IS THE THIRD TIME REALLY A CHARM?

The story about three brothers suffering from adrenoleukodystrophy and about HSCT being a chance to stop the unstoppadle disease.

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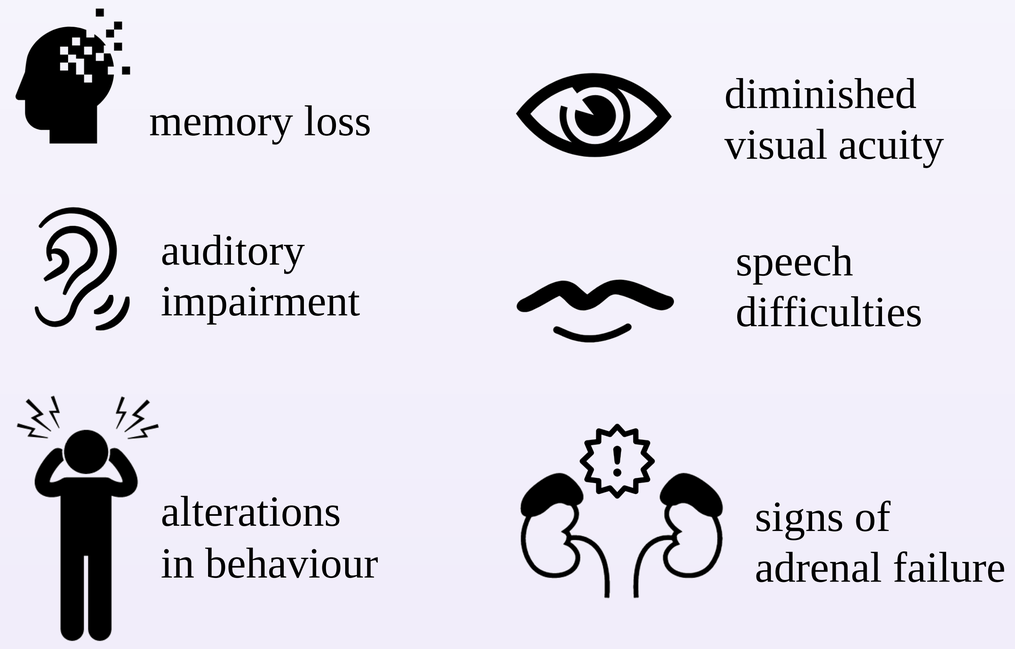
INTRODUCTION

