DATA SHEET

TruSight™ Oncology 500 and TruSight Oncology 500 High-Throughput

Enabling flexible, scalable comprehensive genomic profiling from FFPE samples

- Analyze multiple variant types and key biomarkers in 500+ genes across DNA and RNA in a single assay
- Go from sample to results in 4–5 days using manual or automated workflows that integrate library prep, sequencing, and data analysis with DRAGEN™ secondary analysis
- Generate accurate data and reliable results that meet demanding performance specifications
- Keep samples in house and obtain data that is relevant to the local institution and community

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Introduction

Large-cohort studies show that comprehensive genomic profiling has the potential to identify relevant genetic alterations in up to 90% of samples.[1-6] A single, comprehensive assay to assess a wide range of biomarkers uses less sample and returns results more quickly compared to multiple, iterative tests. To help researchers working with limited tissue supply and time, Illumina offers TruSight Oncology 500 and TruSight Oncology 500 High-Throughput (Table 1).

Analyze multiple tumor types and biomarkers with a single workflow

TruSight Oncology 500 and TruSight Oncology 500 High-Throughput are next-generation sequencing (NGS) assays that simultaneously analyze both DNA and RNA (Figure 1) in one integrated workflow (Figure 2). Panel content includes multiple variant types and key biomarkers (Figure 3) across 523 cancer-relevant genes from DNA and 55 genes from RNA (Table 2, Table 3, and Table 4), eliminating the need to spend time and precious sample, such as formalin fixed, paraffin embedded (FFPE) tissue blocks, on iterative testing.

Table 1: TruSight Oncology 500 and TruSight Oncology 500 High-Throughput

Parameter	TruSight Oncology 500	TruSight Oncology 500 High-Throughput	
System	NextSeq 550 System or NextSeq 550Dx Instrument (research mode)	NovaSeq 6000 System or NovaSeq 6000Dx Instrument (research mode)[a]	NovaSeq X Series[a]
Sample throughput	8 samples per run	16–192 samples per run	Single flow cell: 32–480 samples per run Dual flow cell: 32–960 samples per run
Panel size	1.94 Mb DNA, 358 kb RNA	1.94 Mb DNA, 358 kb RNA	1.94 Mb DNA, 358 kb RNA
DNA input requirement	40 ng	40 ng	40 ng
RNA input requirement	40 ng	40–80 ng	40–80 ng
FFPE input requirement	Minimum recommendation of 2 mm^3 from FFPE tissue samples	Minimum recommendation of 2 mm^3 from FFPE tissue samples	Minimum recommendation of 2 mm^3 from FFPE tissue samples
Total assay time	4–5 days from nucleic acid to variant report	4–5 days from nucleic acid to variant report	4–5 days from nucleic acid to variant report
Sequence run time	24 hr	19 hr (SP and S1), 25 hr (S2), or 36 hr (S4)	18.5 hr (1.5B), 20 hr (10B), or 33 hr (25B)
Sequence run	2 × 101 cycles	2 × 101 cycles	2 × 101 cycles
Software version	DRAGEN TruSight Oncology v2.5.2+	DRAGEN TruSight Oncology v2.5.2+	DRAGEN TruSight Oncology v2.5.2+
Limit of detection	5% VAF for small variants 5 copies per ng RNA input for fusions CNVs: 2.2× fold-change for amplifications 0.5× fold-change for deletions	5% VAF for small variants 5 copies per ng RNA input for fusions (80 ng input) CNVs: 2.2× fold-change for amplifications 0.5× fold-change for deletions	5% VAF for small variants 5 copies per ng RNA input for fusions (80 ng input) CNVs: 2.2× fold-change for amplifications 0.5× fold-change for deletions
Analytical sensitivity	> 96% (for all variant types at 5% VAF)	> 96% (for all variant types at 5% VAF)	> 96% (for all variant types at 5% VAF)
Analytical specificity	> 99.9995%	> 99.9995%	> 99.9995%

a. Requires separate, standalone DRAGEN server if secondary analysis with an on-premises server is desired.

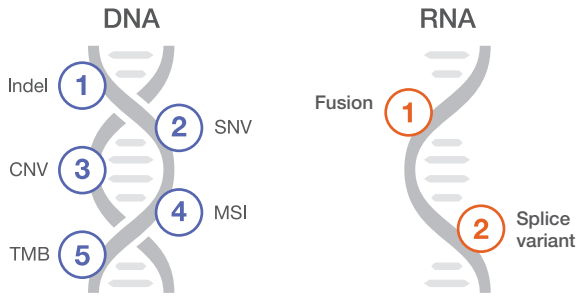

Figure 1: Variant types detected by TruSight Oncology 500 and TruSight Oncology 500 High-Throughput.

Comprehensive content design

Illumina partnered with recognized authorities in the oncology community to design TruSight Oncology 500 and TruSight Oncology 500 High-Throughput content. The resulting panels provide comprehensive coverage of biomarkers commonly mutated in numerous cancer types (Figure 3), including 523 genes for single nucleotide variants (SNVs), insertions/deletions (indels), copy number variations (CNVs); and 55 genes for known and novel fusion and splice variants (Table 2, Table 3).

