Rare disease

Achalasia cardia in a premature infant

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Summary

Achalasia cardia is defined as a neuromuscular disorder of the oesophagus with abnormal motility and failure of relaxation of the distal oesophagus. It is an uncommon but well-recognised entity in infants and children. However, achalasia in a preterm baby has not been previously described. We report the condition in a premature infant with unusual presentation, treated successfully with Heller's oesophagomyotomy and fundoplication.

BACKGROUND

Achalasia cardia in a preterm baby has not been previously described in the literature. This is also an unusual presentation in that the baby had no prior symptoms and the diagnosis was suspected following an incidental chest x-ray finding. Many neonatal units in the UK and worldwide use high flow nasal cannula oxygen (Vapotherm) to provide respiratory support. In our case, the lower oesophageal dilation seen on chest x-ray was initially thought to be secondary to high flow respiratory support. After a thorough literature search, we found a paper describing physiological gaseous upper oesophageal distension in premature infants receiving nasally delivered non-invasive respiratory support. However, gaseous lower oesophageal distension in this group of babies has not been reported. Our case illustrates the need to investigate babies on non-invasive respiratory support who are noted to have lower oesophageal dilation. Also early diagnosis of this condition reduces the risk of aspiration pneumonitis and provides the opportunity to achieve prompt surgical relief.

CASE PRESENTATION

A premature male infant weighing 1200 g was delivered at 29 weeks' gestation by emergency caesarean section due to worsening maternal pre-eclampsia. Pregnancy was by in vitro fertilisation with egg donation and embryonic implantation. Antenatal scans showed dilatation of the cerebral lateral ventricles. The mother had received two doses of steroids 3 days before delivery and there were no risk factors for infection.

The baby was intubated at birth due to poor respiratory effort and received one dose of surfactant. He was extubated at 13 h of age on to high flow nasal cannula oxygen (Vapotherm). At 36 h of age, a chest radiograph performed to check the position of a long line, was incidentally noted to show lower oesophageal dilation (figure 1). The oesophageal dilation was initially thought to be secondary to the high flow respiratory support; however, an oesophagogram performed at 2 weeks of age confirmed achalasia cardia (figure 2). Management involved insertion of a nasogastric tube for feeding and an oro-oesophageal tube for suctioning

to reduce the risk of aspiration. Surgery was planned for when the baby reached term, but was brought forward to 36 weeks postconceptual age due to recurrent aspirations. A Heller's oesophagomyotomy and fundal wrap was performed. The postoperative recovery was uneventful. Dilatation of cerebral lateral ventricles was confirmed on postnatal cranial ultrasound and brain MRI but was non-progressive. On follow-up to 12 months of age, the baby has been feeding and thriving well with some delay in motor milestones noted.

INVESTIGATIONS

1. Chest x-ray, which showed lower oesophageal dilation at 36 h of age while the baby was on high flow nasal cannula oxygen (Vapotherm).
2. Contrast oesophagogram showed lower oesophageal dilation with a bird beak appearance at the gastro-oesophageal junction confirming achalasia cardia.

TREATMENT

Heller's oesophagomyotomy and fundal wrap was performed.

OUTCOME AND FOLLOW-UP

On follow-up to 12 months of age, the baby has been feeding and thriving well with some delay in motor milestones noted.

DISCUSSION

Achalasia cardia is an uncommon but well-recognised entity in infants and children. The reported incidence is 0.5 to 1/100 000, with 3–5% of cases occurring in children and less than 0.5% in neonates. The youngest infant with achalasia reported in the more recent literature was a 2-week-old term baby who presented with symptoms of continued vomiting and bilateral aspiration pneumonitis.¹ He was diagnosed to have achalasia after a contrast study and oesophagomyotomy was performed at 3 months of age. We report the first premature infant with achalasia cardia. This was also a novel presentation with lower oesophageal dilation noted on a chest x-ray on day 2 of life.

Figure 1 Chest x-ray showing lower oesophageal dilation at 36 h of age while the baby was on high flow nasal cannula oxygen.

