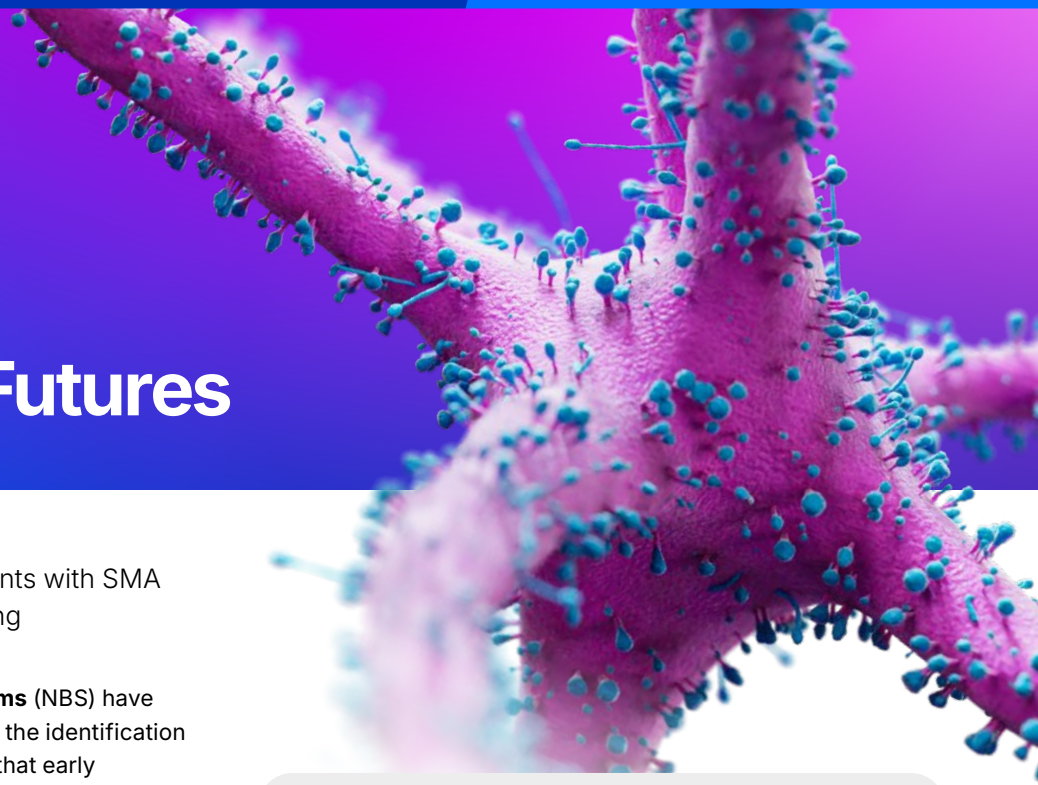
bio-techne diagnostics™



Empowering Futures

Enabling Accurate Diagnosis of Patients with SMA Identified through Newborn Screening

Newborn screening programs (NBS) have been deployed to accelerate the identification of children with SMA, given that early diagnosis and treatment leads to significantly better health outcomes

~97–98% of patients with SMA can be diagnosed with a simple qualitative test, yet *SMN2* copy number assessment requires **quantitative** methodologies that are not easily implemented in most laboratories[1]

The SMA-testing community has reported **~40%** discordance between laboratories in the quantification of *SMN2* copy numbers[2]

Alignment of clinical practices for *SMN2* copy number determination is needed to assist in accurate diagnosis and optimal patient care. Accurate characterisation of *SMN2* copies is critical for treatment decision making.

Currently in wider Europe, 83% of newborns are screened for SMA.[3] As the NBS programs get established, more labs will be performing molecular testing for SMA, thus highlighting the need for a robust and streamlined test.

