

Spinal Muscular Atrophy With Scoliosis: The Juvenile Kugelberg-Welander Type

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Introduction

This is an autosomal recessively inherited degeneration of the anterior horn cells of the spinal cord resulting in symmetrical weakness of trunk and proximal muscles including those of the shoulder and pelvis girdle in 60% of cases, there is a positive family history and both genders are equally affected. The age at onset and the extent of clinical involvement are the most important prognostic factors. There are two main areas of concern- the hips and spine. Neuromuscular scoliosis is either balanced (down to a square pelvis) or unbalanced (a long C-shaped curve going down to an oblique pelvis).

Report

A 17 years old boy referred from tertiary hospital. He was diagnosed as Spinal Muscular Atrophy type II with neuromuscular scoliosis with Cobb's angle 53° and long C shaped thoracolumbar curve (Fig. 1). Clinically, he presents with hypotonia, progressive symmetric and proximal weakness affecting the legs more than the arms, sparing of the facial muscles with bulbar muscle weakness. There is also weakness of the intercostal muscles with relative sparing of the diaphragm, which results in the typical "bell-shaped" chest and paradoxical breathing pattern. He is admitted for posterior instrumentation and fusion from T4 to sacro-alar iliac (Fig. 2). Post operatively no neurological deficits and able to sit straight (Fig. 3).

Conclusion

For many years, only one infantile form of muscular atrophy described by Werdnig and Hoffmann. Then, a later onset form with a better prognosis was described by Kugelberg and Welander. Therefore, while the condition is

now regarded as one continuum it is still useful to consider infantile (Wernig-Hoffmann) and juvenile (Kugelberg-Welander) forms. The juvenile type has an onset between 2 years and 17 years. The diagnosis is made by the clinical and family history, the physical examination and three investigations-serum enzyme studies, electrophysiology and nerve and muscle biopsy. Scoliosis surgery is commonly complicated by pulmonary problems therefore we did a posterior instrumentation with fusion. As for this case, surgery is the best indicated in the wheelchair sitter who lost spinal stability.



References

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