DIAPHRAGMATIC HERNIA AND LIMB REDUCTION DEFECTS
CASE REPORT AND REVIEW

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ABSTRACT

The association of diaphragmatic defects and limb anomalies is rare. The case of a male newborn, product of consanguineous marriage with severe left upper limb reduction and diaphragmatic eventration on the same side is reported. Earlier cases with the same association were reviewed and possible relation with mesodermal disruption sequences is evoked.

INTRODUCTION

The association of diaphragmatic defects and limb anomalies is rare. A new case, product of consanguineous marriage is reported. Possible relation with mesodermal disruption sequences is evoked.

CASE REPORT

A 3 days old male newborn who was born at home by spontaneous vaginal delivery was admitted to the neonatal unit because of respiratory distress, hyperbilirubinemia and limb anomalies. The patient was the second infant of a Palestinian couple from Hebron area. Parents were first paternal cousins, mother's age was 27 years and father's 29. They had a normal living female daughter. Pregnancy was uneventful and no medications were taken during pregnancy. The baby was full term, birth weight was 2970 gm, height 50 cm and head circumference 34 cm. Family history could be traced to the end of the last century and no major limb abnormality was reported.
Initial chest X-ray confirmed the clinical impression of left diaphragmatic hernia (Figure 1).

The face showed a minimal left sided asymmetry with smaller palpebral fissure and smaller cheek compared to the right . No mouth asymmetry on crying was noted. Ears were not low set but have mild asymmetrical aspect (Figure 2, 3). Eye aspect and motility were normal and no ptosis was noted.

The left chest wall was hypoplastic with apparent absence of the left pectoralis major muscle. The left nipple was present.

There was severe reduction of the left upper limb in the form of phocomelia. The only remanant of it was a 3-4 cm appendage inserted directly into the left shoulder, it was grossly triangular in shape.

Skeletal X-rays showed: absent left humerus and scapula but persistence of acromion and spine, absent coracoid process, 2 miniature bones at the place of ulna and radius and other 2 residual metacarpal bones and probably the first phalange of the thumb (Figure 1). No abnormality of the cervical, thoracic or lumbar spine was noted. Hips and lower limbs were normal. No abnormality was noted on the upper right limb. Skull and face bones were normal.

ECG was normal. Echocardiography showed no evident abnormalities, abdominal, renal and liver ultrasound examination were normal. Transfontanellar ultrasound examination of the brain was normal. Routine blood investigations were normal except for high bilirubinemia due to ABO incompatibility which required exchange transfusion. A transient post exchange thrombopenia was noted.

The infant was operated on the 10th day. Upon operation the left diaphragm consisted only of a thin membrane like structure. A hypoplasia of anterior chest wall muscle were noted. Different biopsies of the hernia sac consisted of fibrin structure without any muscular tissue. The immediate post operative course was smooth. On discharge at one month of age the patient was still in need of small amount of oxygen by nasal catheter and considered as a case of bronchopulmonary dysplasia. His facial asymmetry was not evident and no abnormality of cranial nerves was noted.

DISCUSSION

Many abnormalities had been described in association with diaphragmatic hernia (1, 2). Our discussion will be limited to cases of diaphragmatic hernia associated with limb defects not related to the use of certain drugs during pregnancy (3).

The association of diaphragmatic defects and limb anomalies on the same side was rarely reported. McCredie and Reid (4) reported four single cases, one of whom had an eventration of the diaphragm, two had a posterior-lateral defect and one had aplasia of the diaphragm. The limb defects ranged from a radial hypoplasia to a transverse deficiency, with a block of bone distal to the humerus. The authors postulate that both the upper limb and the diaphragm might be involved together in prenatal injury of the cervical neural crest. The distribution of limb deformities lend themselves to interpretation by "sclerotome substraction" according to the hypothesis put forth by Inman and Saunders (5).

