Images in Clinical Practice

Schwartz-Jampel Syndrome

An eight year old boy presented with short stature and dysmorphic features. His face was flat with fixed sad apearance. He had hypertelorism, short palpebral fissure and blepherophimosis which was marked on the left side. His mouth was small with pursed lips and neck very short. He had gross pectus carinatium, right sided small inguinal hernia and small testes (*Fig.*). He had several skeletal abnormalities. These included a small mandible, widened metaphysis of long bones, scoliosis in thoraco-lumbar spine, coxa vara, genu valgus and contractures at all bigger joints. All muscles were in a state of contraction giving an appearance of hypertrophied muscles. He had IQ of 70. A clinical diagnosis of Schwartz-Jampel syndrome was made.

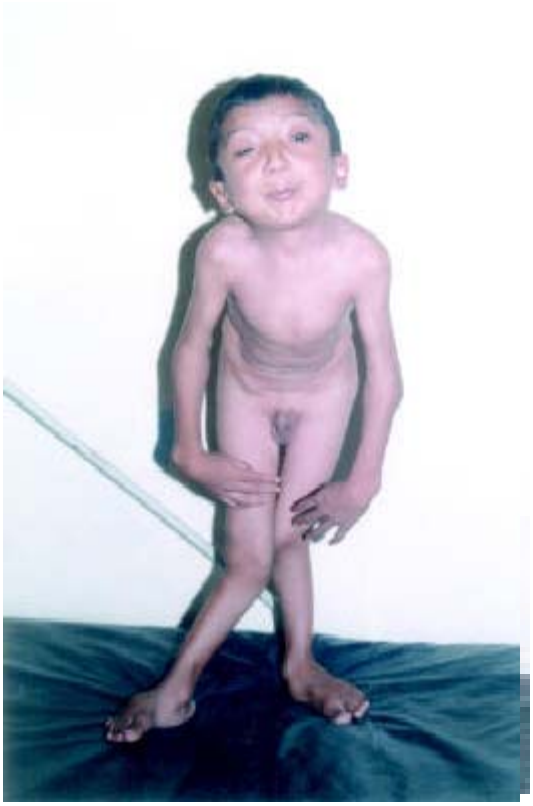

Fig. 1. Clinical appearance of Schwartz-Jampel syndrome.

Schwartz-Jampel syndrome (Syn: mytonic myopathy, chondro-dystrophic myotonia) is a rare autosomal recessive disorder caused by a defect in chromosome one. The characteristic feature of the syndrome include short stature, flat face with sad fixed appearance, full cheeks and low hair line. Eyes have narrow palaberal fissure, blepharophimonis and long irregular eye lashes. Mouth is small with pursed lips. Short neck, pectus carinatum, umbilical hernia, inguinal hernia, small testes are other features.

Skeletal features include small mandible, spinal anomalies, hip contracture, fragmenting of femoral epiphysis, widened metaphysis, joint contractures., osteoporosis and delayed bone age. Muscle have characteristic myotonic features with weakness and wasting. Mental retardation is seen in 25% and voice is high pitched. Contractures progress with age. CPK and aldolase may be mildy raised in some cases, EMG might show continous discharge and muscle biopsy shows nonspecific myopathic changes.

Physiotherapy and medication like phenytoin, carbamazepine, mexiletin and procainamide may help myotonia.

M.L. Kulkarni,
Rajappan Pillai,
Department of Pediatrics,
J.J.M. Medical College, Davangere,
Karnataka, India.