DOI: 103126/JNPS.V4113

Case Report

Infantile Hypertrophic Pyloric Stenosis in an Infant with Tetralogy of Fallot: Association or Coincidence?

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Article History
Received on - 2023 Aug 24
Accepted on - 2023 Oct 17

Keywords:
Infantile hypertrophic pyloric stenosis; tetralogy of fallot; pyloromyotomy

Online Access

DOI: https://doi.org/10.60086/jnps477

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Abstract

Infantile hypertrophic pyloric stenosis is an important cause of vomiting in apparently healthy infants, typically between two to eight weeks of life. It mostly occurs as an isolated abnormality but there are cases reported to be associated with congenital anomalies. We report a case in a two month old infant who presented with non-bilious vomiting associated with feeding for 15 days. He was already diagnosed to have pink tetralogy of Fallot. He was diagnosed as a case of hypertrophic pyloric stenosis on the basis of ultrasonography and barium studies and underwent pyloromyotomy. Post-operative recovery was uneventful and he has no associated complications on follow-up.

Introduction

Infantile Hypertrophic Pyloric Stenosis (IHPS) is caused by gradually increasing hypertrophy of the pyloric sphincter muscle leading to gastric outlet obstruction (GOO).[1] Its incidence is estimated to be 1 - 3.5 infants per 1000 live births.[2] Symptoms appear between two to 12 weeks of age; and include non-bilious projectile emesis, repeated episodes of dehydration and failure to thrive.[1,2] The etiology of IHPS is not very well-understood; and multiple environmental / genetic factors play role in its pathogenesis. Risk factors for IHPS include male gender, premature birth, first-born children, bottle-feeding, smoking history during gestation or young age of mother, and treatment of young infants with macrolides.[2] A positive family history has been described in up to 47.9% of infants with IHPS.[3] IHPS has also been associated with CHARGE association, Edward's Syndrome, Cornelia-de-Lange Syndrome, Turner's Syndrome and Smith-Lemli-Opitz Syndrome.[4] Association of congenital heart disease (CHD) and isolated IHPS is not well-established. However, a recent analysis demonstrated that infants with CHD have a relative risk of 2.6 of suffering from IHPS.[1] We are reporting a rare case of a two months old male who had IHPS with tetralogy of Fallot (TOF). To the best of our knowledge, no such association has been reported in South Asian region so far.

Case report

A two months old male presented at Paediatric Medical Emergency with complaints of five to six episodes of projectile non-bilious vomiting per day associated with feeding for 15 days. There was no associated fever or loose stools. He was a known

case of TOF, diagnosed during early infancy during routine examination. He, however, has remained acyanotic since birth and was kept on follow-up as a case of pink TOF. He was born to consanguineous parents with insignificant antenatal or birth history. He had three siblings with no significant family history including CHD. Two weeks before presentation, he started vomiting for which he had been given oral / injectable medications at local hospitals, and was started on anti-regurgitation formula with no response. He was active and alert with normal anthropometry (Weight 3.5 kg, length 57 cm, OFC 37 cm) and stable vitals. Abdominal examination on test-feeding revealed a palpable mass in the epigastrium. Precordial examination revealed an ejection-systolic murmur at left-lower sternal border; while rest of systemic examination was unremarkable.

Lab investigations showed hemoglobin 11.4 g / dL, TLC 8600 / µL and platelets count 7,34,000 / µL. Serum electrolytes and blood gases revealed hypochloremic metabolic alkalosis (pH-7.470, serum sodium 143 mmol / L, potassium 4.0 mmol / L, chloride 98 mmol / L, HCO_3 32.5 mmol / L). His liver, renal function tests and coagulation profile were all normal. Echocardiography revealed TOF-like anatomy with 50% over-riding of sub-aortic ventricular septal defect, bidirectional shunt and RVOT gradient of 35 mmHg. Ultrasound (USG) abdomen showed pylorus length of 17 mm and thickness of 13 mm. Contrast meal studies performed were suggestive of gastro-esophageal reflux with hold-up of contrast at the level of pylorus. No contrast could be passed into duodenum after half hour (Figure-1). A diagnosis of IHPS was made and pyloromyotomy was undertaken. Post-op recovery was uneventful. He remained admitted for three days post-operatively and nasogastric / oral feed was built gradually. On follow-up after one month, patient had improved symptoms, good weight and length gain, and no surgery-related complications were observed.