Content comprises genes listed in current guidelines with significant coverage of key guidelines for multiple tumor types (Figure 4) and genes involved in over 1000 clinical trials. In addition, the TruSight Oncology 500 panels include the microsatellite instability (MSI) biomarker, with known correlations to responses,[7-9] and the tumor mutational burden (TMB) biomarker (Table 4).[10]

Integrated workflow

Implementing CGP in house is simplified with the availability of a comprehensive, streamlined workflow that spans from sample input to final report (Figure 2). Using automated library preparation kits and methods, variant calling tools, and interpretation and reporting software enables a smooth workflow that can be completed in as few as four days.

Start with DNA or RNA

The TruSight Oncology 500 assays can use DNA or RNA extracted from the same sample as input material. If using DNA, sample preparation starts with shearing the genomic DNA (gDNA). If starting from RNA, the first step is to reverse transcribe the sample into cDNA. Sequencing-ready libraries are prepared from sheared gDNA and cDNA simultaneously.

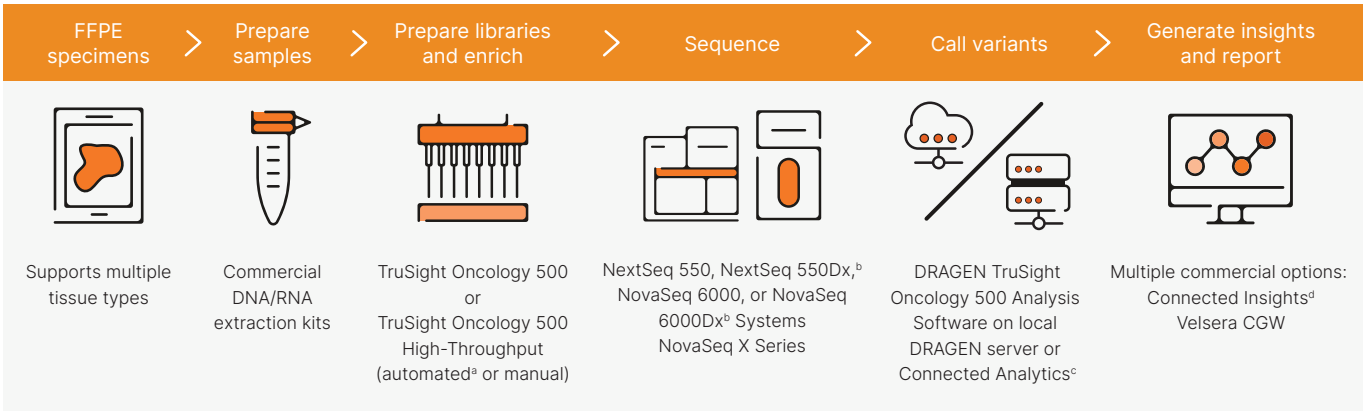

Figure 2: TruSight Oncology 500 workflow—TruSight Oncology 500 and TruSight Oncology 500 High-Throughput integrate into current lab workflows, going from nucleic acids to a variant calls in four days.

a. TruSight Oncology 500 and TruSight Oncology 500 High-Throughput kits are available in automation-compatible versions.
b. NextSeq 550Dx and NovaSeq 6000Dx Instruments in research mode.
c. Local Run Manager TruSight Oncology 500 Analysis Module is available on the NextSeq 550 System only.
d. Not available in all countries. Illumina Connected Insights supports user-defined tertiary analysis through API calls to third-party knowledge sources.

