



DNA TEST REPORT

IDS Mutation Investigational Analysis Report

Subject Information

Sample Information

Order no./Sample ID: 45024/130199;45025/130200

Sample Type: Blood (2.5 ml) in EDTA tube

Receipt Date & Time: 18th Dec., 2018; 10.08 AM

Order Date: 17th Jan., 2018

Clinical indications: The index patient, Mr. Suman Mallick presented with clinical indications of mild developmental delay, speech issues, coarse facies, hepatosplenomegaly, left inguinal hernia (operated), coronary heart disease (operated at 18 years for thickened aortic valve) and presence of diverticula in urinary bladder. Two of his younger brothers are similarly affected and have been diagnosed with mucopolysaccharidosis II (Hunter syndrome). Mr. Suman Mallick is suspected to be affected with mucopolysaccharidosis II (Hunter syndrome) or mucopolysaccharidosis IV (Maroteaux–Lamy syndrome). He was found to harbor a hemizygous variation in IDS gene. His siblings are being evaluated for the same variation.

Requested by: Dr. Sana Islam, Institute Of Child Health, Kolkata.

Report Date & Time: 19th Feb., 2018; 3.10 PM

Result Summary

Analysis for: Variation detected by Next Generation Sequencing in the IDS gene of Mr. Suman Mallick (130279)

Gene Name: IDS (Exon 9)

Sl. no.	Sample ID	Patient Name, Gender, Age	Relationship to index patient	Variation reported in the index patient	Variation detected in family member*	Clinical condition of family member
1.	130199	Master Biswajit Mallick, Male, 12yrs	Sibling	chrX:148564581_148564585del TCCTC; (HEMI) c.1345_1349del; p.Glu449SerfsTer6	Present (Hemizygous)	Symptomatic
2.	130200	Master Surojit Mallick, Male, 17yrs	Sibling	chrX:148564581_148564585del TCCTC; (HEMI) c.1345_1349del; p.Glu449SerfsTer6	Present (Hemizygous)	Symptomatic

* The variant analysis in Sanger sequencing is based on the IDS reference sequence nomenclature NG_011900.3 REGION: 4981..33345 in the NCBI GenBank (https://www.ncbi.nlm.nih.gov/nuccore/NG_011900.3?from=4981&to=33345&report=genbank) database. The exon number and nucleotide numbers will differ based on the reference file chosen and the database used. The cDNA-based allele calls (c.) for the variation and the corresponding amino acid change is based on IDS RefSeq sequence ENST00000340855.

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Test Details & Interpretation

Test

Exon 9 of *IDS* gene was PCR-amplified and the product was sequenced using Sanger sequencing. The detection limits of Sanger sequencing for presence of variation is ~12%. The sequence was aligned to available reference sequence NG_011900.3 (REGION: 4981..33345) in NCBI GenBank database to detect variation using variant analysis software programs.

Result

A hemizygous 5 base pair deletion in exon 9 of the *IDS* gene (**chrX:148564581_148564585delTCCTC; c.1345_1349del**) that results in a frameshift and premature truncation of the protein 6 amino acids downstream to codon 449 (**p.Glu449SerfsTer6**) was detected in the index patient, Mr. Suman Mallick (Sample ID: 130279) by NGS and was further validated by Sanger sequencing.

The same pathogenic variation was detected in hemizygous condition in the affected siblings of the index patient, Master Biswajit Mallick (Fig.1 A) and Master Surojit Mallick (Fig.1 B).

The variations detected in this test and their significance needs to be carefully correlated with the clinical indications of the individuals tested.

