

Neonatal Schwartz-Jampel Syndrome With Dense Bones

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Citation

P Manikoth, A Nair, S Al Khusaiby, A Rajab. *Neonatal Schwartz-Jampel Syndrome With Dense Bones*. The Internet Journal of Pediatrics and Neonatology. 2004 Volume 5 Number 1.

Abstract

Neonatal Schwartz-Jampel Syndrome is a rare disorder with abnormal facial appearance, respiratory distress, feeding difficulty, myotonic chondrodystrophy and postnatal short stature. We report a neonate with the above syndrome but having unusually dense bones and congenital heart disease, a feature not reported earlier.

INTRODUCTION

The Schwartz-Jampel Syndrome (SJS) is a rare condition characterized by constant findings such as typical facial appearance, muscle hypertrophy and continuous muscle activity. Other associated findings include skeletal abnormalities and growth and developmental delay. The authors report a newborn with type 2 SJS having dense bones and congenital heart disease and review the literature about this condition analyzing the clinical picture, the recent genetic findings and the electrophysiological studies.

CASE REPORT

A baby girl was born by spontaneous vaginal delivery to a gravida 5, para 3 mother at term with normal Apgar scores. The antenatal scan had showed short limbs and the baby had a birth weight of 3300 gm, length of 47 cm and head circumference of 36 cm. The parents were first cousins and a previous female child had died undiagnosed at 4 months of age with short limbs and ventilated for respiratory distress at birth at the local hospital and had similar appearance.

The present baby was admitted to the Special Care Baby Unit of our hospital with respiratory distress. She showed nasal flaring and grunting respiration with a rate of 80 per minute, but was not cyanosed. Air entry was equal on both sides of the chest. She had obvious dysmorphic features. The head was large with a short neck and low set ears. The chest was broad and short and she had overlapping fingers and toes with bilateral single palmar crease and camptodactyly. The facies had a typical grimace with ptosis and narrow palpebral fissures due to blepharophimosis (Fig 1).

Figure 1

Figure 1: Typical facies with myotonia and blepharophimosis



The limbs were short with bowing of femur and tibia. The abdomen was normal and examination of cardiovascular system revealed normal heart sounds with a short grade 2/6 systolic murmur. Muscles showed evidence of myotonia, more prominent on the face.

Xray of the chest revealed a short thorax with normal heart and no lung infiltrates. Xray of the long bones revealed bowing and shortness of both femur and tibia with increased density and poor corticomedullary differentiation (Figure 2).

Figure 2

Figure 2: X-ray of lower limbs showing bowing and shortening of femur and tibia with increased bone density.



Similar features were seen in the upper limbs also but were less prominent. Ultrasound examination of the head was normal. The echocardiogram showed fenestrated atrial septal defect, moderate patent ductus arteriosus and spongiform right ventricle. Chromosomal analysis revealed a female karyotype with 15 p+. The baby was given incubator oxygen, intravenous fluids and later nasogastric tube feeds. Antibiotics were given to cover for possible sepsis, although blood counts and cultures were normal. The respiratory symptoms resolved after 2 days but the baby was unable to feed by mouth due to facial myotonia. She was also noticed to have minimal inspiratory stridor and had bouts of fever unrelated to any infection. She was discharged home on nasogastric tube feeds with advice to prevent aspiration of feeds and dehydration.

DISCUSSION

Oscar Schwartz and Robert S Jampel in 1962 first reported in the Archives of Ophthalmology, two siblings having congenital blepharophimosis associated with a unique generalized myopathy (1). The above syndrome is characterised by myotonic myopathy, bone dysplasia, joint contractures and growth delay resulting in dwarfism. It is caused by a genetic abnormality mapped to one or more regions of the first chromosome. Most cases are of autosomal recessive inheritance although a few dominant

cases have also been reported (2).

Affected children have a characteristic facies with a fixed expression due to tonic contraction of the facial muscles. The eyes show a narrow palpebral fissure and ptosis due to blepharophimosis. Prominent double eyelashes, cataracts and myopia occur in some. The ears are low set with folded helices. There is micrognathia with high arched palate and microstomia occurs due to contraction of the perioral muscles and pursing of the lips. Spine shows kyphoscoliosis, lumbar lordosis, platyspondyly and cleft vertebrae. Hip dysplasia with acetabular flattening, congenital hip dislocation, joint contractures, bowing and metaphyseal widening of long bones, osteopenia and limitation of joint movement may also occur. Pectus carinatum, inguinal hernia, small testes and generalized hirsutism are other features. There is generalized myotonic myopathy with muscular hypertrophy and hyporeflexia. There is drooling of saliva with feeding difficulty and a high pitched indistinct speech. Growth and development is delayed and mental retardation occurs in 25%. Anesthesia may be complicated by difficult intubation due to microstomia and risk of malignant hyperthermia.

Two types of the disorder have been described based on clinical features and age of onset (3). Type 1 which is the classical form of the disorder has two subtypes. Type 1 A which is less severe and manifests later in childhood with moderate bone dysplasia. The original descriptions of Schwartz and Jampel correspond to this type. Type 1 B which is manifest at birth and is more severe clinically with pronounced bone dysplasia. Both these subtypes appear to be derived from the same chromosomal region. Type 2 is similar to type 1 B and manifest at birth, more severe clinically and associated with a high mortality, but mapped to a different chromosome. These infants have respiratory and feeding problems and bouts of hyperthermia in addition to joint contractures, bone dysplasia and short stature. The neonatal Schwartz-Jampel syndrome (SJS type 2) is probably the same as Stuve-Wiedemann syndrome based on clinical, radiological and histological similarities (4). The neonate described above had features typical of SJS type 2, but had unusually dense bones and congenital heart disease, features not reported earlier.

The gene defect in SJS type 1 is located in the 1p34 - p36 region of chromosome 1, whereas it is different in Type 2 (5,6). Perlecan the major proteoglycan of basement membranes is altered in patients with Schwartz-Jampel

syndrome (7). Electrophysiological studies of patients with the Schwartz-Jampel syndrome show normal nerve conduction and spontaneous activity myotonic discharge, which implies that the spontaneous activity originates in the muscle membrane. However single motor unit action potential pattern of repetitive neuronal discharges, suggests that a defect in the muscle membrane is not the only reason for the abnormality (8). Muscle biopsy shows histological evidence of a myopathy.

Treatment of this syndrome is mainly symptomatic and supportive. Early intervention is essential for these children to develop their full potential. Neonates with the severe form of the disorder may require oxygen therapy, nasogastric tube feeds and other supportive measures. Episodes of hyperthermia need proper treatment as well as prevention. Musculoskeletal abnormalities require specialised orthopaedic management. Visual abnormalities need corrective glasses to improve vision. Some children may be prone to recurrent respiratory infections. When surgical procedures are contemplated, there is risk of malignant hyperthermia on exposure to anaesthetics and muscle relaxants. Muscle stiffness and cramps can often be prevented by massage and slow warming up before exercise. Medications like phenytoin, carbamazepine and quinine have been tried for the myotonic symptoms. Genetic counselling should be offered to affected patients and their families.

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