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Metachromatic Leukodystrophy: A Case Report

Authors : Mary Rose Eunice S. Gundayao, Manolo M. Fernandez

Abstract : Metachromatic leukodystrophy (MLD) is a rare autosomal recessive lysosomal storage disease that causes progressive demyelination of the central and peripheral nervous system. Lysosomal storage disorders tend to be serious, have a progressive neurodegenerative course, and can cause multiorgan failure and, ultimately, death within 5-6 years (early-onset form). 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
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