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

Status: Recruiting

Eligibility Criteria

Age: Not specified

This study is NOT accepting healthy

Healthy Volunteers: volunteers

Inclusion Criteria:

- age 0 to 100 - patient or family member diagnosed with ALD (confirmed by positive VLCFA testing and/or genetic mutation - patient or family member with known or presumed mutation with ALD based on pedigree or confirmed mutation in ABCD1 gene - living in the United States and territories

Exclusion Criteria:

- have undergone BMT or other cellular therapy - not fluent in English who are unable to consent in-person - people who are unable to read or write

Conditions & Interventions

Conditions:
Rare Diseases
Keywords:
Adrenoleukodystrophy, ALD, Cerebral Adrenoleukodystrophy

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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