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Service information: Spinal Muscular Atrophy (SMA)

Gene/Locus: *SMN1* (5q13.2) **OMIM:** #253300 (SMA1), #253550 (SMA2), #253400 (SMA3) #271150 (SMA4) (*600354 – *SMN1* gene)

Referrals: Clinical Geneticists, Neurologists, Paediatric Neurologists.
Testing: Diagnostic testing offered in clinically affected patients.
Target Reporting Times:

• Routine diagnostic	28 calendar days	
• Diagnostic testing in neonates	5 working days	
• Carrier testing	28 calendar days	From Clinical Genetics only
• Urgent testing*	3 calendar days	Prenatal diagnosis/carrier testing affecting pregnancy

***Please contact the laboratory for urgent testing.**

Test Details:

- **Diagnostic testing: Multiplex ligation-dependent probe amplification (MLPA analysis)** to detect homozygous and heterozygous deletions of exons 7 and 8 of the *SMN1* gene (MRC Holland P060 kit).
- If a single copy of *SMN1* exon 7 is detected by MLPA, DNA can then be sent to another laboratory for *SMN1* point mutation testing. The funding for this testing will need to be provided by your department.
- **Carrier testing: Multiplex ligation-dependent probe amplification (MLPA analysis)** to detect heterozygous deletions of exons 7 and 8 of the *SMN1* gene (MRC Holland P060 kit).
- **Prenatal diagnostic testing: PCR and restriction digest analysis of *SMN1* exon 7 and 8** to detect homozygous deletions of exon 7 of the *SMN1* gene.

Information:
Homozygous deletions of *SMN1* exon 7 are predicted to detect >95% cases of SMA, regardless of clinical severity (Lefebvre et al. Cell (1995) 80:155-165). Extended testing including *SMN1* sequencing analysis is predicted to detect a further 4% of SMA cases.

Sample Requirements:

- **EDTA** blood sample (1-4 ml), labelled with patient's **full name**, **date of birth** and **NHS number**, or genomic DNA (≥20 μl at ≥50 ng/μl)
- Samples should be accompanied by a **fully** completed referral card which should include the patient's full name, date of birth and NHS number.
- Please also include details of the test, relevant clinical details and full details of the referring clinician/centre.

Consent:
Please note that in submitting a sample, it is the responsibility of the clinician to ensure that consent has been taken i) for testing, ii) for storage, and iii) for the use of this sample and the information generated to be shared with the patient's relatives and their health professionals. Following testing, a sample of the patient's DNA may also be used anonymously to validate new tests and for internal quality control purposes.

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