TruSight™ Portfolio—New Possibilities for Next-Generation Sequencing

Expert-designed sequencing panels enable laboratories to realize the benefits of next-generation sequencing.

Highlights

- **Expert-Designed Content**
  Content chosen by recognized healthcare experts at leading institutions to target specific diseases or conditions
- **High-Quality Data**
  Designed for use with the widely adopted and proven Illumina next-generation sequencing (NGS) technology
- **Low Input DNA Requirement**
  Begin with as little as 50 ng DNA for reliable results

Expert-Designed Content Leverages Next-Generation Sequencing

For cost-effective, streamlined, targeted next-generation sequencing of specific genetic diseases or conditions, Illumina offers the TruSight sequencing panels. Designed by recognized healthcare experts at leading institutions, the TruSight panels comprise oligo probes targeting genes and regions thought to be relevant for particular diseases or conditions. The panels are for research use only and not intended for diagnostic use; however, labs can potentially leverage this content to develop their own unique targeted tests, in their own laboratories, in accordance with CLIA regulations.

Six TruSight Panels Currently Available

Working closely with recognized experts in the healthcare community, Illumina developed six TruSight sequencing panels (Table 1).

TruSight One—Targets > 4,800 genes, enabling labs to existing menus, streamline workflows, or create an entire portfolio of sequencing options

Developed by Illumina, the TruSight One Sequencing Panel provides comprehensive coverage of 4,813 genes with an associated clinical phenotype. Laboratories can analyze all of the genes on the panel or choose to focus on a specific subset. In this way, a single panel can effectively represent an entire next-generation sequencing portfolio.

TruSight Tumor—Access a deeper view of variation in solid tumors

The TruSight Tumor Sequencing Panel includes 26 genes and 175 amplicons of relevant content from CAP and NCCN guidelines and late-phase pharmaceutical clinical trials carefully selected for their involvement with solid tumors[1–3]. TruSight Tumor provides coverage of exon coding regions where variation has been cataloged in the COSMIC database[4] in oncogenes, and coverage of all exons in tumor suppressor genes. This provides a more comprehensive view of somatic variation in solid tumors, including lung, colon, melanoma, gastric, and ovarian.

TruSight Cancer—Targets genes previously linked to a predisposition towards cancer

Developed in collaboration with Dr. Nazneen Rahman and team at the Institute of Cancer Research (ICR), London, the TruSight Cancer Sequencing Panel targets over 90 genes known to play a role in cancer, including genes associated with both common (e.g., breast, colorectal) and rare cancers. In addition, the set includes SNPs found to correlate with cancer through genome-wide association studies (GWAS).

Table 1: TruSight Sequencing Panels

Sequencing Panel	Collaborators	Genes per Set
TruSight One	Illumina leveraging content from the Human Genome Mutation Database (HGMD), Online Mendelian Inheritance in Man (OMIM) catalog, GeneTests.org, Illumina TruSight panels, and other commercially available sequencing panels	4,813
TruSight Tumor	Illumina leveraging content from CAP and NCCN guidelines and late-phase clinical trials	26
TruSight Cancer	Dr. Nazneen Rahman and team at The Institute of Cancer Research (ICR), London	94
TruSight Cardiomyopathy	Dr. Heidi Rehm and team at Laboratory for Molecular Medicine (LMM), Partners Healthcare Center for Personalized Genetic Medicine (PCPGM), Harvard Medical School	46
TruSight Inherited Disease	Dr. Stephen Kingsmore and team at Children's Mercy Hospital (CMH) for Pediatric Genomic Medicine; Dr. Carol Saunders at CMH; Dr. Hilger Ropers at Max Planck Institute	552
TruSight Autism	Dr. Jonathan Pevsner and team at Kennedy Krieger Institute (KKI)	101

TruSight Cardiomyopathy—Focuses on identifying inherited causes of cardiomyopathy

