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Research Paper

Medical Science

KEYWORDS : Autosomal, inherited, chest deformity

A Study of Schwartz Jampel Syndrome

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ABSTRACT

Schwartz-Jampel syndrome (SJS) is a term now applied to 2 different inherited, autosomal recessive conditions, sometimes termed SJS type I and SJS type II. Both are very rare. SJS type I has 2 recognized subtypes, IA and IB, which are similar, except that type IB manifests earlier and with greater severity. 3 yr old boy is brought by his mother to neuro OPD with chief complaints of Chest deformity since 2 yrs, Intermittent abnormal facies since 1.5yr and Abnormal gait since 1yr

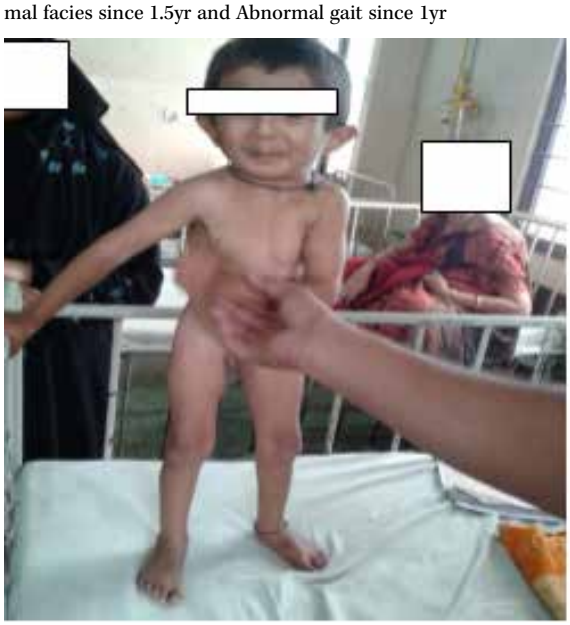
INTRODUCTION

Schwartz-Jampel syndrome (SJS) is a term now applied to 2 different inherited, autosomal recessive conditions, sometimes termed SJS type I and SJS type II. Both are very rare. SJS type I has 2 recognized subtypes, IA and IB, which are similar, except that type IB manifests earlier and with greater severity [1].

This condition also called as chondrodystrophic myotonia was first described in the year 1962 by Schwartz and Jampel as congenital blepharophimosis associated with unique generalized myopathy. The prevalence is <1/million and total reported cases are <100. This is inherited as autosomal dominant and autosomal recessive. Most patients become symptomatic between 3 years and 10 years. They present with typical phenotypic features, characterized by short stature, mask-like facies, epicanthic fold, receding chin, upturned nose, long philtrum, short neck, pinched face, low-set ears, high arched palate, prominent muscles, and high pitched voice. The diagnostic findings are typical facial appearance, muscle hypertrophy, and continuous muscle fiber activity [2].

CASE REPORT

3 yr old boy is brought by his mother to neuro OPD with chief complaints of Chest deformity since 2 yrs, Intermittent abnormal facies since 1.5yr and Abnormal gait since 1yr



Child was apparently asymptomatic till 1 yr of age then he developed progressive deformity of chest.

Associated with recurrent respiratory tract infections, feeding difficulties.

- Abnormal facies :
- Puckered face.
- Difficulty in eye opening.
- Perioral stiffness.
- Abnormal facies relieved gradually over a period of 15- 20 mins spontaneously.
- Several episodes per day.
- Occur even during sleep
- Difficulty in walking- stiff limbs.
- No diurnal variation.
- No h/o of regurgitations, autonomic disturbance, seizures , constipation ,
- h/o not gaining of weight

ANTENATAL HISTORY : Booked case, not significant

- No h/o decreased fetal movements.

BIRTH HISTORY :

- 1st in birth order/Term /LSCS / birth wt. 3.3 kg
- No h/o any NICU admission.

DEVELOPMENTAL HISTORY:

- Achieved according to age .
- VACCINATION HISTORY:
- Child immunised according to UIP

FAMILY HISTORY:

died due to neural tube defects and congenital hydrocephalus at the age of 9 months.

SOCIOECONOMIC :

Lower middle class

GENERAL EXAMINATION :

- Microcephaly
- low set ears
- narrow palpebral fissure
- (blepharophimosis)
- Puckered facies
- Pectus carinatum
- Generalised Muscle Hypertrophy
- Joint Contractures
- Short Stature
- Short neck

VITALS :

- Temp normal
- Pulse 88/min normal volume
- RR 22/min
- BP 80/50 mm Hg rt arm

ANTHROPOMETRY

- Wt 8 kg < 3rd centile
- Ht 83 cm <3rd centile
- HC 46.5 < 3sd

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- MUAC 12.5 cm
- Us /Ls 1.2

Dwarf, proportionate
Microcephaly
- Nervous System
- Higher functions normal
- Speech: muffled
- Cranial nerves normal
- Motor system
- Hypertrophy of all proximal muscles of UL and LL, Chest , Abdomen
- TONE hypertonia in all group of muscles
- REFLEXES AJ + +
- other DTR absent
- POWER :> 3/5 in all proximal and distal muscle and is ambulatory without support
- child not co-operative for tongue myotonia
- Myotonia of forearm muscles present.
- Sensation - normal
- GAIT waddling gait loss of associated movements
- Spine kyphosis
- Short neck

Investigation
- CPK – slightly elevated{351}
- EMG s/o myotonia
- S.Calcium , Phosphorus ,ALP, Vit D : Normal
- T3,T4,TSH: Normal
- ECG, 2D Echo: Normal

