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Research Article

Noonan Syndrome and Paravertebral Schwannoma: About A Rare Association

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Abstract

Noonan syndrome is a rare autosomal genetic multisystemic disorder. Extramedullary tumor is rarely associated. Neurological manifestations are rare, with neurofibroma being the leading condition. No cases of paravertebral schwannoma associated with Noonan syndrome have been described in the literature. We report the case of a 19-year-old male with a history of Noonan syndrome who presented to the emergency department with progressively developing paraplegia. A spinal MRI was performed, revealing a paravertebral mass with radiological characteristics suggestive of a schwannoma, which was confirmed on histopathological examination after surgical excision.

Keywords: Paravertebral mass; Shwanoma; Noonan syndrome; MRI

Introduction

Noonan syndrome is an autosomal genetic disorder [1]. It is characterized by multisystemic involvement with intellectual disability, distinctive facial features, and multiple cardiovascular, neurological, and lymphatic anomalies [2]. Rarely, these patients develop extramedullary neurofibromas [4]. No cases of paravertebral schwannoma associated with Noonan syndrome have been described in the literature. We report the case of a 19-year-old male with a history of Noonan syndrome who presented to the emergency department with progressively developing paraplegia secondary to paravertebral schwannoma confirmed on MRI and after surgical excision.

Case Report

We report a case of 19-year-old male with a history of Noonan syndrome and no specific prior medical conditions presents to the emergency department with progressively developing paraplegia. Clinical examination reveals flaccid paraplegia with the absence of osteotendinous reflexes. An urgent MRI has been performed, showing a paravertebral mass with hyperintense T2 signal, intense and heterogeneous enhancement after gadolinium injection, along with harmonious foraminal extension and widening (Figures 1 & 2).

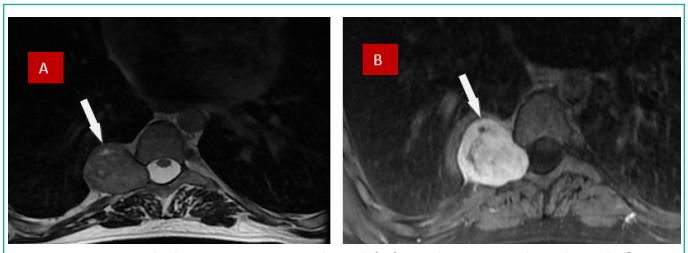

Figure 1: Axial slices in T2-weighted (A) and T1-weighted with fat saturation (B) of a spinal MRI before (A) and after the injection of the contrast agent (B).

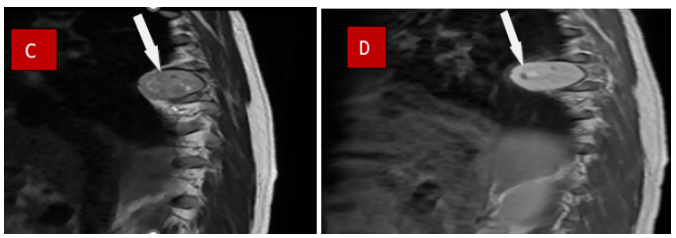

Figure 2: Sagittal slices in T2-weighted (C) and T1-weighted with fat saturation (B) of a spinal MRI before (A) and after the injection of the contrast agent (B).

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Discussion

Noonan syndrome is a multisystemic genetic disorder, autosomal dominant in nature, with an estimated prevalence of 1000-2500 cases, involving genes encoding proteins that play roles in the RAS-MAPK pathway [1]. It was first described in 1963 by Jacqueline Noonan [2] and is characterized by distinctive facial features, developmental delay, and a wide range of cardiovascular, lymphatic, and neurological impairments [3]. More rarely, neurogenic tumors such as neurofibromas have been identified in Noonan syndrome with multiple pigmented spots, formerly known as LEOPARD syndrome [4].

Schwannomas or spinal neurinomas account for nearly a third of primary spinal tumors [5]. These are benign tumors that develop from Schwann cells of the spinal roots, with a strong predilection for the lumbar [5] and cervical regions [6]. Extradural or mixed "hourglass" forms are relatively rare (15%), unlike intradural forms, which remain more frequent (70-75%) [5]. When they are located in multiple sites, neurofibromatosis type II should be systematically investigated [5]. No cases of spinal schwannoma associated with Noonan syndrome were described in the literature.

Clinically, they manifest as a slow spinal cord compression picture, with symptoms depending on the level of involvement, including a lesion-related syndrome (radicular pain, hypoesthesia, and even motor deficits), a below-lesion syndrome (motor, sensory, and sphincteric disturbances), and a spinal syndrome (spinal pain, abnormal posture) [7].

MRI is the diagnostic tool of choice. It helps to diagnose a schwannoma when there is a paravertebral mass with hyperintense T2 signal, intense and heterogeneous enhancement after gadolinium injection, along with harmonious foraminal extension and widening, without the "dural tail sign" frequently found in meningiomas [8].

The treatment of hourglass-shaped schwannomas is typically surgical with complete excision, sometimes requiring both anterior and posterior approaches. In rare cases with a risk of functional deficit, the supporting root may be preserved [7].

Conclusion

While neurological symptoms are rare in individuals with Noonan syndrome, neurofibroma is the most commonly reported neurological condition. It's worth noting that there have been no documented cases of paravertebral schwannomas associated with Noonan syndrome in the available literature.

Author Statements

Declaration of Interests

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

Patient Consent Statement

Written informed consent for publication was obtained from patient.

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