

HGPS (Progeria) *LMNA* Exon 11 Testing/ *LMNA* Known Mutation Single Exon Sequencing

Hutchinson-Gilford Progeria syndrome (HGPS) is a rare genetic disorder characterized by features reminiscent of marked premature aging. Recently, sequencing of exon 11 of the *LMNA* gene, revealed that 18 out of 20 classical cases of HGPS harbored an identical de novo (that is, newly arisen and not inherited) single-base substitution, C1824T (GGC > GGT), within exon 11. One additional case was identified with a G608S (GGC > AGC) within the same codon. Both of these mutations result in activation of a cryptic splice site within exon 11, resulting in production of a protein product that deletes 50 amino acids near the carboxy terminus. Identification of the common C to T mutation has potential clinical significance for presymptomatic diagnosis in this population allowing the clinician to determine which patients carry the common mutation and therefore have an increased risk for developing HGPS. Testing of the additional exons of the *LMNA* gene is done in patients without this “common” mutation. This testing is described elsewhere. In addition, we offer exon specific screening of other *LMNA* exons to confirm known familial mutations in additional family members.

A positive result reports defining a *LMNA* gene mutation indicates only the potential for the development of HGPS. Further testing is required to confirm the diagnosis of HGPS in this patient.

It is important to note that a negative result for this assay does not rule out other rarer mutations either in other non coding regions of the *LMNA* gene or in other as yet non defined genes that may be contributory to HGPS.

Specimen Requirements

- Collect 2-5 ml of whole blood in EDTA (purple top tube) or ACD (yellow top tube). 5 ml is the preferred volume.
- Only one blood tube is required for multiple tests.
- Ship whole blood specimens at room temperature.
- Do not freeze blood.
- During hot weather, include a frozen ice pack in the shipping container. Do not allow the ice pack to come in direct contact with the specimen tube.
- In cold weather, include an unfrozen ice pack to help moderate extremes in temperature. The DNA in whole blood is stable for at least 48 hours at 21°C, 5-7 days at 4°C.

Single exon Sequence analysis screening for the presence previously identified mutations in the *LMNA* gene **\$230.00.**

Molec Diag, Ascertainment	83890	
Molec Diag, Isolation	83891	
Molecular Diag, Amplif	83898	Turnaround time is 1~2 weeks
Mutat Id By Seq, Single Seg	83904	
Molecular Diag, Separation	83894	
Interpretation and Report	83912	

Accreditation Info. CLIA ID #: 52D1027685 (expires 1/18/07) (CAP#: 7185561, AU ID: 1407125 expires 12/20/06)

Ship to:

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