SMA NBS Alliance

SMN2 Testing Challenges

- Accurate *SMN2* copy number determination is correlated with disease severity, informative for diagnosis
- Reporting of disease-severity modifying variants:[4] These can account for 40% of discrepancies between *SMN2* copy number and expected phenotype[5]

***SMN2* Copy Number**	**c.859G>C Variant Status**	**Interpretation**
1	Negative	SMA (Type 0 probable)
2	Negative	SMA (Type 1 probable)
2	Detected in one copy	SMA (Type 2/3 probable)
2	Detected in two copies	SMA (Type 3/4 probable)
3	Negative	SMA (Type 2/3 probable)
3	Detected in one copy	SMA (Type 3 probable)
≥4	Negative	SMA (Type 3/4 probable)

It is recommended that *SMN2* copy number is evaluated in conjunction with disease-modifying variants to refine prognosis[6]

Key Strengths of AmplideX® SMA Plus Kit*

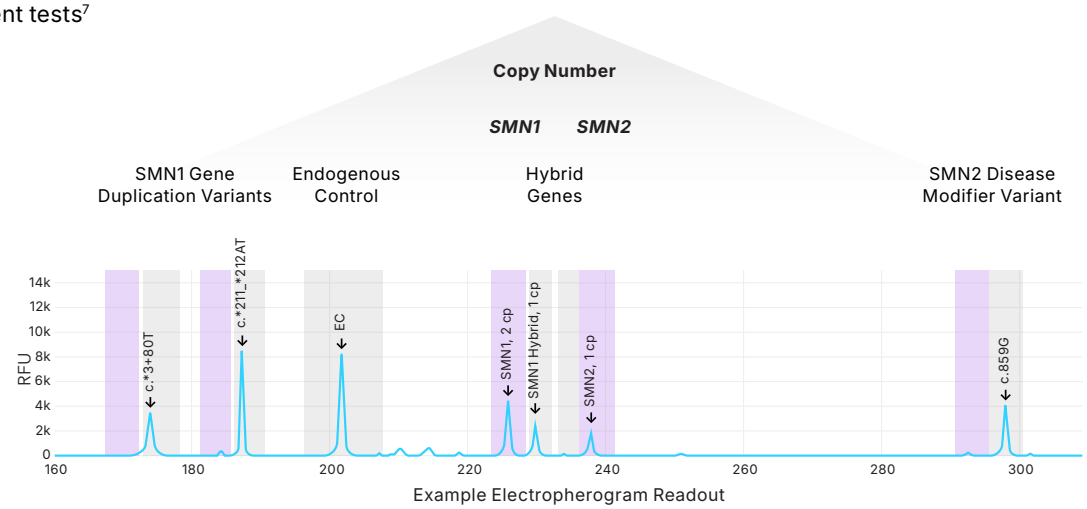
Streamlined Workflow for Laboratories:

- Complete kit which includes all reagents and offers flexible use on widely installed CE equipment
- AmplideX® Reporter Software automates analysis
- Delivers results in <4 hours to enable time-sensitive treatments

>98% concordance with multiple reference methods (MLPA, ddPCR, Sanger seq) in multisite independent tests[7]

Confidence for Clinicians:

- Reliable results for SMA genotyping
- Accurate quantification of ***SMN2* copy number**
- Includes batch control in kit with 3 copies of *SMN2* and additional optional controls with high *SMN2* copy numbers in line with ENMC recommendations[1]
- **Disease modifying variant detection** to inform SMA prognosis



Example electropherogram readout. Complete genotyping in a single trace

Deeper Insights with AmplideX Nanopore Carrier Plus Kit†

The AmplideX Nanopore Carrier Plus Kit expands further on *SMN1/2* characterisation leveraging nanopore long-read sequencing. The assay enables detection and phasing of pathogenic variants, along with the identification of additional disease-modifying variants.

1. Abiusi E, et al. Neur Dis. 2024;
2. Schorling DC, et al. Neurology. 2019; 93 (6):267-269;
3. SMA Europe. Available from: https://www.sma-screening-alliance.org/map (last accessed, June 2026);
4. Calucho M, et al. Neuromuscul Disord. 2018.; Prior TW, et al. Am J Hum Genet. 2009; Vezain M, et al. Hum Mutat. 2010;
5. Cuscó I, et al. Neurol Genet. 2020;
6. Milligan JN, et al. Genes 2022, 13 (9); 1657; 7. Miligan JN, et al. J Mol Diagn, 2021

*CE-IVD. For US Export only.

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

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