Infantile Thoracic Dystrophy

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Infantile thoracic dystrophy, which is a generalized chondrodystrophy, was first described by Jeune et al in 1955.[1] Although the number of published cases up to the present time is not great, the increase during the past few years indicates that the condition may not be so rare as was at first thought. The purpose of this paper is to present a case, which we believe to be the first reported from Turkey.

Case Report

The patient was a six-hour-old female infant, the product of an eight and a half months pregnancy, who was noticed, soon after delivery, to be extremely cyanotic. The history of the pregnancy (according to records of the maternity hospital) showed that the mother had had polyhdramnios, and that because of weak contractions during labor she was given medications to induce the birth. The family history revealed that the mother was 34 and the father 35 years old, and that there was no consanguinity between them. Two female sibs, aged five and eight years, were reported to be alive and in good health, but another sib was lost at the age of two due to some unknown illness.

Physical examination showed the infant to be 46 cm long and 2.5 kg. in weight. Her appearance was interesting in that she seemed to have a long narrow body with relatively short extremities. The most marked finding was that the chest was extremely narrow. She had relatively severe

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tachypnea and dyspnea with cyanosis of the lips and the nails, and the shoulders were high set. The patient expired from asphyxia at the age of three days.

X- Ray Examination: The ribs were hypoplastic,and the rib ends did not pass the axillary lines medially. Both clavicles were located higher than is usual (Figure 1). There was also hypoplasia of the iliac bones, and the ossification was irregular in the Y cartilage region, but there was no evidence of any metaphysial irregularities in the enchondral ossification.

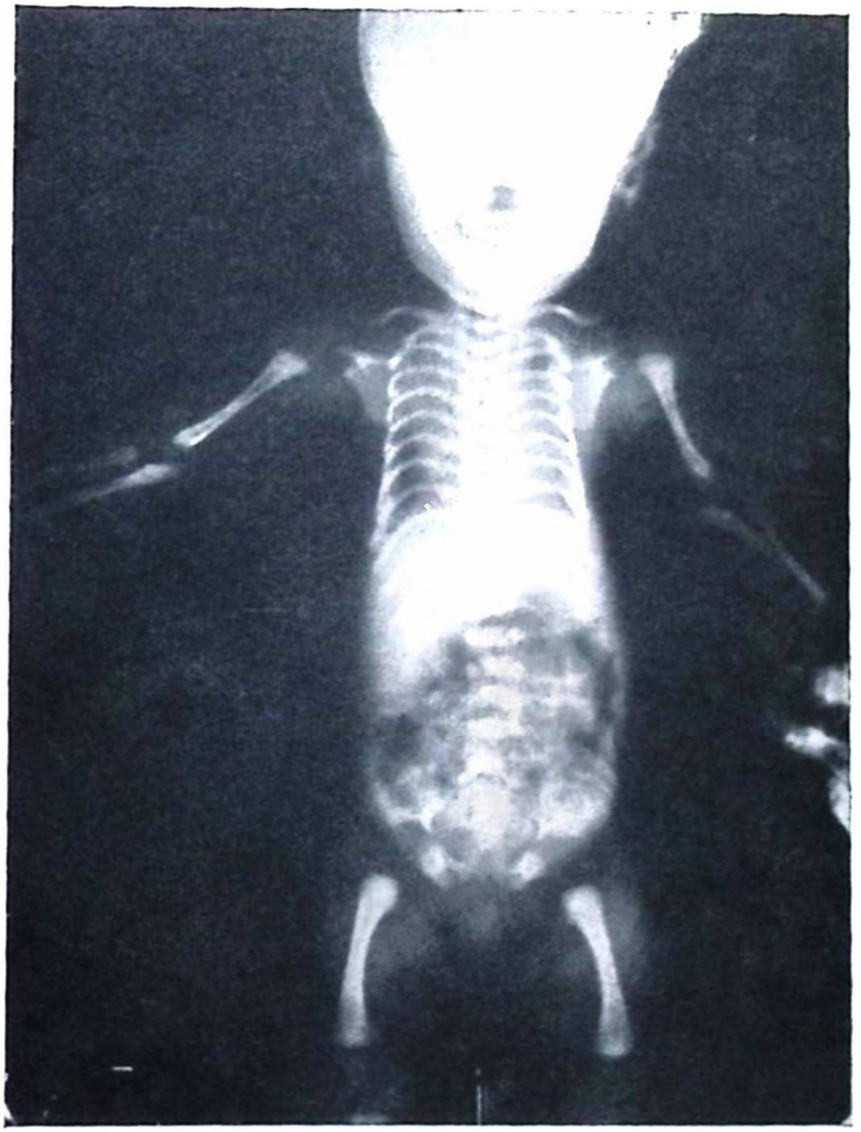

Figure 1

Discussion

According to a recent review of the world literature on infantile thoracic dystrophy by Langer,[2] the number of cases reported until 1968 was 48. We failed to find any case reported from Turkey. The condition has been

described under different names, such as asphyxiating thoracic dystrophy,[1] asphyxiating thoracic dystrophy of the newborn,[3] Jeune's disease,[4] infantile thoracic dystrophy, and thoracic phalangial dystrophy[2].

