Newborn Screen Follow-up for Spinal Muscular Atrophy

SMNDX / Spinal Muscular Atrophy Diagnostic Assay, Deletion/Duplication Analysis, Varies
Includes exon 7 analysis for SMN1 and SMN2

- SMN1 zero functional copies
  - SMN2 zero to 2 functional copies
    - Spinal muscular atrophy (SMA) type 1 confirmed
      - Refer to specialist. Treatment available for all SMA types
  - SMN2 three or more functional copies
    - Intermediate to mild SMA

- SMN1 one or more functional copies
  - SMA unlikely. However, 5% of patients are undetectable by this method.