

# AmplideX<sup>®</sup>

## PCR/CE SMN1/2 Plus Kit\*

### 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<sup>®</sup> 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

- Similar workflow to AmplideX PCR/CE *FMR1*<sup>††</sup> kit eases implementation and training
- Multiplexed, scalable design allows analysis of single-nucleotide variants, small indels, and copy-number changes from a single PCR reaction
- Assay-specific software automates variant calls and streamlines data analysis

#### OPTIMIZED WORKFLOW

- 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

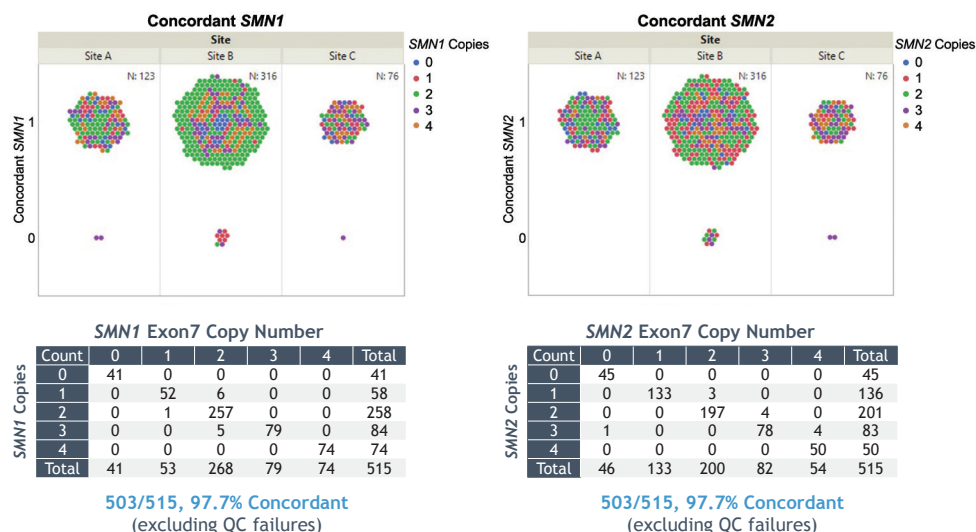
- Ability to differentiate between 0, 1, 2, 3 and  $\geq 4$  copies for both *SMN1* and *SMN2*
- Automated variant and copy-number genotyping
- Accuracy demonstrated through comparisons with multiple orthogonal methods



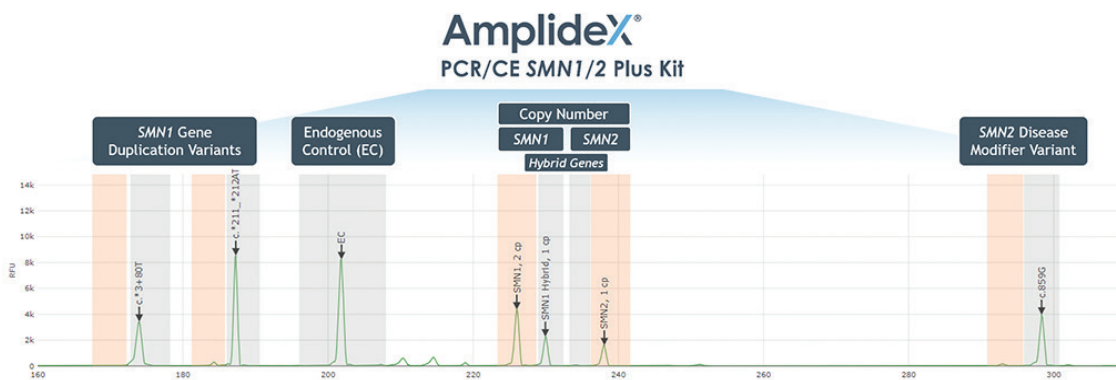
Figure 1. Assay workflow for AmplideX PCR/CE *SMN1/2* Plus Kit\*

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## PCR/CE SMN1/2 Plus Kit \*



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



**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 <sup>®</sup> PCR/CE <i>SMN1/2</i> Plus Kit (RUO)	50	A00050
AmplideX <sup>®</sup> PCR/CE <i>SMN1/2</i> Plus Kit (RUO)	100	A00054



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† CE-IVD for US Export Only.