Neonatal Marfan syndrome - A case report

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Abstract

Neonatal Marfan syndrome is rarely diagnosed at birth due to unusual clinical presentation. The case reported here describes a neonate with typical features.

Keywords: Newborn, Marfan syndrome
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Introduction

Marfan syndrome is one of the most common single gene defects with a prevalence of around 1 in 5000 to 10000 population [1,2]. However, the diagnosis is often missed during the neonatal period. It is on the contrary more important to detect the disease during the perinatal period as the consequences are severe and death is almost inevitable during early life. Here we describe a newborn with Marfan syndrome presenting at birth.

Case report

The female baby was born at 36 weeks gestation to a primi gravida mother with no medical illnesses. There was intrauterine growth restriction and oligohydramnios with no apparent cause for the same. Both parents were of average Indian build and the family history was unre-markable. The baby weighed 1400 grams and was 41 cm long at birth with an upper segment to lower segment ratio of 1.55:1. The arm span was a centimetre more than her length. Hand length to CHL ratio was 13.4% and foot length to CHL ratio was 15.8% (values above 11% and 15% respectively being abnormal [3]. The neonate had wide open fontanelles, cutis laxa, deep set eyes, large ears and pointed chin with a horizontal crease on the cheeks (Fig 1).Long slender fingers and toes with adductor contracture of thumbs, limited extension of the elbow joints were the findings in the upper limbs along with evidence of pes planus, equinovalgus and flexion contracture of the knee joints. The heart was clinically normal but the echo-cardiogram revealed a small ASD. The chest radiography showed emphysematous lungs predominantly in the left lung apex (Fig 2). Her eyes were normal. Her karyotyping was 46XX and had normal serum calcium levels.

The baby was treated for mild respiratory distress on the first postnatal day. She was discharged on Day 7 with a plan for follow up for cardiovascular, ocular and skeletal complications of the condition.

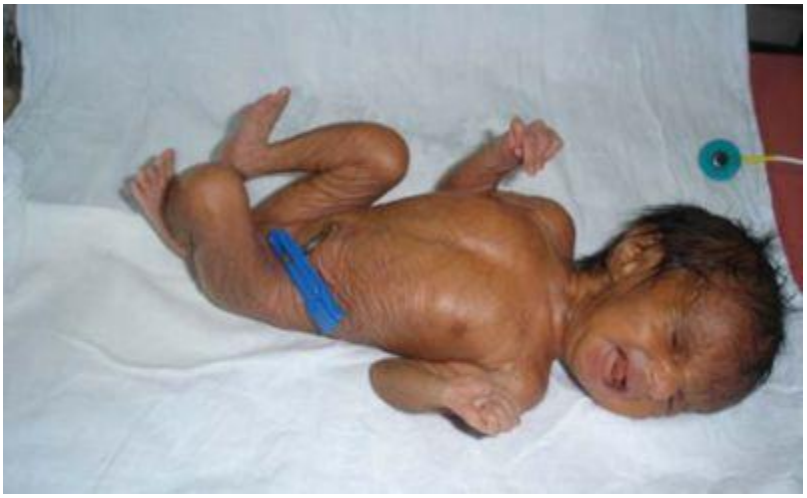

Figure 1. Note the long slender body, cutis laxa, large ears, wrinkled face, deep set eyes, arachnodactyly of toes and fingers and adductor contracture of thumbs

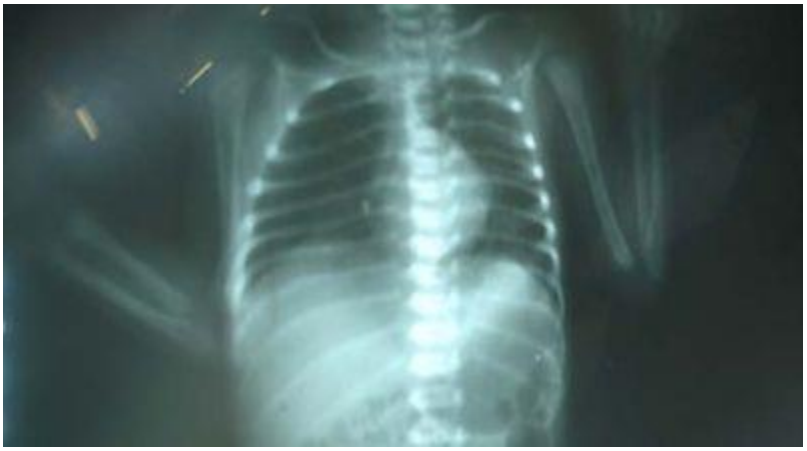

Figure 2. Chest roentgenogram showing left upper zone emphysema

Discussion

Marfan syndrome was named after Antoine Marfan, a French Paediatrician who first described the features in a 5 year old child in 1896 [4]. The presentation in the adults and adolescents is well known. Neonatal Marfan constitutes approximately 14% of the total cases [1].

This autosomal dominant defect in fibrillin synthesis is due to mutations in Chr 15. Many of the cases are sporadic without any family history. Recurring mutation hot spots in exons 24 to 27 and 31 to 32 of the FBN1 gene are found in Neonatal Marfan syndrome [1,5,6]. It is not possible however to predict whether a mutation in exons 24 to 32 will be associated with classic, severe, or neonatal Marfan syndrome [6].

The syndrome should be suspected when a neonate exhibits typical skeletal, ocular or cardiac manifestations. Specific criteria have been laid down for the diagnosis (Berlin criteria now replaced by Ghent criteria) but these are more useful in older children and false negatives are high in neonates [1]. Thin body build, dolichostenomelia, arachnodactyly, hyperextensibility, pectus chest deformity, scoliosis, joint contractures, and high-arched palate are noted at birth [7-9]. Notable features include loose and redundant skin, characteristic facies with micrognathia and abnormal pinnae. Pulmonary emphysema is commonly described and may be found incidentally during a routine skeletal survey [10]. A systematic eye examination by a pediatric ophthalmologist is warranted to detect iridodonesis,

megalocornea and ectopia lentis. A few significant differences exist between adult and neonatal marfan. Newborns present with joint contracture or dislocations, considerable morbidity due to scoliosis, more often than older affected individuals. The cardiac manifestations are also somewhat different from those in adults in the form of multivalvular disease with significant mitral, tricuspid, and pulmonary regurgitations [4]. More than half the patients experience cardiac failure. Cardiovascular problems are responsible for at least 80% of deaths in neonatal MFS [1]. Diaphragmatic hernia and hydrocephalus are unusual and are restricted to rare case reports.

Antenatal counselling is called for when there is evidence of familial affliction. The siblings are to be screened if the parents are found to have the defect. The confirmation by screening for mutation in the FBN gene is required when the clinical diagnosis is in question as usual criteria may be difficult to apply in a newborn. Regular follow up for early detection of scoliosis, lens dislocation and cardiac abnormalities is essential [11]. NeonatalMFS and its high likelihood to cause mortality in infancy or early childhood marks the need for suspicion of the condition clinically. It represents a genuine entity separate from the common adolescent form familiar to all clinicians.

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