

GENETIC DISEASES EVALUATION CENTER

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Test Name & Code:

- **Test Name:** Alzheimer's Disease, *PSEN1* Genetic Analysis, Alzheimer Type 3
- **Test Code:** 4613

Related Diseases & Genes:

- "Acne inversa, familial, 3" (OMIM #: 613737) || Gene: *PSEN1* (OMIM #: 104311)
- "Alzheimer's disease, type 3" (OMIM #: 607822)
- "Alzheimer's disease, type 3, with spastic paraparesis and apraxia" (OMIM #: 607822)
- "Dementia, frontotemporal" (OMIM #: 600274)

Synonyms:

- PRESENILIN 1
- PS1
- S182

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

