INVITAE MOLECULAR GENETICS LABORATORY

Molecular Diagnostics Division

CLIA: 05D2070100 | CAP: 9382104-01 | State Licensed

GENETIC TESTING REPORT

Patient:	Ethan Williams
DOB:	2025-06-10
MRN:	ETHAN_W
Test Date:	2025-09-15
Test Name:	SMN1/SMN2 Copy Number Analysis
Methodology:	MLPA (Multiplex Ligation-dependent Probe Amplification)
Ordering Physician:	Dr. Anna Kowalski

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

Consistent with diagnosis of Spinal Muscular Atrophy. Homozygous SMN1 deletion confirmed. 3 copies of SMN2 detected — associated with SMA Type 1 or Type 2 phenotype.

Laboratory Director: Molecular Genetics Director, MD, PhD, FACMG

Report finalized: 09/15/2025 | This report is confidential and intended only for the ordering provider.