Pan-cancer: *BRAF, NTRK1, NTRK2, NTRK3, RET*, MSI, TMB													
		Genes with biomarkers of clinical significance*											Genes with biomarkers of potential clinical significance†
	Breast	*BRCA1*	*BRCA2*	*ERBB2*	*ESR1*	*PALB2*	*PIK3CA*						**180**
	Colorectal	*ERBB2*	*KRAS*	*NRAS*									**166**
	Bone	*EGFR*	*ERG*	*ETV1*	*ETV4*	*EWSR1*	*FEV*	*FLI1*	*FUS*	*H3F3A*	*HEY1*	*IDH1*	**140**
		MDM2	*NCOA2*	*SMARCB1*									
	Lung	*ALK*	*EGFR*	*ERBB2*	*KRAS*	*MET*	*NUTM1*	*ROS1*					**223**
	Melanoma	*KIT*	*NRAS*	*ROS1*									**172**
	Ovarian	*BRCA1*	*BRCA2*	*FOXL2*									**149**
	CNS‡	*APC*	*ATRX*	*CDKN2A*	*CDKN2B*	*EGFR*	*H3F3A*	*HIST1H3B*	*HIST1H3C*	*IDH1*	*IDH2*	*MYCN*	**140**
		PTCH1	*RELA*	*TERT*	*TP53*								
	Prostate	*AR*	*ATM*	*BARD1*	*BRCA1*	*BRCA2*	*BRIP1*	*CDK12*	*CHEK1*	*CHEK2*	*FANCL*	*FGFR2*	**151**
		FGFR3	*PALB2*	*PTEN*	*RAD51B*	*RAD51C*	*RAD51D*	*RAD54L*					
	Thyroid	*HRAS*	*KRAS*	*NRAS*	*TERT*								**165**
	Uterine & cervical	*BRCA2*	*EPC1*	*ERBB2*	*ESR1*	*FOXO1*	*GREB1*	*JAZF1*	*NCOA2*	*NCOA3*	*NUTM2A*	*NUTM2B*	**138**
		PAX3	*PAX7*	*PHF1*	*POLE*	*SMARCA4*	*SUZ12*	*TP53*	*YWHAE*				
	Other solid tumors	*ALK*	*APC*	*ARID1A*	*ASPSCR1*	*ATF1*	*ATIC*	*BAP1*	*BCOR*	*BRCA1*	*BRCA2*	*CAMTA1*	**152**
		CARS	*CCNB3*	*CDK4*	*CDKN2A*	*CIC*	*CITED2*	*CLTC*	*COL1A1*	*COL6A3*	*CREB1*	*CREB3L1*	
		CREB3L2	*CSF1*	*CTNNB1*	*DDIT3*	*DDX3X*	*DNAJB1*	*DUX4*	*EED*	*EGFR*	*ERBB2*	*ERG*	
		ETV1	*ETV4*	*ETV6*	*EWSR1*	*FEV*	*FGFR2*	*FGFR3*	*FLI1*	*FOXL2*	*FOXO1*	*FOXO4*	
		FUS	*GLI1*	*HEY1*	*HGF*	*HMGA2*	*IDH1*	*KRAS*	*LEUTX*	*MAML3*	*MDM2*	*MYB*	
		MYOD1	*NAB2*	*NCOA2*	*NF1*	*NFATC2*	*NFIB*	*NR4A3*	*NRAS*	*NUTM1*	*NUTM2A*	*NUTM2B*	
		PALB2	*PATZ1*	*PAX3*	*PAX7*	*PDGFB*	*PDGFRA*	*PRKACA*	*PRKD1*	*RANBP2*	*ROS1*	*SDHA*	
		SDHB	*SDHC*	*SDHD*	*SMARCB1*	*SS18*	*SSX1*	*SSX2*	*SSX4*	*STAT6*	*SUZ12*	*TAF15*	
		TCF12	*TERT*	*TFE3*	*TFEB*	*TFG*	*TP53*	*TPM3*	*TPM4*	*TRAF7*	*TSPAN31*	*VGLL2*	
		WT1	*WWTR1*	*YAP1*	*YWHAE*	*ZC3H7B*							

Figure 3: Subset of genomic tumor profiling biomarkers for multiple cancer types—TruSight Oncology 500 and TruSight Oncology 500 High-Throughput include key guideline biomarkers, emerging biomarkers, and pan-cancer biomarkers such as *BRAF*, *NTRK1*, *NTRK2*, *NTRK3*, *RET*, MSI, and TMB. Content analysis provided by Velsera based on software Knowledge Base v8.5 (February 2023).

* Genes with biomarkers of clinical significance linked to current drug labels or guidelines.

† Genes with biomarkers of potential clinical significance based on presence in clinical trials.

‡ CNS, central nervous system.

Table 2: DNA content included in TruSight Oncology 500 and TruSight Oncology 500 High-Throughput

