MT2019-01: Adrenoleukodystrophy National Registry Study (ALD) and Biobank

Status: Recruiting

Eligibility Criteria

Sex: All
Age: Not specified
This study is NOT accepting healthy volunteers
Inclusion Criteria

• Age 0

•100

• ALD patients or family member meeting any of the following criteria:

• Any patient diagnosed with ALD (confirmed by positive VLCFA testing and/or genetic mutation).

• Known or presumed mutation with ALD based on pedigree or confirmed mutation in ABCD1 gene

• Participants living in the United States and territories Exclusion Criteria

• Patients diagnosed with ALD who lack the capacity to consent/assent AND do not have a designated legally authorized representative or guardian.

• Patients who have undergone BMT or other cellular therapy .

• Patients not fluent in English who are unable to consent in-person at the BMT Journey Clinic.

• Patients who are illiterate

• Patient determined by the PI or designee to be unlikely to complete required study components (due to language barriers, compliance issues, etc.)

Conditions & Interventions

Interventions:
Other: Medical Record Abstraction, Other: Biospecimen Sample Collection
Conditions:
ALD (Adrenoleukodystrophy), Adrenoleukodystrophy, Cerebral Adrenoleukodystrophy
Keywords:
Registry, VLCFA, ABCD1, X-chromosome

More Information

Description: In this protocol, we will enroll pediatric, adolescent and adult patients diagnosed with adrenoleukodystrophy (ALD). These patients will include probands diagnosed by newborn screening and their relatives subsequently diagnosed, as well other patients who are diagnosed with ALD due to other presenting signs and symptoms and subsequently were confirmed to have ALD. We will ask consenting subjects to provide a medical history (with verification via medical records), to participate in a semi-annual health survey and provide consent to collect biospecimens. The overarching goal of this work is to engage with families affected by ALD and to assemble a resource of clinical, medical, and biological data that will allow of to better understand the natural history of ALD, and how this is affected by newborn screening. The initial focus will be on patients within Minnesota, but participation will be open to any family interested in the study, as this will be web-based. This registry and biobank, together with other research conducted in tandem, will possibly provide information describing the natural history of ALD and outcomes with interventions. It is anticipated that the data collected will further our understanding of the natural history of the disease, basic biology of adrenoleukodystrophy, diagnosis and outcomes. Ultimately, this research may lead to new avenues for early diagnosis and development of safer and more effective therapies for ALD.
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Phase: NA
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System ID: NCT03789721