The most important finding in these patients for diagnostic purposes seems to be the decrease in chest diameters both antero-posterially and horizontally.[3] Though this is of great diagnostic value, it should be mentioned that the severity of chest findings varies from case to case. Also thoracic dystrophy seems to improve over the years, so that if a patient survives the initial respiratory difficulties diagnosis may not be so easy later as it is at birth.

The other findings, namely the shortness of the extremities, also varies from patient to patient. Reports on a few of the older patients are avaliable and these indicate that they are also significantly below the average height for their age. Another interesting finding is the frequent presence of polydactyly. The extra digit is usually located postaxially on both the hands and the feet. Various types of tooth anomalies have also been described[6]. In a few cases histopathologic examinations of the kidneys have shown tubular atrophy, dilatation and interstitial fibrosis. [7] [8] [9] It is interesting in this respect that even in the terminal stages the glomerules have been found to be free from pathology[7].

During the neo-natal period the most common cause of death is asphyxia, and respiratory difficulties as a cause of death are also encountered in infancy. In a few cases the patient expired due to kidney failure.

Infantile thoracic dystrophy seems to be inherited as an autosomal recessive trait. No sex difference in incidence has been reported; of 49 published cases, including our own, 22 were male and 19 female (in the remainder the sex was not recorded). The condition has been observed in more than one sib in a family.

Radiological Findings: These vary according to the age of the patient; during the neo-natal period and for a few months thereafter x-rays show the ribs to be short and horizontally placed, and the costochrondrial junctions to be irregular. This latter findings is more prominent during infancy since the shortness of the ribs varies markedly from case to case. The iliac bones reveal vertical shortening and flaring of the iliac crests. Both sciatic and pubic bones are shortened, and the upper acetabular borders are oblique. It appears that when both thoracic and pelvic abnormalities are seen in the x-rays they have great diagnostic value; the shortness of the extremities also contributes toward a correct diagnosis. Other findings seen rather frequently, but not always, are poly-

dactyly and brachydactylia. The vertebral and cranial bones reveal no abnormalities in most cases.

In older children metaphypsial irregularity is most prominent in the costo-chondral junctions. By this age the chest no longer appears so narrow, though the pelvic bones are still shortened, but without any flaring of the iliac crest. The most interesting finding at this age involves the hands, in which the epiphyses of the distal and middle phalanges are cone-shaped. Early fusion of the epiphyses with the metaphyses can also be observed in these areas. The distal and middle phalanges are also short. Similar cone-shaped configurations may also be seen in the proximal phalanges, although they are usually less marked here, and in most cases the epiphyses are invaginated into the metarcarps. These findings may also involve the feet. The spine and skull are normal in appearance, and the extremities are shorter than usual, though this latter findings varies markedly from patient to patient.

In summary, in any patient with a narrow chest the possibility of infantile thoracic dystrophy should be considered. Although asphyxia in newborn infants may result from many other causes (such as hypophosphotasia for example), when it is associated with a narrow chest a correct diagnosis is facilitated. X-ray findings are, of course, helpful in distinguishing cases of hypophosophotasia, in which markedly distributed mineralization of the bones is seen, and, except for the central parts, the cranial bones in particular lack ossification, which is not the case in infantile thoracic dystrophy. The level of serum alkaline phosphatase and the appearance of phosphorylethanolamine in the urine also helps in diagnosising hypophosphatasia. The neurological conditions affecting the respiratory muscles may sometimes resemble, both clinically and radiologically, those seen in infantile thoracic dystrophy, but a careful examination of these patients reveals that the ribs are not short, and that the pelvic findings are absent.