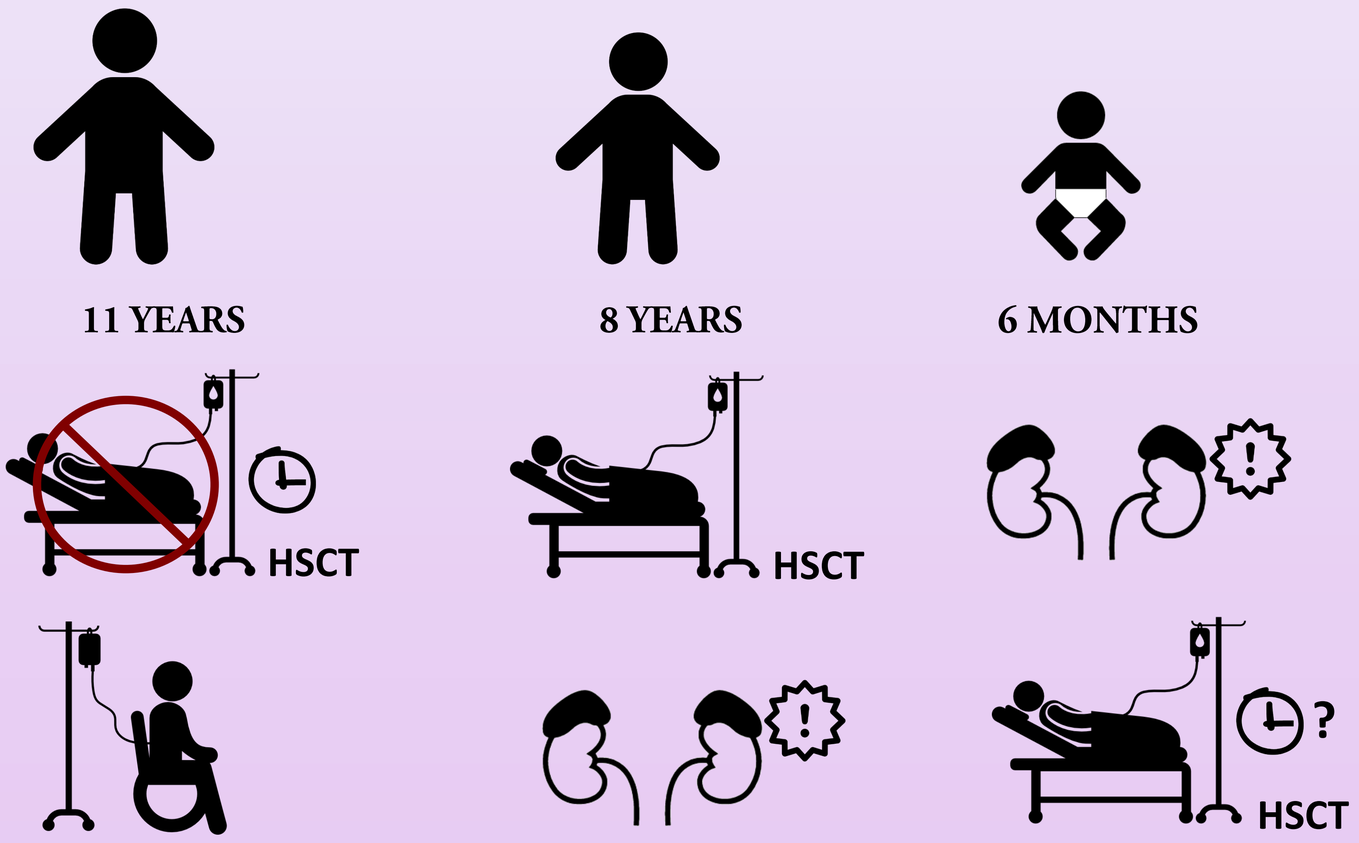
Adrenoleukodystrophy (ALD) is a genetic diseases classified in the group of peroxisomal disorders caused by mutations in ABCD1, gene located on the X chromosome. It is the most common monogenetically inherited neurodegenerative diseases. X-ALD is an inborn error of metabolism characterized by impaired peroxisomal beta-oxidation of very long-chain fatty acids (VLCFA) with a heterogeneous clinical spectrum. VLCFA accumulate principally in the CNS and adrenal glands. It results in a beakdown of the myelin sheath and axons and slowly progressive axonopathy affecting sensory ascending and motor descending spinal cord tracts. No causal treatment for ALD is known, although hematopoietic stem cell transplantation (HSCT) is allowed for early diagnosis. Lorenzo's oil is used in the treatment of asymptomatic patients.

SYMPTOMS OF ALD



CASE REPORT

The study presents a case of 3 boys (siblings) suffering from X-ALD, aged: 6months, 8 and 11 years old, with confirmed mutation in ABCD1 gene. In boys' mother blood, mutation has also been confirmed. The oldest one is currently in poor condition. Due to late diagnosis, he was not qualified for HSCT. His brother, 8 years old is 1.5 year after HSCT. He develops properly with only symptoms of adrenal insufficiency. 6month-old boy has now started to develop adrenal insufficiency without any symptoms from CNS. HSCT is planned for him in 2-3 years. All boys take hydrocortisone and Lorenzo's oil.



CONCLUSIONS

ALD is associated with the severe morbidity and mortality and the symptoms are unspecific. It occurs mostly among young boys and progresses rapidly.

In early stages HSCT gives the best chances of slowing down the progression of disease and allows to alleviate the subsequent consequences of the disease.

Therefore, parents' awareness and early genetic testing (including prenatal testing) are very significant for the progress of ALD.