

Cerebro-costo-mandibular syndrome: Report of two cases

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Cerebro-costo-mandibular syndrome (CCMS) is a rare syndrome that includes a constellation of mandibular hypoplasia and posterior rib defects as its basic features. Additional features can include hearing loss, tracheal cartilage abnormalities, scoliosis, elbow hypoplasia, and spina bifida. Here we report two cases of CCMS and discuss the reported long-term outcome of the disease.

Introduction

CCMS is also known as “rib-gap syndrome,” “rib-gap defect with micrognathia syndrome,” and “Smith-Theiler-Schachenmann syndrome.” It was first described in 1966 by Smith et al (1). It is a rare syndrome; only about 60 cases have been previously reported. The basic features of CCMS are mandibular hypoplasia and gap defects of many ribs that may simulate rib fractures.

CCMS is considered by some authors to be a Pierre Robin sequence associated with the rib gap defect. In Pierre Robin sequence, the main pathology is mandibular hypoplasia that, during intrauterine life, leads to inability of the tongue to migrate caudally, resulting in abnormal positioning of the tongue and hindrance to the normal development of the palate. Because its results are due to a single abnormality, Pierre Robin is considered a sequence rather than a syndrome. However, in 87% of cases it is associated with other syndromic manifestations. Here we report two cases of CCMS. The first case is a neonate, and the second case is 15-year-old patient who was followed up in our institution.

Case 1

A full-term male baby had respiratory distress on day 1 of his life. A chest x-ray was obtained because of respiratory difficulties (Fig. 1).



Figure 1. One-day-old male baby with CCMS. Chest radiograph shows multiple bilateral posterior rib gap defects. The vertebrae are normal.

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The finding of rib gap defects led to further investigation, including a bone survey. Micrognathia with small maxillary bones was found (Fig. 2).



Figure 2. One-day-old male baby with CCMS. Skull x-ray, lateral view, shows micrognathia and small maxillary bones.

CT of the head showed a cleft palate (Fig. 3). There was also bilateral superior semicircular dehiscence. The brain was normal.

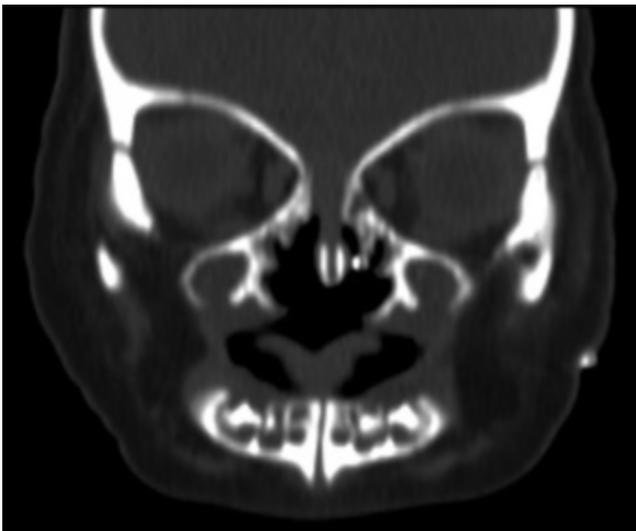


Figure 3. One-day-old male baby with CCMS. Coronally reformatted CT shows a cleft palate.

Because of significant reflux, a gastrostomy tube was placed, and Nissen's fundoplication was performed. A tracheostomy cannula was placed to support respiratory func-

tion, as the patient had respiratory failure. The patient was active and moving all limbs. The family history was negative for prior congenital anomalies. The mother had quit smoking with pregnancy.

Case 2

A male patient was seen at our institution at the age of 15 years. He had a history of mandibular hypoplasia without mandibular angles, multiple bilateral rib gap defects, a cleft of the soft palate, upper airway obstruction, progressive scoliosis, asthma, gastroesophageal reflux, tracheostomy dependence, speech and language disorder, and conductive hearing loss. Chest x-rays of this patient showed multiple rib gaps (Fig. 4).



Figure 4. 15-year-old male with CCMS. Chest radiograph at 13 years of age with ribs gaps noted.

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The CT chest images revealed that the rib gaps were filled with calcified, disorganized tissue (Fig. 5).

Discussion

In 1966, Smith et al reported unusual rib defects in an infant who also had mandibular hypoplasia and abnormal tracheal cartilages and pterygium colli. The infant died at 8 hours of age (1). McNicholl et al reported three siblings with rib deformities (unossified posterolateral segments) with micrognathia, glossoptosis, and mental retardation (2). Two of the three children had also cleft palate. Two of the reported children died as young infants, and the third was followed until the age of 4.



Figure 5. 15-year-old male with CCMS. CT chest image shows the rib gaps to be filled by disorganized calcified tissue (on the right side).

Miller et al reported 2 cases of infants with multiple rib abnormalities and micrognathia. One infant had a high arched palate, and the second had cleft palate. Both died during the early few months of life. The microscopic examination of the rib defects in the first patient showed disorganization of the cartilage and bone. Also noted were renal microscopic cysts lined by flattened cuboidal epithelia in the medulla. The father had cleft palate, which raised the question of a syndrome with variable presentation (3).

The rib defects ranged from a short segment defect that looked like pseudoarthrosis to total absence of the anterior three-quarters of the rib (3). Fibrous tissue and muscle tissue were seen at the site of the rib defect. Some normal cartilage was identified. Although a thorough neurological examination was not carried out, no significant abnormality was noted clinically (3).

Wilcox reported a case with head circumference below the 5th percentile; MRI of the brain showed microcephaly. This patient also had aortic coarctation, a bicuspid aortic valve, and renal cysts. The patient underwent repeated mandibular osteodistraction and was followed until 2 years of age. The father had micrognathia, with a history of cleft palate repair. Interestingly, a chest radiograph of the father showed rib-gap defects. This raised the possibility of an autosomal dominant mode of inheritance (4).

Williams and Sane reported a case of an infant with rib defects (5). In their case report, they mentioned absence of the transverse processes of many of the involved vertebrae. In this case, the ribs united with these vertebrae rather than articulating with them in a normal way. A flask-shaped pelvis with fused sacral segments was also described. This patient was followed up to the age of 5 years, and the rib gap defects diminished with increasing age. The child had limited intelligence.

In CCMS, the posterior aspects of the ribs are absent radiographically because bone is replaced by cartilage or fibrous tissue that may eventually undergo calcification (5). These defects often result in a bell-shaped thorax and may cause flail chest, compounding upper-airway obstruction. Postnatal growth retardation and microcephaly are common findings (4). Mental retardation, at first thought to be inherent, could be a consequence of neonatal hypoxic brain insult due to airway obstruction (4). Reported mortality is 35% to 50% in the first year of life (4). Other less common reported findings include hearing loss, tracheal cartilage abnormalities, elbow and clavicular hypoplasia, scoliosis, pterygium colli, and central nervous system developmental disorders such as spina bifida cystica. Cardiac anomalies are very uncommon (4).

The majority of cases of CCMS are sporadic, yet familial occurrences have been reported, many of which support autosomal recessive inheritance; in other cases, autosomal has been suggested. Because both autosomal dominant and autosomal recessive inheritance has been reported, the disorder could be genetically heterogeneous. Our cases also provide support for the belief that mental retardation in CCMS may occur as a consequence of hypoxic brain injury rather than inherited central nervous system anomalies, since both father and son appear normal in intelligence and gross neurological function (4).

Early diagnosis of this rare syndrome can lead to early intervention in the respiratory tract that could avoid the cerebral effects of prolonged hypoxemia.

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