ABL1	*BCR*	*CHEK1*	*EPHA7*	*FGF23*	*GSK3B*	*IDH2*	*MAP3K1*	*NF2*	*PIK3CA*	*RAD51D*	*SMAD4*	*TGFBR2*
ABL2	*BIRC3*	*CHEK2*	*EPHB1*	*FGF3*	*H3F3A*	*IFNGR1*	*MAP3K13*	*NFE2L2*	*PIK3CB*	*RAD52*	*SMARCA4*	*TMEM127*
ACVR1	*BLM*	*CIC*	*ERBB2*	*FGF4*	*H3F3B*	*INHBA*	*MAP3K14*	*NFKBIA*	*PIK3CD*	*RAD54L*	*SMARCB1*	*TMPRSS2*
ACVR1B	*BMPR1A*	*CREBBP*	*ERBB3*	*FGF5*	*H3F3C*	*INPP4A*	*MAP3K4*	*NKX2-1*	*PIK3CG*	*RAF1*	*SMARCD1*	*TNFAIP3*
AKT1	*BRAF*	*CRKL*	*ERBB4*	*FGF6*	*HGF*	*INPP4B*	*MAPK1*	*NKX3-1*	*PIK3R1*	*RANBP2*	*SMC1A*	*TNFRSF14*
AKT2	*BRCA1*[a]	*CRLF2*	*ERCC1*	*FGF7*	*HIST1H1C*	*INSR*	*MAPK3*	*NOTCH1*	*PIK3R2*	*RARA*	*SMC3*	*TOP1*
AKT3	*BRCA2*[a]	*CSF1R*	*ERCC2*	*FGFR1*	*HIST1H2BD*	*IRF2*	*MAX*	*NOTCH2*	*PIK3R3*	*RASA1*	*SMO*	*TOP2A*
ALK	*BRD4*	*CSF3R*	*ERCC3*	*FGFR2*	*HIST1H3A*	*IRF4*	*MCL1*	*NOTCH3*	*PIM1*	*RB1*	*SNCAIP*	*TP53*
ALOX12B	*BRIP1*	*CSNK1A1*	*ERCC4*	*FGFR3*	*HIST1H3B*	*IRS1*	*MDC1*	*NOTCH4*	*PLCG2*	*RBM10*	*SOCS1*	*TP63*
ANKRD11	*BTG1*	*CTCF*	*ERCC5*	*FGFR4*	*HIST1H3C*	*IRS2*	*MDM2*	*NPM1*	*PLK2*	*RECQL4*	*SOX10*	*TRAF2*
ANKRD26	*BTK*	*CTLA4*	*ERG*	*FH*	*HIST1H3D*	*JAK1*	*MDM4*	*NRAS*	*PMAIP1*	*REL*	*SOX17*	*TRAF7*
APC	*C11orf30*	*CTNNA1*	*ERRFI1*	*FLCN*	*HIST1H3E*	*JAK2*	*MED12*	*NRG1*	*PMS1*	*RET*	*SOX2*	*TSC1*
AR	*CALR*	*CTNNB1*	*ESR1*	*FLI1*	*HIST1H3F*	*JAK3*	*MEF2B*	*NSD1*	*PMS2*	*RFWD2*	*SOX9*	*TSC2*
ARAF	*CARD11*	*CUL3*	*ETS1*	*FLT1*	*HIST1H3G*	*JUN*	*MEN1*	*NTRK1*	*PNRC1*	*RHEB*	*SPEN*	*TSHR*
ARFRP1	*CASP8*	*CUX1*	*ETV1*	*FLT3*	*HIST1H3H*	*KAT6A*	*MET*	*NTRK2*	*POLD1*	*RHOA*	*SPOP*	*U2AF1*
ARID1A	*CBFB*	*CXCR4*	*ETV4*	*FLT4*	*HIST1H3I*	*KDM5A*	*MGA*	*NTRK3*	*POLE*	*RICTOR*	*SPTA1*	*VEGFA*
ARID1B	*CBL*	*CYLD*	*ETV5*	*FOXA1*	*HIST1H3J*	*KDM5C*	*MITF*	*NUP93*	*PPARG*	*RIT1*	*SRC*	*VHL*
ARID2	*CCND1*	*DAXX*	*ETV6*	*FOXL2*	*HIST2H3A*	*KDM6A*	*MLH1*	*NUTM1*	*PPM1D*	*RNF43*	*SRSF2*	*VTCN1*
ARID5B	*CCND2*	*DCUN1D1*	*EWSR1*	*FOXO1*	*HIST2H3C*	*KDR*	*MLL*	*PAK1*	*PPP2R1A*	*ROS1*	*STAG1*	*WISP3*
ASXL1	*CCND3*	*DDR2*	*EZH2*	*FOXP1*	*HIST2H3D*	*KEAP1*	*MLLT3*	*PAK3*	*PPP2R2A*	*RPS6KA4*	*STAG2*	*WT1*
ASXL2	*CCNE1*	*DDX41*	*FAM123B*	*FRS2*	*HIST3H3*	*KEL*	*MPL*	*PAK7*	*PPP6C*	*RPS6KB1*	*STAT3*	*XIAP*
ATM	*CD274*	*DHX15*	*FAM175A*	*FUBP1*	*HLA-A*	*KIF5B*	*MRE11A*	*PALB2*	*PRDM1*	*RPS6KB2*	*STAT4*	*XPO1*
ATR	*CD276*	*DICER1*	*FAM46C*	*FYN*	*HLA-B*	*KIT*	*MSH2*	*PARK2*	*PREX2*	*RPTOR*	*STAT5A*	*XRCC2*
ATRX	*CD74*	*DIS3*	*FANCA*	*GABRA6*	*HLA-C*	*KLF4*	*MSH3*	*PARP1*	*PRKAR1A*	*RUNX1*	*STAT5B*	*YAP1*
AURKA	*CD79A*	*DNAJB1*	*FANCC*	*GATA1*	*HNF1A*	*KLHL6*	*MSH6*	*PAX3*	*PRKCI*	*RUNX1T1*	*STK11*	*YES1*
AURKB	*CD79B*	*DNMT1*	*FANCD2*	*GATA2*	*HNRNPK*	*KMT2B*	*MST1*	*PAX5*	*PRKDC*	*RYBP*	*STK40*	*ZBTB2*
AXIN1	*CDC73*	*DNMT3A*	*FANCE*	*GATA3*	*HOXB13*	*KMT2C*	*MST1R*	*PAX7*	*PRSS8*	*SDHA*	*SUFU*	*ZBTB7A*
AXIN2	*CDH1*	*DNMT3B*	*FANCF*	*GATA4*	*IGF1*	*KMT2D*	*MTOR*	*PAX8*	*PTCH1*	*SDHAF2*	*SUZ12*	*ZFHX3*
AXL	*CDK12*	*DOT1L*	*FANCG*	*GATA6*	*IGF1R*	*KRAS*	*MUTYH*	*PBRM1*	*PTEN*	*SDHB*	*SYK*	*ZNF217*
B2M	*CDK4*	*E2F3*	*FANCI*	*GEN1*	*IGF2*	*LAMP1*	*MYB*	*PDCD1*	*PTPN11*	*SDHC*	*TAF1*	*ZNF703*
BAP1	*CDK6*	*EED*	*FANCL*	*GID4*	*IKBKE*	*LATS1*	*MYC*	*PDCD1LG2*	*PTPRD*	*SDHD*	*TBX3*	*ZRSR2*
BARD1	*CDK8*	*EGFL7*	*FAS*	*GLI1*	*IKZF1*	*LATS2*	*MYCL1*	*PDGFRA*	*PTPRS*	*SETBP1*	*TCEB1*	
BBC3	*CDKN1A*	*EGFR*	*FAT1*	*GNA11*	*IL10*	*LMO1*	*MYCN*	*PDGFRB*	*PTPRT*	*SETD2*	*TCF3*	
BCL10	*CDKN1B*	*EIF1AX*	*FBXW7*	*GNA13*	*IL7R*	*LRP1B*	*MYD88*	*PDK1*	*QKI*	*SF3B1*	*TCF7L2*	
BCL2	*CDKN2A*	*EIF4A2*	*FGF1*	*GNAQ*	*INHA*	*LYN*	*MYOD1*	*PDPK1*	*RAB35*	*SH2B3*	*TERC*	
BCL2L1	*CDKN2B*	*EIF4E*	*FGF8*	*GNAS*	*HRAS*	*LZTR1*	*NAB2*	*PGR*	*RAC1*	*SH2D1A*	*TERT*[b]	
BCL2L11	*CDKN2C*	*EML4*	*FGF9*	*GPR124*	*HSD3B1*	*MAGI2*	*NBN*	*PHF6*	*RAD21*	*SHQ1*	*TET1*	
BCL2L2	*CEBPA*	*EP300*	*FGF10*	*GPS2*	*HSP90AA1*	*MALT1*	*NCOA3*	*PHOX2B*	*RAD50*	*SLIT2*	*TET2*	
BCL6	*CENPA*	*EPCAM*	*FGF14*	*GREM1*	*ICOSLG*	*MAP2K1*	*NCOR1*	*PIK3C2B*	*RAD51*	*SLX4*	*TFE3*	
BCOR	*CHD2*	*EPHA3*	*FGF19*	*GRIN2A*	*ID3*	*MAP2K2*	*NEGR1*	*PIK3C2G*	*RAD51B*	*SMAD2*	*TFRC*	
BCORL1	*CHD4*	*EPHA5*	*FGF2*	*GRM3*	*IDH1*	*MAP2K4*	*NF1*	*PIK3C3*	*RAD51C*	*SMAD3*	*TGFBR1*	

