Molecular Genetics
Institute of Medical Genetics & Genomics
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2025-September **Molecular Lab No: 25F2902/25H270**
Hosp. No: 3627429 Sample Type: Peripheral Blood
Sample collected: 23-09-2025 Sample Received: 23-09-2025 Date of Report: 04-10-2025
Referred by: Dr PRAVEEN SUMAN

Name: KABBIR GULATI DOB:24-03-25 Gender: M

Indication: Floppy infant
Test Requested: MLPA for Spinal muscular atrophy

MOLECULAR GENETIC ANALYSIS FOR SPINAL MUSCULAR ATROPHY (SMA)

Test: The genomic DNA sample of the proband was analysed for the dosage analysis of exons 7 and 8 of the SMN 1 gene and SMN2 gene using Salsa MLPA kit P060 kit from MRC Holland

Results:

Gene	Exon 7 copy number	Exon 8 copy number
SMN1 gene	Zero	Zero
SMN2 gene	Two	Two

Interpretation This confirms the diagnosis of Spinal Muscular Atrophy due to homozygous deletion of exon 7 of SMN1 gene in the proband.

Recommendation

The family carries a 25% risk of having a child with Spinal Muscular Atrophy in every pregnancy. Prenatal Diagnosis can be provided by Chorionic Villi Sampling at 10 -11 weeks of gestation.

Carrier screening can be done for extended family members

Medical consultation is advised- please contact your Doctor as soon as possible for therapeutic options

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.

Dr R Saxena	**Dr S Dubey**	**Dr S Kohli**	**Dr D Gupta**	**Dr Ratna D Puri**
Sr Consultant	*Sr Consultant*	*Sr Consultant*	*Scientist*	*Sr Consultant & Director*

End of Report