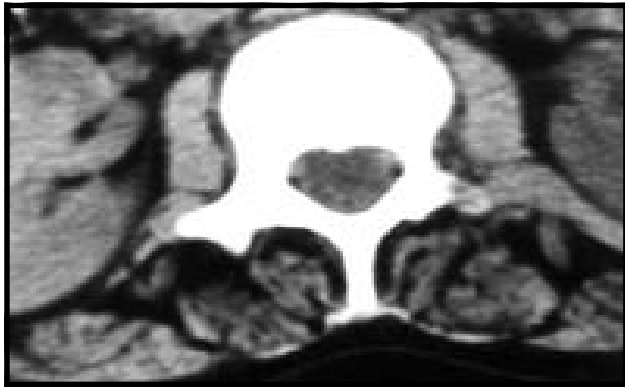


# Radiology Quiz

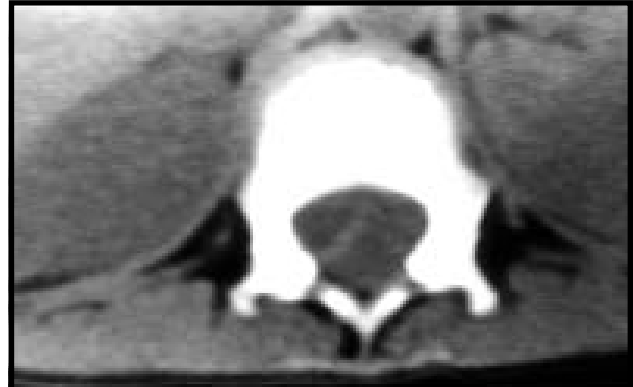
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## Clinical History

Three children, a brother and a sister in one family and another boy from a different family were admitted for evaluation of progressive bent posture, restricted neck movements and gait abnormalities. In addition to serum creatine kinase and muscle biopsy, computerized tomography scan of the lumbar spine was also carried out in all patients.



**Figure 1** - Patient computerized tomography scan.



**Figure 2** - Normal computerized tomography scan.

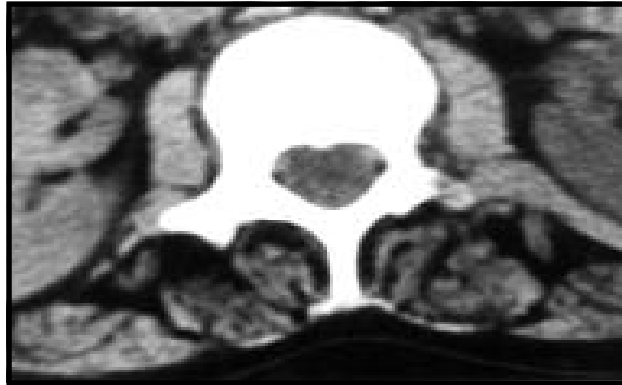
## Questions

1. What are the radiographic abnormalities?
2. What is the diagnosis?

# Radiology Quiz

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## Answer Page



**Figure 1** - Computerized tomography scan of spine shows the loss of muscle substance in Longissimus dorsi. Ilio costalis lumborum and multifidus muscles are being replaced by fibro-fatty tissues. Psoas and quadratus lumborum muscles are normal.

### Findings and Discussion

The clinical features in these 3 children were of Rigid Spine Syndrome (RSS).<sup>1</sup> This is a heterogenous group of disorders. The phenotype has been associated with Merosin Positive Congenital Muscular Dystrophy (MPCMD) and Emery-Dreifuss Muscular Dystrophy (EDMD). Emery-Dreifuss Muscular Dystrophy is often referred to as a RSS and other times X-linked scapuloperoneal syndrome.<sup>2</sup> Contractures and cardiac involvement are the hallmark of EDMD, which links to chromosome Xq28 as a recessively inherited muscular dystrophy. Rigid Spine Syndrome has been demonstrated to be linked to chromosome 1 and gene mapped to x1 is SEPN 1.

Emery and Dreifuss described EDMD in 1966. Dubowitz named the same syndrome RSS in 1973. The main features of the disease are stiffness of back and neck accompanied by elbow contractures followed by progressive scoliosis in teens. The creatine kinase was elevated moderately. Affected individuals often developed arterioventricular block. The gene for EDMD is distal Xq28. The sequence of this gene, termed emerin, is 2100-bp long. Different mutations have been found. In some, no mutations are present indicating genetic heterogeneity. All our patients had elevated creatine kinase. The muscle biopsy in one patient showed features of muscle dystrophy.

### References

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2. Smith SA, Swaiman KF. Muscular Dystrophies. In: Swaiman KF, Ashwal S, editors. *Pediatric Neurology, Principles and Practice*. 3rd ed. St. Louis (MO): Mosby; 1999. p. 1245.