LABORATORY REPORT

Redcliffe labs

Patient NAME	:		
DOB/Age/Gender	:	Report STATUS	:
Patient ID / UHID	:	Barcode NO	:
Referred BY	:	Sample Type	:
Sample Collected	: Sep 14, 2025, 12:47 PM.	Report Date	: Oct 04, 2025, 07:52 PM.

#Huntington Disease Molecular Analysis

Indications :

Referred for Huntington disease.

TEST RESULTS :

Test Name	Result
Huntington Disease Molecular Analysis	HD allele with full penetrance
Ms Mallika Kundu is highly likely to be **affected with Huntington's disease** as the number of CAG repeats on one of the allele at the HD locus fall beyond the normal range of 6-35 repeats.	

	Upper Allele	Lower Allele
Allele Size (bp)	**175**	92
Number of CAG repeats	**44**	16

Note: The estimated error for such analysis is +/- 1-3 repeats

Test Information :

Methodology:

Polymerase Chain Reaction (PCR) using fluorescently labeled primers flanking the CAG repeat region in the HTT gene. The PCR product was run on Genetic Analyser and repeat number estimated using appropriate formulae.

Huntington disease is caused by an expansion of CAG repeat sequences in the HTT gene. The interpretation is based on the following types of repeat sequences:

Range of Repeats	Classification of Pathogenicity
6 - 26	Normal allele
27-35	Mutable normal allele
36-39	HD allele with reduced penetrance
>40	HD allele with full penetrance

Recommendation :

- Genetic counseling is recommended.
- The results should be correlated with clinical and other laboratory findings.

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Limitations :

- Point mutations, Indel and large deletions in HTT gene will not be detected by this test
- This assay could not detect > 150 CTG repeat expansion
- PCR will not amplify genic regions outside primer binding site in HTT gene
- There may be sequence variations in the primer binding site that could give rise to either non-amplification of normal or expanded alleles.
- False positive or negative results may occur for reasons that include somatic or tissue-specific mosaicism, rare genetic variants, blood transfusions, bone marrow transplantation, or erroneous representation of family history.
- Although all precautions are taken during Molecular Genetic testing the currently available data indicate that the technical error rate for all types of Molecular DNA analysis is approximately 1%.
- For test performed on specimens received or collected from non-NRL locations, it is presumed that the specimen belongs to the patient named or identified as labeled on the container/test request and such verification has been carried out at the point of generation of the said specimen by the sender.

References :

- Martin, J. B & Gusella, J. F. (1990). Huntington Disease: pathology and management. NEJM 315,1267-76.
- Jama M, Millson A, Miller CE, Lyon E. Triplet repeat primed PCR simplifies testing for Huntington disease. J Mol Diagn. 2013 Mar;15(2):255-62.
- Bean, L., Bayrak-Toydemir, P. and ACMG Laboratory Quality Assurance Committee, 2014. American College of Medical Genetics and Genomics Standards and Guidelines for Clinical Genetics Laboratories, 2014 edition: technical standards and guidelines for Huntington disease. Genetics in Medicine, 16(12), pp.1-7.
- http://www.ncbi.nlm.nih.gov/books/NBK1305/.

NOTE- **This test is processed at Redcliffe's partnered lab.

*** End Of Report ***

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