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PEDIATRIC ONCALL
CHILD HEALTH CARE

IMAGES IN CLINICAL PRACTICE

MULTIPLE CONGENITAL ABNORMALITIES

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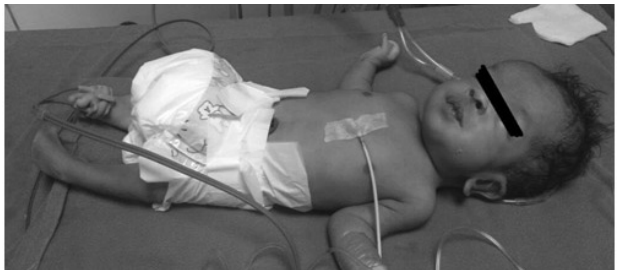
A 7 days old male infant was referred to us with sepsis, cardiovascular shock, and multiple congenital abnormalities. His birth weight was 2.24 kg and APGAR score was not known. At admission to our unit, he was very sick, had severe respiratory distress, shock (blood pressure was 30/18 mm of Hg), hypoxic and had active bleeding from the nasogastric tube. The abdomen was distended with decreased bowel sounds and there was hepatosplenomegaly. On cardiovascular system examination, he had a systolic murmur. He had dysmorphisms in form of micrognathia, flat nasal bridge, high arched palate, congenital talipes equinovarus deformity, polydactyly (bifid right thumb), limb length discrepancy and bilateral undescended testis (Figure 1). He was put on mechanical ventilation. Inotropes, intravenous (IV) fluids and IV antibiotics were started. Echocardiography showed patent ductus arteriosus (PDA) with an atrial septal defect (ASD). Ultrasound (USG) abdomen and scrotum revealed bilateral undescended testis with right-sided hydrocele. USG cranium revealed hypoplasia of the corpus callosum. Blood culture grew Enterobacter cloacae. Coagulation profile was abnormal. He was given multiple platelets, fresh frozen plasma and packed red blood cell transfusions. Gradually he improved and was extubated on day 4 of admission. A repeat echocardiography on Day 9 of admission showed closure of PDA and only presence of ASD 5.5 mm. After stabilization, MRI brain was done that showed lobar holoprosencephaly, hypoplastic corpus callosum, pons and cerebellum with the prominent fourth ventricle. TORCH titers were negative and ophthalmological evaluation was normal. Karyotype analysis proved the diagnosis.

What is the syndrome?



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Karyotype analysis of the patient revealed 46 XY, del (13q22 -32). It is also known as Orbeli syndrome based on the Russian physician who described the syndrome in 1971. It is a partial monosomy involving the long arm of chromosome 13. Patient presents with varying phenotypic features as per deleted band of the chromosome 13q and are classified into 3 groups. Deletions involving chromosome 13 proximal to q32 band can result in phenotypes such as mild mental retardation, growth delay and various dysmorphic features. When loci of q32 gets deleted, it leads to severe congenital malformations including brain anomalies. titers form is deletion distal to q32 which leads to severe mental retardation, but no brain malformation or growth delay.[1,2] The ZIC2 (zinc finger protein of cerebellum 2) gene has been mapped in this region and absence of it by deletion is considered to lead to brain anomaly like holoprosencephaly.

Various clinical features of this syndrome are low birth weight, growth failure, psychomotor retardation, craniofacial abnormalities, microcephaly, brachycephaly, hypertelorism, upslanting palpebral fissures, high arched palate, coloboma, prominent nasal bridge, clinodactyly, syndactyly, dislocation of hips, a , congenital heart disease, renal abnormalities, cryptorchidism, hypospadias and anal atresia.[2] There is no specific treatmentwas given exact an is unknown. Children with 13q deletion syndrome should be followed up continuously by a neurologist, pediatric surgeon, and a rehabilitation physician, who can also coordinate interventions with different specialists.

Compliance with Ethical Standards

Funding: None

Conflict of Interest: None

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