Nebraska Newborn Screening Program
Physician ACT sheet for REPEAT INCONCLUSIVE X-LINKED ADRENOLEUKODYSTROPHY (X-ALD) Result

You Should Do The Following:

☐ Consult with metabolic specialist on call to determine follow up:
____ Dr. C. Baker – Pgr 402-888-0404
____ Dr. W. Rizzo – Pgr 402-888-0742

☐ Notify baby's family of newborn screening results. Assess status of newborn and provide follow up information as discussed with the metabolic specialist.

☐ Arrange for follow up as specified by the metabolic specialist

Screening Test Results

Screening for X-ALD begins with measurement of the very long chain fatty acid lysophosphatidylcholine (C26:0). If the C26:0 remains elevated on the requested repeat newborn screen the lab reflexes to sequencing of the ABCD1 gene. This gene is the causative gene of X-ALD.

Condition Information

X-ALD is a peroxisomal disorder, which is inherited on the X chromosome. Therefore males with an X-ALD causing mutation of the gene are affected. Females with the same X-ALD causing mutations are considered carriers. X-ALD is caused by a deficiency of adrenoleukodystrophy protein (ALDP), which transports very long chain fatty acids (VLCFAs) into peroxisomes. As a result of this deficiency, VLCFAs accumulate in the body. High levels of VLCFA may be toxic to the adrenal cortex leading to insufficient adrenal function. High levels of VLCFAs can also be toxic to myelin resulting in destruction of the myelin sheath.

There is wide variability in severity and age of onset, even among family members.

In **males** there are **three types** of X-ALD and **newborn screening cannot distinguish between them.** The three types and symptoms if untreated, include:

Childhood cerebral—learning and behavior problems begin before age 10 and progress to multiple neurologic issues leading to total disability and death within a few years of symptom onset.

Adrenomyeloneuropathy (AMN)—adults develop progressive stiffness and weakness in the legs, bladder and bowel problems, and adrenocortical insufficiency.

Addison disease only—adrenocortical insufficiency. Later many may develop features of AMN by mid-adult years.

Many **carrier females** will develop symptoms of myelopathy and peripheral neuropathy in their adult years. Cerebral or adrenal disease is very rare.

Treatment

Treatments will be determined in consultation with specialists. Close follow up is important for initiation of treatment at the appropriate time. Treatments can involve replacement of adrenal hormones when insufficiency is present. Hematopoietic stem cell transplantation may also be recommended for the childhood cerebral form of the disease.

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