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CASE REPORT

CEREBRO-COSTO-MANDIBULAR SYNDROME IN A NEONATE-A RARE CASE REPORT

Sowjan.M., Bharani Kumar.R., Rajakumar.P.G and Sai santhosh.R.B

Department of paediatrics Shri sathya sai medical college and research institute, Ammapettai, Thiruporur

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ABSTRACT

Cerebro-costo-mandibular syndrome (CCMS) is a very rare congenital disorder. Approximately only 75 cases has been described in the literature upto date. CCMS is characterized by features typical of pierre robin sequence with specific multiple rib gap defects. It is accompanied by mental deficiency in considerable number of cases. Sometimes, there are associated anomalies and problems, such as spine deformities, brain, heart, kidney or ear anomalies, feeding difficulties, delayed psychomotor development, and growth impairment. Depending on severity of deformities and consecutive respiratory insufficiency, in about 35–50% of CCMS cases, death occurs during the first year of life. These cases are referred to as severe types of CCMS. In this case report we present a newborn with typical features of CCMS. Diagnosis was established on day one, based on micrognanthia and findings of posterior rib-gap defects on the chest X-ray, accompanied by dyspnoea. CCMS should be considered in every infant with micrognanthia and rib-gap defects on chest X-ray.

INTRODUCTION

Cerebrocostomandibular syndrome (CCMS) is a very rare congenital disorder. Only around 75 cases have been reported around the globe. It was first described by Smith et al in 1966.It is also known as “Rib-Gap syndrome”, “Rib gap defect with micrognanthia syndrome” and “Smith-Theiler-Schachenmann syndrome” It is characterised by mainly by growth retardation, Dysmorphic chest, mandibular hypoplasia, glossoptosis and variable palatal abnormalities.Here we describe a neonate with typical features of CCMS

CASE REPORT

A 25 year old mother gave birth to a male child through emergency Lower segment caesarean section. The liquor was thin meconium stained. Apgar score being 6,7and 8 at 1, 5 and 10 minutes respectively. It was a term first born male child, born to non consanguineous parents. The course of pregnancy was uneventful. Family history was unremarkable. Birth weight was 2.750 kgs, birth length was 48cms, head circumference was 35 cms and chest circumference was 29cms. Difference between chest and head circumference was 6cms, no resuscitation was done at birth, routine care was given; the baby had a spontaneous respiration.

On examination baby had tachypnoea and tachycardia. Not maintaining saturations without oxygen. Subcoastal retractions were present. Several malformations were observed: retromicrognathia (fig 1), cleft palate, glossoptosis, hypertelorism, low-set malformed ears, narrow chest (fig 2), wide-set nipples and redunbant skin. Cardiovascular system and central nervous system were normal. Baby had a difficulty in breast feeding. So baby was started on humidified oxygen through nasal cannula at 2liters per minute. Feeding was given through nasogastric tube. These malformations were suggestive of Pierre robin sequence.

Fig 1

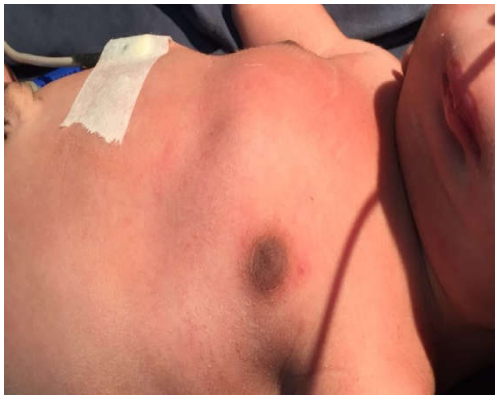

Fig 2

Investigations showed haemoglobin-20.2g/dl, Total Leukocytes Count-20300, Neutrophils-72%, lymphocytes-13%, eosinophils-2% and monocytes-1%, platelets were 3lakhs, renal function test was normal. Echo showed no abnormality .Chest X-ray(fig3) taken on the day one of life showed a narrow, bell-shaped thorax with eleven pairs of ribs, with posterior rib-gap defects of the second to ninth rib on both sides. This finding, accompanied with retromicrognathism, pointed to cerebro-costo-mandibular syndrome.

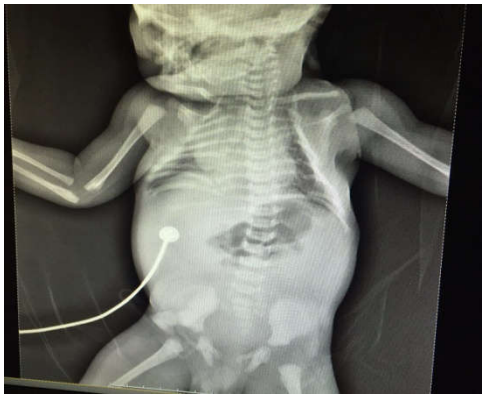

Fig 3

The baby was started on CPAP on day two of life as respiratory distress worsened. Baby maintained saturations thereafter. We were not able to wean off the baby from CPAP even after one week of life. Gills score was applied which was indicative of performing tongue lip adhesion surgery and so referred to a higher centre for surgery.

