

AmplideX® Nanopore Carrier Plus Kit*

Streamlining carrier
screening insights,
even for the most
challenging genes.

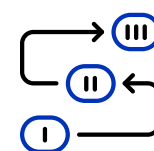
A Long-Range PCR and Long-Read Sequencing Solution for **Carrier Screening Research**

The AmplideX Nanopore Carrier Plus Kit interrogates eleven genes that contribute to high at-risk couple rates in carrier screening.

The design utilizes four mixes across the 11 genes (Figure 1) with modularity to allow any combination of testing.

With a single, flexible workflow designed to consolidate existing assays and methods, Carrier Plus can be used as a primary genetic analysis method or to complement existing NGS workflows for large panels. The use of long-read sequencing provides additional insights and reduces the need for reflex testing compared to traditional methods. The AmplideX One Reporter software further simplifies data analysis and reporting for a seamless, streamlined workflow.

Eleven Genes, Nine High-Prevalence Carrier Screening Conditions, One Workflow



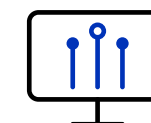
Consolidated Workflow

Single, flexible workflow designed to consolidate existing assays and methods. (e.g. SNV and CNV detection for both *HBA1/2* and *HBB* in a single reaction).



Greater Insights

The use of long-read sequencing provides additional insights compared to existing methods (e.g. AGG interruptions in *FMR1*), reducing the need for reflex testing.



Integrated Data Analysis

The AmplideX One Reporter software further simplifies data analysis and reporting by providing automated quality control and visual outputs to simplify complex genotype interpretations.

FIGURE // 01

Kit configuration - eleven genes across four mixes

Mix A	Mix B	Mix C	Mix D
CFTR	FMR1	HBA1/2	F8 Intron Inversions
SMN1/2		HBB	GBA
Top 3 Genes			CYP21A2 & TNXB
Top 5 Genes			

FIGURE // 02

Two and a half days from extracted DNA to results across multiple samples and genes

	1 Enrichment	2 Quantification & Pooling	3 Library Prep	4 Sequencing	5 Data Analysis	Total
	a. Gene-Specific PCR b. Bead Purification c. Barcoding PCR	a. Quantification of Individual PCR Products and Within-Mix Pooling b. Bead Purification of Pooled Mixes c. Quantification of Pooled Mixes and Between-Mix Pooling	a. End Repair and dA-Tailing b. Adapter Ligation c. Library Quantification & Formulation	a. Flow Cell Preparation and Loading		
Hands-On Time (Hrs:Mins)	1:25	1:15	1:20	0:10	0:05	4:15
Instrument Time (Hrs:Mins)	5:00	0:00	0:00	9:00	1:10	15:10

Hands-on time and instrument time for AmplideX Nanopore Carrier Plus Kit (a 24 sample run across Mixes A, B, and C)

TABLE // 01

Excellent performance across all genes in the assay

Gene	Variant Types	N Measurements	Sample Genotype Percent Agreement
CFTR	SNV, indel, CNV (LED), STR (PolyT/TG)	276	99.3%
SMN1	SNV, indel, CNV	270	97.4%*
SMN2	SNV, indel, CNV	270	97.4%
FMR1	STR (CGG), AGG interrupts	212	99.5%
HBA1/2	SNV, indel, CNV, SV	175	100%
HBB	SNV, indel, CNV	175	100%
F8	SV (intron 1 and intron 22 inversions)	180	100%
GBA	SNV, indel, CNV, SV	178	100%
CYP21A2	SNV, indel, CNV, SV	167	100%

Genotype agreement with orthogonal methods from method comparison and within lab precision studies of the design verification testing (Source - Internal data on file). For SMN1/SMN2, this represents copy number agreement (0, 1, 2, ≥3). For other gene targets, this represents agreement for number of affected alleles (0, 1, ≥2).

*Agreement was 100% at 0 and 1 copies

Note: Five samples with known TNXB structural variants or SNVs were 100% concordant.

