LABORATORY REPORT

Redcliffe labs

CRM ID	: 0000000	Sample Type	: NA
Name	: DUMMY	Date & Time Collected	: DD-MM-YYYY
Sex/Age	: NA	Date & Time Received	: DD-MM-YYYY
Bill. Loc.	: NA	Date & Time Reported	: DD-MM-YYYY
Ref. By	: NA		

Test Report

TEST CONDUCTED	MYOTONIC DYSTROPHY TYPE 1 (PCR, Fragment Analysis)

RESULTS

DMPK Gene CTG Repeats

DETECTED

Interpretation

RESULT	REMARKS
Detected	Indicates >50 CTG repeats in the sample submitted
Indeterminate	Indicates presence of inherent inhibitors in the sample submitted
Not Detected	Indicates 5-34 CTG repeats in the sample submitted

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.

NOTE-**This test is processed and validated at the partner lab of Redcliffe Labs.

***** End of Report *****

Approved by
Dr. Himani Pandey
Postdoc-SGPGIMS Lucknow

Terms and Conditions of Reporting

1. The presented findings in the Reports are intended solely for informational and interpretational purposes by the referring physician or other qualified medical professionals possessing a comprehensive understanding of reporting units, reference ranges, and technological limitations. The laboratory shall not be held liable for any interpretation or misinterpretation of the results, nor for any consequential or incidental damages arising from such interpretation.

2. It is to be presumed that the tests performed pertain to the specimen/sample attributed to the Customer's name or identification. It is presumed that the verification particulars have been cleared out by the customer or his/her representation at the point of generation of said specimen / sample. It is hereby clarified that the reports furnished are restricted solely to the given specimen only.

3. It is to be noted that variations in results may occur between different laboratories and over time, even for the same parameter for the same Customer. The assays are performed and conducted in accordance with standard procedures, and the reported outcomes are contingent on the specific individual assay methods and equipment(s) used, as well as the quality of the received specimen.

4. This report shall not be deemed valid or admissible for any medico-legal purposes.

5. The Customers assume full responsibility for apprising the Company of any factors that may impact the test finding. These factors, among others, includes dietary intake, alcohol, or medication / drug(s) consumption, or fasting. This list of factors is only representative and not exhaustive.

Disclaimer: Method given in report are only indicative and can be changed depending upon type of machine and kit available at time of testing.

Not all tests at all locations are under NABL scope. Availability of tests under NABL scope varies from lab to lab.