SagePath®
Excellence In Health Care

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

CONTINUED....

Dr.RUTURAJ
MD,MICROBIOLOGIST

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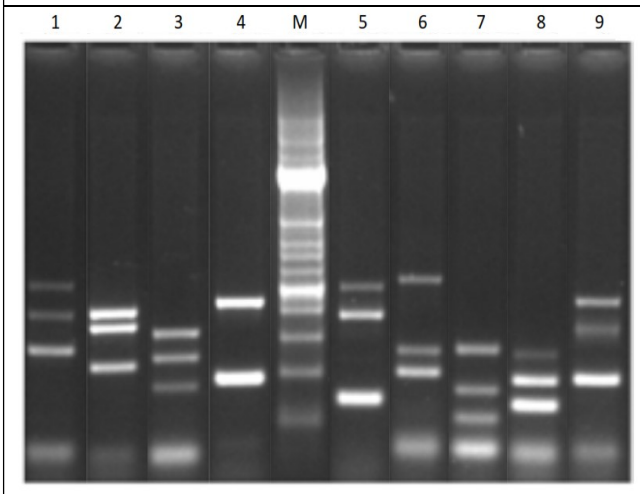
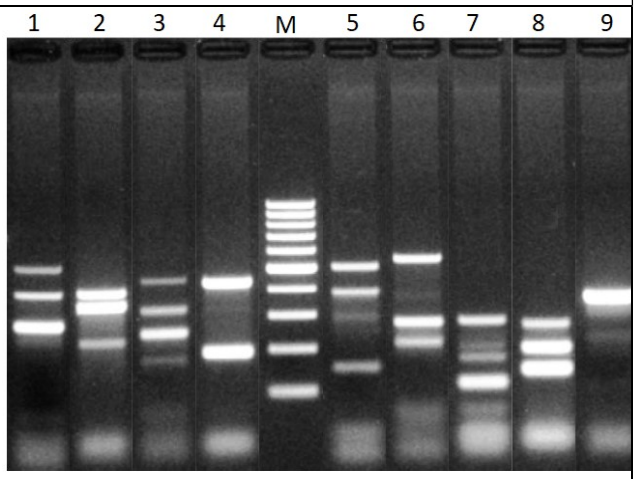
INDICATION	**Dystrophin Gene deletion was not observed in any of the exons studied for Duchenne Muscular Dystrophy**

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

PATIENT **ABHISHEK RATH**	*REFERENCE*
	

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.

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