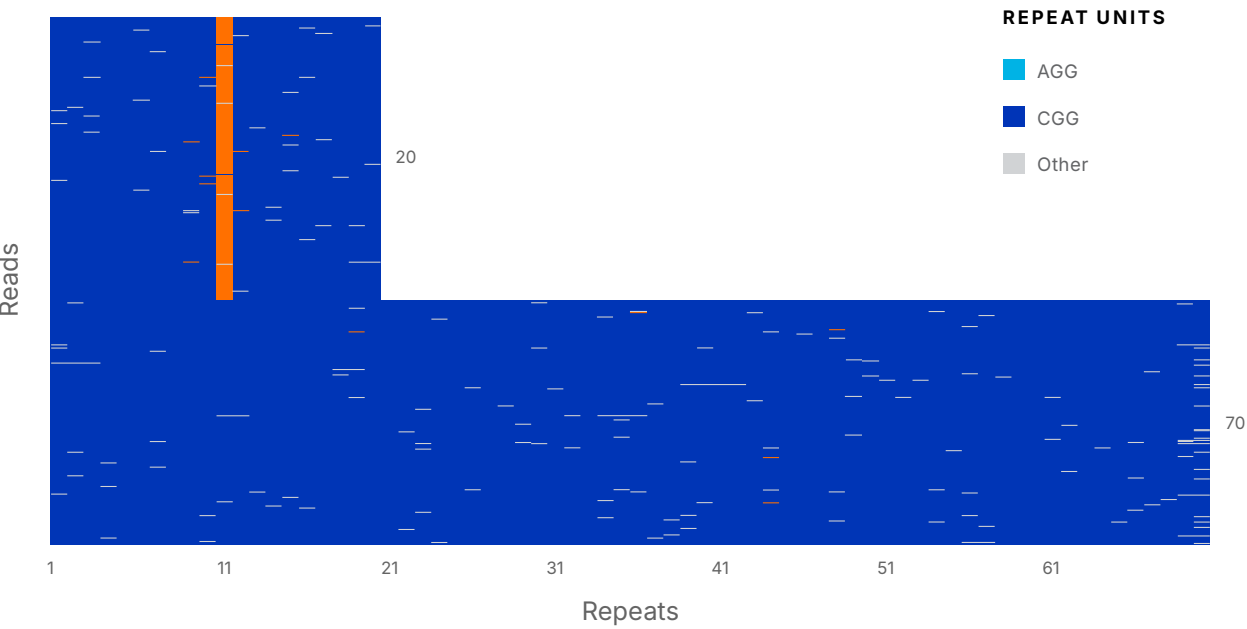
SNV - Single Nucleotide Variant; LED - Large Exon Deletion; STR - Short Tandem Repeats; CNV - Copy Number Variation; SV - Structural Variant

