

# Advancing Genomic Innovation

Novel tools for more efficient genomic analysis

The rise of genomics has revolutionized how genetic diseases are characterized and diagnosed. Innovative tools are necessary that can generate and analyze genomic data, and maximize the impact of genomics on how we understand disease. Here are the latest genomics tools that are fueling breakthroughs in research, the clinic, and industrial applications.

### Sequence Analysis Software

**Congenica**® clinical decision support software, created by the company of the same name, enables healthcare professionals to make rapid and accurate diagnoses based on facilitating accelerated secondary and tertiary analysis of whole genome, whole exome, and gene panel data. Through this platform, diagnostic yield and case throughput is increased, workflow efficiency is maximized, and as a result, clinicians can make diagnoses with more confidence. By automating variant calling, alignment, and prioritization pipelines, the software enables faster data processing and can support the completion of review-to-report workflows in as little as 30 minutes. Congenica was recently expanded to include copy number variation (CNV) calling for exome and gene panel sequencing.



Congenica's clinical decision support platform accelerates the opportunity for diagnosis and characterization of genetic diseases, increasing throughput and diagnostic yield.

The software's user interface facilitates the exploration of phenotypic data and family structures, helping clinicians interpret information and improve their understanding of cases. By bringing together multiple, high-quality reference data sources and

key data from past cases involving patients with matching variants, the platform allows healthcare professionals to accelerate the delivery of clinically actionable results, even in the most challenging cases. The UK government-funded Genomics England is using Congenica to inform medical decisions as part of its national NHS Genomic Medicine Service.

### Targeted Genotyping Panels

The need for reliable detection of informative genetic markers is critical for verifying pedigrees and checking for genetic health in the canine and feline breeding industry. **Thermo Fisher Scientific** recently released three new AgriSeq™ Targeted Genotyping by Sequencing (GBS) solutions for dogs and cats: **AgriSeq Canine Traits and Disorders Panel, AgriSeq Canine SNP Parentage and Identification Panel, and AgriSeq Feline Parentage and ID Plus Traits and Disorders Panel.** These customized GBS screening panels offer comprehensive genetic data that will allow breeders to adopt and switch to highly informative SNP-based genotyping from current parentage testing based on genotyping short tandem repeats.

The AgriSeq GBS panels offer a cost-effective, highly multiplexed and scalable targeted genetic screening tool for companion animals. The ready-to-use panels achieve high call rates with reproducible genotyping results. Simple, high-throughput workflow on the Ion Torrent next-generation sequencing (NGS) platform provides results in two to three days. "Customers will be able to provide more detailed, critical information at comparable

costs and effort which will pave the way for our advancement in targeted GBS testing and screening of companion animals," said Martin Guillet, global head and general manager for AgriBusiness at Thermo Fisher Scientific.

### Metagenomics without Culturing

Advances in the field of metagenomics now allow researchers to probe the microbial world without culturing. **Phase Genomics** recently launched its new **ProxiMeta™ Platform** to accelerate microbiome research at a significantly reduced per-sample cost. The ProxiMeta Platform allows users to assemble hundreds of genomes and associate plasmids, phages, and antibiotic resistance genes with their hosts based on its Hi-C and proximity-ligation technologies, which enable chromosome-scale genome assembly, metagenomic deconvolution, and the analysis of structural genomic variation and genome architecture. This deepens insights into horizontal gene transfer, antibiotic resistance, and the interactions between genetic elements in microbial cells and communities. The eight reaction kit uses improved chemistry, includes all the reagents needed to prepare a sequencing-ready library from a crude

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