INFORMATION FOR PARENTS/GUARDIANS:
My Baby Has a Positive X-Linked Adrenoleukodystrophy (XALD) Newborn Screening Result

What is newborn screening?
These are routine tests done shortly after birth on every baby born in Ontario. A small sample of blood is taken from the heel of your baby and is tested for rare, treatable diseases, including XALD.

What does it mean if my baby has a positive XALD newborn screening result?
This result does **not** mean that your baby has XALD. It means that more testing is needed because your baby **might** have XALD. Babies with XALD are healthier if treatment begins early, so it is important to have follow-up testing done to find out if your baby is at risk of developing XALD.

You may feel worried about your baby's screen positive result. Many parents in this situation feel this way. Remember, we do not know for sure that your baby has XALD until follow up testing has been done.

What is XALD?
XALD is a rare inherited (genetic) disease that can cause problems with the function of the adrenal glands (adrenal insufficiency) and changes in the white matter of the central nervous system (leukodystrophy). Without treatment, children who develop the childhood cerebral form of XALD (CCALD) will begin to have behavioral changes and loss of skills. These symptoms worsen over time, eventually leading to childhood death. Treatment is available. Children with XALD can benefit from being identified early, being monitored for signs/symptoms and getting treatment sooner when needed to help prevent health problems.

XALD affects about 1 in every 15,000 male births in Ontario.

What causes XALD?
XALD is an inherited (genetic) disease. XALD happens when there is a genetic change in the *ABCD1* gene. The job of the *ABCD1* gene is to help the body break down very long-chain fatty acids (VLCFAs). The genetic change causes the *ABCD1* gene to not work properly, resulting in the build-up of VLCFAs in the brain, nervous system and adrenal glands.

The *ABCD1* gene is located on the X-chromosome. Individuals with one X chromosome (typically males) who have a genetic change in their *ABCD1* gene will be at risk of developing XALD. Individuals with two X chromosomes (typically females) are usually not at risk of developing XALD as they have two copies of the *ABCD1* gene, and it is unlikely both copies will have a genetic change.

How do I find out if my baby has XALD?
Blood tests are done to find out if a baby who screened positive has XALD.

When can my baby have these tests?
Your baby's doctor or a health care professional at a Newborn Screening Treatment Centre will call you to talk about the results of your baby's positive newborn screen and arrange for a follow up assessment and blood tests. It can take a few weeks to find out if a baby truly has XALD or not. This waiting period can be hard for families.

Why screen for XALD?
Babies who have XALD look normal when they are born but are at risk of developing problems with their adrenal glands (adrenal insufficiency) and changes in the white matter of the central nervous system (leukodystrophy). Babies identified at a young age through screening can be monitored for symptoms and treated early to help prevent health problems.

How is XALD treated?
Babies with XALD are monitored closely by a team of health care specialists. If tests show signs of the disease, treatment will be started. The goal is to start treatment before symptoms develop. The treatment depends upon what the tests show, such as taking medication if problems with the adrenal glands develop.

Where can I get more information?
For more information on newborn screening, please talk to your local health care provider or visit the Parents section of our website at www.newbornscreening.on.ca

For more information on XALD, please visit the ALD Connect website at https://aldconnect.org/

NOTE TO PARENTS/GUARDIANS: This information is only for parents whose baby has had a positive newborn screening result for X-Linked Adrenoleukodystrophy. Please remember that this fact sheet was written for information purposes only. The fact sheet should not replace professional medical advice, diagnosis or treatment.