FIGURE // 03

Multiple gene/variant level visualizations provide meaningful insights

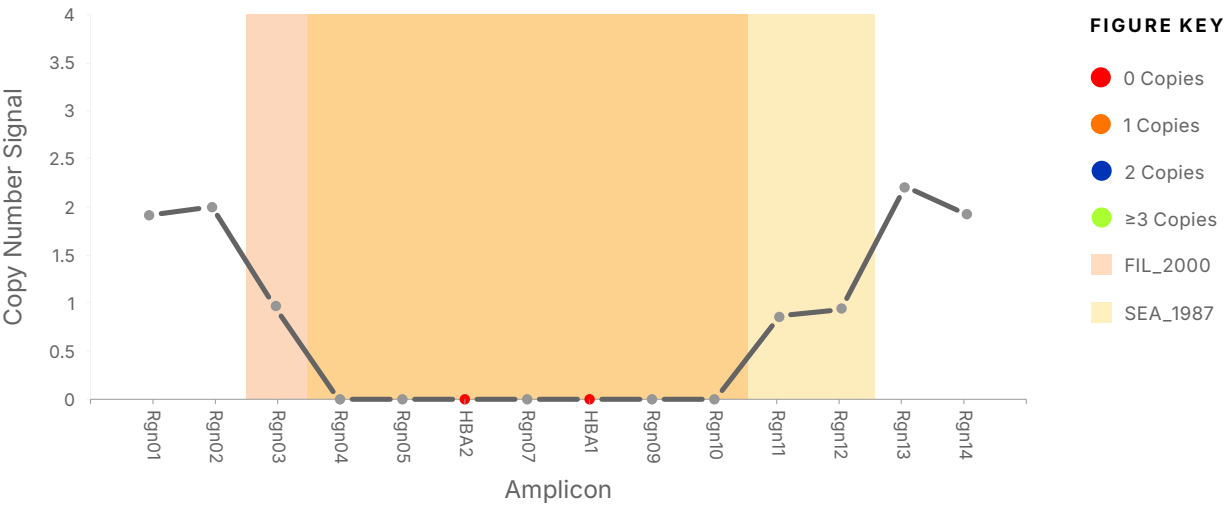
Examples of visual outputs for various gene/mixes:

a. Pre-Mutation (PM) sample with no AGG on PM allele (20(11AGG), 70 CGG)



a. FMR1 output provides CGG sizing and AGG phase and location for each allele

b. NA10797 - compound HET FIL/SEA sample



b. HBA1/2 fold change plot provides accurate copy number and identification of major deletion genotypes

c. NA14733 – 2 copies of CYP21A2 with one of the two copies being a CH-5 fusion

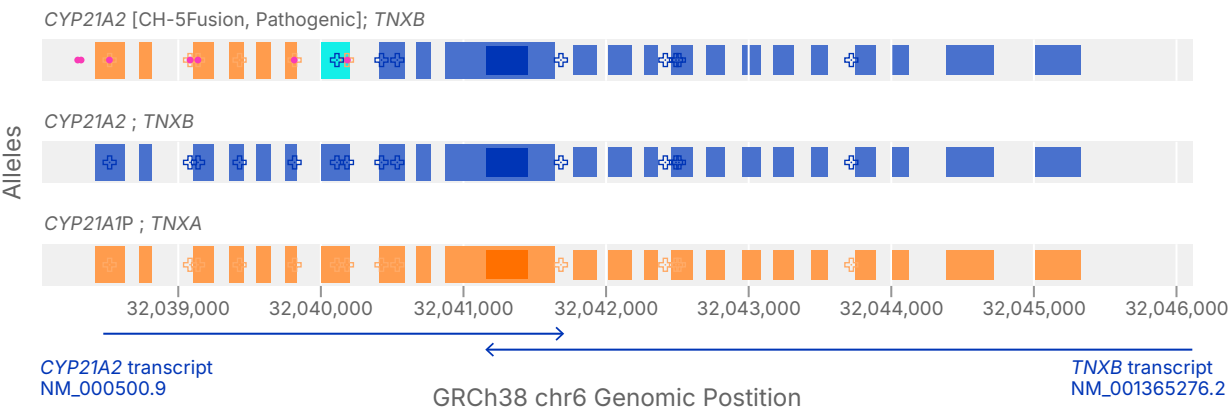


FIGURE KEY

- CYP21A2 Exons
- TNXB Exons
- TNXA-TNXB Exons
- CYP21A1P-CYP21A2 Exons
- CYP21A1P Exons
- TNXA Exons
- Gene PSV
- Pseudogene PSV
- Summarized SNV/INDEL

c. Resolution of CYP21A2/TNXB gene/pseudogene structure down to each exon provides clear visualization of the various changes in this region including phased SNVs.

Part Number	Product	Number of Reactions
A00627	AmplideX Nanopore Carrier Plus Kit A*	96
A00628	AmplideX Nanopore Carrier Plus Kit B*	
A00629	AmplideX Nanopore Carrier Plus Kit C*	
A00630	AmplideX Nanopore Carrier Plus Kit D*	



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