

Evaluating the use of CSF and PET scan biomarkers in detecting progression of Lafora Disease

AUTHORS

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RESEARCH SIMPLIFIED

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AIM

The aim of this study is to investigate if CSF and PET scan biomarkers can be useful in detecting the progression of Lafora disease. A PET scan is an imaging test that can reveal the metabolic activity of your tissues (i.e. how much energy they are generating).

METHODS

The study investigated Lafora disease in six patients from five unrelated families in Italy. Clinical, genetic, and imaging data were collected and analyzed. The patients were monitored regularly for symptoms, including epilepsy, myoclonus, ataxia, and dementia. Imaging studies, including MRI and PET scans, showed distinct patterns of neurodegeneration in the brain. The researchers then took CSF samples to see if they could detect neurodegeneration biomarkers identified for Alzheimer's disease in Lafora patients.

RESULTS

The study identified three progressive electro-clinical stages in the natural history of Lafora disease. These stages were defined based on four main symptoms: epilepsy, myoclonus, ataxia, and dementia. The stages progressed from early onset of epilepsy to worsening epilepsy and myoclonus, followed by the gradual onset of dementia and cerebellar signs, and ultimately severe neurological deterioration. PET imaging highlighted loss of neuronal function in various brain regions, which was most severe in the later stages. The regions of the brain associated with cognitive function were particularly impacted in later disease stages. However, Alzheimer's biomarkers were not present in the CSF samples, indicating the dementia occurs through a different mechanism than the one identified for Alzheimer's.

CONCLUSION

PET imaging improves diagnostic accuracy and provides insights into disease progression. Further studies are needed to replicate these findings and could be used to evaluate new treatments for Lafora disease.