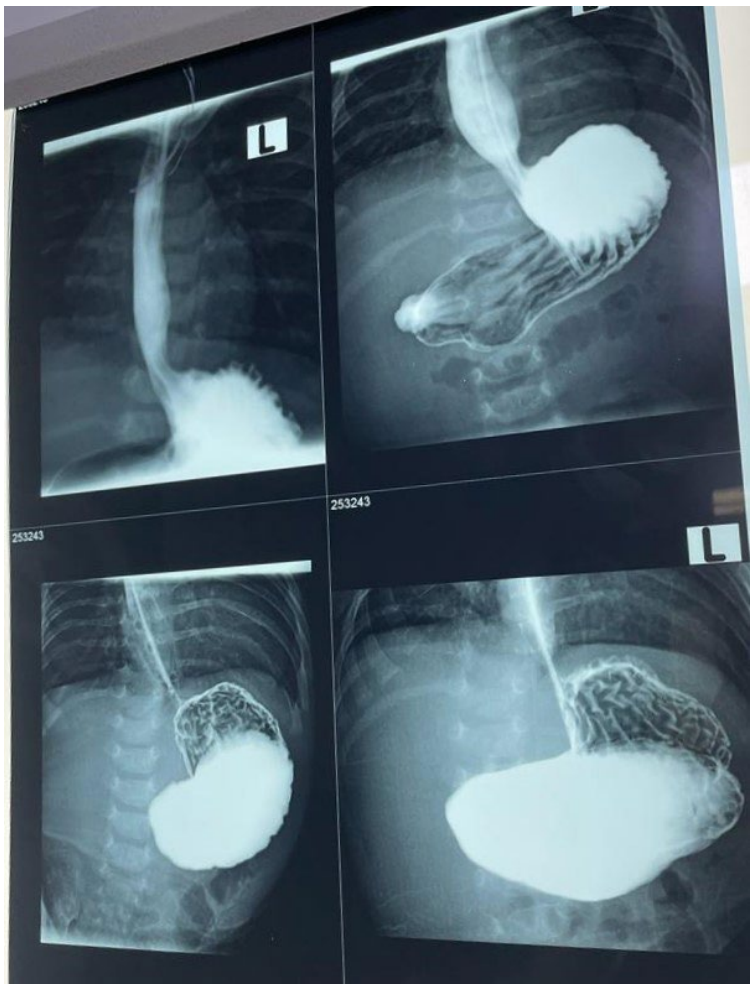

Figure 1: Barium meal of patient showing hold-up of contrast at the level of pylorus

Discussion

IHPS is the commonest cause of GOO in newborns and should be considered in the differential diagnosis of vomiting in newborns. Diagnosis can be frequently delayed and different empirical treatment modalities including H_2-receptor blockers, proton-pump inhibitors and change of feeding formulas. Most of the time IHPS occurs as an isolated abnormality without additional birth defects, however it is reported to have non-causal association with CHD.[1,5] It is therefore suggested that some etiological factors may be common between the two conditions.

Different mechanisms have been postulated to explain co-occurrence of IHPS with TOF or cyanotic CHD in general. Regev et al reported a case of complete GOO in a neonate with TOF. However, USG failed to confirm pyloric muscle hypertrophy and the obstruction was spontaneously relieved. The obstruction was proposed to be a pylorospasm instead of an anatomical pathology.[6] In 2017, Srivastava et al described three cases of cyanotic CHD with post-operative feeding difficulties who were ultimately diagnosed as IHPS based on USG. However all three patients had been given prostaglandin infusions for 13 - 21 days before their respective cardiac surgeries.[7] Prostaglandin-E2 induced foveolar hyperplasia has also been previously described mimicking GOO in infants with cyanotic CHD.[8] However, these theories do not apply to index patient as he was acyanotic, and did not have any history of prostaglandin infusion.

One possible explanation for the occurrence of IHPS in TOF patients is the possible role of NK2 homeobox 5 (NKX2-5) gene. Mutations in the gene have been associated with TOF; while some animal studies have demonstrated that regulatory changes causing change in NKX2-5 expression may also have role in IHPS pathogenesis.[1] However, common genetic variations are yet to be demonstrated in both these conditions.

The differential diagnoses of IHPS in any neonate with GOO include antral polyp / web, idiopathic focal antral hyperplasia of gastric mucosa and pyloric / duodenal atresia.[9] However, the diagnosis is established after USG or upper gastrointestinal contrast studies. Pyloric thickness > 3 mm and pyloric length > 15 mm are considered diagnostic clues for IHPS.[10]

Conclusions

IHPS is a rare entity in infants. However, when diagnosed, association between cardiac anomalies and IHPS should be looked for.

Funding Sources: None

Conflict of Interest: None

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