections, feeding difficulties, and high infant mortality.(Figure 1) Chest X-ray reveals left eventration of diaphragm and mediastinal shift to right side. (Figure 2) On examination patient has highly characteristic

combination of facial anomalies including facial anomalies including typical trigonocephaly/prominent metopic ridge, retrognathia, prominent eyes with hypoplastic supraorbital ridges, upslanting palpebral fissures, depressed nasal bridge, low-set ears, cleft palate and broad alveolar ridges, microcephaly, IUGR and short stature, joint abnormalities, abnormal tone, severe/profound developmental delay, susceptibility to infections, feeding difficulties, and high infant mortality.(Figure 1) Chest X-ray reveals left eventration of diaphragm and mediastinal shift to right side. (Figure 2)

* *Corresponding author.*
E-mail address: drrameshchoudhary@yahoo.co.in (R. Choudhary).

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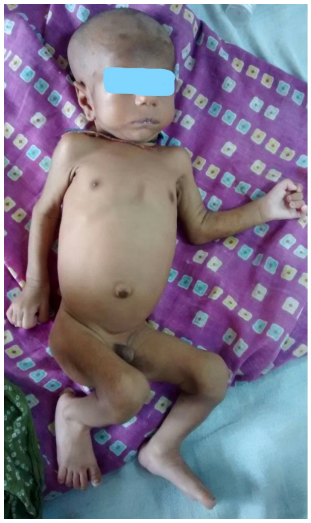

Fig. 1:

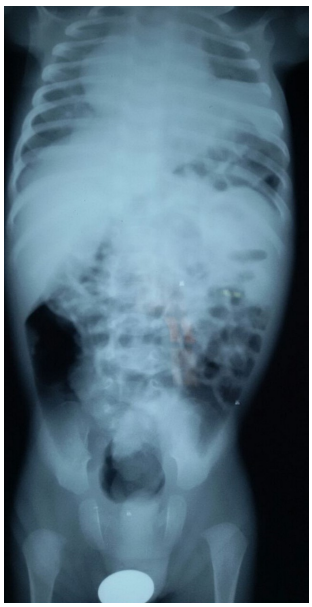

Fig. 2:

Ultrasound findings of kidney bilateral medullary nephrocalcinosis and bilateral cryptorchidism. Echocardiographic findings are patent foramen ovale L TO R SHUNT) and mesocardium due to mediastinal shift to the right side. CT scan, Ultrasound head and Electroencephalogram (EEG) was normal. Baby was managed with oxygen, intravenous antibiotics for infection and subsequently paladai feeding was started. Post discharge baby was enrolled on follow up for nutritional and development assessment.

3. Discussion

From the published patients and our patient, the consistent clinical features of this rare condition are becoming clearer. C syndrome which is also known as Opitz trigonocephaly syndrome is characterized by trigonocephaly, mental retardation, hypotonia, cardiac defects, redundant skin, and dysmorphic facial features.[4] Bohring et al. in 1999 reported four unrelated cases of a syndrome very similar to Opitz trigonocephaly (C syndrome) which differed on the basis of intrauterine growth retardation, cleft lip/palate, exophthalmos, retinal involvement, flexion deformities of upper limbs, dislocation of radial heads, and forehead hirsutism.[1] This syndrome is called Bohring-Opitz or C-like syndrome.[1–4]

Our patient's anomalies were very similar to Bohring-Opitz syndrome in respect of facial anomalies including facial anomalies including typical trigonocephaly/prominent metopic ridge, retrognathia, prominent eyes with hypoplastic supraorbital ridges, upslanting palpebral fissures, depressed nasal bridge, low-set ears, cleft palate and broad alveolar ridges, microcephaly, IUGR and short stature, joint abnormalities, abnormal tone, susceptibility to infections and feeding difficulties.(Figure 1) In addition our patient have eventration of diaphragm (left side).(Figure 2).

This case might be a new syndrome or a variant of Opitz syndrome with facial abnormalities, cleft palate, flexion deformities of the upper limbs, and eventration of diaphragm (left side). There remains a high rate of infant mortality in this condition (40%), most commonly owing to infections. If the early childhood period is survived, then many of the common problems, such as feeding difficulties and recurrent infections, become less problematic. Severe to profound developmental delay is universal, although there is some variability in terms of the level of communication and mobility achieved.[5]

4. Source of Funding

None.

5. Conflict of Interest

None.

Acknowledgments

Department of Pediatrics, S.M.S. Medical College, Jaipur, Rajasthan, India.

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Author biography

Ramesh Choudhary, Associate Professor

Romesh Gauttam, Assistant Professor

Vishnu Pansari, Associate Professor

Anand Kumawat, Junior Specialist

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