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Unusual Presentation of Gastroesophageal Reflux with Corpus Callosum Agenesis, Cleft Palate and Mental Retardation

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Congenital anomalies of upper gastrointestinal tract (GIT) commonly present with frothing and persistent vomiting. We had a neonate who presented with recurrent apnea and cyanosis. On thorough investigation to our surprise the barium studies revealed presence of gastroesophageal reflux (GER). This patient also had other

congenital anomalies like agenesis of corpus callosum, hiatus hernia, cleft of soft palate and mental retardation.

Case Report

A 28-day-old male infant born of non-consanguineous marriage was delivered at full term and weighed 3 kg. He was admitted on 28th day of life with a weight of 2.2 kg and complaints of respiratory distress, feeding difficulties, regurgitation of feed and recurrent apnea. He was treated at Ahmedabad without much improvement. On admission to our hospital, examination revealed cleft of small palate, recurrent apneic spells with cyanosis, bradycardia and evidence of bronchopneumonia which was treated with antibiotics.

However, difficulties in feeding, apnea and cyanosis persisted and the patient was investigated. Apneic spells did not respond to aminophylline and there was no tracheoesophageal fistula on dye studies. Echocardiogram was normal. CT scan to rule out CNS anomalies as a cause of recurrent apnea, revealed agenesis of corpus callosum (*Fig. 1*). Subsequently, barium esophagogram showed Grade IV GER.

Medical treatment in the form of antacid, ranitidine and metoclopramide was given. In view of severity of GER and possibility of associated anomaly, the child was operated (Nissen fundoplication with repair of associated sliding hernia), following which the child was free of all symptoms for a period of 2 months. The above

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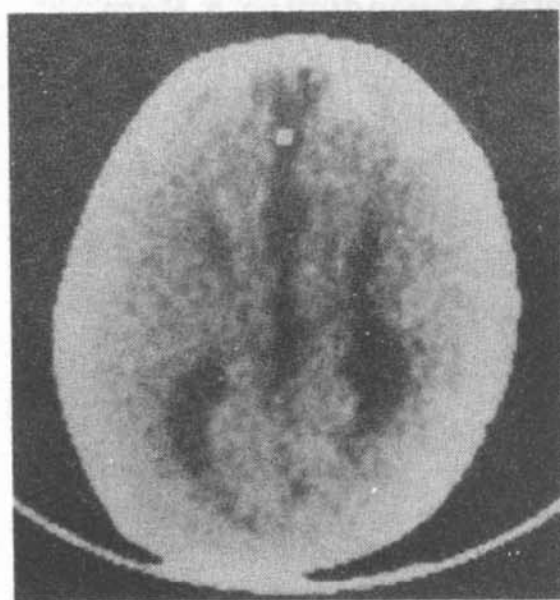


Fig. 1. CT Scan (head) showing corpus callosum agenesis.

symptoms reappeared after 2 months but they were less severe. At this stage, recurrence of GER was ruled out on barium study. At present the child is 11 months old, weighs 8 kg with moderate mental and motor retardation with minimal symptoms of GER. Karyotyping carried out at this stage was normal.

Discussion

Gastroesophageal reflux is very common in the first year of life and subsequently the incidence decreases markedly owing to maturation of lower esophageal sphincter (LES), change to a solid diet and assumption of upright position(1). Though the exact mechanism leading to GER is not very clear; various hypothesis have been advocated(2). Hiatus hernia is frequently associated with GER(3). Nearly 85% of patients presents with excessive vomiting during first week of life and the symptoms abate without treatment in 60% by the age of 2 years(4). Other symptoms noted are failure to thrive (26%), hematemesis and

recurrent pneumonia (18%), apneic spells (10%) and ALTE (apparent life threatening event), *i.e.*, symptom complex of apnea, pallor or cyanosis, altered muscle tone and choking or gagging(5,6). Diagnosis is based on barium esophagogram, esophageal scintiscan and can be confirmed with 24 hour esophageal PH probe studies. On barium studies, pathological reflux can be differentiated(7) and can be graded into 5 grades(8). Technical error leading to false positive and false negative results are known in both investigations. However, lower radiation exposure and follow up of patient over longer are the advantages of scintiscan over barium studies(2,9,10). A drop in esophageal PH to ≤ 4 for at least 8 seconds confirms the diagnosis of GER on PH probe testing which has high sensitivity and specificity. Combination of thickened feeding, positioning, bethenecol, antacid, ranitidine, metoclopramide(14) and cisapride(12) from the mainstay of medical therapy(11,12). Patients with hiatus hernia, severe esophagitis, esophageal stenosis and those who present with apneic spells or aspiration require surgery. Recurrence of GER is known in 3-5% of patients(13). Though association of mental retardation has been known(14), the combination of GER with hiatus hernia, agenesis of corpus callosum with cleft palate has not been documented previously.

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Neonatal Appendicitis: A Rare Cause of Surgical Emergency in Preterm Babies

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Neonatal appendicitis is a rare condition. The clinical diagnosis is difficult and majority of cases are diagnosed on laparotomy. This condition is associated with very high morbidity and mortality. We are reporting two cases of neonatal appendicitis in preterm babies managed in our neonatal intensive care unit (NICU) during the last one year.

Case Report

Case 1: A first of dizygotic twins, was born at 31 weeks of gestation to a primigravida who had pregnancy-induced hypertension. The infant weighed 1360 g (appropriate-for-dates) and did not suffer from any birth asphyxia. He passed small quantities of meconium during the first 3 days, but thereafter passed stools only once during the next six days; and that too after glycerine enema. The baby developed

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