

## Early-Onset Primary Dystonia (DYT1) via *TOR1A* Gene Sequencing

### Sequential (Standard) Test – Test #166; Tier 1 Only – Test #167; Tier 2 Only – Test #168

**Brief Description of Clinical Features:** DYT1 is one of the more common early-onset primary dystonias, typically presenting in childhood or adolescence. DYT1 accounts for 60% of early onset dystonia in the general population and up to 90% of cases in the Ashkenazi Jewish population. Adult onset has been seen with DYT1. DYT1 usually starts in a leg (average age 9 years) or an arm (average age 15 years). Initially, dystonia is often detected in specific actions such as writing or walking. Gradually, foot, leg, or arm posturing caused by dystonic muscle contractions becomes the most common symptom. Disease severity varies considerably even within the same family. For more information, see Raymond and Bressman (Gene Reviews, www.genetests.org, 2008).

**Genetics:** The *TOR1A* gene is the only gene known to be associated with early-onset primary dystonia (DYT1). DYT1 is inherited in an autosomal dominant manner with reduced penetrance. About 70% of individuals who carry a disease-causing allele have no symptoms (Ozelius et al. Nat. Genet. 17:40-48, 1997; Bressman et al. Neurology 59:1780-1782, 2002). Deletion of one highly conserved codon, c.904\_906delGAG (p.delGlu302), has been seen in 99% of DYT1 cases.

**Description of This Particular Test:** This test involves two Tiers. In Tier 1, PCR amplification from genomic DNA and bidirectional sequencing of the protein coding region for exon 5 is done to confirm or deny the presence of the common c.904\_906delGAG (p.delGlu302) mutation. In Tier 2, PCR amplification and bidirectional sequencing of all protein coding regions for exons 1-4 of *TOR1A* is performed. Tier 1 testing may be ordered alone.

**Indications for Test:** Candidates for this test are patients with symptoms consistent with DYT1, and the family members of patients with known mutations. In addition to this DYT1 gene test, PreventionGenetics also offers sequencing of several other dystonia genes.

**Turn Around Time:** Maximum of 40 days, although many tests are completed in less than 2-3 weeks.

**Specimen Requirements:** See page 4 of the Requisition Form.

<b>Price:</b>	<b>Tier 1: Sequencing of exon 5 of the <i>TOR1A</i> Gene</b>	<b>\$190</b>
	<b>Tier 2: Sequencing of exons, 1-4 of the <i>TOR1A</i> Gene</b>	<b>\$290</b>
	<b>Tiers 1 and 2 Sequential</b>	<b>\$440</b>

**CPT Codes:**

Codes	Description	Tier 1 Only	Tier 2 Only	Tier 1 + Tier 2
83890	Ascertainment	\$ 30 (x1)	\$ 30 (x1)	\$ 30 (x1)
83891	DNA Isolation	\$ 40 (x1)	\$ 40 (x1)	\$ 40 (x1)
83898	Amplification	\$ 25 (x1)	\$ 48 (x4)	\$ 89 (x5)
83904	Mutation Ident by Sequencing	\$ 35 (x1)	\$ 82 (x4)	\$171 (x5)
83894	Separation	\$ 15 (x1)	\$ 30 (x1)	\$ 40 (x1)
83912	Interpretation and Report	\$ 45 (x1)	\$ 60 (x1)	\$ 70 (x1)
<b>Totals:</b>		<b>\$190</b>	<b>\$290</b>	<b>\$440</b>

**Single exon sequencing for detection of previously identified familial mutations is also available for \$190.**

**Accreditation Info.** CLIA ID #: 52D1027685 (expires 1/18/13) (CAP#: 7185561, AU ID: 1407125 expires 12/20/12)

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