

GENETIC DISEASES EVALUATION CENTER

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Test Name & Code:

- **Test Name:** DRAVET Syndrome *SCN1A*
- **Test Code:** 4782

Related Diseases & Genes:

- Dravet syndrome (OMIM #: 607208) || Gene: *SCN1A* (OMIM #: 182389)
- “Epilepsy, generalized, with febrile seizures plus, type 2” (OMIM #: 604403)
- “Febrile seizures, familial, 3A” (OMIM #: 604403)
- “Migraine, familial hemiplegic, 3” (OMIM #: 609634)

Synonyms:

- “SODIUM CHANNEL, BRAIN TYPE I, ALPHA SUBUNIT; NAC1”
- “ODIUM CHANNEL, NEURONAL TYPE I, ALPHA SUBUNIT; \diamond SCN1A”
- NAV1.1

Test Details:

- **Methodology:** Next Generation DNA Sequencing
- **Performed:** Everyday
- **Reported:** 6 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

