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Department of Molecular Genetics & Genomics

Patient information	
Patient Name:	Master Mayesh

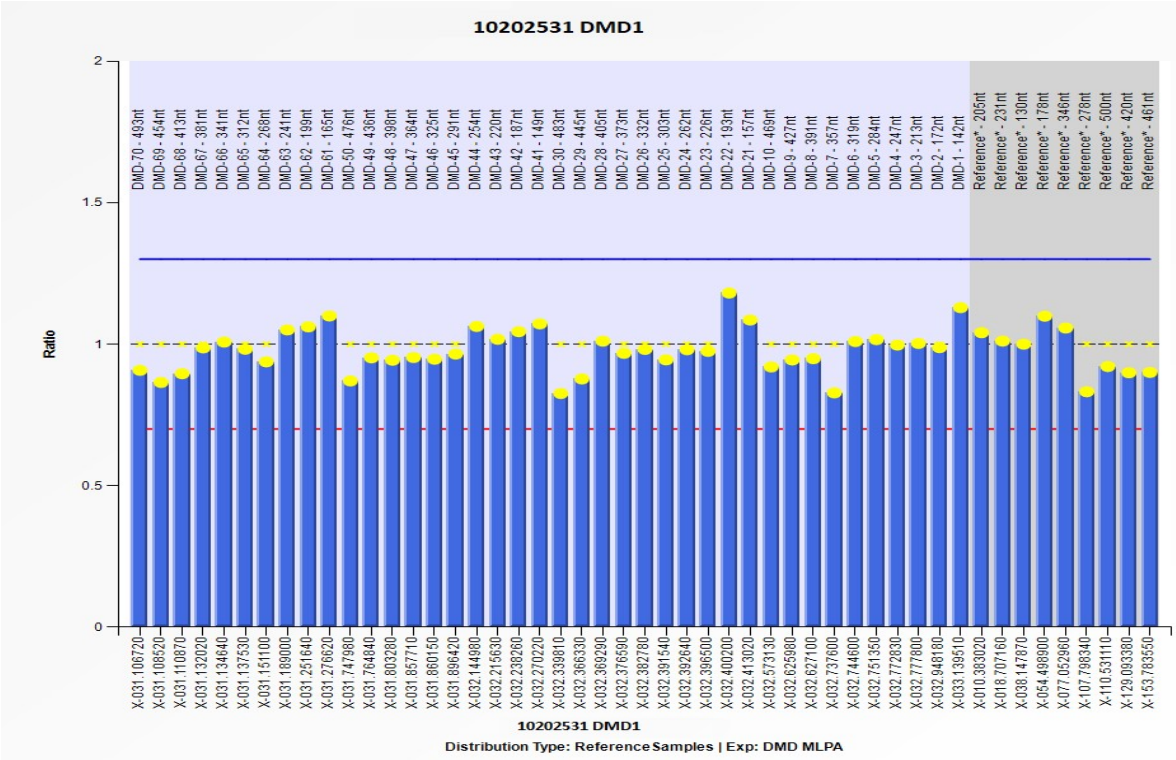
Referral Reason: Clinical Suspicion for DMD

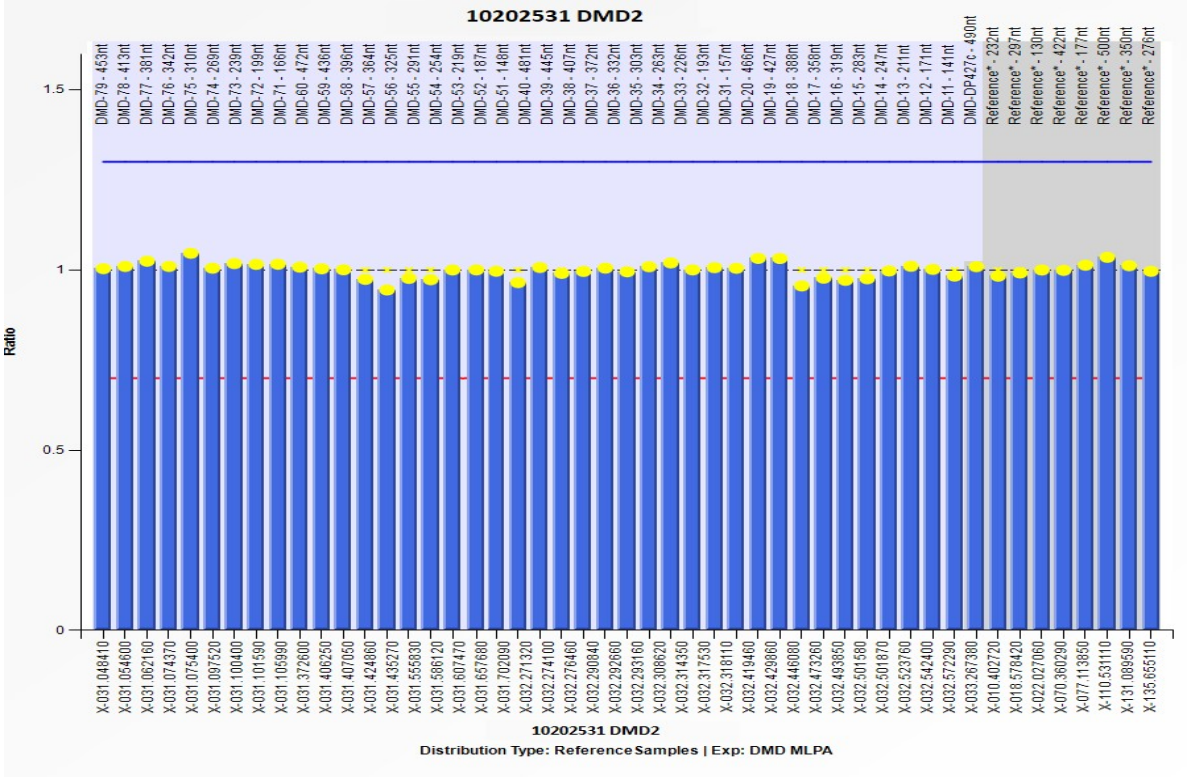
Test Result:

