Schwartz-Jampel Syndrome Type I

**Alternative Names**
SJS1
Schartz-Jampil Syndrome
SJS
Myotonic Myopathy, Dwarfism, Chondrodystrophy, and Ocular and Facial Abnormalities
Schartz-Jampil-Aberfelt Syndrome
SJA Syndrome
Chondrodystrophic Myotonia
Myotonic Chondrodystrophy

**WHO International Classification of Diseases**
Diseases of the nervous system

**OMIM Number**
255800

**Mode of Inheritance**
Autosomal recessive

**Gene Map Locus**
1p36.1

**Description**
Schwartz-Jampel syndrome is characterized by a considerable variety of features with most children first demonstrating signs in late infancy. Symptoms include: skeletal dysplasia, myotonia with mask-like face, blepharophimosis, microstomia, and growth retardation. Two types have been described: The classical form which usually manifests late in infancy and a rare, more severe form with neonatal manifestation.

In Schwartz-Jampel syndrome death occurs because of respiratory compromise. Hypertrophy of the muscle fibers of the diaphragm, tongue, larynx, and perilaryngeal muscle together with repeated aspiration and IgA deficiency could be contributing factors to the respiratory complications.

The diagnosis of Schwartz-Jampel syndrome depends on the typical clinical features, electromyography and characteristic changes on X-ray. The most common X-ray abnormality observed in patients with Schwartz-Jampel syndrome is prominent sternum and hip dysplasia. The characteristic X-ray appearance of the hip and spine becomes more evident with age usually toward the end of the first year.

**Molecular Genetics**
Recessive Schwartz-Jampel syndrome could be caused by mutations in the heparan sulfate proteoglycan type 2 (perlecan) gene (HSPG2). Heparan sulfate proteoglycan is a major component of basement membranes, where the molecule may be involved in the stabilization of other molecules as well as being involved with glomerular permeability to macromolecules and cell adhesion. HSPG2 gene is composed of 97 exons spanning at least 120 kb and probably has multiple transcription start sites. Perlecan gene transcription is upregulated by TGF-beta.

**Epidemiology in the Arab World**

**Egypt**
Meguid et al (1998) reported, for the first time in Egypt, five cases from three different families with Schwartz-Jampel Syndrome (SJS). The five cases were two male sibs from one family, two male sibs from another family, and one sporadic female. Patients were subjected to complete case history, neurological examination, electromyography (EMG), estimation the levels of serum creatinine phosphate kinase (CPK), immunoglobulin (IgG, IgM, IgA) and adenosine deaminase (ADA) enzyme, chromosomal analysis, and clinical assessment before and after treatment with diphenylhydrantoin. They showed all distinctive features of SJS like; mask- like face, short stature blepharophimosis, microstomia, generalized myotonia, and limited mobility of

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the major joints. Spontaneous activity of two males from different families was in the form of persistent typical myotonic discharges, whereas spontaneous activity of the other three cases was atypical myotonic discharges. Meguid et al (1998) indicated that the origin of spontaneous activity in SJS is located in the muscle membrane. Chromosomal analysis was normal for all patients. Two types of SJS were described in each family with two sibs; the classical form that appeared in the late infancy, and the severe form which started at birth and resulted in neonatal death. The female patient had the classical form of SJS. Meguid et al (1998) observed low clinical variability between the five cases, except for the osteoarticular abnormalities; therefore, they suggested that other factors than SJS gene might contribute to the phenotype. Serum IgA was deficient in two males from one family and in the female. Only one male with classical SJS had a slightly elevated CPK level. The cases with neonatal SJS had markedly ADA enzyme deficient and general abnormality of the immune function which may be the cause of death due to recurrent chest infection and respiratory failure. Meguid et al (1998) suggested an autosomal recessive inheritance mode of SJS due to the presence of consanguinity, affected sibs, and young parental ages in their study.

**Oman**

Al-Gazali et al. (1996) reported 11 children in 5 families, four of Omani origin, with severe neonatal Schwartz-Jampel Syndrome. All presented at birth with skeletal abnormalities and feeding difficulties. Five had the typical pursed appearance of the mouth. Nine died from respiratory complications (5 in the neonatal period and 4 before 2 years of age). One (4 months old) remains hospitalized since birth requiring continuous oxygen supplementation and one (5 months old) requires nasogastric tube feeding and has repeated attacks of aspiration. Al-Gazali et al. (1996) suggested that within the group of cases of neonatal Schwartz-Jampel Syndrome there is a subgroup with severe respiratory complications and early death.

**United Arab Emirates**

Al Gazali (1993) reported three sibs, two females and one male, of a family of United Arab Emirates origin with severe manifestation of Schwartz-Jampel Syndrome. The family consists of unrelated parents with a total of eight children. Two of these children survived until the age of three and four years respectively and one died at two years of age. The two older children, both females, died of overwhelming chest infections at 3-4 years of age. Both had, in addition to the shortening and flaring of the metaphyses, severe spinal deformity (kyphoscoliosis) and fragmented femoral epiphyses with rapid destruction on the right side in the older child. The younger male patient had multiple admissions because of failure to thrive and recurrent respiratory problems with laryngospasm. He died at the age of 2 years. In year 1996, the total number of children with neonatal Schwartz-Jampel Syndrome reported from the United Arab Emirates was estimated to be 14; this was enough reason for Al-Gazali et al. (1996) to suggest that Schwartz-Jampel syndrome is fairly common in the population of the United Arab Emirates [See also: Oman > Al-Gazali et al. (1996)].

**References**


**Contributors**

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