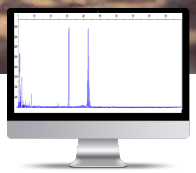
CarrierMax FMR1 Reagent Kit

CarrierMax SMN1/SMN2 Reagent Kit

SeqStudio Genetic Analyzer

3500/3500xL Genetic Analyzer



GeneMapper Software

CarrierMax Software

Carrier screening

CarrierMax FMR1 and SMN1/SMN2 Reagent Kits

Reliable, accurate, and cost-effective kits to complement expanded carrier screening research applications

The Applied Biosystems™ CarrierMax™ FMR1 Reagent Kit and CarrierMax™ SMN1/SMN2 Reagent Kit are designed to detect potential carriers of fragile X syndrome (FXS) and spinal muscular atrophy (SMA), respectively. Both assays utilize either the Applied Biosystems™ SeqStudio™ Genetic Analyzer or the Applied Biosystems™ 3500/3500xL Genetic Analyzer, and the results are analyzed with Applied Biosystems™ GeneMapper™ Software and classification of alleles is reported by Applied Biosystems™ CarrierMax™ Software.

applied biosystems

The workflow of the CarrierMax kits and the throughput of the Applied Biosystems genetic analyzers that support the kits can be seen in Figure 1 and Table 1, respectively.

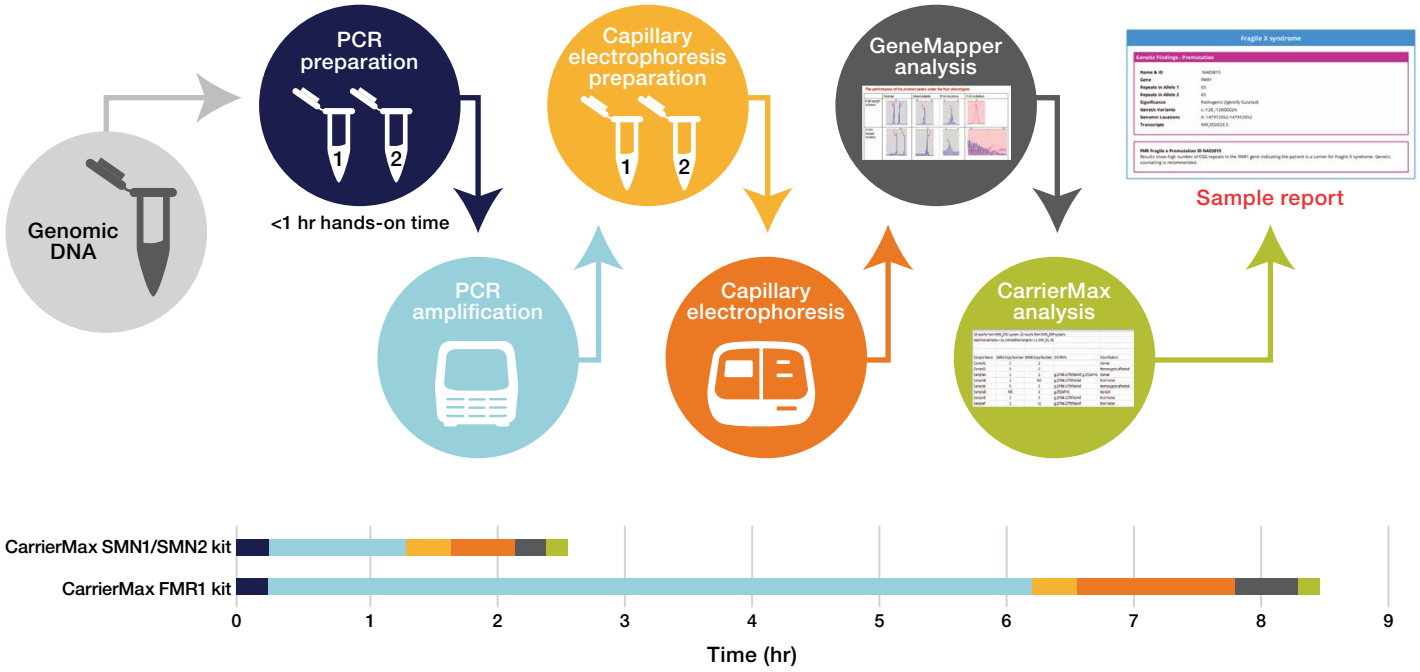


Figure 1. Workflow of the CarrierMax kits.

Table 1. Throughput of Applied Biosystems genetic analyzers that support the CarrierMax kits.

