SYNDROME OF THE MONTH

Schwartz-Jampel syndrome (chondrodystrophic myotonia)

Denis Viljoen, Peter Beighton

Abstract

Schwartz-Jampel syndrome is a rare autosomal recessive disorder. Joint contractures, generalised myotonia, skeletal

anomalies, and facial dysmorphism are common features; malignant hyperthermia is a potentially lethal complication during anaesthesia.

Figure 1 Schwartz syndrome: brothers aged 7 and 10 years with short stature, limb contractures, typical facies, and spinal malalignment. (Reproduced from Beighton P. Clin Genet 1973;4:548-55 with permission.)

Department of
Human Genetics and
MRC Unit for
Skeletal Dysplasias,
University of Cape
Town Medical School,
Observatory 7925,
South Africa.
D Viljoen
P Beighton

Correspondence to
Dr Viljoen.

The Schwartz-Jampel syndrome (SJS) is an autosomal recessive disorder characterised by short stature, skeletal abnormalities, generalised myotonia, ocular anomalies, and a unique facies.¹ Although first described in 1951 by Catel, the eponymic form 'Schwartz-Jampel' syndrome is usually preferred. About 50 cases have now been published. The diagnosis is based upon recognition of the characteristic clinical features and an abnormal, but non-specific, electromyogram. There are no specific biochemical, cytogenetic, or histopathological changes. The disorder is also known as chondrodystrophic myotonia, Aberfeld syndrome, and Schwartz syndrome.

Clinical manifestations

Clinical features may be apparent soon after birth in the form of contractures of major joints, increased muscle tone, microstomia

with feeding difficulties, muscle rigidity, and blepharophimosis.² Death has been reported from asphyxia in the newborn period in a severely affected infant, but this is an infrequent event.³ Height is usually below the 10th centile in all age groups but a single patient has reached normal stature.⁴ Myotonia results in a fixed facial expression with pursed lips and narrowed palpebral fissures. When asked to smile, affected subjects appear about to burst into tears.⁵ The voice may be high pitched. Blepharophimosis or ptosis, micrognathia, low set ears with folded helices and medial displacement of the outer canthi are additional features.³ Persons with SJS have a waddling gait and crouched stance and tiredness results from stiffness of the joints.⁶ Some degree of mental retardation is said to be present in about 25% of patients.³ It is uncertain whether this problem is a primary syndromic component or a consequence of the limitations posed by the severity of the physical handicap.

Figure 2 The brothers depicted in fig 1, now aged 25 and 28 years, showing progression of limb contractures and spinal deformity. (Top right photograph reproduced from Horan F, Beighton P. *J Bone Joint Surg (Am)* 1975;57:542-4 with permission.)

Figure 3 A 6 year old child with Schwartz syndrome. Note the similarity of phenotype to that of the patients in fig 1.

MUSCULOSKELETAL FEATURES

Limitation in the range of movements of major joints is progressive, with flexion at the hips, knees, shoulders, and elbows.⁶ The contractures are maximal by mid-adolescence and thereafter remain static. Pectus carinatum, kyphoscoliosis, lumbar lordosis, bowing of the long bones, pes planus, and valgus ankle deformities are characteristic of SJS. Metaphyseal widening of the long bones may be clinically evident but the digits are usually normal.

Radiographic changes include marked platyspondyly with coronally cleft vertebrae. Failure of development of the anterior half of vertebrae may give rise to severe kyphosis which usually occurs in the thoracolumbar region but is occasionally seen in the cervical spine. Hip dysplasia with acetabular flattening and severe coxa vara is usually present and congenital dislocation of the hip may occur. The diaphyses of the leg bones are bowed anteriorly. The capital femoral epiphyses may be grossly misshapen and the upper tibial and lower femoral epiphyses may be irregular and cupped during early childhood.^{6,7}

Muscle hypertrophy is evident in most patients and myotonia can be shown by thenar percussion. Tendon reflexes are generally depressed although muscle tone is increased.

