

TUMEFACTIVE DEMYELINATING LESIONS IN A YOUNG PATIENT: RARE CASE WITH GOOD OUTCOME

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INTRODUCTION:

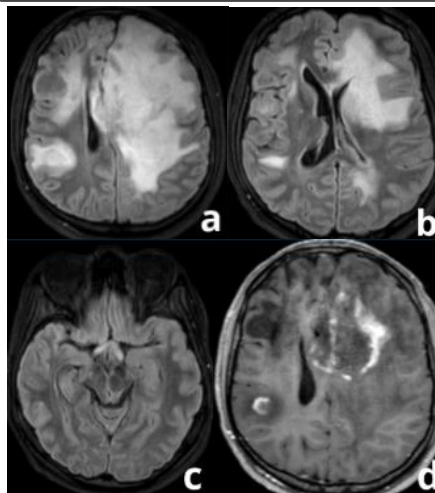
Tumefactive demyelinating lesions are rare inflammatory disorders of the central nervous system that may mimic brain tumors due to their clinical and radiological characteristics. Their diagnosis can be challenging, particularly in young patients with pre-existing neurodevelopmental impairment, requiring careful exclusion of neoplastic, infectious, autoimmune, and genetic conditions. Early recognition is essential, as these lesions may respond significantly to immunosuppressive treatment.

CASE DESCRIPTION:

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.

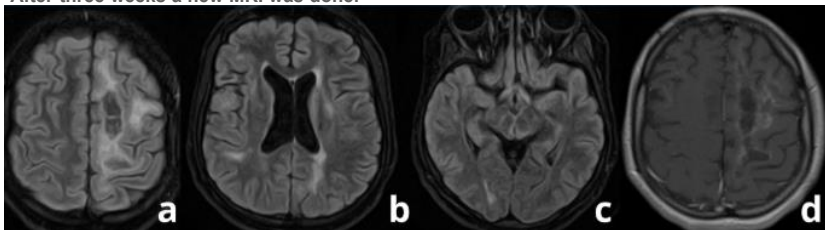
DISCUSSION AND CONCLUSIONS:

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. 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 demonstrating bilateral 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 parenchyma and areas of vacuolization (imaged). Extensive optic neuritis that reached the optic chiasm.