A RARE CASE REPORT OF METACHROMATIC LEUCODYSTROPHY

INTRODUCTION:
Leukodystrophy is defined as a group of genetic disorders which are characterized by the imperfect growth/development of the myelin sheath that covers nerve fibres in the brain. Metachromatic leukodystrophy (MLD), also called Arylsulfatase A deficiency, is a lysosomal storage disease which is commonly listed in the family of leukodystrophies. It is caused by the lack of an important enzyme called Arylsulfatase-A which leads to breakdown of fatty substances (lipids) and causes lipids to build up in the brain, spinal cord and peripheral nerves[1]. They are based on when the symptoms begin:
1. Late infantile MLD symptoms usually begin by ages 1 - 2 years.
2. Juvenile MLD symptoms usually begin between ages 3 - 10 years.
3. Adult (Late-stage juvenile MLD) symptoms may occur over age 16 years.

CASE DISCUSSION:
Here we present a case of 2 years old female child presenting with complaints of deafness and unable to speak since birth. Patient was a term baby with immediate cry preset at the time of birth. No history of perinatal asphyxia, neonatal seizures and neonatal jaundice. No maternal history of fever, rash in the antenatal period, which could be suggestive of toxoplasmosis, rubella, cytomegalovirus, and herpes simplex (TORCH) infection. No family history of similar complaints.

Her vaccination is adequate up to her age. Her motor milestones were normal for her age.

On MRI, multiple confluent T2/FLAIR hyperintensity was noted typically involving the bilateral periventricular white matter symmetrically with sparing of sub-cortical U fibres.

Metachromatic leukodystrophy (MLD), a rare neurodegenerative metabolic disorder occurs with incidence of 1 in 40,000 to 1,60,000 individuals, worldwide[2].

Metachromatic leucodystrophy (MLD) occurs due to cerebrosidesulfatide accumulation in the extraneural and neural tissues. Arylsulfatase A (ARSA) enzyme deficiency leads to progressive focal or generalized demyelination.

CONCLUSION:
MLD is a severe disease that gets worse over time. Eventually patient loses all muscle and mental function. Life span varies depending on what age the condition started, but the disease course usually runs 3-20 years. Thus, prompt diagnosis is required for improving the quality of life for the patient. The important diagnostic modalities used to confirm this degenerative disorder are arylsulfatase-A enzyme activity, molecular genetic testing of arylsulfatase A, estimation of urinary sulfate and detecting metachromatic lipid depositsin the nervous tissue [3]. Gene sequence analysis of arylsulfatase A is an important tool for prenatal diagnosis. As MLD progresses with age and the neurodegeneration worsens with time, there is no definitive treatment tilldate.

Newer treatment modalities include stem cell transplantation, bone marrow transplantation along with genetic engineering and these might halt the progression of neurologic dysfunction [4]. Recombinant human ARSA administration, an experimental treatment can be a promising option in future, although it lacks universal recommendation and adaptation.
MR Spectroscopy at TE 33 reveals decreased Choline : Creatinine ratio with increased NAA/Creatinine ratio.

**Treatment**—prophylactic antiepileptics with symptomatic treatment with counselling. In individuals with late symptomatic and juvenile forms of disease bone marrow or cord blood transfusion may stabilise neurocognitive functions[5].

**REFERENCES:**