### MYOTONIC DYSTROPHY TYPE 1

* (PCR, Fragment Analysis)

<table>
<thead>
<tr>
<th>DMPK Gene CTG Repeats</th>
<th>INDETERMINATE</th>
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**Interpretation**

<table>
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<th>RESULT</th>
<th>REMARKS</th>
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<tbody>
<tr>
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<td>Indicates &gt;50 CTG repeats in the sample submitted</td>
</tr>
<tr>
<td>Indeterminate</td>
<td>Indicates presence of inherent inhibitors in the sample submitted</td>
</tr>
<tr>
<td>Not Detected</td>
<td>Indicates 5-34 CTG repeats in the sample submitted</td>
</tr>
</tbody>
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**Note**

1. Results must be interpreted in context with clinical findings, family history and other relevant laboratory data.
2. Genetic Counselling is recommended.
3. This is an in-house developed assay.
4. Test conducted on EDTA whole blood.
5. Exact number of repeats is not elucidated by this assay.

**Comment**

Myotonic dystrophy is an autosomal dominant disorder characterized mainly by muscular dystrophy, cataracts, hypogonadism, frontal balding, and ECG changes. Myotonic dystrophy Type 1 (DM1), has a severe congenital form and a milder childhood-onset form as well as an adult-onset form. The genetic defect in DM1 results from an amplified trinucleotide repeat in the 3-prime untranslated region of DMPK gene. Disease severity varies with the number of repeats: normal individuals have 5 to 37 repeats, mildly affected persons have 50 to 150 repeats, patients with classic DM have 100 to 1,000 repeats, and those with congenital onset can have more than 2,000 repeats.

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**End of report**  

**IMPORTANT INSTRUCTIONS**

*Test results released pertain to the specimen submitted.*  
*All test results are dependent on the quality of the sample received by the Laboratory.*  
*Laboratory investigations are only a tool to facilitate in arriving at a diagnosis and should be clinically correlated by the Referring Physician.*  
*Sample repeats are accepted on request of Referring Physician within 7 days post reporting.*  
*Report delivery may be delayed due to unforeseen circumstances. Inconvenience is regretted.*  
*Certain tests may require further testing at additional cost for derivation of exact value. Kindly submit request within 72 hours post reporting.*  
*Test results may show interlaboratory variations.*  
*The Courts/Forum at Delhi shall have exclusive jurisdiction in all disputes/claims concerning the test(s) & or results of test(s).*  
*Test results are not valid for medico legal purposes.*  
*Contact customer care Tel No. +91-11-39885050 for all queries related to test results.*