a. Large rearrangements (exon-level CNVs) detected for *BRCA1* and *BRCA2*.

b. TERT promoter region only covered for variant calling.

CNV calling is available for all genes except: *HIST2H3A*, *HIST2H3C*, *HLA-A*, *HLA-B*, *HLA-C*, *KMT2B*, *KMT2C*, *KMT2D*, *TERT*

Table 3: RNA content in the TruSight Oncology 500 TruSight Oncology 500 High-Throughput panels

ABL1	*EGFR*	*FGFR2*	*MLL*	*PAX3*
AKT3	*EML4*	*FGFR3*	*MLLT3*	*PAX7*
ALK	*ERBB2*	*FGFR4*	*MSH2*	*PDGFRA*
AR	*ERG*	*FLI1*	*MYC*	*PDGFRB*
AXL	*ESR1*	*FLT1*	*NOTCH1*	*PIK3CA*
BCL2	*ETS1*	*FLT3*	*NOTCH2*	*PPARG*
BRAF	*ETV1*	*JAK2*	*NOTCH3*	*RAF1*
BRCA1	*ETV4*	*KDR*	*NRG1*	*RET*
BRCA2	*ETV5*	*KIF5B*	*NTRK1*	*ROS1*
CDK4	*EWSR1*	*KIT*	*NTRK2*	*RPS6KB1*
CSF1R	*FGFR1*	*MET*	*NTRK3*	*TMPRSS2*

All genes listed are assessed for known and novel fusions; content shaded in gray is analyzed for splice variants.

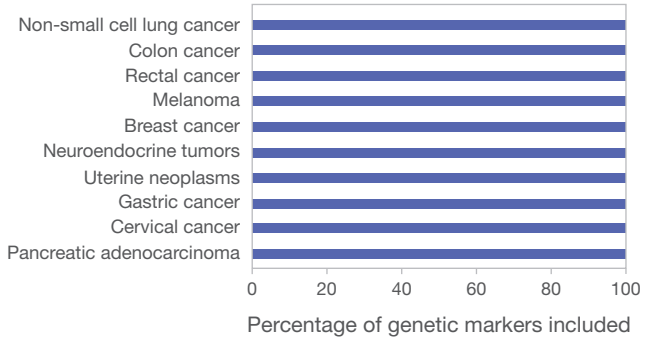

Figure 4: TruSight Oncology 500 content alignment to key guidelines by cancer type—The graph provides examples of content alignment; it is not meant to be all-inclusive.

