

Table 12: SMA

Overall Result	Disorder	Screening Result	Screening Result Note
Normal	SMA	Normal	
Abnormal	SMA	Abnormal	Probable Spinal Muscular Atrophy. Deletion of SMN1 exon 7 detected. Zero copies of SMN2 detected. Recommend rapid molecular confirmation including SMN1 and SMN2 copy number and telephone consultation and referral to a neurologist or neurogeneticist within 24 hours.
Abnormal	SMA	Abnormal	Probable Spinal Muscular Atrophy. Deletion of SMN1 exon 7 detected. One copy of SMN2 detected. Recommend rapid molecular confirmation including SMN1 and SMN2 copy number and telephone consultation and referral to a neurologist or neurogeneticist within 24 hours.
Abnormal	SMA	Abnormal	Probable Spinal Muscular Atrophy. Deletion of SMN1 exon 7 detected. Two copies of SMN2 detected. Recommend rapid molecular confirmation including SMN1 and SMN2 copy number and telephone consultation and referral to a neurologist or neurogeneticist within 24 hours.
Abnormal	SMA	Abnormal	Probable Spinal Muscular Atrophy. Deletion of SMN1 exon 7 detected. Three copies of SMN2 detected. Recommend rapid molecular confirmation including SMN1 and SMN2 copy number and telephone consultation and referral to a neurologist or neurogeneticist within 24 hours.
Abnormal	SMA	Abnormal	Probable Spinal Muscular Atrophy. Deletion of SMN1 exon 7 detected. Four copies of SMN2 detected. Recommend rapid molecular confirmation including SMN1 and SMN2 copy number and telephone consultation and referral to a neurologist or neurogeneticist within 72 hours.
Abnormal	SMA	Abnormal	Probable Spinal Muscular Atrophy. Deletion of SMN1 exon 7 detected. Four or more copies of SMN2 detected. Recommend rapid molecular confirmation including SMN1 and SMN2 copy number and telephone consultation and referral to a neurologist or neurogeneticist within 72 hours.
Abnormal	SMA	Abnormal	Probable Spinal Muscular Atrophy. Deletion of SMN1 exon 7 detected. Five or more copies of SMN2 detected. Recommend rapid molecular confirmation including SMN1 and SMN2 copy number within 24 hours and referral to a neurologist or neurogeneticist within 7 days.
Abnormal	SMA	Abnormal	Probable Spinal Muscular Atrophy. Deletion of SMN1 exon 7 detected. SMN2 copy number cannot be determined. Recommend rapid molecular confirmation including SMN1 and SMN2 copy number and telephone consultation and referral to a neurologist or neurogeneticist within 24 hours.
Abnormal	SMA	Abnormal	Probable Spinal Muscular Atrophy. Deletion of SMN1 exon 7 detected. This result is consistent with the previous newborn screening specimen results for the same baby. If not already completed, recommend rapid molecular confirmation including SMN1 and SMN2 copy number and telephone consultation and referral to a neurologist or neurogeneticist within 24 hours.
Abnormal	SMA	Abnormal	free text
Normal	SMA	Revised Result	free text
Abnormal	SMA	Revised Result	free text
Unsatisfactory	SMA	Revised Result	free text