Instrument	Capillaries	CarrierMax FMR1 Reagent Kit		CarrierMax SMN1/SMN2 Reagent Kit	
		Run time (per sample)	Samples per 8 hr	Run time (per sample)	Samples per 8 hr
3500	8	75 min	24 samples	30 min	64 samples
3500xL	24		72 samples		192 samples
SeqStudio system	4		12 samples		32 samples

The CarrierMax FMR1 and SMN1/SMN2 Reagent Kits have been shown by customers to successfully run on Applied Biosystems™ 3730 DNA Analyzer and are fully compatible to run on the Applied Biosystems™ SeqStudio™ Flex Genetic Analyzer.

CarrierMax FMR1 Reagent Kit

FXS and premature ovarian failure (POF) are two distinct diseases with distinct causes: FXS is associated with >200 CGG repeats and POF is associated with 55–199 CGG repeats. FXS is associated with only development delay and not with POF. The vast majority (99%) of detected aberrations resulting in FXS arise from silencing of the fragile X messenger ribonucleoprotein (*FMR1*) gene through an increase in the number of CGG repeats in the gene.

The CarrierMax FMR1 Reagent Kit offers the ability to reliably and accurately screen multiple samples to determine the *FMR1* CGG repeat status of both *FMR1* copies for females and of the one *FMR1* gene for males. This kit uses a dual PCR system combining full length and triplet-primed PCR amplification (TP-PCR), followed by fragment analysis to accurately determine up to 200 CGG repeats and detect alleles of >200 CGG repeats.

Key features of the CarrierMax FMR1 Reagent Kit

- Accurately and consistently detect and classify alleles as normal (<45 CGG repeats), intermediate (45–54 CGG repeats), and premutation (55–199 CGG repeats)
- Identify full mutation alleles of >200 CGG repeats
- Determine the presence of AGG-interrupting sequences
- Seamlessly report allele classification using included CarrierMax Software

CarrierMax SMN1/SMN2 Reagent Kit

SMA resulting from a loss of function of the *SMN1* gene is one of the leading causes of neurodegenerative disorders; therefore, *SMN1* is a critical target for preconception carrier screening. The majority (95%) of SMA cases result from a loss of function of the *SMN1* gene through either a deletion of the gene or a point mutation in exon 7. Alternatively, a genetic recombination results in two copies of the *SMN1* gene on a single chromosome resulting in a carrier status (2+0) that is often not detected using conventional testing methods. An equally important gene to include in the screen is the *SMN2* gene, as an increase in the number of copies of *SMN2* can modify the phenotype associated with SMA.

The CarrierMax SMN1/SMN2 Reagent Kit offers a quick and comprehensive screen to accurately determine the copy number of exon 7 in *SMN1* as well as an indication of potential 2+0 carriers through haplotype assessment. The CarrierMax SMN1/SMN2 Reagent Kit uses multiplex PCR amplification of genomic DNA and capillary electrophoresis to detect deletions in exon 7 of the *SMN1* gene. In addition, the kit detects two variants associated with the silent carrier (2+0) haplotype.

Key features of the CarrierMax SMN1/SMN2 Reagent Kit

- Accurately and consistently detect exon 7 copy number states of 0, 1, or ≥2 for both *SMN1* and *SMN2* genes
- Determine if *SMN1* to *SMN2* exon 7 gene conversion has occurred
- Detect haplotype variants associated with SMA silent (2+0) carrier status
- Seamlessly report sample carrier classification using included CarrierMax Software

Product specifications

	CarrierMax FMR1 Reagent Kit	CarrierMax SMN1/SMN2 Reagent Kit
Description	Accurate, reliable, and cost-effective analysis of *FMR1* gene CGG repeat units	Accurate, reliable, and cost-effective analysis of copy number of *SMN1* and *SMN2* genes, including single nucleotide polymorphisms (SNPs) for 2+0 risk assessment
Research applications	Primary and secondary single gene testing	
Method	Capillary electrophoresis	
Platform	SeqStudio and 3500/3500xL genetic analyzers	
Content	A dual PCR system combining full-length and triplet-primed PCR amplification (TP-PCR) to detect the number of CGG repeats in the *FMR1* gene	Multiplex PCR amplification and fragment analysis to detect deletions in exon 7 that result in truncated SMN protein and silent carrier (2+0) haplotype detection
Throughput	36, 72, or 216 samples/run	48, 184, or 384 samples/run

Ordering information

Description	Quantity	Cat. No.
CarrierMax FMR1 Reagent Kit	48 samples	952362
CarrierMax SMN1/SMN2 Reagent Kit	50 samples	952363
CarrierMax A5D Matrix Standard Kit	8 samples	952364
CarrierMax FMR1 Control DNA Kit	22 samples	952414
CarrierMax SMN Control DNA Kit	22 samples	952445

Learn more at **thermofisher.com/carriermax-fmr1** and **thermofisher.com/carriermax-smn1**