

# INVITAE MOLECULAR GENETICS LABORATORY

Molecular Diagnostics Division  
CLIA: 05D2070100 | CAP: 9382104-01 | State Licensed

## GENETIC TESTING REPORT

Patient:	Sofia Reyes
DOB:	2026-01-15
MRN:	SOFIA_R
Test Date:	2026-01-28
Test Name:	SMN1/SMN2 Copy Number Analysis
Methodology:	MLPA (Multiplex Ligation-dependent Probe Amplification)
Ordering Physician:	Dr. Michael Torres

## RESULTS

Gene/Marker	Result
SMN1 Exon 7 Copy Number	0 copies
SMN1 Mutation Type	Homozygous deletion of SMN1 exon 7
SMN2 Copy Number	4 copies

## INTERPRETATION

Biallelic SMN1 deletion confirmed. 4 copies of SMN2 detected. Higher SMN2 copy number is generally associated with milder phenotype (SMA Type 3 or Type 4), though clinical correlation is necessary. Presymptomatic treatment is recommended based on genetic diagnosis regardless of predicted phenotype.