

Investigating Leukodystrophies in Adults

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Problems faced at present:

- Lack of funds for investigating
- Nil or no medication

Routes to a Leukodystrophy Diagnosis

- MRI scan
- Clinical features often do not relate
- Imaging
- Blood work
- Nerve conduction studies
- White cell enzyme testing
- Compound heterozygosity for MLD
- Brain biopsy
- Skin fibroblast
- Genetic studies

What is needed for the future:

- NHS Genomic Medical Service
- Aims being:

To provide consistent care

Build a National Data Base for all Leukodystrophies