Automate for efficiency

TruSight Oncology 500 and TruSight Oncology 500 High-Throughput offer manual and automated options to support scalable library prep. Illumina has partnered with Hamiton and Beckman Coulter Life Sciences, leading liquid-handling manufacturers, to produce fully automated workflows for TruSight Oncology 500 assays that support a range of throughput needs. These automated workflows achieve the same high-quality results produced by manual protocols, while reducing hands-on time by ~50%, enabling labs to save on labor costs and improve efficiency.

Add tags for analytical specificity

During library preparation, unique molecular identifiers (UMIs)[11] are added to the gDNA or cDNA fragments. These UMIs enable detection of variants at low variant allele frequency (VAF) while simultaneously suppressing errors, providing high analytical specificity.

Enrich libraries to focus efforts

Library preparation is based on proven hybrid–capture chemistry to purify selected targets from DNA- and RNA-based libraries. Biotinylated probes hybridize to regions of interest, which are pulled down using streptavidin-coated magnetic beads, and then eluted to enrich the library pool. Hybridization-based enrichment is a useful strategy for analyzing specific genetic variants in a given sample and reliably sequencing exomes or large numbers of genes (eg, > 50 genes). It delivers dependable results across a wide range of input types and quantities. Hybrid–capture chemistry offers several advantages over amplicon sequencing, including yielding data with fewer artifacts and dropouts. Additionally, hybrid-capture chemistry is fusion agnostic, enabling detection and characterization of known and novel fusions. Unlike amplicon-based approaches, which require confirmatory tests as false-positives can arise, the hybrid-capture method is highly sensitive and can accurately characterize gene fusions with both known and novel partners.

Table 4: Simultaneous analysis of multiple lung cancer biomarkers using DNA and RNA in the same sample

Biomarker	DNA content	RNA content
MSI	✓	
TMB	✓	
Biomarker genes	**Small variants**	**Fusions**
AKT1	✓	
ALK	✓	✓
BRAF	✓	✓
DDR2	✓	
EGFR	✓	✓
ERBB2	✓	✓
FGFR1	✓	✓
FGFR3	✓	✓
KRAS	✓	
MAP2K1	✓	
MET	✓	✓
NRAS	✓	
NTRK1	✓	✓
NTRK2	✓	✓
NTRK3	✓	✓
PIK3CA	✓	✓
PTEN	✓	
RET	✓	✓
TP53	✓	

Sequence 8–960 samples

TruSight Oncology 500 and TruSight Oncology 500 High-Throughput follow the same sample and library preparation workflow. The primary difference between the assays is scale. TruSight Oncology 500 runs on the NextSeq™ 550 or NextSeq 550Dx* Systems, which can batch up to eight samples at a time. TruSight Oncology 500 High-Throughput provides scalability to higher sample throughput. When run on the NovaSeq™ 6000 or NovaSeq 6000Dx* Systems, customers can batch from 16 to 192 samples. When run on the NovaSeq X Series, customers can batch from 32 to 480 samples in a single flow cell run and 64 to 960 samples in a dual flow cell run. This broad flexibility across platforms is enabled by the availability of 192 unique indexes for TruSight Oncology 500 High-Throughput and NovaSeq flow cells that accommodate varying throughput levels (Table 5). Each sample index performs consistently to produce sequencing metrics above quality control (QC) expectations.

Analyze data

Variant calling for TruSight Oncology 500 and TruSight Oncology 500 High-Throughput is available with DRAGEN secondary analysis, either on premises using a local DRAGEN Server or in the cloud using Illumina Connected Analytics, now with data streaming and autolaunch ca-

* NextSeq 550Dx or NovaSeq 6000Dx Instruments in research mode.

Table 5: Scalable solution

Assay	TruSight Oncology 500	TruSight Oncology 500 High-Throughput						
System	NextSeq 550 or NextSeq 550Dx System (research mode)	NovaSeq 6000 or NovaSeq 6000Dx System (research mode)				NovaSeq X Series[a]		
Flow cell	High-output	SP	S1	S2	S4	1.5B	10B	25B
No. samples	8	16	32	72	192	32	192	480

a. Throughput shown is for a single flow cell on the NovaSeq X System. The NovaSeq X Plus System offers a dual flow cell option with twice the capacity listed.

pabilities. Both versions take advantage of sophisticated proprietary algorithms that remove errors, artifacts, and germline variants, resulting in highly accurate variant calling performance with an analytical specificity of > 99.9995%. This level of specificity is particularly beneficial when it is critical to know the exact number of mutations per Mb, as in TMB evaluation with a tumor-only workflow. DNA variant data analyzed with the TruSight Oncology 500 Local App† and DRAGEN TruSight Oncology 500 show concordant results (Figure 5C, Figure 6C); however, analysis with the DRAGEN pipeline is completed 2–4× faster than with the local app (Table 6), reducing the time to final results.

To learn more about Illumina Connected Analytics, read the Security, privacy, and compliance with Illumina Connected Analytics technical note.

