Case Report

A Rare Differential Diagnosis of Air under the Diaphragm on Abdominal X-ray – Chilaiditi’s Syndrome in a Child with Skin Wrinkle Syndrome

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INTRODUCTION

In 1910, 15 years after the discovery of X-rays, a Viennese radiologist named Dr. Chilaiditi first noted a condition characterized by the presence of the hepatodiaphragmatic interposition of the intestine which was visualized in abdominal X-ray (Chilaiditi’s sign) and associated with various symptoms including nausea, vomiting, abdominal pain, constipation, and respiratory distress (Chilaiditi’s syndrome). Since then in children, only 30 cases were reported in the literature. We report a pediatric case of Chilaiditi’s syndrome in a case of a child with skin wrinkle syndrome and also discuss the importance of this diagnosis from a pediatric surgical perspective in the light of available literature.

CASE REPORT

A 5-year-old male child who was a known case of homozygous variant in the ATP6V0A2 for skin wrinkle syndrome along with corpus callosum agenesis with global developmental delay presented to pediatric emergency with the chief complaint of persistent vomiting (occasional coffee ground color) and not passed stools for 6 days, which was also associated with abdominal distension. There was no history of fever, cold, and cough. Past history of constipation was present which was managed with laxatives and similar episodes of vomiting managed conservatively. On examination, child was afebrile, not in distress, had mild dehydration signs, warm peripheries, pulses well felt and chest was clear. On per abdomen examination, abdomen was distended, diffuse vague tenderness present but no guarding or rigidity. Initial investigations showed that hemoglobin was 10.2 g/dl, white blood cells – 14.6 × 10^3 (neutrophils – 12.6 × 10^3), and renal function tests were within normal limits. X-ray of the abdomen erect was done which showed a picture of bilateral air under the diaphragm on first impression and the child was urgently referred to pediatric surgery [Figure 1]. Clinically as the child was afebrile and stable vitals and classical signs of guarding and rigidity were not present on physical examination of the abdomen, the radiographs were reviewed closely which revealed haustra in the air under the diaphragm. The child subsequently underwent noncontrast computed tomography (CT) of the abdomen which revealed the hugely dilated transverse colon and no free air in the peritoneum [Figure 2]. The diagnosis of Chilaiditi’s syndrome was made. The child successfully managed conservatively with bowel enemas. The child improved and was discharged on full oral feeds in a stable condition after 3 days on laxatives. The child was kept

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on follow-up and treated for constipation with oral laxatives.

**DISCUSSION**

The incidence of Chilaiditi’s syndrome is around 0.025%–0.28% in the general population. There is an increased prevalence in the elderly which might suggest that it is an acquired rather than a congenital condition.\(^1\)

The cause of Chilaiditi’s syndrome is currently unknown but may include intestinal, diaphragmatic, or hepatic factors.\(^4\) The lax hepatic suspensory ligaments of the liver are thought to be one of the etiological factors. Interestingly, the skin wrinkle syndrome, which is seen in this child, has been associated with lax ligaments in general. This association may add to the understanding of the etiological factors of Chilaiditi’s syndrome in children which are more congenital in origin.

Only 30 pediatric cases were reported, of which 19 (63%) cases were male.\(^2\) The median age of diagnosis was 4.5 years old with an interquartile range of 2.0–10.0 years. The most common predisposing factors in children are aerophagia (12/30 cases) and constipation (13/30 cases). Ninety percent of the cases presented with complete intestinal interposition, in which 100% of which the colon was involved. While most cases can be managed conservatively, a few cases required surgical intervention. The index case which is a 5-year-old male child with constipation and complete interposition variant, which was successfully managed conservatively, also closely resembles the above characteristics.

There are two important implications of knowing this syndrome from the clinician’s perspective. The first one is that the knowledge of this uncommon syndrome might prevent unnecessary emergency surgical intervention as it closely mimics pneumoperitoneum in an abdominal X-ray. As shown in the index case, the presence of haustra or plica circularis between the liver and diaphragm helps in distinguishing the Chilaiditi’s sign from the former condition. The other differential diagnoses for Chilaiditi’s sign on radiographic film are subphrenic abscess, Morgagni hernia, and posterior hepatic lesions. The presence of pleural effusions and basilar atelectasis and a smaller air–fluid level in the upper quadrant are characteristic features of subphrenic abscess. In case of any further confirmation required or to rule out other differentials depending on the clinical presentation, a noncontrast abdominal CT scan will be helpful. The second important implication is that the correct diagnosis helps in avoiding inadvertent injury to the bowel during percutaneous transhepatic approaches such as percutaneous liver biopsy, percutaneous transhepatic cholangiography, or biliary drainage that the children may be subjected in future and the usage of real-time ultrasound for these procedures will be helpful in avoiding the injury to the intestine.

**Declaration of patient consent**

The authors certify that they have obtained all appropriate consent forms. The patients/guardians understand that the patient names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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**Conflicts of interest**

There are no conflicts of interest.

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