

# AmplideX<sup>®</sup>

## SMA Plus Kit\*



Spinal muscular atrophy (SMA) is a debilitating illness of the central nervous system and is a leading genetic cause of infant death. The disease is associated with a loss of functional copies of the *SMN1* gene and its severity is, largely, inversely correlated to the number of copies of *SMN2*. The availability of novel treatments requires early diagnosis to effectively combat the disease. Furthermore, nearly 1 in 50 people are asymptomatic carriers of SMA, prompting leading clinical organizations to recommend carrier screening for all populations.

The AmplideX<sup>®</sup> SMA Plus Kit is an *in vitro* nucleic acid amplification kit intended to aid in the screening of carriers of and diagnosis of spinal muscular atrophy (SMA). The kit quantifies the number of copies of exon 7 of both *SMN1* and *SMN2* reported as 0, 1, 2, 3, or  $\geq 4$  genomic copies. The assay also detects variants *SMN1* c.\*3+80T>G and *SMN1* c.\*211\_\*212del, which are associated with *SMN1* gene duplication and “silent carrier” status, as well as variant *SMN2* c.859G>C, which is associated with a milder disease phenotype.



### REDUCED COMPLEXITY

#### Ease of data analysis and reporting

- One kit to identify SMA patients, carriers (including detection of variants associated with silent carriers), and refine disease prognosis - all from a single PCR reaction
- Similar workflow to AmplideX PCR/CE *FMR1*\*† kit eases implementation and training
- Assay-specific software automates results reporting and streamlines data analysis

### OPTIMIZED WORKFLOW

#### Reduces valuable operator hands-on-time and overall turnaround time

- Diagnostic and screening results are reported in less than four hours with only 60 minutes of hands-on-time
- Scalable workflow supports high sample throughput testing
- Optimized for use on widely installed CE equipment
- Fully-kitted solution sourced from a single vendor

### QUALITY PERFORMANCE

#### Comprehensive analysis of *SMN1* and *SMN2* genes for diagnosis and screening of SMA

- High resolution of *SMN1/2* copy number across a broad range improves accuracy in identifying SMA patients and carriers
- Excellent concordance of copy number and variant results compared to multiple orthogonal test methods





# AmplideX® SMA Plus Kit\*

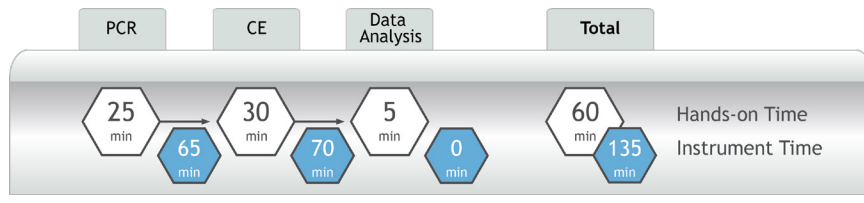


Figure 1. Assay workflow for AmplideX SMA Plus Kit\*

## 120+ Samples

AmplideX vs	MLPA	AmplideX vs	ddPCR	AmplideX vs	MLPA AND	ddPCR
<i>SMN1</i>	<i>SMN2</i>	<i>SMN1</i>	<i>SMN2</i>	<i>SMN1</i>	<i>SMN2</i>	
126/128	126/128	122/123	119/121	121/122	119/121	
98.4%	98.4%	99.2%	98.3%	99.2%	98.3%	

Figure 2. Excellent concordance of results between the AmplideX SMA Plus Kit\* and other methodologies, including MLPA and ddPCR, across more than 120 unique blood and cell line samples

## AmplideX® SMA Plus Kit

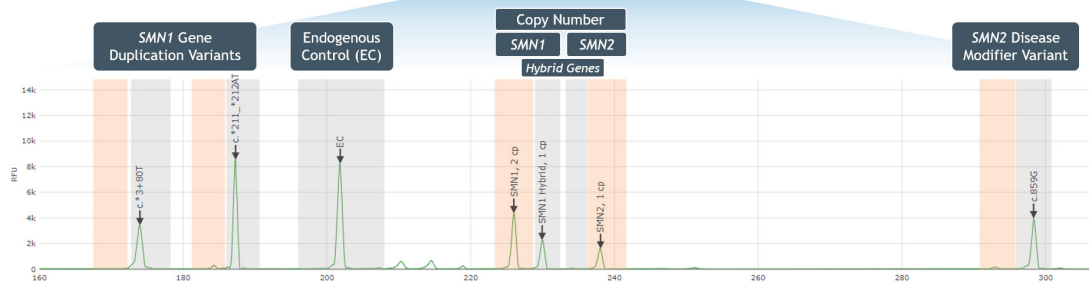


Figure 3. AmplideX SMA Plus Kit\* Example Electropherogram Output - One reaction provides information on *SMN1* and *SMN2* copy number, Disease Modifier Variant and Gene Duplication Variants

Product Name	Number of Reactions	Catalog Number
AmplideX SMA Plus Kit*	50	A00055
AmplideX SMA Plus Kit*	100	A00056

2000-0142 rev. A

\* CE-IVD for US Export Only

† For Research Use Only. Not for use in diagnostic procedures.