

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

- Similar workflow to AmplideX PCR/CE *FMR1*⁺ 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

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 ≥4 copies for both *SMN1* and *SMN2*
- Automated variant and copy-number genotyping
- Accuracy demonstrated through comparisons with multiple orthogonal methods





Constant Improvement

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

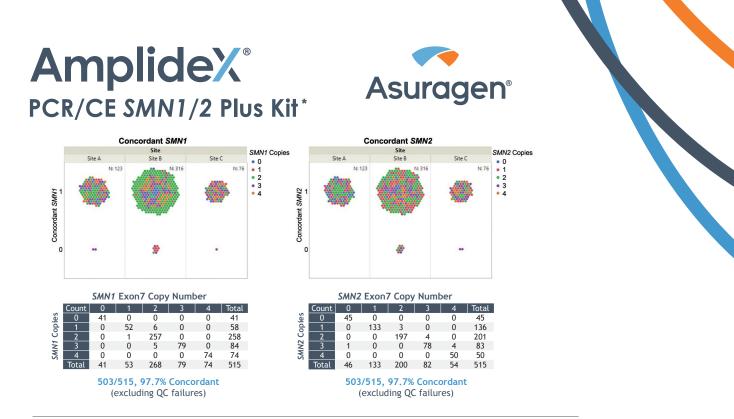


Figure 2. Excellent concordance of SMN1 and SMN2 copy number across sites for 500+ measurements using the AmplideX PCR/CE SMN1/2 Plus Kit*

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

* For Research Use Only. Not for use in diagnostic procedures. † CE-IVD for US Export Only.

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

