Tumefactive Demyelinating Lesions in a Young Patient: Rare Case with Good Outcome

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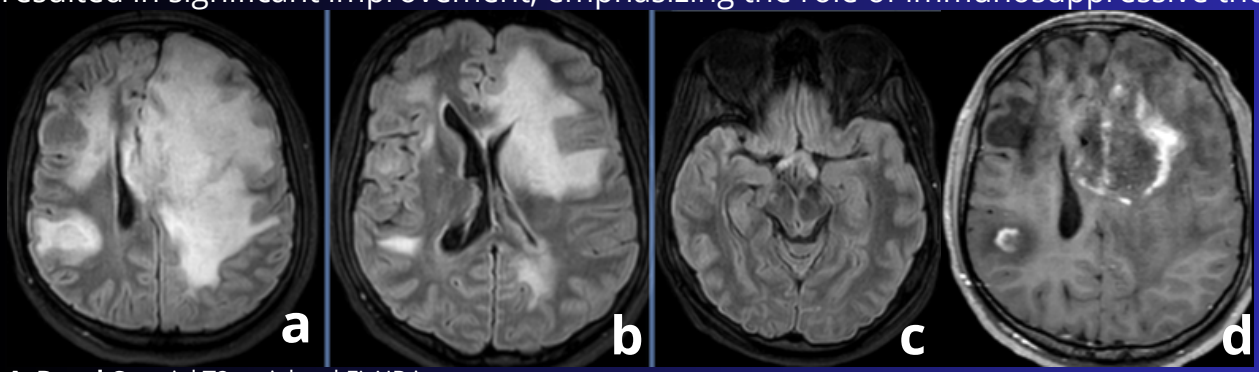
Brief Description of the Objectives: To report a rare case of tumefactive demyelinating lesions initially misdiagnosed as tumors in a young patient with neuropsychomotor developmental delay and parental consanguinity, emphasizing the differential diagnosis and treatment response.

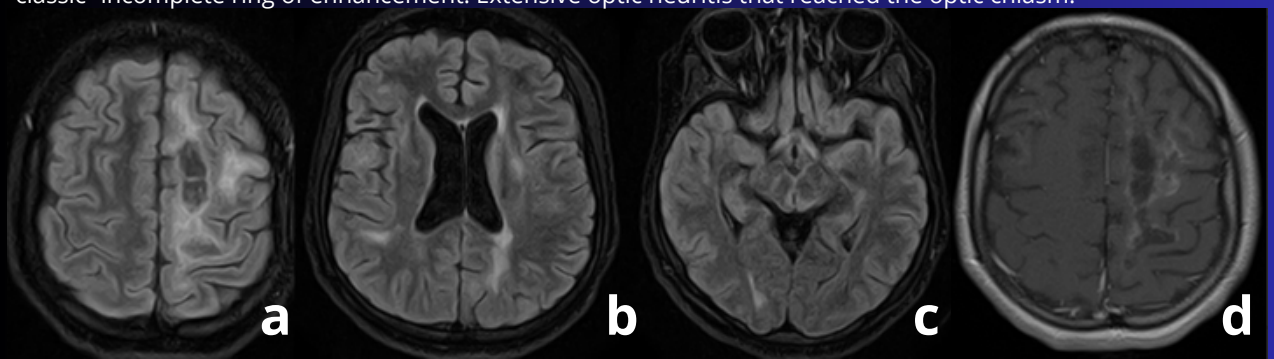
Clinical History: A 19-year-old female with neuropsychomotor developmental delay since age 4 and severe intellectual disability. Her consanguineous parents are cousins. In 2023, she developed apathy, hallucinations, a 15-kg weight loss, and progressive inability to walk. Admitted to the emergency department in January 2024 via critical priority ("vaga zero"), imaging revealed multiple tumefactive brain lesions, extensive demyelination in the brainstem and spinal cord, absence of longitudinally extensive transverse myelitis (LETM), and bilateral optic neuropathy. Treatment included corticosteroid pulse therapy, plasma exchange, and cyclophosphamide, resulting in significant improvement. Neurology and genetics follow-up was scheduled post-discharge.

Discussion and Diagnosis: Initially suspected as brain neoplasia, further investigation ruled out infections and autoimmune diseases, leading to a diagnosis of tumefactive demyelinating lesions. MRI findings were not consistent with leukodystrophy. Parental consanguinity prompted involvement of the Genetics department. The patient did not attend follow-up appointments.

Conclusions: This case highlights the need to consider tumefactive demyelinating lesions as a differential diagnosis in young patients with progressive neurological symptoms. Parental consanguinity and neurodevelopmental delay suggest possible genetic factors. Early, multidisciplinary treatment resulted in significant improvement, emphasizing the role of immunosuppressive therapy.



A, B and C - axial T2-weighted FLAIR images
D - axial T1 weighted post gadolinium demonstratingbilateral tumefactive demyelinating lesions with the "classic" incomplete ring of enhancement. Extensive optic neuritis that reached the optic chiasm.



After three weeks a new MRI was done:
A, B and C - axial T2-weighted FLAIR images
D - axial T1 weighted post gadolinium demonstrating regression of the bilateral tumefactive demyelinating lesions. Shrinkage of brain parenchima and areas of vacuolization (imaged). Extensive optic neuritis that reached the optic chiasm.