FRYNS SYNDROME: DIAGNOSIS AND MANAGEMENT

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Abstract

Fryns syndrome is a rare autosomal recessive condition characterized by multiple malformations. Congenital diaphragmatic hernia is a cardinal feature, recorded in more than 80% of cases. This is a case report of a newborn with right congenital diaphragmatic hernia, high anorectal malformation with recto-urethral fistula, a sacral dimple, left undescended testis, low set ears, microphthalmia and bilateral syndactyly. The literature on the subject is also reviewed and awareness of this condition is important for proper diagnosis and parenteral counselling.

Keywords: Fryns syndrome, congenital diaphragmatic hernia

Introduction

Fryns syndrome is an autosomal recessive syndrome characterized by multiple congenital malformations including congenital diaphragmatic hernia, pulmonary hypoplasia, a coarse face, a cleft palate / lip and cloudy corneae, distal limb hypoplasia and central nervous system, cardiovascular, gastrointestinal, urogenital and skeletal anomalies [1- 6]. The exact incidence of Fryns syndrome is unknown and many cases die in utero, born as stillborn or go unrecognized. It is important for physicians caring for these patients to be aware of this. This is a case report of Fryns syndrome and review of the literature highlighting aspects of diagnosis and outcome. This is the first case of Fryns syndrome to be reported from the Middle East.

Case report

A 38 hours old male, a product of full term caesarian section delivery was referred to our hospital because of mediastinal mass and anorectal agenesis. He was a product of 37 weeks’ gestation and his birth weight was 2.5 kg. The Apgar score 8 and 9 at 1 and 5 minutes. He had tachypnea immediately after delivery and had a chest x-ray which showed a large opacity on the right side of the chest. CT scan of the chest showed a large mass on the right side of the chest pushing the mediastinum to the other side (Fig. 1). Clinically, he was in mild respiratory distress with decreased air entry on the right side. He looked dysmorphic with microphthalmia, bilateral syndactyly of the fingers, and low set ears. The heart had a normal rate and regular rhythm. He was also found to have imperforated anus, a sacral dimple and left undescended testes. A repeat chest x-ray at our hospital showed right-sided congenital diaphragmatic hernia with herniation of bowel loops into the right side of the chest (Fig. 2). His abdominal and cross table lateral X-ray showed high anorectal agenesis. He was stabilized and underwent insertion of a central line through the right external jugular vein, laparotomy and repair of right congenital diaphragmatic hernia as well as posterior sagittal anorectoplasty. This revealed high anorectal agenesis with recto-urethral fistula. Post-operatively, he did well, started on oral feeds which he tolerated well and started to pass stools. He was discharged home.
in a good general condition to be followed in the clinic.

Discussion
Fryns syndrome is an autosomal recessive condition characterized by multiple congenital anomalies with major involvement of the midline structures. In 1979, Fryns et al. reported this syndrome in 2 stillborn sisters with multiple congenital anomalies including coarse faces with cloudy corneae, diaphragmatic defects, absence of lung lobulation, and distal limb deformities [7, 8]. Fitch et al. reported a single infant, born of second-cousin parents, who had absent left hemidiaphragm, hydrocephalus, arhinencephaly, and cardiovascular anomalies [9]. They claimed that they were the first to describe this syndrome in 1978. In 1990, Cunniff et al. described 3 other cases, bringing the total reported cases of Fryns syndrome to 25 [10]. In 2007, a total of 83 cases of Fryns syndrome were reported in the literature [2].

Fryns syndrome is a condition that affects the development of many parts of the body and this explains the clinical variability of this syndrome. The diagnosis of Fryns syndrome is based on the presence of the clinical manifestations. The features of this syndrome however vary widely among affected individuals and overlap with the signs and symptoms of several other disorders. These factors can make Fryns syndrome difficult to diagnose and many cases go unrecognized. Fryns syndrome is a genetic disorder in which congenital diaphragmatic hernia is a cardinal feature, recorded in more than 80% of cases [11]. It has been estimated that Fryns syndrome makes up about 10% of all cases of congenital diaphragmatic hernia but many of these patients die in utero or are born as stillborn which makes it difficult to accurately determine the incidence of Fryns syndrome [4, 6]. The estimated prevalence of Fryns syndrome is 0.7 per 10,000 births [12]. There are however reports of Fryns syndrome with no diaphragmatic defects [11, 13, 14]. The reported anomalies in Fryns syndrome are variable but the most frequent anomalies were diaphragmatic defects, lung hypoplasia, cleft lip and palate, cardiac defects, including atrial and ventricular septal defects and aortic arch anomalies, renal cysts, urinary tract malformations, and distal limb hypoplasia [1-5]. Most patients with Fryns syndrome also had hypoplastic external genitalia and anomalies of internal genitalia, including bifid or hypoplastic uterus or immature testes. The gastrointestinal anomalies encountered in patients with Fryns syndrome include duodenal atresia, pyloric hyperplasia, malrotation and common mesentery. More than 50% of patients with Fryns syndrome have malformations of the brain including Dandy-Walker anomaly and/or agenesis of the corpus callosum. In 2004, Pierson et al. described 3 new patients with Fryns syndrome, reviewed 77 reported patients and summarized the abnormalities of them had unilateral microphthalmia and cloudy cornea like our patient. A few patients demonstrated cloudy cornea. Histologically, these abnormalities include retinal dysplasia with rosettes and gliosis of the retina, thickness of the posterior capsule of the lens, and irregularities of Bowman membrane. Our patient had
In conclusion, Fryns syndrome is a rare syndrome characterized by multiple congenital malformations with congenital diaphragmatic hernia present in more than 80% of the cases. It is familial and inherited as an autosomal recessive. The diagnosis of Fryns syndrome is clinical and physicians caring for these patients should be aware of this for proper diagnosis and parental counselling.

REFERENCES