VDH VIRGINIA DEPARTMENT OF HEALTH

Division of Child and Family Health- Newborn Screening Follow-Up Program

# Adrenoleukodystrophy (ALD)- Females

*Parent* Fact Sheet

A newborn screening test is a **screen** and not diagnostic testing. An "abnormal" or "critical" result on a newborn screen indicates the baby may be at a higher risk of having a disorder; however, it does not diagnose the baby with the condition. Follow-up testing is **vital** to determine if the baby has the disorder indicated. In the event the condition is diagnosed, timely follow-up testing will result in earlier treatment and better outcomes.

**Disorder Indicated:** Adrenoleukodystrophy (ALD) occurs when the body is unable to break down certain fats (very long chain fatty acids, VLCFA). The VLCFAs build up and affect normal bodily functions, largely the nervous system and adrenal glands. When an individual has ALD, the buildup of VLCFAs may disrupt the fatty covering (myelin) of the nerve cells in the brain and spinal cord causing the myelin to breakdown, which reduces the ability of the nerves to relay information to the brain and disrupt some bodily functions. X-linked ALD is caused by a mutation of the ABCD1 gene. Although X-ALD is most often inherited through females, there can be novel variants that appear for the first time in an affected individual and were not inherited.

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| Incidence | 1 in every 17,000 newborns. |
| Signs and Symptoms<br><br>*Please note:*<br>*Most newborns are asymptomatic* | **Females** with elevated VLCFA and that have a mutation of the *ABCD1* gene are carriers of ALD. Carriers tend to not develop symptoms during childhood but may develop symptoms similar to Adrenomyeloneuropathy (AMN) as an adult. Confirmatory testing is vital to rule out a peroxisomal disorder and confirm ALD carrier status. If the female is found to be a carrier of ALD, extended familial genetic testing is recommended to identify at risk males.<br><br>Symptoms of Adrenomyeloneuropathy (AMN):<br>• Usually appear later in life (30-60 years)<br>• Urinary and genital tract disorders<br>• Progressive stiffness and weakness in the legs (paraparesis) |
| Next Steps ***may*** include: | **Follow up with your child's pediatric provider**<br>Clinical assessment<br>Laboratory Testing |
| Treatment ***(if indicated)*** | Follow up with your child's pediatric provider and Genetic Specialist |
| Additional Resources | VDH Newborn Screening *http://vdhlivewell.com/newbornscreening*<br>Baby's First Test *www.babysfirsttest.org*<br>American College of Medical Genetics (ACMG) ACT Sheets *www.ACMG.net*<br>Genetics Home Reference *https://ghr.nlm.nih.gov/*<br>ALD Connect *https://aldconnect.org/* |

*Educational content adapted from www.babysfirsttest.org*

109 Governor St. 9th Floor, Richmond VA 23219
Phone: 804-864-7711 Fax: 804-864-7807