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Case Report

Imaging Utility in the Diagnosis of Dejerine-Sottas Disease: Case Report

Halfi Mohamed Ismail*, Zahi Hiba, Fikri Meriem, Jiddane Mohamed and Touarsa Firdaous

Neuroradiology Department, University Hospital of Ibn Sina, Mohamed V University, Rabat, Morocco

***Corresponding Author:** Halfi Mohamed Ismail, Neuroradiology Department, University Hospital of Ibn Sina, Mohammed V University, Rabat, Morocco.

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Abstract

Dejerine-Sottas disease (DSD) also called progressive hypertrophy interstitial neuropathy is a congenital polyneuropathy with results in sensory and motor symptoms. It affects the peripheral nerves of the extremities as well as cranial nerves in 15% of cases. The role of imaging isn't well documented in recent literature due to the important scarcity of this disease. We report the case of a woman with a confirmed diagnosis of DSD as well as the MRI's role in the process. Even though the imagining features aren't specific they may strongly direct the diagnosis into a smaller pool of neuromotor diseases.

Keywords: *Dejerine-Sottas Disease; Neuropathy; MRI; Imaging; Congenital; Motor; Sensory*

Abbreviations

DSD: Dejerine-Sottas Disease; MRI: Magnetic Resonance Imaging; STIR: Short-TI Inversion Recovery; CSF: Cerebrospinal Fluid

Introduction

Dejerine-Sottas disease is rare and had been classified under the umbrella of Charcot-Marie-Tooth neuropathies which are multi-faceted conditions with a vast constellation of clinical, and histologic findings. DSD damages the myelin sheath, therefore gradually causing distal sensory and motor leg impairment [1]. Genetic studies have revealed it autosomal recessive trait [2]. In this case report we will discuss imaging's role generally and MRI's input more specifically in the diagnosis of this neuropathology.

Case Report

A 32 years old female with no particular relevant history, suffering from the age of 11 of the heaviness of all limbs associated with trouble walking. Neurological examination revealed a spastic walk, paresthesis, and hyporeflexia with distal amyotrophic limbs. An electrophysiological study of the motricity and sensitivity of the nervous conduction of the upper extremities detected a significant increase in the latency of distal motricity in the ulnar and median nerve. Also, a decrease in the amplitude of the motor and sensitive action potential associated with a significant lengthening of the F-wave latency has been noticed. On the other hand, in the lower extremities, there was no trace of neurological muscle activity on the graphs all in favor of demyelinating neuropathy.

MRI showed a hypertrophic brachial (Figure 1) and lumbar nerve plexus (Figure 2) on T2 weighted and STIR sequences more accentuated on the left-hand side with noticeable amyotrophic glute muscles. A nerve biopsy performed at the ankle revealed several 'onion

bulbs' and enlargement of nerve roots in addition to a segmental loss of myelin all pathognomic pathological findings corroborating the diagnosis of DSD.

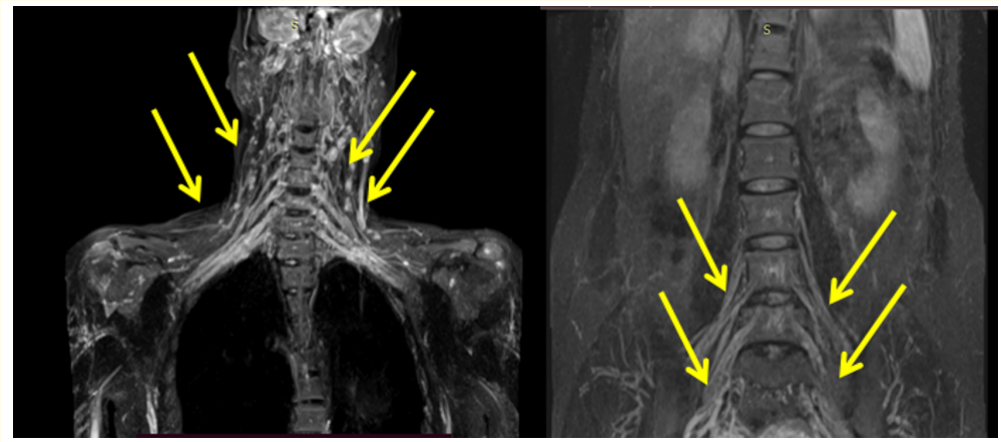

Figure 1: *3D SPACE STIR MRI sequences showing a clear hypertrophy of plexus brachial lumbar and sacral nerves more significant on the left at their emergence from their respective cervical foramina (Yellow arrows).*

