Adrenoleukodystrophy: A Rare Case

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Adrenoleukodystrophy (ALD) is an X-linked neurodegenerative disorder which is characterized by progressive demyelination of nervous system and primary adrenal cortical insufficiency (Addison’s disease). This peroxisomal disorder is caused by the defect of ABCD1 gene on q28 segment of X-chromosome (Xq28). It has incidence of 1:20,000 males. X-ALD leads to the impairment of peroxisomal β-oxidation and biochemical signature is the accumulation of saturated, unbranched, very long chain fatty acids (VLCFA) especially hexacosanoic acid (26:0) and tetracosanoic acid (24:0) because of deficiency of acyl CoA synthetase. Inability of ALD patients to catabolize these fatty acids results in formation of membrane lipids and lipidated proteins. Neuronal myelin sheath is destabilized by these abnormal molecules, as a result inflammatory response occur which leads to demyelination. Seven different presenting phenotypes have been reported. Two most common are Adult onset adrenomyeloneuropathy (AMN) and progressive neuro-inflammatory Childhood Cerebral ALD. Others include adolescent cerebral ALD, adult cerebral ALD, Addison’s disease only, asymptomatic and heterozygous women. Childhood cerebral adrenoleukodystrophy was described by Siemerling and Creutzfeldt in 1923. The onset of neurological symptoms heralds the rapid progression of the disease and this leads to vegetative state in 2 years.

Case Report

A Seven years old boy resident of Rawalpindi was admitted in Holy Family Hospital with complaint of uncontrolled fits for last 15 days. One year ago, he started having complaint of progressive weakness of left side of body, walking difficulty and generalized pigmentation of skin. For last 4 months, child is bedridden, not walking, not responding and not moving left side of body.

Physical examination: Height=110.5cm, Weight=25kg. Blood pressure was within normal range. His Glasgow Coma Scale was 5/15. There was generalized pigmentation of skin and there was sensory-neural hearing loss in left ear (confirmed by audiometry). Muscle tone was increased in both upper and lower limb of left side. Left sided Deep tendon reflexes were brisk and planters were upgoing. Pupils were round and reacting to light. Fundoscopic examination was normal.

Discussion

In the cerebral form of X-ALD, the first neurological manifestations most commonly occur at the age of 4-8 years. Neurological manifestations include impaired auditory discrimination, visual disturbances, poor coordination, spatial disorientation, behavioral disorder such as abnormal withdrawl or aggression, poor memory and school performance. Progression of disease leads to vegetative state in 2 years and death.
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Treatment of ALD is symptomatic. Steroids are used for adrenal insufficiency while psychotropics for psychiatric symptoms. Lorenzo’s oil which is a mixture of oleic acid and erucic acid, when administered before the symptoms onset can delay the appearance of childhood cerebral form. Statins can reduce VLCFA level. Switching to a diet that contains low levels of VLCFAs is also beneficial. Long term benefit can be provided to boys with early evidence of childhood cerebral form of X-ALD by bone marrow transplantation but is not done in those symptoms are severe or who have adult onset or neonatal forms. Genetic counselling of family members may be advisable. An amniocentesis or chorionic villus sampling can be done during pregnancy to determine if unborn child is affected.

References

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