

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.