There are, however, other conditions which are characterized by short extermities fully manifested at birth, which include:

1. Achondroplasia;[10]
2. Chondrodystrophy calcificans congenita;[11]
3. Diastrophic dwarfism;[12]
4. Spondylo-epiphysial dysplasia congenita;[13]
5. Metatrophic dwarfism;[14]
6. Cartilage-hair hypoplasia;[10]

7. Chondro-ectodermal dysplasia (Ellis-van Creveld syndrome);[10]

8. Tricho-rhino phalangeal dysplasia.[10]

The real difficulty in differential diagnosis concerns Ellis van Creveld syndrome, as the findings in this condition are very similar to those seen in infantile thoracic dystrophy both during the neo-natal period and in infancy. In both cases there is early ossification of the head of the femur, which may even be present during the first days of life. Polydactyly of the hands is almost a constant finding in chondro-ectodermal dysplasia, and may also be present in infantile thoracic dystrophy. The shortness of the middle phalanges and relative hypoplasia of the distal ones are seen in both conditions, but are more prominent in Ellis-van Creveld's syndrome. Also in chondro-ectodermal dysplasia anterior-posterior x-rays show the capitata and hamata to be either fused, or overlapping. Sometimes the hamatum is triangular, with its long axis in a transverse position. As the child grows the characteristic finding of the knees appear: the proximal metaphyses are slightly displaced, and as a result genu valgum deformity appears, which is not seen in patients with infantile thoracic dystrophy.

Other differentiating factors include congenital heart disease which is present in about two-thirds of patients with Ellis-van Creveld's syndrome, while it is seldom seen in infantile thoracic dystrophy, and the chest abnormality is much milder in the former condition.

There are also certain findings which are seen only in Ellis-van Creveld syndrome; these include hypoplastic and deformed nails, involving both fingers and toes, shortening of the upper lip, which is also fused to the alveoli or connected by bands, in most cases, and the presence of teeth at birth, or early eruption of them (Table I). When an older patient with infantile thoracic dystrophy is seen for the first time the chief complaints are usually either shortness of stature and brachydactylia or concern the kidneys, since the respiratory findings are at a minimum at this stage. When there are signs of renal involvement skeletal - x-ray studies are of great diagnostic assistance, since infantile thoracic dystrophy is the only disease in which this is combined with shortness and brachydactylia.

An interesting disease entity recently reported for the first time, tricho-rhino phalangial dysplasia, also shows some resemblance to infantile thoracic dystrophy. As the name indicates, patients with this condition have extremely fine hair. The nose somewhat resembles that of a clown, i. e. it is round and blunt, and many of the phalangeal epiphyses show conification. However, these changes do not involve all the epiphyses of the distal and middle phalanges, being present in only a few of

TABLE I

DIFFERENTIAL DIAGNOSIS BETWEEN I.T.D. AND C.E.D

	Infantile Thoracic Centre Dystrophy	Ellis Van Creveld Synd.
Ectodermal Pathology	None Or Slight	Severe
Tooth Anomalies	Rare	+
Short Upper Lips With Multiple Frenula	—	+
Nail And Hair Dysplasia	Rare	+ (% 50)
Congenital Heart Disease	—/+	+ (% 60)
Inheritance	Autosomal Recessive	Autosomal Recessive
Renal Disease	+	—
Chest Abnormalities	+ + +	+
Shortness of Ribs	+ + +	—/+
Long Bone Changes	—	+
Knee Deformities	—	+
Polydactyly	—	+

the proximal ones, and much milder in those where they are found. The narrow chest, which is so characteristic in infantile thoracic dystrophy is not, of course, seen in these patients.

Histopathologic Findings

These findings were investigated by Razzi and his colleagues in the one case[4], who found that the most significant changes are seen in the ribs, especially in the cartilaginous tissue of the chondro-costal junctions, and, to a lesser degree, in the spinal cord. They were unable to find any important changes in the other organs, but they observed that in the chondro-costal junctions the ends of the costals were widened, and the bone tissues in these areas were tapered forming a beak-like projection, which was shown to be of cartilaginous origin, though most of it was produced by the perichondrium. Razzi et al also found an impairment of the enchondral ossification while the periostium was normal in appearance. The thinness of the ribs was not associated with any significant changes in their cortices, but there was marked atrophy of the intercostal muscles. In the spinal cord, especially in the anterior horn cells, there was a significant decrease in the number of ganglion cells, as well as certain structural abnormalities involving them, but there were no important findings in those of the posterior horn. The most significant findings in the renal pathology were degenerative changes in the tubules and the presence of multiple epithelial cysts in the cortices.

Summary

Infantile thoracic dystrophy is a familial disease with autosomal recessive inheritance. It is most frequently confused with Ellis-van Creveld's syndrome, though certain findings help to differentiate these two entities. Early diagnosis of these cases seems to be of great importance, because by proper management of pulmonary complications which commonly occur during infancy, there is a good change of survival, and once past this period the symptoms improve steadily. Certain other clinical and x-ray findings, especially the extremely narrow chest that is commonly seen in these patients, are also of great diagnostic value, even at birth.

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