KENTUCKY NEWBORN SCREENING PROGRAM

PARENT TEACHING SHEET

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X-Adrenoleukodystrophy (X-ALD)

X-ALD is a complex inherited disorder that affects boys and young men, rarely male infants, and in later life can affect adult women. The condition can damage nerve cells in the brain and other parts of the body as well as disturb the function of the adrenal gland. The condition is due a defect in a cell organelle called a peroxisome that is present in most cells. Symptoms are not often apparent at birth but may begin to show in boys later in childhood or even into adolescence. Expression in women may not be apparent until the third to six decade of life.

Spectrum of Findings in X-ALD		
Age of Onset	**Type**	
Boys 4-12 years	Affected Brain About 35% of boys	• Learning/behavior issues • Vision/hearing problems • Movement difficulties • Seizures, progressive changes of Brain MRI
Boys 13-50 years	Adrenomyeloneuropathy AMN About 65%	• Clumsiness/motor problems • Weakness, shaky movements • Brain complications
Women 40-70+ years	AMN About 85%	• Clumsiness/motor problems • Weakness, shaky gait
Boys and Men 8+ years	Addison Disease	• Adrenal failure • Bronze skin • Problems of loss of minerals • From blood and hypoglycemia (these findings may be present with other symptoms above)

X-ALD is a serious disorder but very variable and severity cannot be predicted with gene or biochemical tests.

X-ALD Treatment

Male patients with X-ALD are followed closely in childhood with monitoring by physical and neurological examination and MRI studies. Any brain changes of X-ALD are searched for by regular MRI studies and at the first sign of alternations in brain white matter the boy is a candidate for stem cell transplant

Other treatments for boys/men/women are based upon symptoms.