

## GENETIC DISEASES EVALUATION CENTER

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### Test Name & Code:

- **Test Name:** Lafora Disease (*NHLRC1* Gene Sequence Analysis)
- **Test Code:** 4617

### Related Diseases & Genes:

- Epilepsy, progressive myoclonic 2B (Lafora) (OMIM #: 254780) || Gene: *NHLRC1* (OMIM #: 608072)

### Synonyms:

- MYOCLONIC EPILEPSY OF LAFORA
- LAFORA BODY DISEASE; LBD
- Epilepsy, progressive myoclonic 2B
- *NHLRC1*

### Test Details:

- **Methodology:** DNA Sequencing
- **Performed:** Everyday
- **Reported:** 2-3 weeks

### Specimen Details:

- **Specimen Collection:** Peripheral blood
- **Container:** Tube with EDTA
- **Specimen Volume/Amount:** > 3 ml
- **Storage/Transport Condition:** Refrigerated
- **Unacceptable Specimens/Conditions:** Serum; Frozen specimens; Hemolyzed specimens; Icterus; Lipemia

### Testing Area(s):

- Neurogenetics

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