A CASE OF REFRACTORY SEIZURES IN JUVENILE CANAVAN DISEASE.

Introduction: Canavan disease is a rare, progressive neurological disorder classified as a leukodystrophy, caused by mutations in the ASPA gene on chromosome 17, resulting in a deficiency of the enzyme aspartoacylase. This leads to the accumulation of N-acetyl aspartic acid (NAA) in the brain. There are two forms of the disease: a severe infantile/neonatal form and a milder juvenile form, both presenting symptoms such as hypotonia, intellectual disability, feeding difficulties, paralysis, and seizures, with severity varying between forms. Diagnosis is achieved by detecting increased NAA levels in blood, urine, and cerebrospinal fluid, and MRI shows characteristic bilateral white matter changes. Prognosis is poor for infantile form, while juvenile form patients have better outcomes .

Case Report :A 22-year-old male presented with a history of seizures, experiencing nine episodes over three days, each lasting 1-2 minutes and involving all four limbs, mouth frothing, upward eye rolling, and involuntary urination. He had a normal early childhood but began having seizures at age 10, followed by loss of motor skills and speech difficulties. He experienced seizures approximately every two months, especially when off anti-epileptic medications. His parents were related (second-degree consanguinity), but there's no family history of seizures or intellectual disability. The patient has moderate mental retardation, cannot walk, exhibits spasticity and exaggerated deep tendon reflexes, has a positive bilateral Babinski reflex, and myoclonus in the right lower limb, while the sensory system remains intact. MRI findings showed diffuse white matter leukodystrophy, and MR spectroscopy indicated increased N-acetyl aspartic acid (NAA) peaks.

CONCLUSION

Any young male presenting with refractory seizures should be evaluated for genetic disorders.