An overview of Leukodystrophy (LD)


Leukodystrophy (LD) means deterioration of white matter of the brain function. This deterioration manifests itself by regression of previously acquired skills which in majority of cases leads to devastating neurological function. Leukodystrophy (LD) are genetic diseases causing degeneration of myelin sheaths in the CNS and occasionally associated with peripheral nerves involvement. In this types of neurologic diseases, basic defect is directly related to the synthesis of myelin membranes and its maintenance. Most of the time clinical manifestation of Leukodystrophy (LD) appear during childhood or adolescence and almost all the times leads to progressive course, untreatable and premature death. Early diagnosis helps supportive and experimental therapies, family counseling and family members screening to detect unaffected individuals who currently are nonsymptomatic. The few treatments which have been proposed are effective in the early stages. Making definite diagnosis of leukodystrophy (LD) requires good knowledge of clinical features and having neuroimaging data. Being aware of typical age of onset of different clinical types and brain MRI patterns of leukodystrophies (LD) are essentials tools to the correct diagnosis. Precise diagnosis may need specific laboratory tests and other investigations to make leukodystrophy (LD) and undoutable entity.

Clinical Features

Age of onset
The main symptoms of LD are neurological which mostly appear in children who previously enjoyed a good health. Progressive deterioration of motor skills and mental symptom in a child every or an adolescent, should arouse a high index of suspicion towards a leukodystrophy (LD).

Physical features
Except some of the patients who are macrocephalic, others do not show any physical stigma. Few of them have dysmorphic features, skeletal anomalies and dental abnormalities.

Neurologic Symptoms
Neurologic Symptoms of LD. Consist of increasing motor signs and symptoms and change of cognition and language. MRI of the head, nerve conduction studies, Evoked potentials, evaluation of other organs function likes: eye, heart and G.I are those investigating tools which help clear cut diagnosis of LD.

Keywords: Leukodystrophy; Early diagnosis; Neurologic symptoms
References


