

Reproductive health

# Advance carrier screening in your research laboratory now

An important step toward informed family planning through research studies



Given the increasing ethnic diversity of couples around the globe, many molecular testing laboratories are evaluating or revising their approach to carrier screening research. Advances in genetic analysis tools are revolutionizing reproductive health and transforming the way carrier screening research is being conducted.

Whether you are looking for a targeted panel or a broad, pan-ethnic expanded carrier screening (ECS) research solution, we can help devise the correct workflow—for essential genes like *FMR1*, *CFTR*, and *SMN1*—to meet the unique needs of your laboratory.

## You've got options—find the right tools for your lab

Choose from our comprehensive portfolio of carrier screening research solutions.

Assay	Method	Instrument	Description
<b>Ion Torrent™ CarrierSeq™ ECS Kits</b>	Ion Torrent™ next-generation sequencing (NGS)	Ion GeneStudio™ S5, S5 Plus, or S5 Prime System	<ul style="list-style-type: none"> <li>Rigorously designed specifically for ECS research</li> <li>420-gene panel to analyze SNVs and CNVs for 28,000 nonbenign ClinVar variants in a consolidated NGS assay</li> <li>Customizable solution that includes targeted content, optimized reagents, and intuitive data analysis software to simplify and streamline implementation</li> </ul>
			
<a href="https://thermofisher.com/carrierseq">thermofisher.com/carrierseq</a>			
Assay	Method	Instrument	Description
<b>Applied Biosystems™ CarrierMax™ reagents</b>	Capillary electrophoresis (CE) fragment analysis	Applied Biosystems™ SeqStudio™ Genetic Analyzer	<ul style="list-style-type: none"> <li>Accurate, reliable, and cost-effective testing for <i>FMR1</i> and <i>SMN1</i> genes</li> <li>Detect the number of CGG repeats in the <i>FMR1</i> gene using a dual PCR system combining full-length and triplet-primed PCR amplification (TP-PCR)</li> <li>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</li> </ul>
			
<a href="https://thermofisher.com/carriermax-fmr1">thermofisher.com/carriermax-fmr1</a>			
Assay	Method	Instrument	Description
<b>Applied Biosystems™ CarrierScan™ Assay</b>	High-throughput, microarray-based platform	Applied Biosystems™ GeneTitan™ Multi-Channel Instrument	<ul style="list-style-type: none"> <li>High-throughput, cost-effective, pan-ethnic panel with simple data analysis, reporting, and exporting</li> <li>Combine CNVs for the research of challenging diseases such as SMA, DMD, and thalassemias, with sequence variants for consolidation of assays</li> <li>Tailor content from our database of &gt;6,000 variants, including &gt;1,000 verified variants with true-positive samples</li> </ul>
			
<a href="https://thermofisher.com/carrierscan">thermofisher.com/carrierscan</a>			
Assay	Method	Instrument	Description
<b>Applied Biosystems™ TaqMan™ Assays</b>	Real-time PCR for <i>CFTR</i> gene mutation analysis	Applied Biosystems™ QuantStudio™ 12K Flex Real-Time PCR System	<ul style="list-style-type: none"> <li>Fully qualified content with an extensive menu of targets; design custom research panels from a menu of 199 <i>CFTR</i> gene targets, including full coverage of the ACMG-23 panel and majority coverage of the <i>CFTR2</i> mutation list</li> <li>Flexible formats provide breadth in content and choice of four assay formats, including prespotted 384-well arrays and Applied Biosystems™ OpenArray™ plates</li> </ul>
			
<a href="https://thermofisher.com/cftr">thermofisher.com/cftr</a>			

Find out more about our comprehensive reproductive health portfolio at [thermofisher.com/rh](https://thermofisher.com/rh)