



Sir Ganga Ram Hospital

Molecular Genetics
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2015- AUGUST

Molecular Lab No: 15F2511/15E587

Hosp No: 1699839, G15-3393
Sample collected: 03-08-15
Referred by: Dr RATNA PURI

Sample Type: Peripheral blood
Sample Received: 03-08-15

Date of Report: 19-08-15

Name: YASHVI BHARDWAJ

DOB: 20/10/10

Gender: M

C/O:

Indication: Clinically DMD, motor dev delay
Test requested: DYSTROPHIN GENE DEL/DUP

DELETION- DUPLICATION ANALYSIS OF DYSTROPHIN GENE

Test The DNA was tested for dosage analysis of 79 exons of the Dystrophin gene to investigate for deletion/ duplication in the gene.

Result A deletion in the dystrophin gene involving multiple exons (46-52) was observed (exons 45 and 53 were present)

Interpretation **This result is consistent with the diagnosis Dystrophinopathy** (out of frame deletion)

Recommendation

- # Prenatal diagnosis can be provided to family members at risk by testing for the deleted exons. This is done by chorionic villi sampling at 10-11 weeks of high-risk pregnancy.
- # Carrier screening can be done for female relatives of this patient.
- # Genetic counseling is advised

Please note:

- Although all precautions are taken during Molecular Genetic testing the currently available data indicate that the technical error rate for all types of Molecular DNA analysis is approximately 2%. It is important that all clinicians or persons requesting Molecular Genetic diagnostic tests are aware of these data before acting upon these results.
- The results assume that all patient information provided is correct.

Consultant / Scientist

Senior Consultant

Senior Consultant & Director

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End of Report