LABORATORY REPORT

Redcliffe labs

Name	: Dummy		
Lab No.	: XXX	**Age**	: 29 Years
Ref By	: Dr.XXX	**Gender**	: Male
Collected	: 29/01/2024	**Reported**	: 8/2/2024
A/c Status	: P	**Report Status**	: Final
Collected at	:XXX		

Test Report

TEST CONDUCTED	MYOTONIC DYSTROPHY TYPE 2 (PCR, Fragment Analysis)

RESULTS		
Myotonic Dystrophy Type 2		
NOT DETECTED		

Interpretation

RESULT	REMARKS
Detected	Indicates >75 CTG repeats in the sample submitted
Indeterminate	Indicates presence of inherent inhibitors in the sample submitted
Not Detected	Indicates 11-26 CTG repeats in the sample submitted

Note

1. Results must be interpreted in context with clinical findings, family history and other relevant laborator data.
2. Genetic Counselling and clinical correlation 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 2 (DM2), is rare and generall manifests with milder signs and symptoms than DM1. DM2 is caused by heterozygous expansion of a CCT repeat in intron 1 of the ZNF9 gene (3q21.3). *CNBP* (formerly *ZNF9*), the gene encoding cellular nuclei acid-binding protein (zinc finger protein 9), is the only gene in which mutation is known to cause DM2. *CNB* intron 1 contains a complex repeat motif, (TG)n(TCTG)n(CCTG)n. Expansion of the CCTG repeat cause DM2. Individuals with DM2 have muscle pain and stiffness, progressive muscle weakness, myotonia, mal hypogonadism, cardiac arrhythmias, diabetes, and early cataracts. Other features may include cognitive dysfunction, hypersomnia, tremor, and hearing loss.

*** End of Report ***

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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.