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Case Report on Late Infantile Type Metachromatic Leukodystrophy

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Abstract:
Background: Metachromatic leukodystrophy is a genetic disorder characterized by the accumulation of fatty substances (lipids) in cells, particularly in the brain, spinal cord, and peripheral nerves. A lack of sulfatides, which are enzymes that aid in the breakdown of lipids, causes this buildup. The brain and nervous system gradually lose function as the substance that covers and protects nerve cells (myelin) is damaged.
Case presentation: This case involves a male child of 2.5 years with a known case of Metachromatic leukodystrophy. He was brought by his parents to the tertiary care hospital with the complaint of Inability to walk for 6-month, Inability to talk for 6-month, neurological regression, unable to roll sit and stand. As narrated by his parent's child had a history of delayed development, the patient also lost his language milestones along with walking. Computed tomography and magnetic resonance imaging of the brain was done. And later he was treated with multivitamins and muscle relaxants and continued with physiotherapy.
Conclusion: A rare condition of myelin metabolism is known as metachromatic leukodystrophy. The myelin sheath of the central and peripheral nerve systems accumulates cerebroside sulfatide as a result of aryl sulfatase-A enzyme deficiency, which causes this degenerative condition.

Keywords: Metachromatic leukodystrophy, Arylsulfatase, Periventricular

INTRODUCTION:

A rare condition of lysosomal sphingolipid storage known as metachromatic leukodystrophy (MLD) results from arylsulfatase A deficiency (ASA).(1) Incidence estimates range from one in 40 000 to 160 000 people globally. (2)The effect of this insufficiency is an accumulation of sulfatides in the gallbladder and other organs, including the central nervous system.(3)To rule out other clinical problems and to approximate a diagnosis that will later be confirmed by the proper molecular studies, magnetic resonance plays a significant role in the characterization of underlying abnormalities. (4)

CASE PRESENTATION:

This case involves a male child of 2.5 years with a known case of Metachromatic leukodystrophy. He was brought by his parents to the tertiary care hospital with the complaint of Inability to walk for6-month, Inability to talk for6-month,neurological regression,unable to roll sit and stand. He was born through normal vaginal delivery. He had a breech presentation, delayed crying after birth, and had a regression in milestones.

As narrated by the parents' child was the apparently asymptomatic 6 months back. Gradually, the child who was able to walk with support slowly was unable to do so. The patient also lost his language milestones. He had a history of delayed development. He had Head control on 6month, rolling over 6 months, crawling on 7-8month, and supported walkingfora 1year, Independent walkingis not achieved yet.The child is unable to Roll sit and stand unable to speak.

On physical examination, the child is not able to perform daily activities or impaired mobility is observed. Loss of intellectual, cognitive, and memory abilities. Motor skills such as walking, moving, speaking, and swallowing are lost. Muscle stiffness and rigiditywere found with increased tone in all four limbs.

On radiological examination, MRI shows meta-chromaticleukodystrophyof periventricular white matter. The brain's CT revealed bilateral periventricular and deep cerebral white matter hypodensities, which are indicative of neurodegenerative disorders, as well as hypodensities in the corpus callosum.

The patient is treated with syrup multivitamin 2.5 ml orally, tab. Baclofen 10mg orally. He had given daily physiotherapy where he received bilateral movement of the right and left upper and lower extremities with, a rolling facilitator.Levator stretching.

DISCUSSION:

Metachromatic leukodystrophy can be categorized as late-infantile (onset before 3 years of age), juvenile (onset before 16 years of age), or adult (onset after 16 years of age).(2)Usually, between the ages of 14 months and 2 years, the IMLD appears as flaccid weakness or an ataxic walk with absent tendon reflexes. The motor and mental regression advance quickly.(5-15)

Zafeiriou et al suggested, that the primary clinical features of the late infantile MLD form are gait disturbance with weakness or ataxia, developmental delay, loss of speech, optic atrophy, and progressive spastic tetraplegia, and the age of start is often between 1 and 2 years. When the early clinical symptoms of late-infantile MLD start to manifest, the white matter changes detectable by MRI can already be present.(16-23)
D.F. van Rappard et al. suggested, that for all MLD clients, currently there is no curative treatment available. Gene therapy, enzyme replacement treatment, and hematopoietic stem cell transplantation (HSCT) have all been thoroughly investigated in mice models. Clinical trials to test the effectiveness of HSCT, gene therapy, and enzyme treatment have been initiated as a result of the promising outcomes from several animal studies.(24-28)

CONCLUSION:

A rare condition of myelin metabolism is known as metachromatic leukodystrophy. The myelin sheath of the central and peripheral nerve systems accumulates cerebroside sulfatide as a result of aryl sulfatase A enzyme deficiency, which causes this degenerative condition. In MLD, a severe storage illness, the central and peripheral nervous systems accumulate sulfatides due to a lack of the lysosomal enzyme arylsulfatase A. Depending on when the disease manifests, there are three types of onsets: late-infantile, juvenile, and adult. Clinical signs of brain damage, such as slurred speech, hearing, vision, and gait, develop progressively over time until becoming deadly.

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