Variant insights and report generation are available through integration with Illumina Connected Insights and other commercial providers, such as Velsera Clinical Genomics Workspace. Variant calling files produced locally or via the cloud with Illumina Connected Analytics can be uploaded into the preferred tertiary analysis tool. From potentially thousands of variants, biologically relevant variants can be filtered and prioritized into a final, customizable report.

Table 6: Faster analysis using DRAGEN TruSight Oncology 500 Analysis Software

No. tissue biopsy samples	Average time for analysis to complete[a]	
	Local App[b]	DRAGEN pipeline[c]
8	5.5 hr	2 hr
16	12 hr	3 hr
32	18 hr	5 hr
72	24 hr	10 hr

a. Analysis times are based on actual runs and will vary from run to run.
b. Local server specifications: Amazon EC2, c5.9xlarge instance (36 vCPU, 72 GiB memory); analysis time will vary with server specifications.
c. Time for the DRAGEN pipeline run on the DRAGEN Server v3.

† Previous generation of TruSight Oncology 500 software (not based on the DRAGEN pipeline).

Proven, reliable results

Although TruSight Oncology 500 and TruSight Oncology 500 High-Throughput were designed to run on separate sequencing platforms with different throughput options, the assays have the same genomic content and performance expectations for variant calling. Both assays demonstrate high concordance when detecting MSI, TMB, CNVs, small variants, and fusions.

Accurate assessment of TMB and MSI

TruSight Oncology 500 and TruSight Oncology 500 High-Throughput are well suited to interrogate MSI and TMB, which rely upon analysis of multiple genomic loci. Traditionally, MSI status has been analyzed with PCR (MSI-PCR) and immunohistochemistry. While other methods deliver a qualitative result describing samples as either MSI-stable or MSI-high, NGS-based assessment with the TruSight Oncology 500 assays interrogates 130 homopolymer MSI marker sites to calculate an accurate quantitative score for MSI status (Figure 5).[12]

Obtaining a precise and reproducible TMB value at low mutation levels can be challenging with smaller panels. TruSight Oncology 500 panels combine comprehensive genomic content with sophisticated informatics algorithms to provide accurate TMB estimation that is highly concordant with whole-exome studies (Figure 6, Table 7).[12] The addition of UMIs during library preparation coupled with proprietary Illumina informatics reduces sequencing error rates by 10–20 fold.[11] Removing FFPE artifacts (such as deamination, oxidation) enables analytical sensitivity as low as 5% VAF from low-quality DNA samples.

Table 7: High concordance between whole-exome sequencing (WES) and TruSight Oncology 500 for TMB classification at 10 mutations/Mb

Metric	Value
Percent positive agreement	94.7%
Percent negative agreement	96.1%
Overall percent agreement	95.4%

Based on TMB values from 108 FFPE tissue samples; percent agreement is shown for TMB-high or TMB-low classifications, with 10 mutations/Mb as the threshold value.

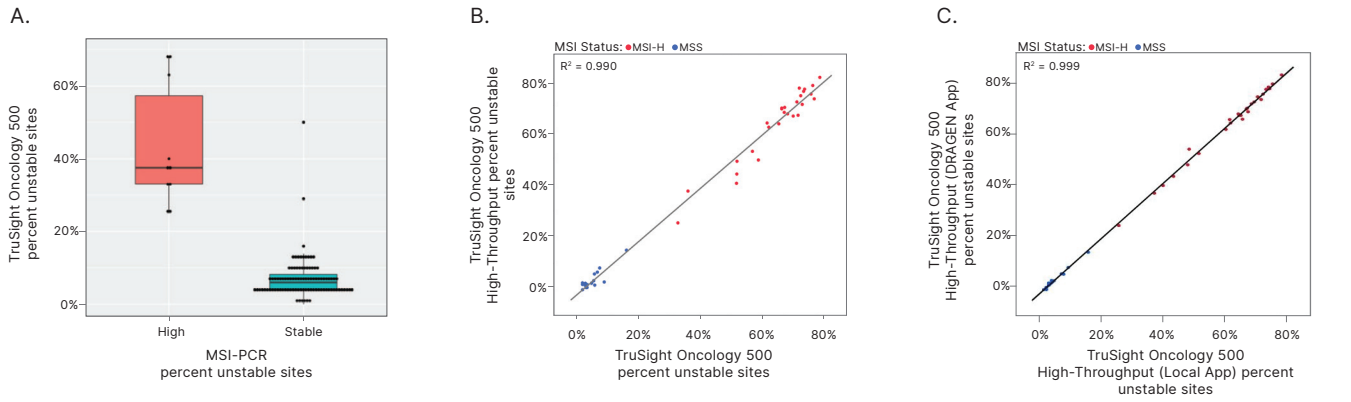

Figure 5: Accurate assessment of MSI status—(A) FFPE tissue samples analyzed using TruSight Oncology 500 produce a quantitative score (Y-axis) compared to a qualitative score using MSI-PCR (X-axis). (B) High concordance of MSI analysis between TruSight Oncology 500 and TruSight Oncology 500 High-Throughput. (C) High concordance between TruSight Oncology 500 data analyzed using DRAGEN TruSight Oncology 500 v2 software and the TruSight Oncology 500 Local App v2.2.