Lerone et al. reported a further case of left congenital diaphragmatic hernia associated with ipsilateral thumb hypoplasia (6). The patient had also a right inferior facial palsy which was traumatic and the possibility of Moebius syndrome was excluded. Parents were not consanguineous. A similar case of congenital left diaphragmatic hernia with ipsilateral thumb hypoplasia and absent radius was reported by Wallerstein et al. (7).

Schinzel described two sisters, the offspring of unrelated parents, with phocomelia and other abnormalities (8). One of the sisters had diaphragmatic hernia, an absent gallbladder, phocomelia of both lower limbs and left upper limb.

Gershoni-Baruch et al. described a male infant with diaphragmatic hernia, an exomphalos, an absent thumb on one side and a finger-like thumb on the other, bilateral radioulnar synostosis, malar hypoplasia, down-slanting palpebral fissures, a unilateral malformed ear and micrognathia(9).

At least 2 cases of Poland anomaly associated with diaphragmatic hernia and limb defect were reported. In the first there was absence of pectoralis major, aplasia of radius, thumb, hemidiaphragm and lung on the same side (10). The second was reported in a female infant with dextrocardia, diaphragmatic hernia and herniation of the liver (11). The authors suggested that a disruption of the lateral embryonic plate mesoderm may have been responsible for the observed lesions. Other syndromes including facio-auriculo-vertebral sequence received a similar interpretation (12).

Diaphragmatic defects, a coarse face, distally hypoplastic phalanges and nails, are cardinal features of Fryns syndrome (13). This syndrome has sufficient distinctive features to be confused with isolated limb defects and diaphragmatic hernia like this patient.

The limb abnormalities in Holt Oram syndrome are variable. The thumbs are most commonly involved, other fingers might be absent, pectorals and the humerus might be hypoplastic. Heart lesions are predominantly an ASD, or more rarely a VSD, but other lesions have also occurred (14).

Three generations of a Jordanian family with a deformity of appendicular skeleton and shoulder associated with congenital heart disease were described (15). Jordanian and Palestinian families might have common origins but no relation was found between this family and the case under consideration.

Congenital diaphragmatic defects are variable in size, location as well as the presence or absence of a hernial sac. In congenital eventration of the diaphragm there is no actual hernial orifice in the diaphragm. The leaf of the diaphragm is stretched out, weakened by an apparent reduction of normal muscular elements which permit the diaphragm to rise high up. It may be of neurogenic origin or it may be a congenital defect in the development of the central tendon or diaphragmatic muscle. Eventration may affect part or the entire diaphragm. When the entire diaphragm is affected, as in our case, distinction between diaphragmatic hernia with intact pleuroperitoneal membranous hernia sac and congenital eventration of the diaphragm is arbitrary (16).

The upper limb defect in our patient can be classified as type I (defect of the humerus, including deficiencies of other parts of the limb,if the humerus is also affected) according to Foster and Bird (17). In a large population study, these authors had reported 11 cases of such type; none was associated with diaphragmatic hernia, one with asymmetric face and one with asymmetric ears (17).

Using the Spanish Collaborative Study of Congenital Malformations Martinez-Frias (18) concluded that the association of congenital diaphragmatic hernia and upper limb defects is specific or preferential, and constitutes a primary polytopic developmental field defect.

There is certain homogeneity between the cases described by McCredie and Reied (4) , Lerone et al. (6) and the present case. Consanguinity, the total absence of the scapula associated with the persistence of the spine, the agenesis of the pericardium in this case constitute some particularities. The hypothesis of a mesodermal disruption or malformation sequence can be reasonably advanced as an alternative explanation, to the interpretation by "sclerotome substraction", for cases of diaphragmatic hernia-limb defect.

