

Surgical Outcomes and Radiologic Features of Scoliosis in Escobar Syndrome: A Retrospective Review

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INTRODUCTION:

Escobar Syndrome is a rare genetic condition classified as a non-lethal form of Multiple Pterygium Syndrome. It is caused by a mutation in the cholinergic receptor nicotinic gamma subunit gene, leading to impaired fetal movement, joint contractures, and pterygium. One of the most common orthopedic manifestations is scoliosis, reported in up to ninety-three percent of affected patients. Despite this high prevalence, spinal management in Escobar Syndrome remains poorly understood due to limited data.

METHODS:

This study aims to report the clinical presentations, radiographic characteristics, surgical treatments, and outcomes in patients with Escobar Syndrome and scoliosis. A single-center retrospective review was conducted on patients diagnosed between 2000 and 2024. Of the twenty-one patients identified, twenty met inclusion criteria.

RESULTS:

Fourteen underwent spine surgery at an average age of seven years. Surgical techniques included growing rods, magnetically controlled rods, posterior spine fusion, and vertical expandable prosthetic titanium rib. Radiographic evaluation showed congenital spinal anomalies and high rates of intraspinal abnormalities, including tethered cords in forty percent of patients. A total of eleven complications were recorded, with the highest rate observed in patients treated with vertical expandable prosthetic titanium rib. Only two patients were successfully managed conservatively.

DISCUSSION AND CONCLUSION:

Our findings demonstrate that scoliosis in Escobar Syndrome often presents early, progresses rapidly, and is associated with a high incidence of intraspinal anomalies, especially Tethered Cord Syndrome. The results suggest vertical expandable prosthetic titanium rib may be contraindicated in this population due to frequent complications. Preoperative imaging and neurological evaluation are crucial to guide safe and effective scoliosis treatment in these patients. This represents the largest reported cohort of patients with Escobar Syndrome and scoliosis management to date.