Achalasia cardia is an oesophageal motor disorder characterised by incomplete relaxation of the lower oesophageal sphincter and a lack of normal oesophageal peristalsis. The aetiology remains unclear. Various hypotheses have been postulated, including loss of ganglion cells in the lower oesophagus, Wallerian degeneration of extraoesophageal nerve fibres and a reduction in the dorsal motor nuclei of the vagus nerve.² Symptoms of achalasia in infancy vary from regurgitation of feeds to recurrent aspiration pneumonia. The symptoms are often confused with gastro-oesophageal reflux disease, delaying correct diagnosis and treatment.³ A high index of suspicion and an awareness that achalasia of the oesophagus may occur in the very young patient are necessary for timely diagnosis. To date all case reports on oesophageal achalasia in the literature have been diagnosed after presentation with clinical symptoms. In our case the baby had no clinical symptoms of achalasia initially and the diagnosis was suspected following an incidental chest x-ray finding.

Nasal continuous positive airway pressure (nCPAP) has been used since 1975 as a treatment for respiratory distress syndrome in very premature infants. A large retrospective study has shown a common occurrence of gaseous upper oesophageal distension secondary to use of nCPAP in a

population of premature infants.⁴ The authors considered the cause to be dilation of the proximal cervical oesophagus in association with closure of the upper oesophageal sphincter. They implicated lower tone in the cervical oesophageal musculature, especially in the premature infants. Many neonatal units in the UK and worldwide use high flow nasal cannula oxygen (Vapotherm) to provide respiratory support.⁵ This also delivers a component of nCPAP. Evidence from the literature and from UK neonatal units suggests that Vapotherm is an effective and well-tolerated method of providing babies with respiratory support.⁶ Despite its widespread use, Vapotherm has not been proven to be better than other forms of respiratory support. In our case, incidental lower oesophageal dilation was noted when the baby was started on Vapotherm. The mechanism may be attributed to gaseous distension of the lower oesophagus with the pathological closure of the lower oesophageal sphincter seen in achalasia.

Multiple associations with achalasia have been described, most commonly with alacrima and adrenocortical insufficiency also known as achalasia-addisonianism-alacrima syndrome or Allgrove's syndrome.⁷ Multiple congenital anomalies associated with infantile achalasia were described in a case report of two newborn infants who

Figure 2 Contrast oesophagogram showing lower oesophageal dilation with a 'bird beak' appearance at the gastro-oesophageal junction.

both had non-rotation of the small bowel.⁸ In our case, dilatation of the cerebral lateral ventricles had been noted prenatally, but this was non-progressive on imaging postnatally.

Diagnosis of achalasia cardia is made on contrast oesophagogram which shows a dilated oesophagus with smooth narrowing of the distal oesophagus and oesophago-gastric junction, also called the 'bird-beak' sign. Diagnosis may be aided by the use of manometry in which there is a failure of relaxation of the lower oesophageal sphincter with swallowing. Manometry is performed very infrequently in children and requires experience in interpretation.⁹ Due to lack of equipment for a 1200 g baby and the lack of diagnostic value in our case, we did not pursue this test. The relative safety of Heller's oesophagomyotomy with fundoplication and its excellent long-term results make this the most attractive form of treatment for this disorder in young patients.¹⁰

In conclusion, achalasia cardia should be kept in mind in the differential diagnosis of lower oesophageal dilation in

the newborn, particularly in babies receiving nasally delivered non-invasive respiratory support. Early diagnosis reduces the risk of aspiration pneumonitis and provides the opportunity to achieve prompt surgical relief.

Learning points

- ▶ Achalasia cardia should be kept in mind in the differential diagnosis of lower oesophageal dilation in newborns, particularly in babies receiving nasally delivered non-invasive respiratory support.
- ▶ Early diagnosis reduces the risk of aspiration pneumonitis and provides the opportunity to achieve prompt surgical relief.

Competing interests None.

Patient consent Obtained.

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