Adrenoleukodystrophy (ALD)
Patient Clinical Journey

EveryLife Foundation for Rare Diseases

DELAYED DIAGNOSIS

Items in red may be prevented with a Timely Diagnosis.

PRE-DIAGNOSIS (ages 0-4)

Typical Accompanying Diagnoses

- Seizures/convulsions in boys
- Cognitive impairment, Speech impairment
- Adrenal crisis – *can be fatal*
- ADHD – *misdiagnosis*
- Autism
- Ataxia
- Behavioral issues
- Vision problems
- Anemia – *secondary finding to adrenal crisis*
  - Glucocorticoid deficiency
  - Mineralocorticoid deficiency

Specialists

- Behavioral Therapist
- Developmental Pediatrician
- Ophthalmologist
- Optometrist

With no treatment, expected mortality is within 1-5 years of symptom onset

YEAR 1

TIME OF DIAGNOSIS (ages 4-10)

Typical Accompanying Diagnoses

Previous diagnoses may remain present, in addition to the following listed diagnoses

- Hearing Loss
- Primary adrenal insufficiency
- Vision Loss

The following may present at the time of diagnosis or just after

- Gross motor problems/gait issues
- Dysphagia

Specialists – *Children may present to any of these specialists. These specialists will care for the patient throughout their life*

- Counseling Support Services
- Developmental Pediatrician
- Endocrinologist
- Geneticist
- Hematologist
- Neurologist
- Physical Therapist (PT) / Occupational Therapist (OT)

Testing & Procedures – *generally lead to diagnosis*

- Gene sequencing (ABCD1 gene) – *definitive diagnostic test*
- Multiple brain MRIs (also neck, spine, and CT of head/brain)
- Routine blood chemistry
- Leukocyte lysosomal enzymes
- Chromosome karyotyping
- Fatty acid profile (very long chain fatty acid analysis)

POST DIAGNOSIS

Typical Accompanying Diagnoses

Previous diagnoses may remain present, in addition to the following listed diagnoses

- Gross motor problems/gait issues/loss of muscle control
- Seizures
- Sensory impairment (blindness, deafness)

Testing & Procedures

- Stem Cell Transplant

Treatments – *can prevent or dramatically delay many complications*

- Allo transplant – *if eligible*
- Gene therapy
- Hormone replacement
- Glucocorticosteroid replacement therapy
- Mineralocorticoid replacement therapy
- Pain Management
- Physical therapy

Specialists

- Counseling Support Services
- Endocrinologist
- Hematologist
- Neurologist
- Palliative care
- Total care team – *once all loss of function occurs*

TIMELY DIAGNOSIS – NEWBORN SCREENING (NBS) OR PRENATAL SCREENING

Items in green are typically only seen with a Timely Diagnosis.

This clinical map is intended for males with cerebral X-linked ALD, ICD-10 code E71511. Women with X-linked ALD may be symptomatic later in life and are typically not at risk for cerebral issues.

With the introduction of new treatments and the changing natural history of the disease, care guidelines for patients post-diagnosis are continuing to evolve. Visit this pdf for 2022 diagnosis and care recommendations: https://n.neurology.org/content/neurology/99/21/940.full.pdf

TIME OF DIAGNOSIS

Testing & Procedures

- Newborn Screening – *tandem mass spectrometry*
- Routine adrenal screening – *occurs every 6 months and can prevent hospitalizations*
- Routine MRI screening – *occurs every 6 months after one year of age and will detect lesions before symptoms occur*

Events

- Hospitalizations

Specialists

- Genetic counselor
- Counseling Support Services
- Endocrinologist
- Developmental Pediatrician
- Neurologist

Typical Accompanying Diagnoses – *Most of the following disease presentations will be mitigated through a successful bone marrow transplant*

- Adrenal crisis – *can be fatal*
- Primary adrenal insufficiency
- Autism
- Anemia – *secondary finding to adrenal crisis*
  - Glucocorticoid deficiency
  - Mineralocorticoid deficiency

POST DIAGNOSIS

Specialists

- Genetic Counselor
- Counseling Support Services
- Endocrinologist
- Geneticist
- Hematologist – *to perform allo transplant or gene therapy only if active cerebral disease is present*
- Neurologist
- Physical Therapist (PT) / Occupational Therapist (OT)

Testing and Procedures

- Multiple brain MRIs (also neck, spine, and CT of head/brain) – *occur every 6 months until age 3, then every 12 months after*
- Gene sequencing (ABCD1 gene)
- Routine adrenal screening – *occurs every 3-4 months the first few years of life, then every 6 months*
- Routine MRI screening of the brain
- Routine blood chemistry
- Chromosome karyotyping
- Fatty acid profile (very long chain fatty acid analysis)
- Stem Cell Transplant as the final bullet

Treatments

- Gene therapy
- Allo transplant – *only if active cerebral disease is present*
- Glucocorticosteroid replacement therapy
- Mineralocorticoid replacement therapy
- Physical therapy
- Steroid replacement – *if adrenal insufficiency is present*

**This guide is not intended as a clinical decision-making tool. Please consult your specialists for any medical decisions.*