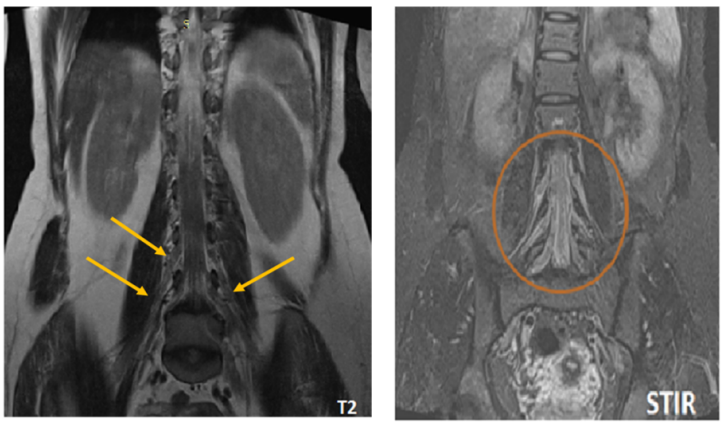

Figure 2: *Coronal T2 weighted and STIR MRI sequences of the same patient demonstrate the same nerve enlargement at the lumbar level without enhancement after gadolinium injection (Orange arrows and circle).*

Results and Discussion

First characterized in the 19th century, DSD was described as a recessive sickness affection sibling in the pediatric population [1]. It is a demyelinating neuropathy manifested by a mild constellation of neuro-motor impairments such as progressive weakness and atrophy of

the four extremities with a variant chronology of installation (mostly the upper limbs are affected first followed a few years after by their lower counterparts), causing the patient to suffer from trouble walking and standing up [2].

In addition to severe sensory changes causing both numbness and electrical-like pain [1].

Similar to our patient, the clinician should look for palpable peripheral firm nerve hypertrophy which is evident, and light-near dissociation, muscle atrophy, foot deformities, cranial nerve deficits, tremors, and general ataxia; an overlapping symptom across the different types of Charcot-Marie-Tooth diseases [2].

Terminologically, it wasn't until recently that the disease was called hereditary motor sensory neuropathy type 3 (HMSN type III) since type I and II correspond to Charcot-Marie-Tooth disease which both are segmental demyelination with few or no enlarged nerve roots, both consequences of dominant inheritance [2]. Further points of comparison are its prevalence and demography: the first type is the most common in children, the second much less but most likely occur in patients of the second decade, and the third, as we mentioned, is rare and starts in early life with progressive aggravation during following decades and can accompany several conditions such as diabetes, Refsum's disease, Guillain Barré syndrome and metachromatic leukodystrophy [3].

Genetic research incriminated multiple mutations in the myelin genesis genes, the periaxin gene, and the early growth response gene. However, even if the genes responsible for this syndrome have been thoroughly studied and identified, the genetic component in various cases of DSD remains a mystery [4]. In addition, molecular research identified a form of Dejerine-Sottas syndrome given the code name CMT-IV. It is revealed to have both components in play recessive and dominant genes, on top of the not fully elucidated physiopathology it is relevant to underline that additional molecular and genetic research is yet to be complete for a full understanding of this disease [5].

