World Academy of Science, Engineering and Technology
International Journal of Medical and Health Sciences
Vol:19, No:05, 2025

Metachromatic Leukodystrophy: A Case Report

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Abstract : Metachromatic leukodystrophy (MLD) is a rare lysosomal storage disorder with an autosomal recessive inheritance pattern. Lysosomal storage disorders are often severe, follow a progressively neurodegenerative path, and may result in multi-organ failure, potentially leading to death within 5 to 6 years in cases of early-onset forms. There are limited data regarding cases of MLD in Filipino children. This is the case of a 2-year-old Filipino girl who presented with progressive neurological deterioration and was diagnosed with metachromatic leukodystrophy by molecular genetic testing. This case report aims to present this patient's clinical history, neurological findings, diagnosis and novel genetic mutations causing MLD. A concise review of updated literature on MLD will be discussed.

Keywords : metachromatic leukodystrophy, ARSA gene, peripheral neuropathy, case report, demyelinating disease
Conference Title : ICNE 2025 : International Conference on Neurology and Epidemiology
Conference Location : Amsterdam, Netherlands
Conference Dates : May 13-14, 2025