



Identify causal variants with confidence

Two complementary technologies—one seamless workflow

Clinical research labs can choose from multiple options to study human genetic diseases based on the levels of phenotypic heterogeneity observed.

Options include analyzing single genes by Sanger sequencing or targeted gene panels using next-generation sequencing (NGS).

What technology best fits your clinical research needs?

Choose Sanger sequencing when you need to:

- Study diseases with clearly defined phenotypes
- Sequence 1 or 2 genes or up to 96 targets
- Sequence 1–96 samples at a time without barcoding
- Confirm NGS variants with up to 99.99% accuracy
- Get longer read lengths (up to 1,000 bp)

Choose NGS when you need to:

- Study diseases with higher levels of phenotypic heterogeneity
- Sequence more than 2 genes or more than 96 targets
- Sequence more than 96 samples for multiple targets
- Discover novel variants



End-to-end genetic analysis solutions for your clinical research lab

To make it easier to confirm Ion Torrent™ NGS variants with Sanger sequencing, we provide direct links from the Torrent Suite™ Variant Caller plug-in into the Primer Designer Tool, so ordering primer pairs for your final list of variants is easy and seamless.

NGS workflow

Library preparation Hands-on time: 15 minutes*	Template preparation Hands-on time: 15 minutes	Run sequence Hands-on time: less than 15 minutes	Analyze data Hands-on time: N/A
			XXXX
Use predesigned panels or design your own at ampliseq.com	Automated overnight run on lon Chef™ System	NGS using Ion S5™ System	Torrent Suite™/Ion Reporter™ software for variant identification

^{*} Automated library preparation of any 2-pool Ion AmpliSeq panel on the Ion Chef System.

More targets or samples



Orthogonal validation of NGS variants

Sanger sequencing workflow

Perform PCR	Generate products	Run sequence	Analyze data
\$ \\ \\ \\ \\ \\ \\ \\ \\ \\ \\ \\ \\ \\			XXXX
Use predesigned primer pairs or design your own	Cycle sequencing	Sanger sequencing using the Applied Biosystems™ 3500 Genetic Analyzer	Minor Variant Finder Software

Talk to your sales representative about how you can empower your clinical research lab with our complete suite of sequencing solutions.

Learn more at thermofisher.com/sequencing