No deletion/duplications are detected in the DMD gene of this individual.

Interpretation: This individual is found to be normal for the deletions/duplications of all the 79 exons tested for DMD gene

Probe distribution ratio for 10202531





Recommendations:

- Genetic Counselling and clinical correlation is recommended
- Parental carrier testing

Methodology:

Multiplex Ligation-dependent probe amplification (MLPA) method is used for detection of deletion and duplication in 79 exons on DMD gene using SALSA MLPA probe mix available from MRC (Holland). Coffalyser.Net software is used for data analysis. The exon numbering used in this P034-B2 DMD-1 and P035-B1 DMD-2 product description is the exon numbering from the RefSeq transcript NM_004006.2, whichis identical to the LRG_199 sequence.

Introduction to DMD:

Duchenne muscular dystrophy (DMD) is a neuromuscular disease caused by a mutation of the dystrophin gene (DMD) (OMIM*300377) on Xp21. DMD gene encodes the protein called dystrophin that is a part of dystroglycan complex of the membrane that provides structural stability to muscle tissues. duplications, and point mutations at this gene locus may cause DMD, Becker muscular dystrophy or cardiomyopathy. The lack of functional dystrophin results in progressive damage and degeneration of muscle fibers characterized by abnormal dystrophin on muscle biopsy, abnormal electrocardiogram, calf and thigh cramping muscle pains, calf muscle pseudohypertrophy, dilated cardiomyopathy, flexion contractures, high serum creatine kinase, hyporeflexia, hypotonia, increased lordosis, positive Gowers sign, pulmonary hypoventilation, respiratory failure and scoliosis. Deletions and duplications in DMD gene account for 60–70% of the DMD cases.

Incidence: DMD: 1 in 3,500 male births
Inheritance: X-linked; de novo mutations occur in one-third of cases.
Penetrance: Males: 100 percent. Females: Varies with X-chromosome inactivation.
Cause: Pathogenic DMD mutations.
Clinical Sensitivity: DMD: 55-75 percent, BMD: 75-90 percent.

Limitations: *DMD* base pair substitutions, small deletions/duplications, deep intronic, and regulatory region mutations will not be detected. Breakpoints for large deletions/duplications will not be determined. Diagnostic errors can occur due to rare sequence variation.

Disclaimer:

- Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete.
- This test doesn't detect SNPS in the DMD gene.
- Although all precautions are taken while conducting these tests, there is a standard error rate of approximate 1% in all genetic tests and this should be taken into consideration before any clinical decision.
- It is presumed that the specimen used to perform the test belongs to the patient specified above, such verification having been carried out at the collection level of sample.
- Diagnostic errors can occur due to rare sequence variations. Regulatory region mutations, large gene deletions/duplications and some deep intronic mutations will not be detected.

References:

1. *Katharina J Hoff (2009): The effect of sequencing errors on metagenomic gene prediction.* BMCGenomics*, 10:520.*

2. *Hoffman, E. P. et al. Characterization of dystrophin in muscle-biopsy specimens from patients with Duchenne's or Becker's muscular dystrophy. N. Engl. J. Med. (1988). doi:10.1056/NEJM198805263182104*

3. *Muntoni F. et al. (1993). Deletion of the Dystrophin Muscle-Promotor region associated with X- linked dilated cardiomyopathy. N EnglJ Med 329:921-925.*

4. *Dastur RS et al. (2011). Identification of deletions and duplications in the DMD gene and female carrier status in western India using combined methods of multiplex polymerase chain reaction and multiplex ligation-dependent probe amplification. Neurol India 59:803-809.*

5. *McKusick V.A., Mendelian Inheritance in Man. A Catalog of Human Genes and Genetic Disorders. Baltimore: Johns Hopkins University Press (12th edition), 1998*

--- End of Report ---

Jagadeesh Babu S	**Dr. Naushad SM**	**Dr. Kiran Kumar**
Technical Lead -Genomics	Chief Scientific Officer	Consultant Genomics