# AmplideX®

## 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® 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 ≥ 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

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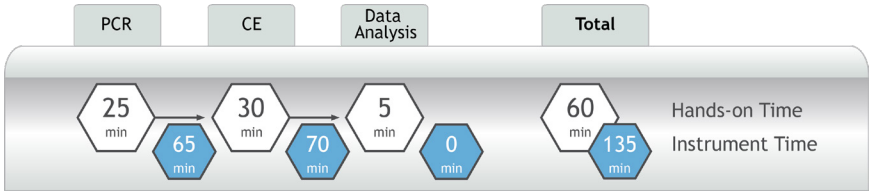


**Figure 1.** Assay workflow for AmplideX SMA Plus Kit*

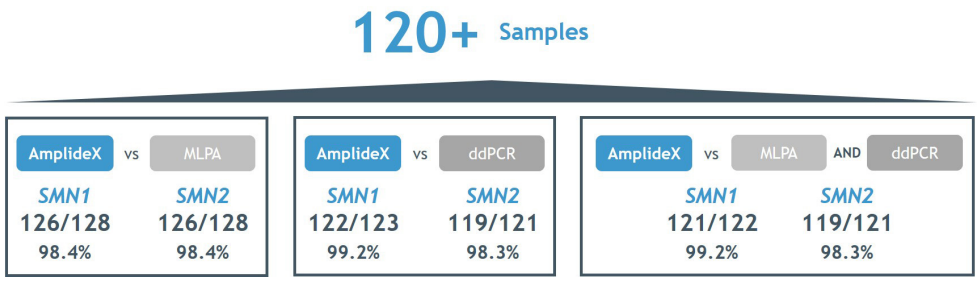


**Figure 2.** Excellent concordance of results between the AmplideX SMA Plus Kit* and other methodologies, including MLPA and ddPCR, across more than 120 unique blood and cell line samples

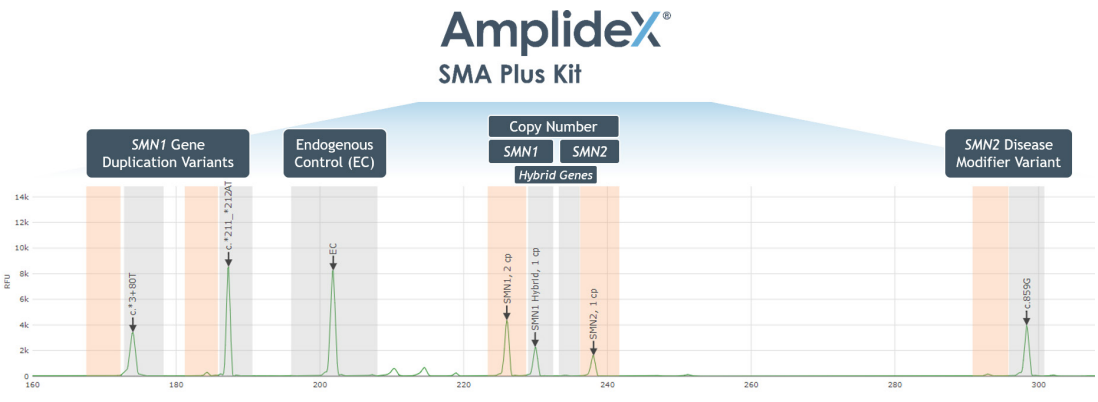


**Figure 3.** AmplideX SMA Plus Kit* Example Electropherogram Output - One reaction provides information on *SMN1* and *SMN2* copy number, Disease Modifier Variant and Gene Duplication Variants

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

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