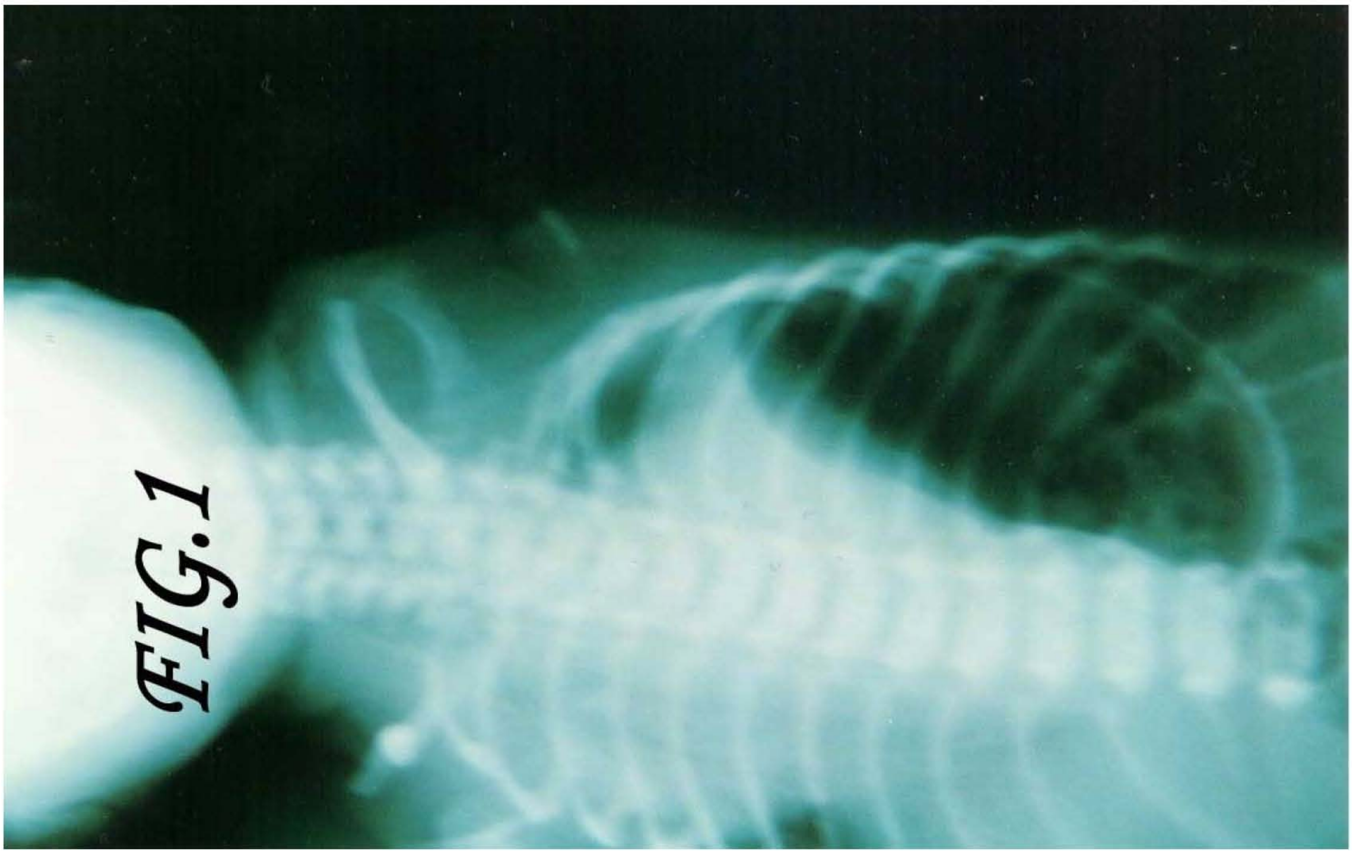

FIG.1

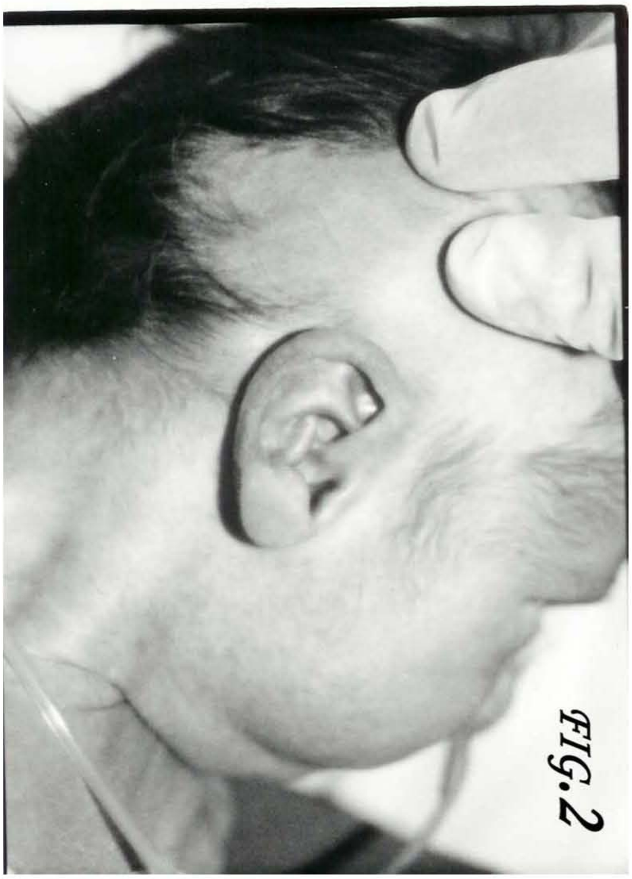

FIG.2

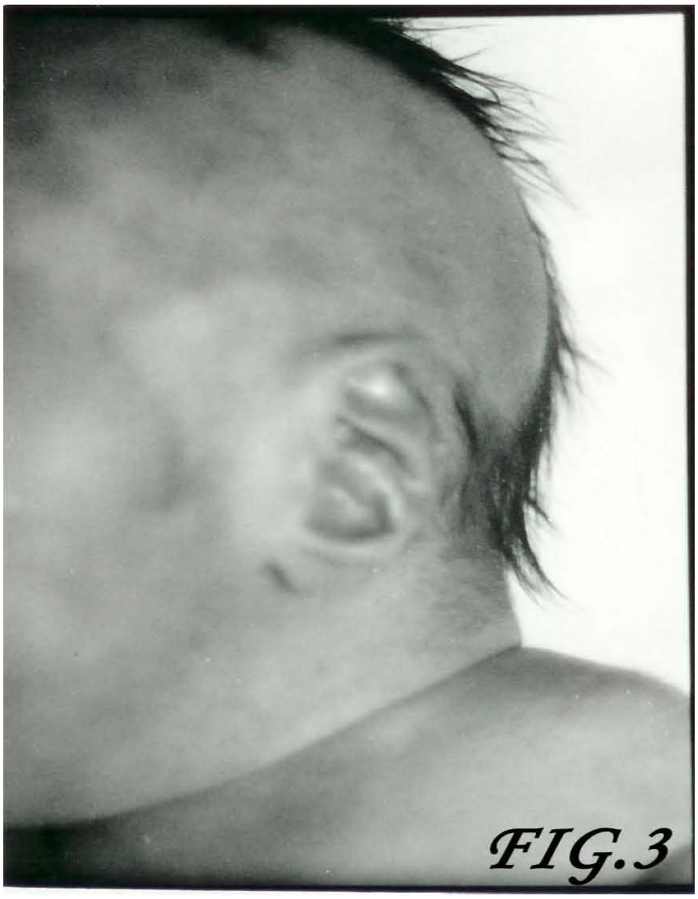

FIG.3

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PUBLICATIONS RELATED TO CHILDREN

Note from the editor

Presented in this issue two articles that we considered important for all child carers.

The first is a regular publication from the Palestinian Central Bureau of Statistics(PCBS). We have only introduced minimal editing modifications to facilitate its reading for all. It is important to mention that the PCBS has achieved excellent professional work in all fields related to health and other aspects of social interest.

The second is an intervention that was made by Dr. Sarraj and which had for subject the first Intifada consequence related to children and the issue of violence. As we are now in the disastrous condition of the second Intifada it is time to think about consequence knowing the social and political differences between the two events. I think that consequence will be more serious and disastrous for both Israeli and Palestinian children and their common future.