

Table 12: SMA

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte 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.

Table 12: SMA

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
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			free text
Normal	SMA	Revised Result			free text
Abnormal	SMA	Revised Result			free text
Unsatisfactory	SMA	Revised Result			free text