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Letter to the Editor

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Complex spinal case associated with Angelman syndrome

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Dear Editor,

We would like to share a rare and challenging case of rapidly progressive scoliosis in a pediatric patient with Angelman syndrome, which may contribute to the clinical understanding of spinal deformities associated with neuromuscular disorders. A 14-year-old female patient diagnosed with Angelman syndrome presented with a rapidly worsening spinal deformity over the past three months. Her gait, previously ataxic and unsteady, deteriorated significantly, and she developed marked weakness in the right lower extremity. Caregivers also reported a decline in her motor performance at her special education school, and she became unable to ambulate without assistance. On neurological examination, the patient was non-verbal and appeared apathetic, with severe cognitive impairment. Muscle strength was assessed using the medical research council (MRC) 0–5 scale. Preoperatively, the upper extremity strength was 5/5 bilaterally, the left lower extremity was 4/5, and the right lower extremity was 2/5.

Angelman syndrome (first described by Dr. Harry Angelman in 1965) is a neurogenetic disorder caused by abnormalities in the maternal chromosome 15q11-q13 region (loss of maternal UBE3A gene function), with an estimated incidence of approximately 1 in 15.000-30.000 live births.[1] Clinically, Angelman syndrome is characterized by severe developmental delay, profound speech impairment (minimal to no spoken language, though receptive comprehension is often better), gait and balance difficulties (ataxic movements), microcephaly, abnormal electroencephalography findings, disordered sleep, and a characteristically happy demeanor with frequent laughter.[2] Notably, scoliosis is a common comorbidity in Angelman syndrome, reported in roughly 10–20% of affected children and up to 30–50% of adults as they age.[3,4] Given the progressive nature of her scoliosis and associated neurological deficits, surgical intervention was considered. In such complex spinal cases, intraoperative neuromonitoring plays a critical role in reducing the risk of neurological complications.

Intraoperative neuromonitoring included somatosensory evoked potentials (SEP), motor evoked potentials (MEP), and free-run electromyography, with total intravenous anesthesia (TIVA) used to optimize signal quality, avoiding inhalational agents and minimizing neuromuscular blockade. Baseline SEP and MEP responses were obtained in the supine position after induction and intubation, and monitored continuously after turning the patient prone and throughout all stages of correction. Postoperatively, posterior segmental pedicle-screw instrumentation resulted in correction and improved coronal alignment (**Figure**).

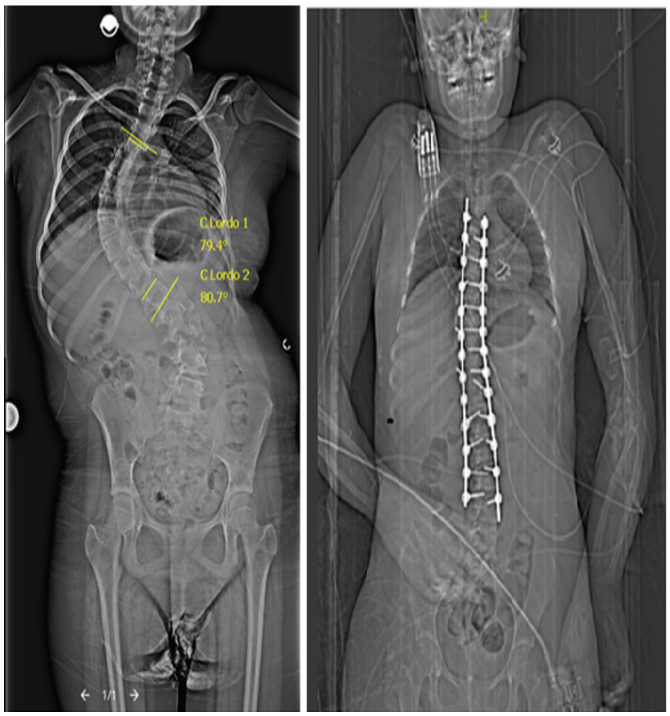

Figure. Pre- and immediate postoperative whole-spine radiographs in a 14-year-old girl with Angelman syndrome. (**A**) Standing AP film shows severe double-curve neuromuscular scoliosis with Cobb angles of 79.4° and 80.7°. (**B**) Postoperative AP film demonstrates posterior segmental pedicle-screw instrumentation with correction and improved coronal alignment

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This case underscores the need for early recognition and individualized surgical planning for spinal deformity in Angelman syndrome. Intraoperative neuromonitoring is indispensable—baseline values should be obtained in the supine position, re-verified after prone positioning, and neurophysiological signals interpreted throughout corrective maneuvers in close coordination with anesthesia—to minimize risk in these high-risk patients.

ETHICAL DECLARATIONS

Informed Consent

The patient signed and free and informed consent form.

Referee Evaluation Process

Externally peer-reviewed.

Conflict of Interest Statement

The authors have no conflicts of interest to declare.

Financial Disclosure

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Author Contributions

All of the authors declare that they have all participated in the design, execution, and analysis of the paper, and that they have approved the final version.

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