Developed in collaboration with Dr. Heidi Rehm and team at the Laboratory for Molecular Medicine (LMM) and Partners Healthcare Center for Personalized Genetic Medicine (PCPGM), Harvard Medical School, the Cardiomyopathy Sequencing Panel targets 46 genes linked to Hypertrophic Cardiomyopathy (HCM), Dilated Cardiomyopathy (DCM), Arrhythmogenic Right Ventricular Cardiomyopathy/Catecholaminergic Polymorphic Ventricular Tachycardia (ARVC/CPVT), and Left Ventricular Noncompaction Cardiomyopathy (LVNC). Content was chosen based on careful review of the literature and LMM's nine years of experience testing many of these genes[5]. Additional content is included from syndromes, such as Danon, Fabry, etc., that present with isolated cardiomyopathies.

TruSight Inherited Disease—Focuses on severe, recessive pediatric onset diseases

Developed in collaboration with Dr. Stephen Kingsmore and team at Children's Mercy Hospital (CMH) for Pediatric Genomic Medicine in Missouri, the Inherited Disease Sequencing Panel was initially based on a 448 disease panel designed for preconception carrier testing for severe, recessive childhood diseases published in Science Translational Medicine. The original content was revised by Dr. Carol Saunders, FACMG, at CMH (following ACMG guidelines for testing ultra-rare genetic diseases) to reflect the needs of medical geneticists with a primary focus on severe recessive diseases with childhood onset. Intellectual disability genes were added by Dr. Hilger Ropers at the Max Planck Institute.

TruSight Autism—Assists in the evaluation of characteristics associated with autism

Developed in collaboration with Dr. Jonathan Pevsner and team at Kennedy Krieger Institute (KKI), the TruSight Autism Sequencing Panel targets approximately 100 developmental delay genes linked specifically to autism. The set includes genes reported in the Online Mendelian Inheritance in Man (OMIM) database on autism; genes with recurrent mutations resulting in developmental delays as noted in Nature publications[6–8]; genes with reported mutations as found in case studies involving developmental delay characteristics; genes from other publicly available autism panels; and genes from summaries of autism-relevant genes (e.g., AutismKB[9]). Genes neighboring strong association signals were excluded in the absence of published reports of mutations.

Learn More

To learn more about the TruSight sequencing panels and Illumina next-generation sequencing technology, visit www.illumina.com/trusight.

Ordering Information

Sequencing Panel	Catalog No.	TG Catalog No.*
TruSight One (9 samples)	FC-141-1006	TG-141-1006
TruSight One (36 samples)	FC-141-1007	TG-141-1007
TruSight Tumor (48 samples)	FC-130-2001	TG-130-2001
TruSight Cancer (4 enrichment rxns)	FC-121-0202	TG-141-1002
TruSight Cardiomyopathy (4 enrichment rxns)	FC-121-0204	TG-141-1004
TruSight Inherited Disease (4 enrichment rxns)	FC-121-0205	TG-141-1005
TruSight Autism (4 enrichment rxns)	FC-121-0203	TG-141-1003

* TG-labeled consumables include features intended to help customers reduce the frequency of revalidation. They are available only under supply agreement and require customers to provide a binding forecast. TruSight sequencing panels are available for evaluation purposes prior to executing a supply agreement. Please contact your account manager for more information.

References

1. National Comprehensive Cancer Network (www.nccn.org/professionals/physician_gls/f_guidelines.asp#site)
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3. ClinicalTrials.gov (www.clinicaltrials.gov)
4. Forbes SA, Bindal N, Bamford S, Cole C, Kok CY, et al. 2011 COSMIC: Mining COMplete cancer genomes in the catalogue of somatic mutations in cancer. Nucl Acids Res. 39: D945–50.
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6. Marshall CR, Noor A, Vincent JB, Lionel AC, Feuk L, et al. (2008) Structural variation of chromosomes in autism spectrum disorder. Am J Hum Genet. 82(2): 477–488.
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8. Vendeweyer G and Kooy RF (2009) Balanced translocations in mental retardation. Hum Genet. 126(1): 133–147.
9. Autism KB (autismkb.cbi.pku.edu.cn)

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