**TEST CONDUCTED**  
**DRPLA (DENTATORUBRAL-PALLIDOLUYSIAN ATROPHY) GENE ANALYSIS #**  
(PCR, Fragment Analysis)

<table>
<thead>
<tr>
<th>RESULT</th>
<th>REMARKS</th>
</tr>
</thead>
<tbody>
<tr>
<td>Detected</td>
<td>Indicates &gt;=48 CAG 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 6-35 CAG repeats in the sample submitted</td>
</tr>
</tbody>
</table>

**Note**
1. Results showing homozygosity for normal alleles of the same size should be confirmed by an appropriate alternate method (triplet primed -PCR or southern blotting). For homoallelic patients, parental testing is recommended.
2. Results must be interpreted in context with clinical findings, family history and other relevant laboratory data.
3. Genetic Counselling and clinical correlation is recommended.
4. This is an in-house developed assay.
5. Test conducted on EDTA whole blood.

**Comment**
Dentatorubral-pallidoluysian atrophy (DRPLA) is a progressive disorder of ataxia, myoclonus, epilepsy, and progressive intellectual deterioration in children and ataxia, choreoathetosis, and dementia or character changes in adults. DRPLA is caused by a heterozygous expanded trinucleotide repeat (CAG) in the ATN1 gene on chromosome 12p13. Onset ranges from before age one year to age 72 years; mean age of onset is 31.5 years. The clinical presentation varies depending on the age of onset. The cardinal features in adults are ataxia, choreoathetosis, and dementia. Cardinal features in children are progressive intellectual deterioration, behavioral changes, myoclonus, and epilepsy.
**LPL - LPL-ROHINI ( NATIONAL REFERENCE LAB)
SECTOR - 18, BLOCK -E ROHINIDELHI 110085**

<table>
<thead>
<tr>
<th>Name</th>
<th>DUMMY N167</th>
<th>Collected</th>
<th>29/7/2020 12:00:00AM</th>
</tr>
</thead>
<tbody>
<tr>
<td>Lab No.</td>
<td>153672607</td>
<td>Received</td>
<td>29/7/2020 6:27:46PM</td>
</tr>
<tr>
<td>A/c Status</td>
<td>P</td>
<td>Collected</td>
<td>29/7/2020 12:00:00AM</td>
</tr>
<tr>
<td>Age</td>
<td>24 Years</td>
<td>Gender</td>
<td>Male</td>
</tr>
<tr>
<td>Ref By</td>
<td>UNKNOWN</td>
<td>Report Status</td>
<td>Final</td>
</tr>
</tbody>
</table>

Dr Atul Thatai  
PhD, Biotechnology  
HOD Molecular Diagnostics  
NRL - Dr Lal PathLabs Ltd

**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.  
* Sample drawn from outside source.