Progressive scoliosis in central core disease

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Central core disease (CCD) is a rare congenital myopathy with autosomal dominant inheritance. Here, we report on two cases of progressive scoliosis in CCD, pointing out the value of a muscle biopsy to establish the correct diagnosis. The first case involves a 13-year-old boy with severe progressive scoliosis and joint contractures. The patient was initially diagnosed with arthrogryposis multiplex congenita. The second case involves a 45-year-old man with severe scoliosis that had slowly progressed over the years. Both patients suffered from unexplained muscle weakness and severe restriction of pulmonary function. The correct diagnoses were established through muscle biopsies taken from the paravertebral musculature during scoliosis surgery. Correction of the spinal deformities was achieved through posterior instrumentation in both patients, with prior anterior release in one patient. Although scoliosis is a common feature in CCD, the correct diagnosis can be missed in scoliosis patients. Therefore, we recommend a muscle biopsy in patients with scoliosis, unexplained muscle weakness and multiple joint problems.