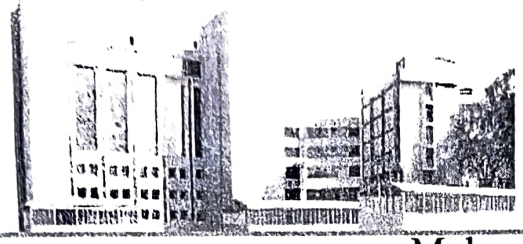

Sir Ganga Ram Hospital

Molecular Genetics

Institute of Medical Genetics & Genomics
Phone No: 91-11-42252115, Fax 91 11 25862206

Molecular Lab No: 18F1782/18D126

2018-May

Hosp No:
Sample Type: Peripheral Blood
Sample collected: 30-05-18 Sample Received: 31-05-18
Date of Report: 23-06-18
Referred by: Dr. SHEELA NAMPOOTHIRI

Name: B/O ASWATHY DOB: 20/12/2016 Gender: M

C/O:

Indication: hand flapping, seizures, generalized tremulousness
Test Requested: Angelman Syndrome

MOLECULAR DIAGNOSIS OF ANGELMAN SYNDROME

Test	The genomic DNA was analyzed copy number changes as well as to analyse the CpG island methylation of the PWS/AS region at 15q11 region using SALSA MS-MLPA probemix ME028 from MRC Holland	
Result	Micro deletion 15q11	Positive
	SNRPN gene	Hypomethylated
Interpretation	**This is consistent with the diagnosis of Angelman syndrome** due to microdeletion of chromosome 15.	
Recommendation	These results should be correlated with clinical and other laboratory findings.	
	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**

End of Report

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