OTHER CLINICAL FINDINGS

Additional clinical findings commonly include generalised hirsutism, blepharospasm, myopia

in 50% of cases, umbilical and inguinal hernias, and small testes. Myotonia may result in drooling and indistinct speech.³

Investigations

In addition to the radiographic appearances, electromyography is a useful confirmatory procedure; no pathognomonic biochemical or histopathological tests are available. It has been claimed that SJS is not a true myotonic disorder as the continuous repetitive discharges recorded on myography can be abolished with curare.⁴ Other investigations have shown independent repetitive discharges from individual muscle fibres which are unaffected by curare.⁸

Apart from diffuse atrophy, light microscopic studies of muscle are normal in SJS. Vacuolation in muscle fibres seen on electron microscopy has been occasionally documented.⁴

Reduced numbers of B and T cells in the haemopoietic system of some patients with SJS⁹ have suggested that an immunological deficiency is associated with the disorder, but there is little clinical evidence to support this.

Pathogenesis

The basic defect in SJS is unknown, and it is uncertain whether the short stature and bone malalignment seen in affected persons is a primary growth defect or a malformation owing to persistent muscle contractions from myotonia. New light has been shed on pathogenesis with the finding that a sodium channel defect may be responsible for the hyperexcitability and associated slowed relaxation of muscle fibres in affected subjects.¹⁰

Genetics and prevalence

SJS is inherited as an autosomal recessive trait and affected sibs have been documented in more than a dozen kindreds.^{3,4} Parental consanguinity was noted in four of these families and the male:female ratio is equal. Reproductive fitness in affected persons is unknown as few subjects have reached adulthood and none is known to have reproduced.

The disorder is said to be infrequent but it is possibly less rare than supposed. SJS has been reported in diverse population groups including persons of Italian, Irish, Dutch, German, Portuguese, and Brazilian descent.⁴ We have encountered six affected persons in southern Africa during a 20 year period; two were brothers of mixed ancestry and the others were sporadic subjects in the black population (figs 1 to 4). These latter persons were members of different tribal groups and it is therefore evident that the gene is widespread, although of relatively low frequency, in this community.

Prenatal diagnosis by means of ultrasound has been achieved in the midtrimester of pregnancy in a female fetus whose male sib in a previous pregnancy had severe features of SJS. Constant flexion of the fingers, decreased fetal activity, and mild shortening and bowing of

*Figure 4 Platyspondyly is evident with anterior wedging of a vertebral body in the thoracolumbar region. The long bones are gracile with bowing of the tibiae and metaphyseal cupping. (Top left photograph reproduced from Horan F, Beighton P. *J Bone Joint Surg (Am)* 1975;57:542-4 with permission.)*

the femora were noted.¹¹ However, ultrasonic prenatal diagnosis cannot always be made with certainty except in severely affected fetuses.

Differential diagnosis

The disorder may be difficult to differentiate clinically from Freeman-Sheldon and Marden-Walker syndromes. In the former, the 'whistling face' is characteristic and the lips are puckered with microstomia, a long philtrum, and dimpled chin. Ulnar deviation of the elongated fingers produces a windmill vane configuration. Marden-Walker syndrome is a multiple congenital anomaly disorder consisting of immobile facies, blepharophimosis, ptosis, multiple congenital joint contractures, failure to thrive, and mental retardation.

Other conditions to be differentiated from SJS are Seckel syndrome, familial myotonia, myotonic dystrophy, and various congenital contractural disorders and arthrogryposes.

Complications

Major complications of SJS may arise during anaesthesia. These include mechanical difficulties during intubation owing to microstomia and jaw muscle rigidity. Malignant hyperthermia is a well documented and potentially lethal hazard.¹²

The capacity of affected subjects to perform manual work is considerably reduced because of their joint contractures. Progressive crippling may require orthopaedic surgical intervention and cosmetic repair of their narrowed

palpebral fissures may be necessary to facilitate normal vision. Ophthalmological treatment of myopia and juvenile cataract is occasionally warranted.

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