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Extensive cervical spine and foregut anomaly in 'serpentine syndrome'

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ABSTRACT

INTRODUCTION: We report an extremely rare and challenging combination of congenital anomalies. Only five similar cases have been described in the English language medical literature to date.
PRESENTATION OF CASE: A male infant was born at 30^{+5} weeks gestation by emergency caesarian section. Cervical spine rachischisis, shortened oesophagus, intrathoracic stomach, atretic duodenum and absent spleen were noted, in addition to respiratory insufficiency. Gastrointestinal re-anastomosis, particularly oesophageal lengthening, was not feasible at the initial thoracotomy. Surgical stabilization of the cervical spine was unlikely to be successful until two years of age. Asplenia predisposed the infant to sepsis from encapsulated organisms, and recurrent respiratory infections occurred.
DISCUSSION: A close relationship exists between the upper gastrointestinal tract and cervical spine during embryonic development. An embryonic aberration at this level could account for all the deformities present in this infant. Tethering of the embryonic cervical oesophagus to the somites in the first trimester, preventing foregut elongation, and producing ischaemia at the coeliac axis, is suggested as the aetiology.
CONCLUSION: This case presented a challenge to the multi-disciplinary team involved in his management and prompted extensive consultation with international experts. After considerable counseling of the parents, care was directed towards palliation.

1. Case report

The authors obtained consent for this case report from the parents of the infant described.

A 37 year old healthy primigravida was admitted to a district hospital with antepartum haemorrhage. Both parents (non-consanginous) had been counselled antenatally following detection on ultrasound of a probable congenital diaphragmatic hernia. Amniocentesis at 22 weeks was normal and the pregnancy was otherwise uncomplicated. Chromosome analysis confirmed a normal male karyotype. No microdeletions or chromosomal duplications were found on further testing.

A male infant was born by emergency caesarean section at 30^{+5} weeks gestation. He was not dysmorphic and had a birthweight of 1.7 kg (50th–75th centile). Respiratory insufficiency at birth necessitated intubation, surfactant administration and mechanical ventilation prior to transfer to the regional neonatal intensive care unit. A chest radiograph demonstrated curling of an orogastric tube within the thorax, and cervical vertebral anomalies (Fig. 1). Upper gastrointestinal contrast studies confirmed a total intrathoracic stomach, and no contrast passed distal to the stomach (Fig. 2). Laparotomy and right thoracotomy on day 3 of life confirmed the presence of a shortened oesophagus, atretic proximal duodenum and absent spleen. Biliary structures were not observed. Both hemi-diaphragms were intact. The stomach was tethered by the shortened oesophagus and could not be mobilized into the abdomen at the time of the original surgery, therefore a proximal small bowel stoma was formed to facilitate enteral feeding. The blood supply to the stomach and duodenum was from a caudad direction arising from below the diaphragm and therefore assumed to be from the coeliac axis.

Patency of the distal small bowel and colon was demonstrated by contrast studies. Asplenia required phenoxymethylpenicillin prophylaxis from encapsulated organisms, which was delivered via the stoma. Absence of splenic tissue was confirmed later by the presence of Howell-Jolly bodies on blood film.

Enteral feeding proved difficult to establish, and the infant developed cholestasis with parenteral nutrition. Introduction of a hydrolysed formula with addition of aspirated bilious gastric contents to the feed enabled tolerance of full enteral feeding by day 36 of life.

Echocardiography showed a structurally normal heart. Ophthalmological examination was normal. Ultrasound of abdomen and renal tracts revealed normal kidneys and bladder, but absent gallbladder and pancreas. Although the porta hepatis was not accurately defined, the liver was present, with no significant intrahepatic bile duct dilatation.

Spinal investigation was prompted by an abnormal plain radiograph which reported a widened spinal canal and by a case series of

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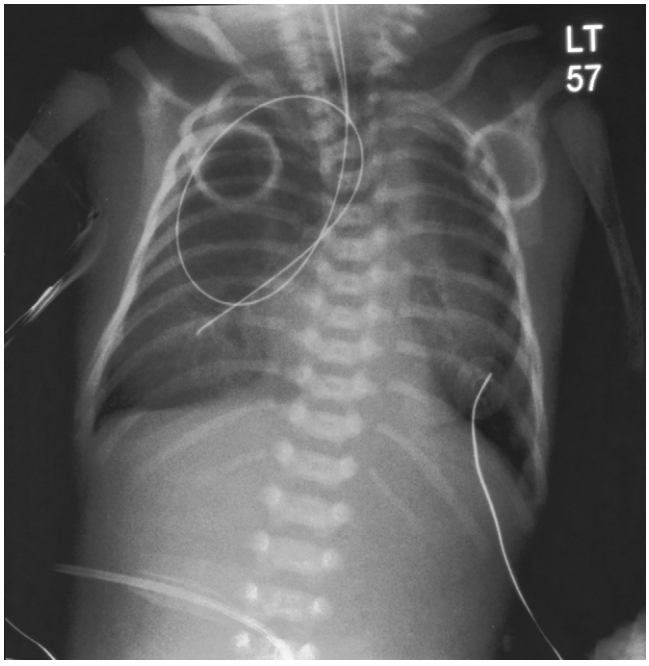

Fig. 1. Chest radiograph with feeding tube curled up in right hemithorax. Cervical vertebral body ossification centres are absent.

two infants with intrathoracic stomach who had associated spinal abnormalities.[1]

The extent of the spinal abnormalities was determined using magnetic resonance imaging (MRI) (Fig. 3) and computed tomography (Fig. 4). The cervical vertebral bodies were markedly hypoplastic, with anterior rachischisis, and the ossification centre of the odontoid peg was absent. The spinal canal was capacious and displaced anteriorly in keeping with a possible cervical neuroenteric fistula. The posterior arches were not fused in the midline, but several levels were fused to each other. The right-sided pedicles and laminae were larger than those on the left, and the facets were perched. MRI of the brain was normal.

