Joshua Hellmann Foundation

Newborn Metabolic Screening Program

CUHK-BCM Joint Centre for Medical Genetics

The Chinese University of Hong Kong

Information for Parents

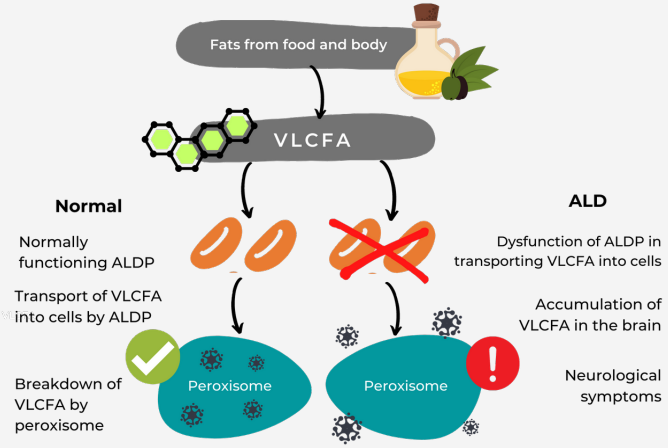
Screening for Adrenoleukodystrophy (ALD)

Enquiries:
(852) 5569 6412 (office hours from 9:00-17:00)
(852) 3505 4219 (voicemail service during non-office hours)
Website: http://www.obg.cuhk.edu.hk/fetal-medicine/fetal-medicine_services/iem/

If you wish to join this screening program, please contact your obstetrician during antenatal period or contact your paediatrician within 7 days after delivery.

What is adrenoleukodystrophy?

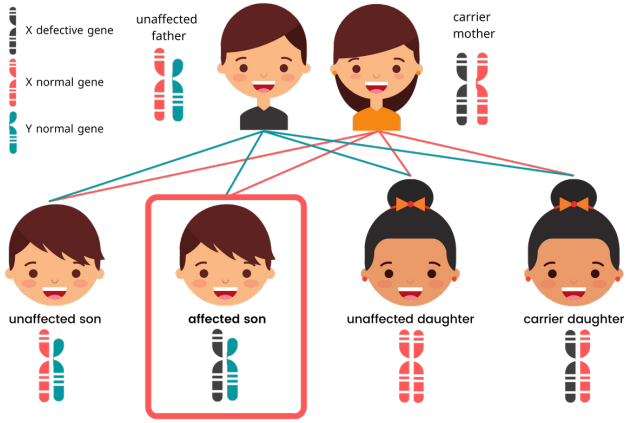
Adrenoleukodystrophy (ALD) is caused by mutation in the ABCD1 gene on the X chromosome, leading to dysfunction of the adrenoleukodystrophy protein (ALDP) which transports long-chains of fatty acids (VLCFA) to peroxisomes in the cell for proper metabolism. The accumulation of excess VLCFA in the brain invades the protective myelin around nerve cells, leading to degeneration of the central nervous system. The adrenal cortex (small glands on each kidney) may also be affected.



What are the symptoms of ALD?

The symptoms of ALD vary among patients and may begin at different ages. The onset of the most severe type, cerebral ALD, usually occurs between the ages of 4 and 8, with initial symptoms of learning or behavioral abnormalities, such as deterioration in school performance, dyslexia, orientation disorder, visual and hearing impairment, epilepsy, etc. The disease usually deteriorates rapidly within 6 months to 2 years after its onset, causing the patient to lose motor ability or even early death. Patients with a milder form of ALD, adrenal spinal neuropathy (AMN), develop symptoms between the age of 20 and middle age. Other types of ALD include Addison Disease, adrenal insufficiency, and female symptomatic heterozygote.

How is ALD inherited?



Adrenoleukodystrophy (ALD) is an X-linked recessive inherited disease. A male carrying such mutated gene is affected as he carries only one X chromosome. A female carrier of the mutation in one of her X chromosomes will not have symptoms as it is dominated by the other normal X chromosome. Among the offspring of an asymptomatic female carrier, boys have a 50% chance of developing ALD whilst girls have a 50% chance of becoming a carrier; among the offspring of a male patient, all girls are asymptomatic carrier. However, some female carriers may have neurological symptoms in adulthood.

ALD screening and its aim

The ALD screening aims to detect babies at risk of severe ALD by measuring the concentration of VLCFA such that follow up and treatment can be given before severe brain and nerve damage or disability. Patients with milder signs of ALD are not the target of the current programme. A few drops of blood are collected onto a card by pricking the baby's heel in its first 24 hours to 7 days of life. All babies with a positive screening result will have to undergo further investigation to confirm whether they are affected by ALD or not.

Outcomes of ALD patients

For ALD patients who have not developed symptoms, their outcomes can be improved by early treatment including steroid replacement therapy and bone marrow transplant.