

UCLA ORPHAN DISEASE TESTING CENTER

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<http://www.odtc.ucla.edu>

**Please send/deliver all specimens to: UCLA Pathology Outreach Services,
 10833 Le Conte Avenue, Room 1P-201 CHS, Los Angeles, CA 90095-1732**

Patient Identification / Label				ODTC #: _____ Wsh #: _____		
				Patient ID #: _____		
				DOB: _____		
				Sex: Male Female		
				Specimen Collection		
				Date	Time	By
Ordering Physician/ Ordering PI	(LAST)	(FIRST)	(Initial)	Physician's ID #	Contact #:	Loc/Clinic/Lab:
Physician's Address:				City:	State:	Zip:

<p>Specimen Information: Please select one and specify the amount.</p> <ul style="list-style-type: none"> Blood (2cc in Lav top required for each test) Blood spot Amniotic fluid Paraffin block / section: DNA (1 µg minimum) Buccal brush Tissue: (≥0.2g) Source: Culture, type: 	<p>Test ordered:</p> <ul style="list-style-type: none"> Cystic Fibrosis, Full Gene Sequencing Cystic Fibrosis, One Exon Cystic Fibrosis, Two Exons Nijmegen Breakage Syndrome, Full Gene Sequencing Nijmegen Breakage Syndrome, One Exon Nijmegen Breakage Syndrome, Two Exons Nijmegen Breakage Syndrome, 657del5 Mutation Analysis Hereditary Inclusion Body Myopathy (HIBM) M712T Mutation Analysis Familial Mediterranean Fever, Full Gene Sequencing Familial Mediterranean Fever, One Exon Familial Mediterranean Fever, Two Exons Custom Mutation Analysis, One Exon/Mutation Custom Mutation Analysis, Two Exons/Mutations
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Pertinent Family History/Ethnicity:
Primary Counseling Issue for Genetic Disease:
Proband Diagnosis Prenatal Diagnosis Carrier Screening Presymptomatic Diagnosis Other:

<p>Billing Information: <i>Billing to Physician/Referring Laboratory only. Third party payments must be made in advance.</i></p> Billing Contact _____ Hospital/Laboratory Name _____ Address _____ Address 2 _____ City _____ State _____ Zip _____ Country _____ Phone _____ Fax _____
<p>Payment to be payable to: Regents of UC</p>

If Included, Please Mark:

A diagram or sequence of the gene with primer and mutation sites indicated.
The sequencing autoradiogram or chromatogram of the amplicon, if required.
A photo of the agarose gel containing the amplicon and the size markers.
Positive and negative DNA controls.
Forward and reverse primers for amplification, if required.
References.

Technical Information Requested by ODTG:

1- Name of gene to be tested and GDB accession #:

2- Detailed description of the mutation(s):

Heterozygous Homozygous Hemizygous Compound Heterozygous

3- The PCR conditions, in detail:

4- Cycling conditions:

5- The anticipated size of the amplicon:

6- Forward and reverse primers:

Forward sequence:

Reverse sequence:

Concentrations:

7- Contact names and numbers:

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