

HGPS (Progeria) Testing via *LMNA* Gene 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. In addition, we offer exon specific screening of other *LMNA* exons to confirm known familial mutations in additional family members. This testing is described elsewhere.

Testing of the 10 additional exons of the *LMNA* gene is done in patients without this “common” mutation.

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.

Sequence analysis of the <i>LMNA</i> gene	\$890.00
Molec Diag, Ascertainment	83890
Molec Diag, Isolation	83891
Molecular Diag, Amplif x11	83898
Mutat Id By Seq, Single Seg x11	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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