

Report Text for Newborn Screening Reports-SMA

State Hygienic Laboratory at The University of Iowa

Disorder	Substance Measured	Result Interpretation	Test Trailer/Comment
Spinal Muscular Atrophy	SMN1 exon 7	Within Normal Limits	
		Presumptive Positive	A program medical consultant will be contacting you regarding the abnormal test results. In Addition to their recommendation, please resubmit a repeat blood spot
		Indeterminate-Resubmit	Resubmit: Collect another complete filter paper specimen and send to INMSP. If further action is required a program medical consultant will contact you.
		Poor Quality-Resubmit	Resubmit: Collect another complete filter paper specimen and send to INMSP. If further action is required a program medical consultant will contact you.

Report Trailer

Spinal Muscular Atrophy screening uses a deletion in exon 7 of the survival motor neuron 1 (SMN1) gene to assess risk. 95% of SMA cases are caused by deletion of SMN1 exon 7. Thus, the screening assay may miss up to 5% of SMA cases that are not caused by SMN1 exon 7 deletion.