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Case Report

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Demyelinization Lesions in a Patient with Type 1 Neurofibromatosis

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Introduction
Neurofibromatosis type 1 (NF1) is an autosomal dominant genetic disorder that predisposes individuals to the development of benign or malignant tumors of nervous tissue, as well as various non-tumor-related neurological conditions. The association between NF1 and multiple sclerosis (MS) is rare.

Observation
This is a 20-year-old female patient with type 1 neurofibromatosis of paternal origin, which manifested in childhood with cutaneous (café-au-lait spots) and ophthalmological (bilateral Lisch nodules) manifestations. She was hospitalized for right eye adduction palsy, resulting in divergent strabismus, which occurred postoperatively following right ileo-renal bypass surgery. Neurological examination revealed involvement of the third cranial nerve on the right. Brain MRI showed abnormalities in the supratentorial and infratentorial white matter, while spinal MRI revealed a 9 mm focal demyelinating lesion of the cervical spinal cord at the C6 level. The infectious, inflammatory, and immunological workup was negative, and cerebrospinal fluid (CSF) analysis showed normal cytobiochemistry without intrathecal synthesis. Measurement of the kappa light chain remains essential to confirm the diagnosis.

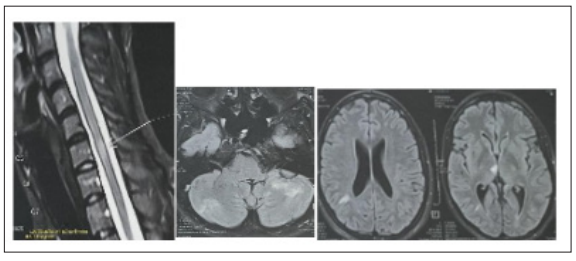

Figure 1: Cerebrospinal Cord Demyelinating Lesions

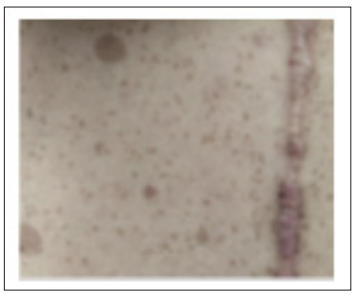

Figure 2: Coffee-Milk Stains

Discussion
The association between NF1 and MS remains exceptional. Whether this is a fortuitous coincidence or an underlying genetic link is still debated. Indeed, the mutation of the NF1 gene could influence immune regulation and increase susceptibility to demyelination processes.

Conclusion
A better understanding of the interactions between the NF1 gene and demyelination mechanisms could help to elucidate a common pathophysiology and improve the diagnostic and therapeutic management of these patients [1-5].

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