



University of Pittsburgh

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We are conducting a research study at the University of Pittsburgh to characterize the genetic mutations that cause Autosomal Dominant Leukodystrophy (ADLD). The symptoms of ADLD usually begin in adulthood around the age of 30-50 years. However, children with LMNB1 duplications and having symptoms have also been rarely reported. The genetic cause of ADLD has been identified as a mutation involving the gene Lamin B1. Patients have an extra copy of the gene as compared to normal individuals. As the mutation has only been relatively recently identified it is unclear as to whether other kinds of mutations involving Lamin B1 are also responsible for the disease. This study aims to identify and characterize the different types of mutations that cause ADLD. We will use custom designed molecular tools to carry out genetic testing and may identify ADLD mutations that have been missed by previous testing methods. The results of this study will be important for the development of accurate genetic tests and thus will aid in the better diagnosis of ADLD patients.

Who can participate?

The study is limited to those who fulfill any of the following criteria:

1. Individuals who have a genetic diagnosis of ADLD,
2. Individuals who have symptoms that fit ADLD but who have not undergone genetic testing.
3. Individuals who have symptoms that fit ADLD but for whom genetic testing for ADLD mutations has turned out to be negative. These individuals are especially encouraged to participate in the study.

What will be required by the study:

As a part of this study you will be asked to fill out a questionnaire about your medical and family history, donate blood or a saliva sample. You may also be asked to provide relevant health information including any previous MRI scans.

There are no costs to participate in the study and participants will not be financially compensated.

If you are interested in either participating or learning more about the study, you may contact us directly by phone, fax or email. Thank you.

Sincerely

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