Report prepared by:

Results verified by:

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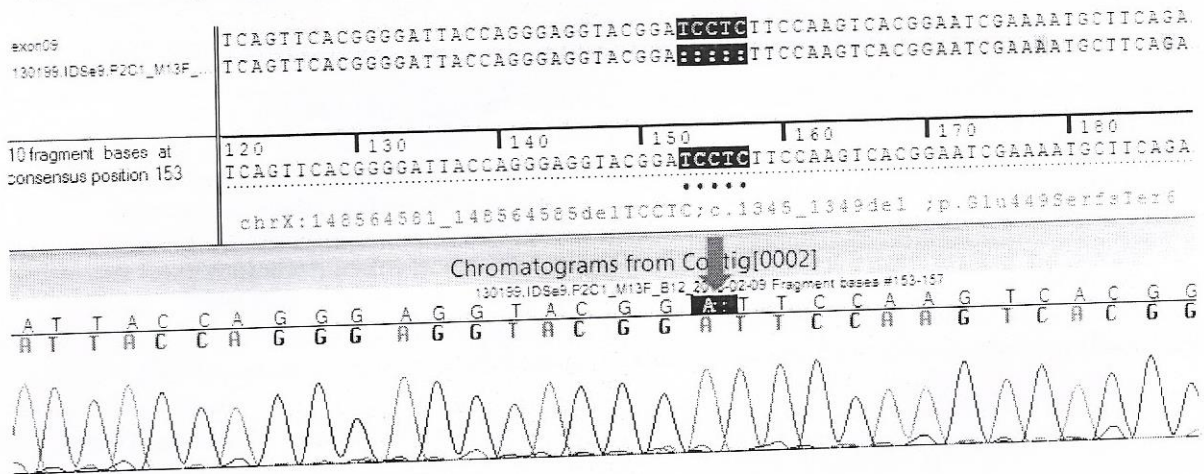
V L Ramprasad, PhD
Chief Operating Officer



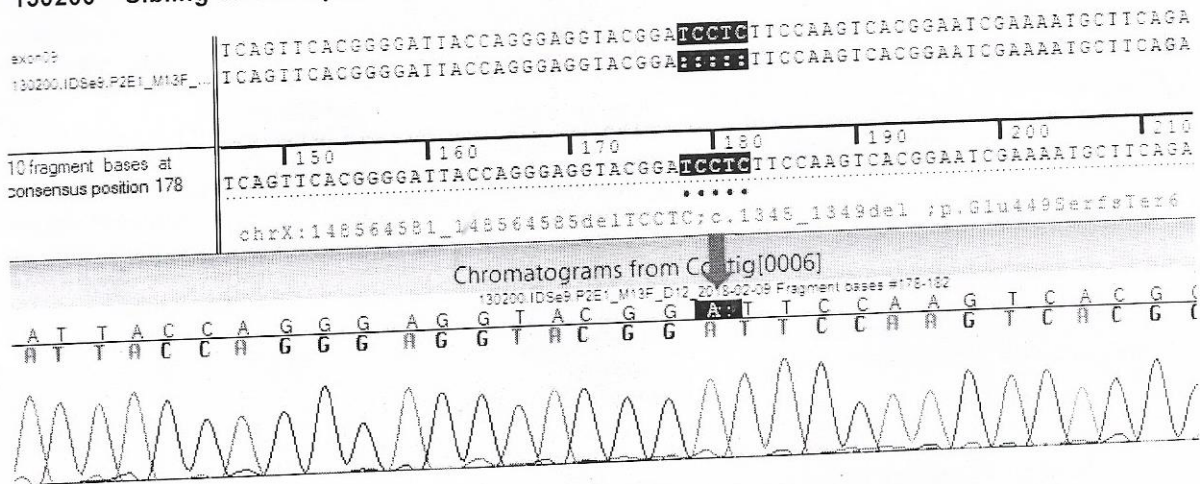
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Fig1: Sequence chromatogram and alignment to the reference sequence showing the variation in exon 9 of IDS gene (chrX:148564581_148564585delTCCTC; c.1345_1349del; p.Glu449SerfsTer6) detected in hemizygous condition in the siblings of index patient, Master Biswajit Mallick (A) and Master Surojit Mallick (B).

A. 130199 – Sibling of index patient (Master Biswajit Mallick)



B. 130200 – Sibling of index patient (Master Surojit Mallick)



End of Report