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A CASE OF THE CEREBRO-COSTO-MANDIBULAR SYNDROME*

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The cerebro-costo-mandibular syndrome is characterized by defective costal development, cerebral maldevelopment and the features of the Pierre Robin syndrome[1-4].

This syndrome was reported for the first time in 1966. Between then and 1980, 24 cases were reported in the literature[5,6]. We have not been able to find any reports of this syndrome since 1980.

As this syndrome is very rare and as it has not been reported previously from our country we wanted to present our case.

Case Report

A girl baby, born at term and weighing 2100 grams, was referred to Hacettepe University Hospital for evaluation when she was three days old. The mother had not been exposed to any drug during pregnancy but the parents were first cousins. The patient was the second child of the family and her brother had been operated on for a cleft palate.

Physical examination revealed marked micrognathia, complete cleft palate, epicanthal fold, low–set ears and bilateral clubfeet. There was no microcephaly, but there was marked respiratory distress with inspiratory retractions.

Laboratory investigations, including hemoglobin, white blood cell count, peripheral blood smear, urinalysis and amino acid analysis of urine and blood, were all within normal limits. Total bilirubin level was 11.9 mg / dl and direct bilirubin 0.5 mg / dl. In the chest X-ray, fifteen pairs of ribs were identified, but an

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accurate count was not possible. There were multiple rib defects such as bifid rib and bilateral rib gap (Figure 1).

The patient was treated in an incubator with frequent pharyngeal suction, antibiotics and gavage, but she died on the second hospital day. Postmortem examination showed a small thymus, intraalveolar hemorrhage, dilated lateral ventricles, gliosis and the presence of a 5th ventricle. There was microvesicular fatty degeneration and extramedullary hematopoiesis in the liver. As the cerebro-costo-mandibular syndrome was not under consideration during postmortem study, the dorsal ribs were not examined.

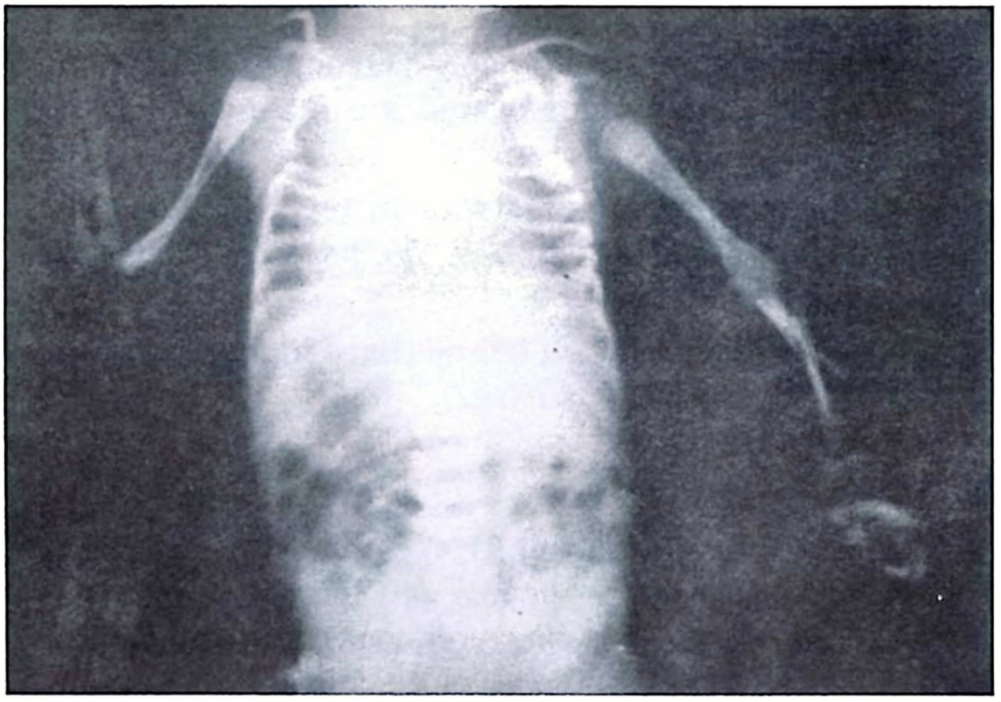

Figure 1
Bilateral rib gaps in the chest X–ray

Discussion

The possibility of this patient having the cerebro-costo-mandibular syndrome was considered because of the presence of micrognathia, cleft palate, rib defects, clubfeet and central nervous system abnormalities. In this syndrome, as in our case, the rib defects may be bilateral but not symmetric[4]. While cerebral involvement is expected, normal findings of the central nervous system are possible in this syndrome[6]. Some pathological findings such as a large unossified gap and segmental defect in the primitive mesenchymal tissue may be seen in a microscopic examination of the dorsal ribs[6]. However, as we had not considered the possibility of this being an instance of the cerebro-costo-mandibular syndrome

before the infant died, histological examination of the dorsal ribs was not carried out. Similar cases have also been reported in the literature[7].

Spondylothoracic dysostosis was not considered in the case of our patient in spite of the cleft palate and central nervous system abnormalities. This syndrome, which is characterized by malformation and abnormal fusion of several thoracic vertebrae and ribs associated with a short trunk[8], has been referred to by various terms, including costovertebral dysplasia, occipito-facial-cervico-thoracic-abdomino-digital dysplasia, spondylothoracic dysostosis and the Jarcho-Levin syndrome of vertebral anomalies[8-11].

The cerebro-costo-mandibular syndrome is a rare pattern of dysmorphic intrauterine growth and defective postnatal growth. Besides the cerebral and costal abnormalities, the Pierre Robin syndrome, clubfoot, hip dislocation, ventricular septal defect, kidney abnormalities and elbow dysplasia may also be seen in this syndrome[6]. Most of these cases are diagnosed at birth with the findings of severe respiratory distress. With one patient, however, the diagnosis was not made until 14 years of age when chest films were obtained prior to surgery for a cleft palate[12]. In one case, X-rays of the infant *in utero* showed multiple rib defects in the seventh month of gestational age[1]. This last finding is very important in the prenatal diagnosis of the cerebro-costo-mandibular syndrome.

It is interesting that our patient's sibling had a cleft palate. Similarly, another case has been reported in which there was an older sibling who had a cleft palate and mental retardation but no rib defects[13].

It is necessary to bear in mind the association between the Pierre Robin syndrome and rib dysplasia when such an infant presents with nonspecific respiratory distress. Radiological examination of the chest is advised in neonates with the Pierre Robin syndrome and respiratory distress.

Summary

A 3-day-old girl baby with the cerebro-costo-mandibular syndrome is presented. It is suggested that radiological examination of the chest should be performed in neonates with the Pierre Robin syndrome and respiratory distress.

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