Biswajit

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MEDGENOME

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:	Results verified by:	Report released by:
Neetha A	**Lakshmi Mahadevan, PhD**	**Udhaya H. Kotecha, MD (Paediatrics),**	**V L Ramprasad, PhD**
Genome Analyst	Principal Scientist	Fellowship in Medical Genetics. Consultant - Clinical Geneticist	Chief Operating Officer

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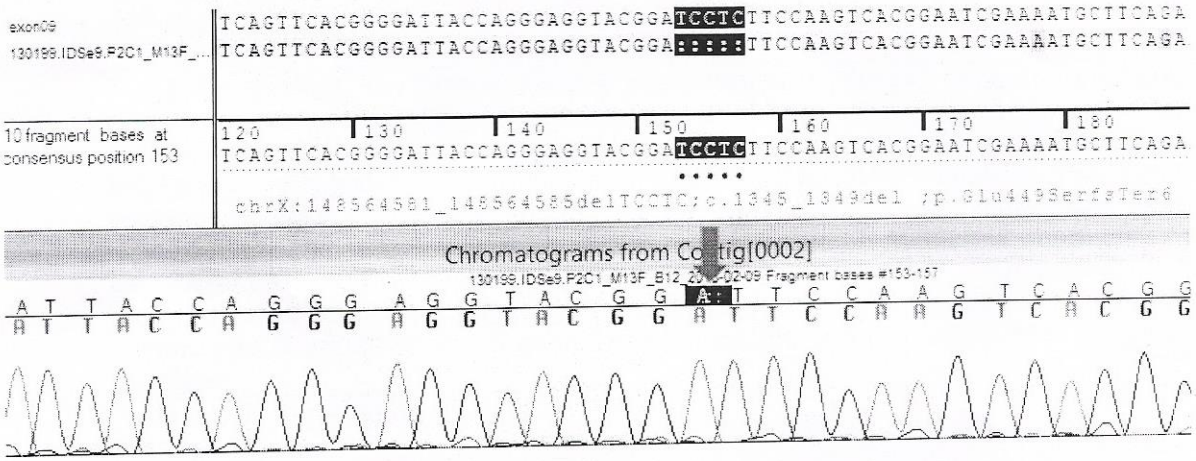
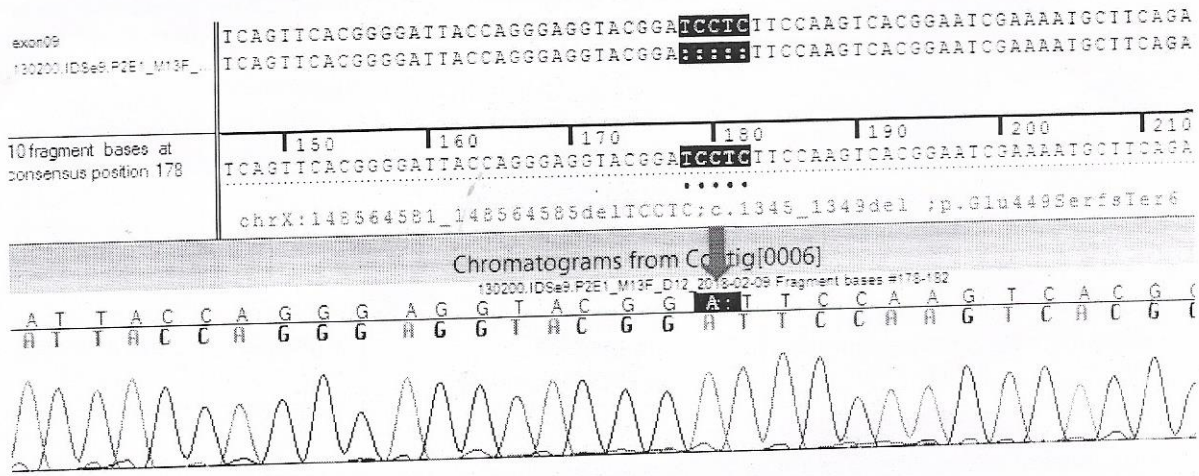
Fig1: Sequence chromatogram and alignment to the reference sequence showing the variation in exon 9 of *IDS* gene (chrX:148564581_148564585delTCCTC; c.1345_1349del; p.Glu449SerfsTer6) detected in hemizygous condition in the siblings of index patient, Master Biswajit Mallick (A) and Master Surojit Mallick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

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



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



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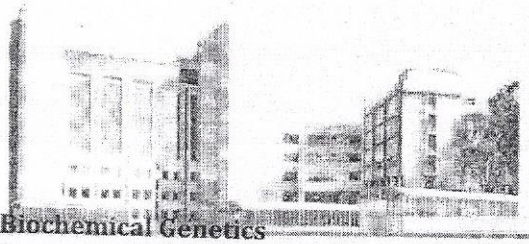

Sir Ganga Ram Hospital

Biochemical Genetics
Institute of Medical Genetics & Genomics
(An NABL Accredited Laboratory)

ERNDIM
QUALITY ASSURANCE IN LABORATORY TESTING FOR RMA
EQA Participant

Sponsored By: Genzyme
Sample Received: 9.11.17
Lab .No: 5546-17
Name: Suman Mallick
Referred by: Dr. Sana Islam
Clinical details: Available

Reporting Date: 11.11.17
Sample condition: visually ok
DOB/ Sex: 7.7.95/M
Specimen: Dried blood filter paper
Consent form: Available

Sample: Dried blood spots

Assay for α- Iduronidase enzyme (Hurler- MPS I) - Fluorometry method using artificial substrate

Patient	**2.7 nmol / hr/ml**

Biological reference intervals:

Normal subjects (n=25):	2.4 - 12.0 nmol/ hr/ ml
Patients (N=11):	< 0.68 nmol/hr/ ml

Comment: The value of alpha iduronidase enzyme is in the normal range.

Precautionary measures:

1. Blood specimen should be air dried at RT not by dryer/ blower. It should not be exposed to sunlight or high temperature.
2. The blood sample should be uniformly spread without caking / repeated spotting / droplets.
3. At least one circle should be saturated with blood sample on testing card.

Ms. Sandeepika Sharma
Tech assist.

Dr. Jyotsna Verma
Sr. Consultant

Dr. Ratna Puri
Chairperson

Biochemical Genetics laboratory, Tel. +91-11-42252112

*Although all precautions are taken during test, the currently available data indicate that technical error rate for biochemical assays is approx. 2%. It is important that all clinicians & persons requesting the tests are aware of these data before acting upon these results.
* *The results assume that all patient information provided are correct.*