Spinal Deformity in Neurofibromatosis

Early-onset scoliosis (EOS) or kyphosis is common in patients with neurofibromatosis (NF) and is characterized by rapid progression of deformity.

Scoliosis is the most common spinal deformity observed in NF1 and approximately 2% of all pediatric scoliosis cases are associated with NF1 [1].

»: Among patients with NF type 1 (NF1), morphologic characteristics of the spinal deformity are different in those with paraspinal neurofibromas than in those without paraspinal tumors.

»: Patients with NF1 are at low risk for developing malignant peripheral nerve sheath tumors during childhood (<1%) and their lifetime (8% to 12%), and routine imaging surveillance for malignancy in the absence of symptoms should be clinically directed.

»: Further investigation is needed to standardize screening for EOS in children with NF1 and to develop guidelines for ideal imaging modalities, including their frequency and a timeline [2].

Classification

Scoliosis in NF1 can be further sub-classified as either non-dystrophic or dystrophic, based on the presence of various radiographic findings.

Pathogenesis

Despite the high prevalence and significant morbidity of spinal anomalies in neurofibromatosis type 1 (NF1), the pathogenesis of these defects remains largely unknown.

Rhodes presents two murine models: Nf1flox/-; PeriCre and Nf1flox/-;Col.2.3Cre mice, which recapitulate spinal deformities seen in the human disease. Dynamic histomorphometry and microtomographic studies show recalcitrant bone remodeling and distorted bone microarchitecture within the vertebral spine of Nf1flox/-; PeriCre and Nf1flox/-; Col2.3Cre mice, with analogous histological features present in a human patient with dystrophic scoliosis. Intriguingly, 36-60% of Nf1flox/-; PeriCre and Nf1flox/-; Col2.3Cre mice exhibit segmental vertebral fusion anomalies with boney obliteration of the intervertebral disc (IVD). While analogous findings have not yet been reported in the NF1 patient population, we herein present two case reports of IVD defects and interarticular vertebral fusion in patients with NF1. Collectively, these data provide novel insights regarding the pathophysiology of dystrophic spinal anomalies in NF1 and provide the impetus for future radiographic analyses of larger patient cohorts to determine whether IVD and vertebral fusion defects may have been previously overlooked or underreported in the NF1 patient population [3].

Treatment

Spinal Deformity in Neurofibromatosis treatment.

Case series

The natural history, associated anomalies, and response to operative and nonoperative treatment were reviewed in 102 patients with neurofibromatosis and spine deformity. Eighty patients were found to have curvatures associated with dystrophic changes in the vertebrae and ribs. The presence of dystrophic changes such as rib penciling, spindling of the transverse processes, vertebral scalloping, severe apical vertebral rotation, foraminal enlargement, and adjacent soft-tissue neurofibromas was found to be highly significant in prognosis and management. Brace treatment of dystrophic curves was unsuccessful. Posterior fusion, with or without internal fixation, was the procedure of choice for problems due purely to scoliosis. Patients with dystrophic kyphoscoliosis required both anterior and posterior fusion to achieve stability. Sixteen patients had compression of the spinal cord or cauda equina [4].

1)
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