

INVITAE MOLECULAR GENETICS LABORATORY

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

GENETIC TESTING REPORT

Patient:	Aiden Foster
DOB:	2022-03-10
MRN:	AIDEN_F
Test Date:	2022-12-15
Test Name:	SMN1/SMN2 Copy Number Analysis
Methodology:	MLPA (Multiplex Ligation-dependent Probe Amplification)
Ordering Physician:	Dr. Amanda Liu

RESULTS

Gene/Marker	Result
SMN1 Exon 7 Copy Number	0 copies
SMN1 Mutation Type	Homozygous deletion of SMN1 exon 7
SMN2 Copy Number	3 copies

INTERPRETATION

Biallelic SMN1 deletion confirmed. 3 copies of SMN2 detected. Clinical presentation consistent with SMA Type 2.

Laboratory Director: Molecular Genetics Director, MD, PhD, FACMG
Report finalized: 12/15/2022 | This report is confidential and intended only for the ordering provider.