

## SMN1 and SMN2 Copy Number, Silent Carrier, and Disease Modifier Genotyping—in Less Than Four Hours

Copy number variations in SMN1 and SMN2 are, respectively, associated with the onset and severity of spinal muscular atrophy (SMA), a debilitating and life-threatening illness of the central nervous system. Recent studies have demonstrated that transmission risk and disease severity may be impacted by the presence of additional variants, such as SMN1 gene duplication events and disease modifier in SMN2.

The AmplideX® PCR/CE SMN1/2 Plus Kit\* revolutionizes the analysis of these two genes by delivering comprehensive results in less than four hours. Powered by AmplideX technology, the assay accurately quantifies SMN1 and SMN2 exon 7 copy number and also detects SMN1 gene duplication and SMN2 disease modifier variants - all from a single reaction. The assay shares a common workflow with other assays in the AmplideX product portfolio and is optimized for use on widely established laboratory equipment.

## REDUCED COMPLEXITY OPTIMIZED WORKFLOW

- Similar workflow to AmplideX PCR/CE FMR1<sup>+</sup> kit eases implementation and training
- Multiplexed, scalable design allows analysis of singlenucleotide variants, small indels, and copy-number changes from a single PCR reaction
- Assay-specific software automates variant calls and streamlines data analysis
- DNA-to-data in less than four hours with only 60 minutes of hands-on-time
- Optimized for use on commonly installed CE equipment
- Fully-kitted solution sourced from a single vendor

## **QUALITY PERFORMANCE**

- Ability to differentiate between 0, 1, 2, 3 and ≥4 copies for both SMN1 and SMN2
- Automated variant and copy-number genotyping
- Accuracy demonstrated through comparisons with multiple orthogonal methods









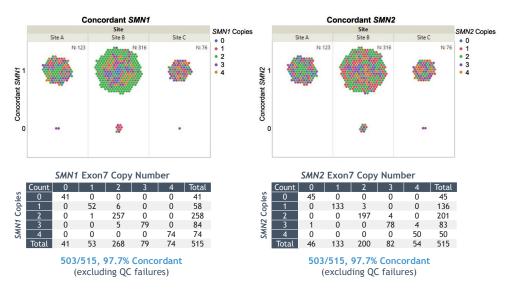
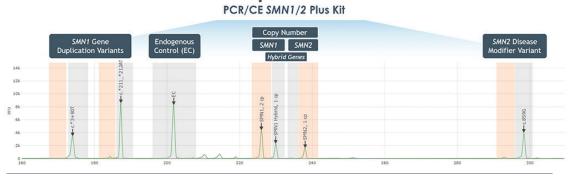


Figure 2. Excellent concordance of SMN1 and SMN2 copy number across sites for 500+ measurements using the AmplideX PCR/CE SMN1/2 Plus Kit\*

**AmplideX**°



**Figure 3.** Example Electropherogram Output - One reaction provides information on SMN1 and SMN2 copy number, disease modifier variant and gene duplication variants

Product Name	Number of Reactions	Catalog Number
AmplideX® PCR/CE SMN1/2 Plus Kit (RUO)	50	A00050
AmplideX® PCR/CE SMN1/2 Plus Kit (RUO)	100	A00054



 $<sup>^{\</sup>star}$  For Research Use Only. Not for use in diagnostic procedures. † CE-IVD for US Export Only.