Spinal precautions were taken within the limits of his size following detection of the spinal abnormality, utilizing a custom

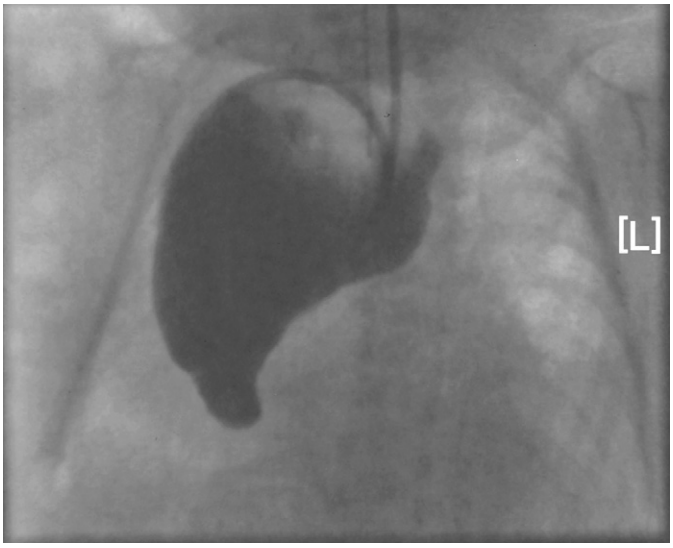

Fig. 2. Contrast study via orogastric tube demonstrating intrathoracic stomach, prior to laparotomy. The greater curvature of the stomach lies superiorly to that of the lesser curvature, suggesting rotation around the blood supply.

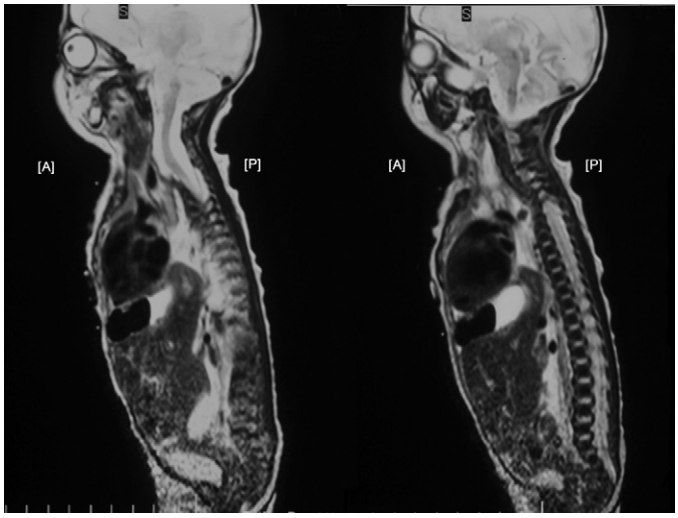

Fig. 3. Sagittal T2 weighted MRI demonstrating a widened spinal canal and multiple level posterior cervical fusion.

cervical orthosis. Weakness of the shoulder girdles was present, although he was otherwise neuro-developmentally appropriate for an infant of his gestational age.

Following a prolonged period of mechanical ventilation and postnatal steroids, the infant was weaned and extubated on day 40 to continuous positive airway pressure (CPAP). He subsequently managed 11 days with no respiratory support before a further deterioration requiring nasopharyngeal CPAP and admission to intensive care. At this stage, ultrasound examination suggested he had no movement of his left hemi-diaphragm and a further short trial off CPAP resulted in a rapid deterioration.

Early re-anastomosis of his gastrointestinal tract was indicated in order to establish oral feeding. A staged technique was considered for oesophageal lengthening, with multiple procedures and a high risk of failure. Stabilization of the cervical spine could be attempted at two years of age, however no precedent existed. The success of spine surgery would rely on sufficient ossification of the skull to allow rib grafting, and safe immobilization until then. Further respiratory support was anticipated, and asplenia posed a higher risk of sepsis from respiratory infections. The constellation of difficulties faced by this infant was considerable.

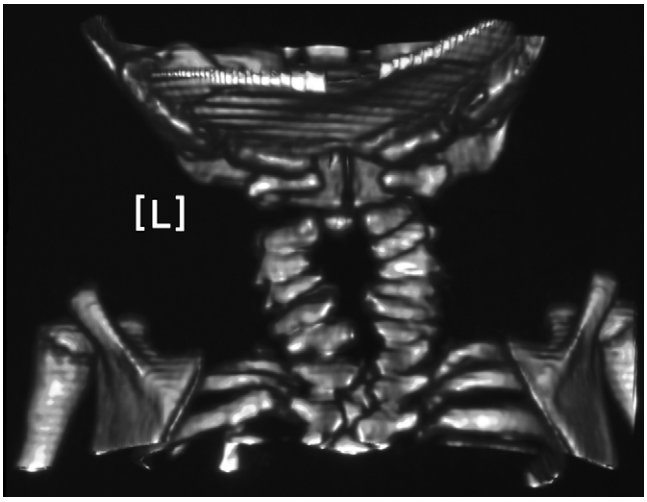

Fig. 4. Posterior view of three-dimensional reconstruction of cervical spine computed tomography. Asymmetrical laminae, perched facets, and anterior and posterior rachischisis are present.