DISCUSSION

The association of Robin anomaly and posterior rib abnormalities appear to constitute a specific syndrome, which has been defined as cerebro-costo-mandibular syndrome. Clinical manifestation of CCMS may vary, but rib-gaps defect and micrognanthia are mandatory findings. There is a wide range of rib anomalies in children with CCMS. The severity of rib abnormalities in CCMS can vary from a few affected dorsal rib segments to complete absence of all and may not be symmetric. Vertebral changes may also be associated with the rib abnormalities [1]. Posterior rib-gap defects are typical finding in CCMS [2][3]. Rib abnormalities originate from replacement of the bone tissue between costovertebral junction and the lateral arch with undifferentiated fibrous and muscle tissue, or cartilage [8][9].As the name implies cerebral features are not always consistent with this syndrome. It was reported by plotz et al. after reviewing 50 cases he reported that more than one-half of the patients had no cerebral abnormality [4]. Typical clinical features and X-ray findings are sufficient to diagnosis CCMS.

X-ray chest may imitate multiple costal fractures at birth, especially when resuscitation is applied, as in our case. Spontaneous rib fractures are also reported in some vaginally delivery neonate [11]. Micrognanthia in association with posterior rib-gap defects, which is present on subsequent chest X-rays without callus formation provides clues for diagnosis of CCMS. Pierre Robin sequence is characterized by micrognanthia glossoptosis and cleft palate, but without accompanied rib abnormalities. More than 50% of infants born with Pierre Robin sequence have associated syndrome or anomalies, including skeletal dysplasias [14], but posterior rib-gap defects which is specific to CCMS. Atelosteogenesis type I and campomelic dysplasia infants will also present with, narrow thorax at birth [10, 12], but very specific other clinical and radiological features.

Review of literature shows in 35–50% of CCMS infants, death occurs during the first year of life [15]. Nagasawa et al. [16] classified CCMS according to severity into the following three types: lethal CCMS, where death occurs during the first month of life; severe type, where death occurs between the first and 12th month of life; and mild type, where patients live more than a year. Analyzing the descriptions of previously published cases, the same authors concluded that patients with severe type of CCMS had respiratory infections significantly more often than patients with a mild type [16]. Poor respiratory functions and frequent respiratory infections present the main causes of death in infants with severe CCMS. Their origin lies in under ventilation of lungs, lying position in bed, and often microaspirations which occurs due feeding difficulties. Special issues are infections with multidrugresistant hospital strains in hospitalized patients with CCMS [17]

In several other case reports of CCMS, there were some other anomalies associated: ear anomalies [21], conductive hearing loss [7], occult spina bifida [2], cardiac anomalies [12], central nervous system anomalies [5], microcephaly [5], other bone deformities – except ribs and mandible [5], arthrogryposis [21], and urogenital anomalies [5] renal cysts [12],. All these deformities are periodical and have low incidence [6, 11].Mental subnormality was also associated with CCMS in a considerable number of cases [4, 7, 8, 9, 11], but normal intelligence is also being reported in some infants [8, 12]. At first, it was considered that mental retardation is inherited, and, as such, a part of this syndrome; hence the “cerebro” part of the acronym. Recently, most authors see mental deficiency as a consequence of neonatal hypoxic brain injury [8], with the exception of cases accompanied with microcephaly, cerebral abnormalities or extensive perivascular calcification as a basis for mental retardation [5, 20].

The mode of inheritance in CCMS in heterogeneous. Both Dominant and Recessive patterns have been postulated. Many case reports have been reported to have parental consanguinity, whereas some others exhibit autosomal dominant transmission [5] [6]. Drosseou *et al*. In 1991 reported two pairs (four sibs) of twins with CCMS which supports high predominance of autosomal recessive inheritance [7].

The only mode of prenatal diagnosis is through ultrasonographic examination. Megier *et al*. reported a case

with CCMS diagnosed prenatally for the first time [13]. Polyhydramnios and a very unusual shape of the ribs were documented as ultrasonographic findings of CCMS [13] [14].
Maintainig the airway patent is a key point in early treatment of infants with CCMS. Depending on the nature and severity of respiratory distress, one should try conservative methods first – prone positioning, nasopharyngeal airway, laryngeal mask [24], or continuous positive airway pressure via nasal mask, as applied in our patient. If and when these measures become insufficient, surgical interventions must be considered. In several other reported cases, tracheotomy was performed [12, 13, 21, 25]. In some infants, a tongue-lip adhesion operation [24] and distraction osteogenesis [8] and bilateral mandibular osteostomies were also performed, but in infants who doesn't have an airway narrowing below the tongue base.

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