Mitochondrial Studies: NARP - Neuropathy, Ataxia and Retinitis Pigmentosa

Test Code: 2024

Department: Molecular Genetics

Test Synonyms: Neurogenic Muscle Weakness, Ataxia and Retinitis Pigmentosa

CPT Code(s):
- 83890
- 83892
- 83894
- 83897
- 83898

Background:
Neuropathy, ataxia, and retinitis pigmentosa (NARP) is a condition that causes a variety of signs and symptoms chiefly affecting the nervous system. The NARP assay is clinically indicated for the evaluation of patients with the following clinical criteria (GeneTests Reviews):

- **Neurogenic muscle weakness.** Electromyography (EMG) and nerve conduction studies may demonstrate peripheral neuropathy (which may be a sensory or sensorimotor axonal polyneuropathy).
- **Ataxia.** Cerebral and cerebellar atrophy may be noted on MRI.
- **Retinitis pigmentosa.** The ocular manifestations of NARP are extremely variable and range from a mild salt and pepper retinopathy to bull's eye maculopathy and classic retinitis pigmentosa with bone spicule formation. Ophthalmologic examination may reveal pigmentary retinopathy or optic atrophy. Electroretinogram (ERG) may reveal abnormalities (including small-amplitude waveform) or may be normal. ERG may demonstrate predominantly cone dysfunction in some pedigrees and mainly rod dysfunction in others.

NARP is associated with an T > G mutation at nucleotide 8993 in the mitochondrial ATPase 6 gene. A smaller proportion of cases are associated with an T - C mutation at the same nucleotide 8993.

Reason for Referral:
- Diagnostic Testing
- Prenatal Diagnosis

Methodology:
The two point mutations described above are detectable by PCR and restriction enzyme based analysis.

Specimen Requirements:
- **Muscle:** 50 mg muscle
  - OR-
  - **Blood:** 6.0 mL EDTA (purple-top) or ACD (yellow-top) tube

A REQUISITION FORM MUST ACCOMPANY ALL SAMPLES. Please include detailed clinical information, including ethnicity, clinical history, and family history.
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**Test Performed (Days):**
Weekly

**Turn Around Time:**
14 – 21 Days

**Shipment Sensitivity Requirements:**
Package and ship specimen to remain cold, but not frozen. Ship via overnight express, using the FedEx priority overnight label provided. Contact Client Services for shipping kits and instructions at (855) 535-1522.

**Additional Information:**
This test detects point mutations that are present in levels greater than or equal to approximately 5% of total mtDNA.

**References:**