Genetic Diagnostic and Research Laboratory
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CONFIDENTIAL MOLECULAR GENETIC LABORATORY REPORT.

Name of the patient	Nimal Silva	**Age/sex**	34/Male
Date of collection	Add Date	**Date of reporting,**	1/23/16 4:53 PM
Case ref no	Add Ref No	**Referred By**	Add Referred by
add Referred By : HD			

REPORT ON DUCHENNE MUSCULAR DYSTROPHY

Methodology:10 ml blood was collected from the patient and DNA was extracted from the buffy coat of the EDTA anticoagulated blood. The mutation was characterized by PCR using primers flanking the CAG repeat region and analysed on Gene Scan software.

Results and counseling: On analysis the patient showed

Exon Number	Result (+)/(-)	**Exon Number**	Result (+)/(-)
Ex 19	+	Ex 17	+
Ex 08	+	Ex 44	-
Ex 45	-	Ex 48	-
Ex 51	-	Ex 12	+
Ex 04	+	Pm	+
Ex 03	+	Ex 43	+
Ex 13	+	Ex 06	+
Ex 47	+	Ex 60	-
Ex 52	+	Ex 50	-
Ex 49	+	Ex 53	-

Interpretation:

The patient shows deletion in exons Ex 44, Ex 45, Ex 48, Ex 51, Ex 60,Ex 50, Ex 53,in the dystrophin gene.

Counseling:

1. Duchenne muscular dystrophy is a degenerative disease of muscle, caused due to mutations in Dystrophin gene on the X chromosome.
2. About 60-70% of cases of DMD, are associated with exon deletions whereas the remaining 30% have point mutations in dystrophin gene. The dystrophin gene has 79 exons. Of these, the first 20 exons and exon 45-55 are considered as hot-spots for mutations
3. Carrier detection in female siblings and mother is advised to prevent further occurrences of the disease.
4. However deletion/duplication of a single exon needs confirmation by some other molecular methods
5. Advised Genetic Counseling