Skeletal survey
- Generalised decrease in bone density
- Metaphysial widening
- No bowing of legs
- SPINE : Anterior wedging of few lumbar vertebrae

Differential diagnoses
- Schwartz – Jampel Syndrome
- Myotonia congenita
- Marden –walker syndrome
- Stiff Person Syndrome
- Congenital muscular Dystrophy

Schwartz – Jampel Syndrome
- It is a rare autosomal recessive disorder characterized by permanent myotonia (prolonged failure of muscle relaxation) and skeletal dysplasia, resulting in reduced stature, kyphoscoliosis, bowing of the diaphyses and irregular epiphyses.
- Narrow palpebral fissures with normal eyelid development
- Blepharospasm
- Hypertrichosis of the eyelids – i.e., excessive hair , multiple rows of hair
- Micrognathia
- Unusual, flattened facies with puckered facial appearance

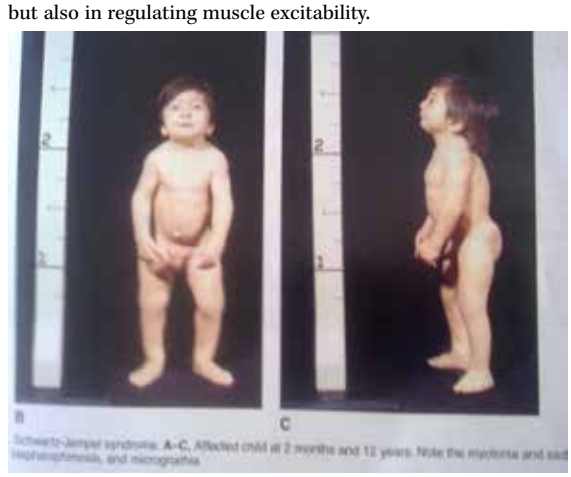
Skeletal and joint
- Short neck
- Pectus carinatum – convex chest; ie, chest is bowed out
- Kyphosis – convex angulation of the spine, giving a humpback appearance
- Coxa valgus– Hip deformity involving an increased neck – shaft angle of the femur
- Irregularity of the capital femoral epiphyses
- Its of two types
- SJS type 1: its resembles myotonic disorders , stiff person syndrome, and Isaacs syndrome
- But in stiff person syndrome stiffness decreases during sleep.
- it is of two types :
- 1A, 1B

Type 1A:
- Classical form that develops in early childhood between first and third year of life.
- Most common
- Muscle stiffness is mild and generally non progressive
- Does not significantly shorten the lifespan.

Type 1B :
It is less common but more severe and appears immediately after birth

Type 2 :
- Similar to 1B but gene on different locus
- Shortens life span, most will not survive till adulthood
- Type 1A, 1B derived from mutations of same gene HSPG2 which codes for perlecan ,a heparan sulfate proteoglycan part of basement membrane of cartilage and muscle.
- Mutations in the perlecan gene (*HSPG2*) cause two classes of skeletal disorders: the relatively mild Schwartz-Jampel syndrome (SJS) and severe neonatal lethal dyssegmental dysplasia, Silverman-Handmaker type (DDSH).

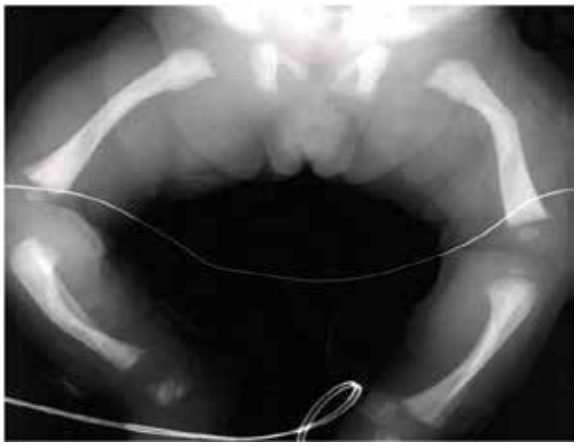
The first human mutations in *HSPG2*, which underscore the importance of perlecan not only in maintaining cartilage integrity but also in regulating muscle excitability.



Schwartz-Jampel syndrome. A–C, Affected child at 2 months and 12 years. Note the myotonia and

Medications used in the treatment of Schwartz-Jampel syndrome (SJS) include the following:

- Anticonvulsants - Phenytoin, carbamazepine
- Antiarrhythmic agents - Mexiletine, procainamide, quinidine – not used in children.
- Antimalarials - Quinine
- Neuromuscular blocking agents - Botulinum toxin
- **The Stüve-Wiedemann syndrome** (SWS) is a rare disorder characterized by respiratory distress, hyperthermic episodes, and early lethality and radiologically by bowing of the long bones with internal cortical thickening and large metaphyses. We report findings in 8 new patients suggesting that this syndrome is clinically homogeneous. All patients had feeding and swallowing difficulties, respiratory insufficiency, abnormal appearance, muscle hypotonia, and postnatal short stature. Recurrent episodes of unexplained fever occurred in all and were the cause of death in 6 of 8 cases. Parental consanguinity and sib recurrence suggest autosomal recessive inheritance. The clinical, radiological, and histological similarities between our patients with SWS and those with the recently delineated "neonatal" Schwartz-Jampel syndrome (SJS type 2) lead us to suggest that SWS and SJS type 2 may be a single entity



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