

PATIENT NAME	Mr. ABHISEK RATH	Barcode No	24201361
Age/Gender	13 Years/ M	Reg. No	0652304200047
Referring by		SPP Code	SPL-OR-063
REF. DOCTOR		Collected On	19-04-2023
Primary Sample	Whole Blood	Received On	21-04-2023
Sample Tested In	Whole Blood EDTA	Reported On	01-05-2023

DUCHENNE MUSCULAR DYSTROPHY DNA PCR (MUTATION ANALYSIS – 21 EXONS)

Test Description	Test Report
DMD mutation analysis	NEGATIVE for DMD

Lane	EXON	Product length	RESULT
1	EXON 01	535 bp	DETECTED
	EXON 03	410 bp	DETECTED
	EXON 16	290 bp	DETECTED
2	EXON 08	360 bp	DETECTED
	EXON 13	238 bp	DETECTED
	EXON 17	416 bp	DETECTED
3	EXON 12	331 bp	DETECTED
	EXON 32	253 bp	DETECTED
	EXON 47	181 bp	DETECTED
4	EXON 42	195 bp	DETECTED
	EXON 49	439 bp	DETECTED
	100 BP MARKER		
5	EXON 46	148 bp	DETECTED
	EXON 48	506 bp	DETECTED
	EXON 51	388 bp	DETECTED
6	EXON 45	547 bp	DETECTED
	EXON 50	271 bp	DETECTED
	EXON 53	212 bp	DETECTED
7	EXON 34	171 bp	DETECTED
	EXON 41	274 bp	DETECTED
	EXON 52	113 bp	DETECTED
	EXON 04	196 bp	DETECTED

COMMENTS: Dystrophin Gene deletion was not observed in any of the exons studied

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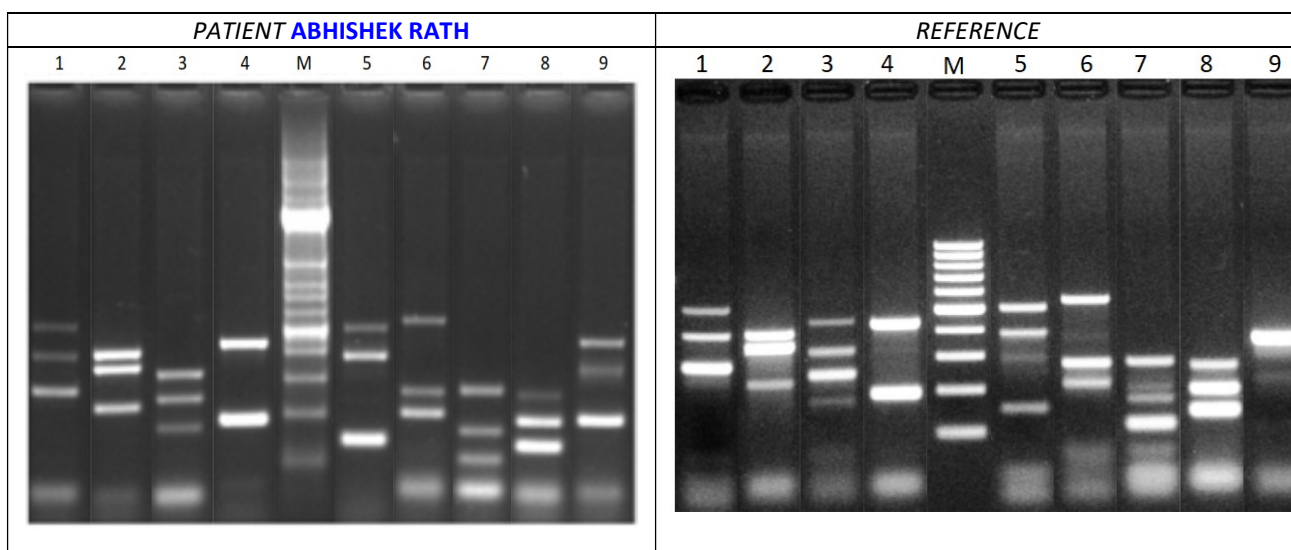


Dr. RUTURAJ
MD, MICROBIOLOGIST

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INDICATION	Dystrophin Gene deletion was not observed in any of the exons studied for Duchenne Muscular Dystrophy
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DUCHENNE MUSCULAR DYSTROPHY DNA PCR (MUTATION ANALYSIS – 26 EXONS)



NOTE:

Although all precautions are taken during DNA tests the currently available data indicate that the technical error rate for all types of DNA analysis is approximately 2%. It is important that all clinicians or persons requesting DNA diagnostic tests are aware of these data before acting upon these results.

Since DMD is an X linked recessive disorder, the women will be normal with respect to all exons related to dystrophin gene, though they are carriers. To know the carriers status of females, genome sequencing is recommended.

DMD (DUCHENNE MUSCULAR DYSTROPHY)

DMD: Duchenne muscular dystrophy is an X chromosome linked recessive genetic disease seen only in males whose mother is a carrier. The affected males manifest degeneration of muscles in legs and shoulder. As they progress into their teens, the legs and hands gets paralysed and they could not walk and cripple. They carry mutant dystrophin gene on their X chromosome with deletion of few or many exons.

MUTATION ANALYSIS: In the present method, mutations on dystrophin gene of the specimen are analyzed by marker specific conventional PCR followed by agarose gel electrophoresis and documentation of gel in gel doc system. A reference control (normal gene) is also carried along with specimen. In the present protocol all 26 exons have been analyzed as the case looked positive for the DMD mutations.



Dr. RUTURAJ

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