Case Report

Right sided eventration of diaphragm with clinodactyly:
A case of Fryns syndrome

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ABSTRACT
Diaphragmatic hernia can be of two types-absence or deficiency of the diaphragm, or eventration resulting in elevation of a portion of the diaphragm as a result of incomplete muscularization. Complete eventration almost invariably occurs on the left side and is rare on the right. 40-50% of individuals with diaphragmatic hernia have complex diaphragmatic hernia i.e. associated with other anomalies. We report a case of right side eventration of diaphragm associated with clinodactyly and digital hypoplasia who was suspected to have Fryns syndrome.

Key words: eventration, diaphragmatic hernia, clinodactyly, digital hypoplasia, Fryns syndrome

INTRODUCTION
Congenital diaphragmatic hernia (CDH) is characterized by: (i) incomplete formation/muscularization of the diaphragm resulting in absence or deficiency of the diaphragm, or (ii) eventration resulting in elevation of a portion of the diaphragm that is thinned as a result of incomplete muscularization resulting in a thin membranous sheet of tissue. About 50%-60% of affected individuals have isolated CDH; the remainder have complex CDH – that is, CDH occurring with additional malformations or as part of a single gene disorder or chromosome abnormality. (1) Complete eventration almost invariably occurs on the left side and is rare on the right. (2)

We report the case of a neonate with right sided eventration of diaphragm with clinodactyly and digital hypoplasia who was suspected to have Fryns’s syndrome.

Case study
A 23-year-old primigravida was referred to our hospital for cesarean section in view of meconium stained liquor. A female baby was born to her by LSCS with a birth weight of 2.25 kg. There was a history of oligohydramnios in the mother. Baby did not cry immediately after birth. Endotracheal suction for meconium was done followed by bag and tube ventilation as per protocol. After three minutes baby developed spontaneous efforts and was extubated. Baby was shifted to the neonatal ICU where she was noted to have respiratory distress in the form of grunting and desaturation. She was reintubated and put on
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intermittent positive pressure ventilation. Physical examination done at that time showed asymmetry of the chest with the right side of the chest much more prominent compared to the left. There was facial dysmorphism in the form of low set ears, flat nasal bridge, micrognathia and hypertelorism. (Fig 1, 2). She had bilateral clinodactyly with mild digital hypoplasia. (fig 3). There were no other obvious congenital malformations. A chest x-ray done at the time showed raised dome of diaphragm on the right side. (fig 4, 5) Ultrasound chest confirmed eventration of diaphragm on right side with? Pulmonary hypoplasia. Baby required high ventilator settings to maintain saturation and had a pulmonary bleed on day two of life. Coagulogram and platelets were within normal limits and there was no evidence of sepsis (septic screen negative and blood cultures sterile). Baby continued to deteriorate and required inotropic support. There was a 10% difference in preductal and post ductal saturations suggestive of pulmonary hypertension. However ECHO could not be done. On day three of life the baby’s parents chose to discontinue treatment and left against medical advice.

Fig 1: photograph showing low set ears, hypertelorism and micrognathia.

fig 2: photograph showing chest asymmetry and prominence of right side.

fig 3: clinodactyly in index case.

fig 4: right sided eventration of diaphragm with raised dome of diaphragm.
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DISCUSSION
The incidence of CDH is between 1/2,000 and 1/5,000 live births, with females affected twice as often as males. Defects are more common on the left (85%) and are occasionally (<5%) bilateral. Right sided defects form a mere 10% of the total and are associated with a higher mortality (3). Chest asymmetry is found in as many as half of individuals with CDH (1). Our patient had a right sided diaphragmatic hernia with chest asymmetry.

A major limiting factor for survival is the associated pulmonary hypoplasia. CDH results in various degrees of pulmonary hypoplasia and severe persistent pulmonary hypertension of the newborn. Despite antenatal ultrasound diagnosis and continuous improvement in neonatal intensive care, these features could not be overcome, and the overall mortality rate in CDH is still reaching 50%. Experimental works during the past 20 years suggest that CDH is a disease of impaired lung development associated with, but not caused by, a structural defect of the diaphragm. Furthermore, there is increasing evidence that the lung in CDH is not only small but that there are numerous disorders (e.g., surfactant deficiency, decreased anti-oxidant activity, increased vascular reactivity with decreased nitric oxide and increased endothelin 1 activity, and left heart hypoplasia) which may be associated with impaired lung development. (4). Our patient had a turbulent course in hospital with inability to maintain saturations even on positive pressure ventilation and features suggestive of pulmonary hypertension.

Congenital diaphragmatic hernia can be an isolated finding or it can be associated with other congenital malformations. Colvin et al. in a retrospective population based study of 116 cases in Western Australia from 1991 to 2002, found an incidence of associated major congenital anomalies in 46.6% of cases and of minor congenital anomalies in 38.8% of nonisolated CDH cases. The anomalies described more frequently are dysmorphic features, genitourinary, musculoskeletal, cardiovascular, neurologic, gastrointestinal and chromosomal malformations (5).

Fryns syndrome (FS) is the commonest autosomal recessive syndrome associated with congenital diaphragmatic hernia (CDH) (6). Congenital diaphragmatic hernia (CDH) and brachytelephalangy or nail hypoplasia are cardinal manifestation of Fryns syndrome (FS; OMIM 229850) (7). It is characterized by diaphragmatic defects (diaphragmatic hernia, eventration, hypoplasia or agenesis); characteristic facial appearance (coarse facies, ocular hypertelorism, broad and flat nasal bridge, thick nasal tip, long philtrum, low-set and poorly formed ears, tented upper lip, macrostomia, micrognathia); distal digital hypoplasia (nails, terminal phalanges); pulmonary hypoplasia; and associated anomalies (polyhydramnios, cloudy corneas and/or microphthalmia, orofacial clefting, renal dysplasia/renal cortical cysts, and/or malformation involving brain, cardiovascular system, gastrointestinal system, genitalia). Survival beyond the neonatal period has been rare. Data on postnatal growth and psychomotor development are limited; however, severe developmental delay and intellectual disability are common.

The diagnosis is based on clinical findings. No genes or loci associated with Fryns syndrome have been identified or mapped (8). Fryns
syndrome is thought to be inherited in an autosomal recessive manner. Because the gene(s) in which disease-causing mutations occur has/have not been identified, carrier testing and prenatal diagnosis using molecular genetic testing are not available. Two- and three-dimensional ultrasonography and fetal magnetic resonance imaging have been used in the prenatal diagnosis of high-risk pregnancies. Our patient fits into the clinical criteria for Fryns syndrome as she had diaphragmatic eventration associated with digital hypoplasia and clinodactyly together with the characteristic facial dysmorphism described previously. However as a chromosomal analysis could not be done therefore other chromosomal anomalies like trisomy 18, which has a known association with CDH, could not be ruled out.

**BIBLIOGRAPHY**