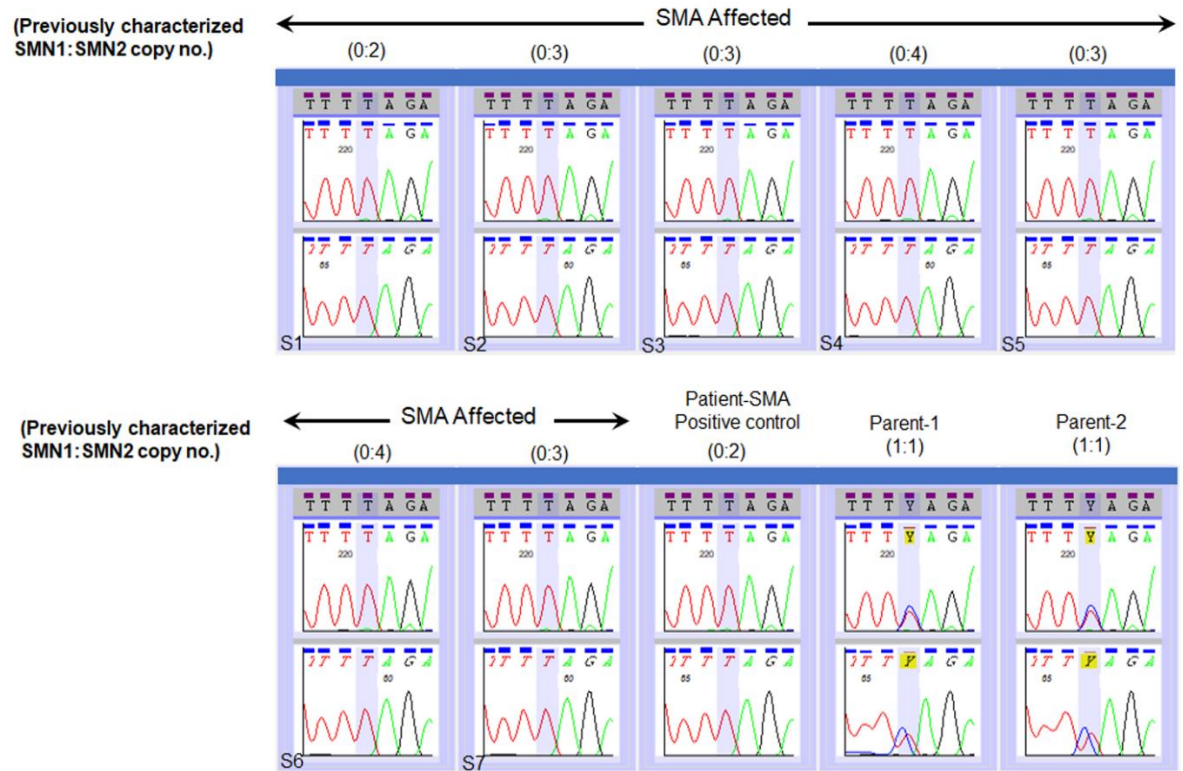
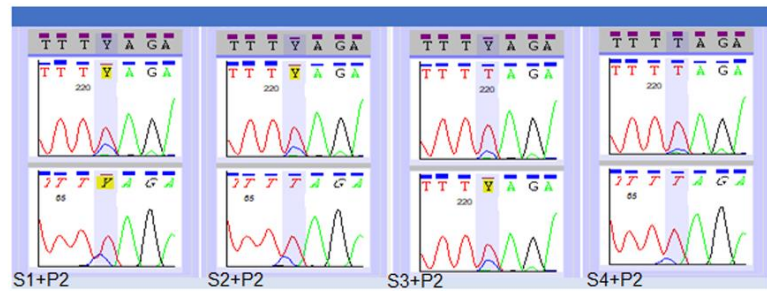


## Supplementary Materials:

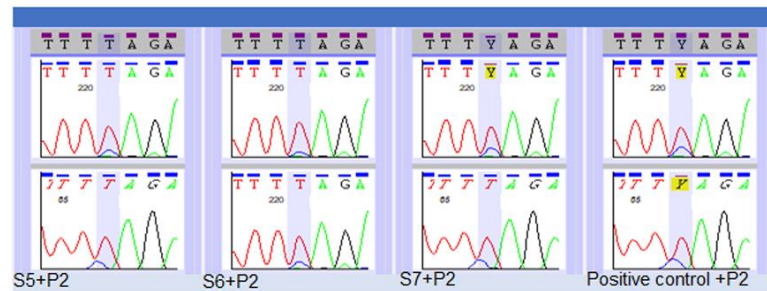


**Figure S1:** Genotype confirmation on the specimens subjected for *SMN2* copy validation. Specimens subjected to the validation study for *SMN2* copy were first confirmed for their genotype using Sanger sequencing as described in methods.

(Previous characterized SMN1:SMN2 copy no.)	(0:2)	(0:3)	(0:3)	(0:4)
Average Copy number by new assay	(0:1.9)	(0:2.8)	(0:3.2)	(0:4 or more)



(Previous characterized SMN1:SMN2 copy no.)	(0:3)	(0:4)	(0:3)	(0:2)
Average Copy number by new assay	(0:3.2)	(0:4 or more)	(0:2.8)	(0:1.9)



**Figure S2:** Determination of *SMN2* copy number assay validation using SMA affected specimens using another spike DNA (parent 2). DNA from SMA affected specimens and a Coriell DNA (positive control) were mixed in equal concentration with spike DNA, and bidirectional sequencing was performed. *SMN1* gene sequence used as reference sequence, and data were analyzed for determine the *SMN2* copy number in the test specimens as described in the methods section.