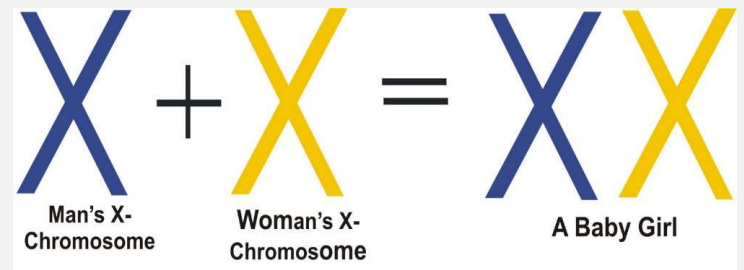
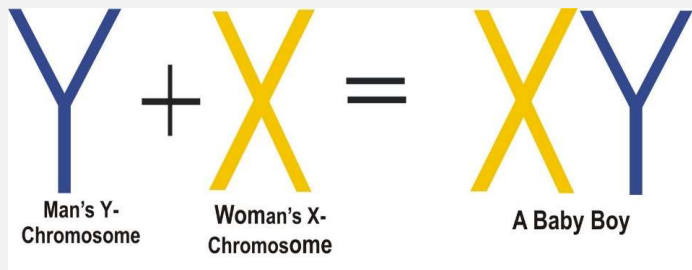


X-LINKED ADRENOLEUKODYSTROPHY (X-ALD)

Deliberative Community Engagement Event for
Newborn Screening
April 7-8, 2018

ADRENOLEUKODYSTROPHY

- Occurs in about 1:20,000 boys
- X-linked means it is a mutation on the X chromosome
- Boys have an X and a Y chromosome and girls have two X chromosomes
- If one of the X chromosomes has the “bad gene” for X-ALD, the other X chromosome (if it’s normal) can “make up” for the bad X chromosome. Boys only have one X chromosome, so don’t have the option of another “good” X chromosome to make up for the bad one.



TYPES OF X-ALD

- Childhood Cerebral ALD (C-ALD) 2-10 years – most lethal form. Damages the protective covering of nerve endings so nerve impulses are slowed or even stopped. Symptoms loss of cognitive ability, vegetative state. Rapidly gets worse until death.
- Adolescent Cerebral ALD 11-21 years
- Adrenomyeloneuropathy (AMN) 20-30 years – most common type. Slow onset of stiffness, weakness in legs, then use of cane, then wheelchair. Defects in spinal cord. May occur in a milder form in female carriers
- Adult C-ALD alone
- Addisonian Disease alone – adrenal insufficiency symptoms may include fatigue, weakness, weight loss, nausea. Stress like infection or trauma can cause severe nausea/vomiting, dehydration, low blood pressure, low blood sugar, low sodium, high potassium. Death may occur with common viral infections
- Asymptomatic – no symptoms



Brian Kelley. From Connecticut Newborn Screening for X-linked ALD, Adrienne Manning, Director

TREATMENT/THERAPY

- SUCCESSFUL TREATMENT IS CRITICALLY DEPENDENT ON STARTING BEFORE ANY SYMPTOMS ARE SEEN
- Any damage that has occurred, cannot be reversed by treatments.
- Bone marrow stem cell transplant – from similar, but not identical donor, like a brother or sister
- Lorenzo's Oil - is a combination of oleic acid and erucic acid, extracted from rapeseed oil and olive oil. In ALD pre-symptomatic boys, Lorenzo's Oil can (but not always) slow down the onset of the disease by stopping the body from producing the very long chain fatty acids, whose buildup leads to the breakdown of the protective cover of nerve endings.

BONE MARROW TRANSPLANT

- To be successful, transplant must occur as soon as possible after disease is diagnosed, and before symptoms start
- Babies can use umbilical cord blood rather than bone marrow.