TruSight™ Portfolio—New Possibilities for Next-Generation Sequencing

Expert-designed, disease-specific content enables 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 content sets. Designed by recognized healthcare experts at leading institutions, the TruSight content sets comprise oligo probes targeting genes and regions thought to be relevant for particular diseases or conditions. The content sets 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.

Five TruSight Content Sets Currently Available

Working closely with recognized experts in the healthcare community, Illumina developed an initial offering of five TruSight content sets (Table 1).

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

Genetically heterogeneous disorders, such as autism, are inherently challenging to molecularly classify with single gene panels. Developed in collaboration with Dr. Jonathan Pevsner and team at Kennedy Krieger Institute (KKI), the TruSight Autism content set 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[1–3]; 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[4]). Genes neighboring strong association signals were excluded in the absence of published reports of mutations.

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

As we learn more about the genetic variants that increase the risk of cancers, clinicians and researchers will be able to provide more comprehensive evaluations of genes that may play a role in disease development. Developed in collaboration with Dr. Nazneen Rahman and team at the Institute of Cancer Research (ICR), London, the TruSight Cancer content set 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). Content selection was based on expert curation of the scientific literature and other high-quality resources.

Table 1: TruSight Content Sets

Content Set	Collaborators	Genes per Set
TruSight Autism	Dr. Jonathan Pevsner and team at Kennedy Krieger Institute (KKI)	101
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 Exome	Illumina leveraging Human Genome Mutation Database (HGMD)	2,761

TruSight Cardiomyopathy—Focuses on identifying inherited causes of cardiomyopathy

By identifying the type of cardiomyopathy, clinicians will be able to better advise their patients, and their patients' families, regarding potential health treatments and lifestyle choices. 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 content set 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

Capture coding exons, intron-exon boundaries, and regions known to harbor pathogenic mutations, some of which are intronic. Developed in collaboration with Dr. Stephen Kingsmore and team at Children's Mercy Hospital (CMH) for Pediatric Genomic Medicine in Missouri, the Inherited Disease content set 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 Exome—Targets rare genetic diseases

Developed by Illumina, the TruSight Exome content set is a subset of the known genes in the full exome. It is focused on disease-causing mutations as curated by the Human Genome Mutation Database (HGMD) and shown to be important in specific inherited conditions.

Learn More

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

Ordering Information

TruSight Content Sets (4 enrichment rxns/kit)	Catalog No.
TruSight Autism	TG-141-1003
TruSight Cancer	TG-141-1002
TruSight Cardiomyopathy	TG-141-1004
TruSight Inherited Disease	TG-141-1005
TruSight Exome	TG-141-1001

References

1. 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.
2. Talkowski ME, Rosenfeld JA, Blumenthal I, Pillalamarri V, Chiang C, et al. (2012) Sequencing chromosomal abnormalities reveals neurodevelopmental loci that confer risk across diagnostic boundaries. Cell **149(3):** 525–537.
3. Vendeweyer G and Kooy RF (2009) Balanced translocations in mental retardation. Hum Genet **126(1):** 133–147.
4. autism.cbi.pku.edu.cn
5. Teekakirikul P, Kelly MA, Rehm HL, Lakdawala N, Funke BH (2012) Inherited cardiomyopathies: Molecular genetics and clinical genetic testing in the post genomic era. J Mol Diag Submitted August 2012.

Note regarding biomarker patents and other patents unique to specific uses of products.

Some genomic variants, including some nucleic acid sequences, and their use in specific applications may be protected by patents. Customers are advised to determine whether they are required to obtain licenses from the party that owns or controls such patents in order to use the product in customer's specific application.

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