Radiologically, MRI is the Gold Standard because it shows triangle-shaped pedicles, pebbling of the vertebral bodies, and larger intervertebral foramina with thicker spinal nerve roots. Further MRI features consist of peripheral nerve hypertrophy and thickening which is graphically shown with aberrant foci that have a high T2 signal intensity thus signaling edema and damage to the myelin sheath [2]. Additionally, nerve root hypertrophy should be checked for in any patient with DSD who exhibits rapid deterioration, especially in the lower extremities, associated with a sensory level and loss of sphincter control. This is because it may cause compression of the conus medullaris and cauda squina, as reported in some cases. In these circumstances, quick spinal canal decompression surgery may be able to restore some of the resulting neurological impairments [6].

Imaging modalities in the diagnosis of DSD may extend to prenatal fetal ultrasound evaluating the nature of the posture, motor pathological tendencies, and effective motions. Occasionally ultrasound is of great assistance while confirming the beginning of a motion defect. It is also used in perinatal care and while delivering the fetus in a breech position when the hypotonia is known to not only avoid complications during vaginal birth but also improve neonatal monitoring of the impaired newborn [7].

Dejerine-Sottas disease shares a lot of similarities with many infectious/inflammatory syndromes. Guillain-Barre syndrome, sarcoidosis, and arachnoiditis, as well as certain congenital conditions, such as neurofibromatosis, can all induce thicker nerve roots and larger enhancing nerves. Moreover, differential diagnosis of DSD also includes neoplastic disorders, subarachnoid seeding, multiple schwannomas, and lymphomatous/leukemic infiltration can be easily ruled out with imaging, electrical nerve stimulation tests, nerve biopsy, and genetic studies [2] (Table 1).

As for its treatment, there is no cure only palliative approaches (to manage the pain and the disability) and interventional surgery in order to limit some complications such as decompression of the conus medulla via techniques such as laminectomy and duraplasty [6]. Although the genetic engineering technique CRISPR-Cas has considerable potential for identifying particular DNA/RNA sequences

Disease	Radiological features
Neurofibromatosis	• The cervical cord is where they are most commonly seen along the spinal nerve roof. This causes the neural exit foramen to enlarge, the pedicle to thin, and the posterior vertebral body to scallops. • T1 hypointense in MRI - T2: A thick center region of the collagenous stroma is assumed to be the cause of the hyperintense rim and core region with a low signal (referred to as the "target sign"). - Heterogeneous amplification in T1 C+ (Gd).
Guillain-Barre syndrome	• MRI: Substrate enlargement and contrast amplification on the cauda equina's nerve pots and conus medullaris. • The anterior nerve root is most frequently concerned.
Leukemic infiltration	• The optic nerve is the most frequent. • MRI: with perineural augmentation of the nerve and intraconal fat infiltration, there is widespread thickening and high signal intensity.

Arachnoiditis	• Associated with the lumbar region where cauda equina usually floats in CSF.
	• MRIz
	  • Type I: Many deformed and twisted nerve roots are seen.
	  • Type II: An empty thecal sac indication results from nerve roots clinging to the theca.
	  • Type IlI: The theca and nerve roots are connected by a unique soft tissue in the middle of the spinal canal.

Table 1: *Radiological features of the main mimickers of Dejerine-Sottas syndrome.*

and controlling gene expression. This encourages potential dynamic partnerships for the treatment of immuno-inflammatory diseases including DSD [8]. These techniques often require a multidisciplinary approach. By extending the results of the current studies into future ethically designed public health-focused studies, it will be possible to gain crucial insights into how to improve the quality of radiological imaging to go hand in hand with the diagnosis and treatment of these diseases [9]. Furthermore, diagnostic spinal magnetic resonance imaging for locating inflammatory tumor core prior to treatment/intervention(s) and non-invasive neuro-radiosurgery targeting of the inflammatory responses in tumors have been crucial in the field of neuro-immuno-oncology [10] but have not yet been implemented in the interventional approaches of DSD, hence we highly welcome more collaborations between the fields.

Conclusion

Though not unique to Dejerine-Sotta's illness, our radiological results may be quite suggestive in young children with congenital neuropathies. DSD should be diagnosed using imaging as well as the clinical presentation, followed by nerve conduction testing, and sural nerve biopsies.

Conflict of Interest

The authors declare that there is no conflict of interest in relation to this research paper.

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