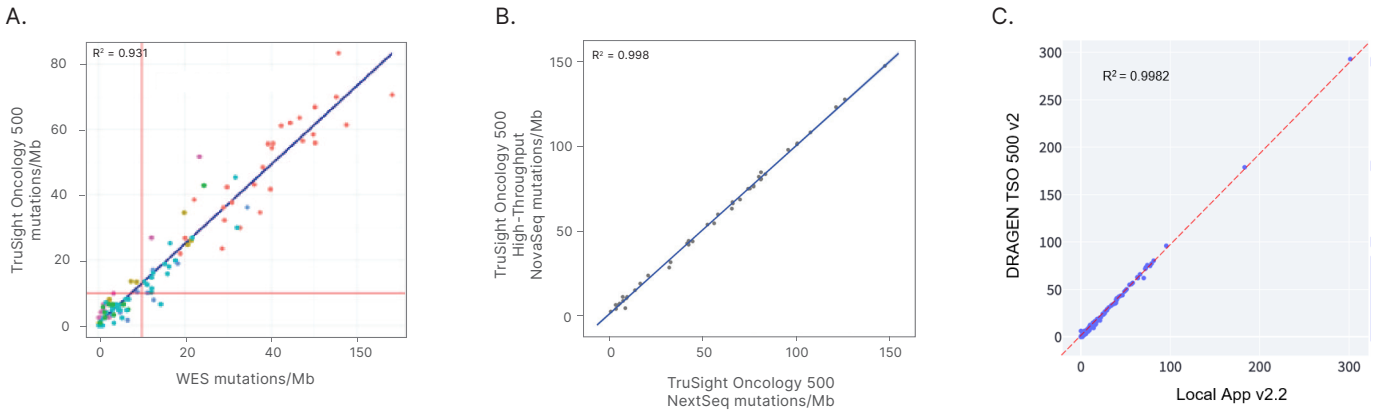

Figure 6: Accurate assessment of TMB status—(A) Analysis of 108 FFPE tissue samples shows high concordance between TMB measurements using WES and TruSight Oncology 500. The red line indicates the threshold value (10 mutations/Mb). (B) High concordance of TMB analysis between TruSight Oncology 500 and TruSight Oncology 500 High-Throughput. (C) High concordance between TruSight Oncology 500 data analyzed using DRAGEN TruSight Oncology 500 v2 software and the TruSight Oncology 500 Local App v2.2.

Sensitive detection of CNVs

Copy-number changes in several genes and tumor types have been associated with tumorigenesis.[13] Both TruSight Oncology 500 assays include analysis of 514 CNV associated genes and can call amplifications with a limit of detection at 2.2× fold change and deletions at 0.5× fold change (Figure 7, Table 8).

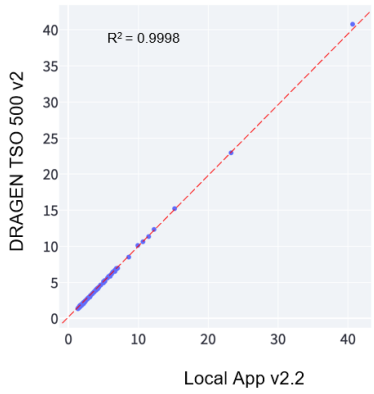

Figure 7: High concordance of CNV detection between TruSight Oncology 500 data analyzed using DRAGEN TruSight Oncology v2 software and TruSight Oncology 500 Local App v2.2. CNV comparison was made with 59 genes.

Detection of *BRCA* large rearrangements

A *BRCA* large rearrangement (LR) step in the DRAGEN TruSight Oncology 500 analysis workflow enables exon-level CNV detection for *BRCA* 1 and 2 genes. For three or more exons, sensitivity is 43%, while for under three exons, sensitivity is 50% on the NextSeq 550 System (Table 9).

Highly sensitive variant detection from FFPE samples

One benefit of target enrichment chemistry is the use of probes designed large enough to impart high binding specificity, but also allow hybridization to targets containing small mutations. This mechanism reduces sample dropouts in the presence of both natural allelic variations and sequence artifacts introduced from FFPE tissue samples. The assay can reproducibly detect variants in FFPE samples as low as 5% VAF (Figure 8, Table 10).

Table 8: Sensitive CNV detection

	Mean fold change	
Gene	DRAGEN TruSight Oncology 500 v2	TruSight Oncology 500 Local App v2.2
AR	2.03	2.17
BRAF	2.09	2.09
BRCA1	1.42	1.42
BRCA2	1.92	1.93
CCND1	4.15	4.14
CCNE1	1.62	1.63
CDK4	3.23	3.24
CDK6	1.85	1.84
CHEK2	1.65	1.68
EGFR	3.55	3.53
ERBB2	8.63	8.66
FGF10	1.60	1.59
FGF19	3.28	3.30
FGFR1	3.57	3.57
KRAS	2.19	2.19
MDM2	2.46	2.47
MDM4	1.65	1.64
MET	1.70	1.69
MYC	1.97	1.98
MYCN	1.45	1.46

The information in this table shows examples of high concordance of data analyzed with DRAGEN TruSight Oncology 500 v2 software and the TruSight Oncology 500 Local App v2.2 and is not a comprehensive list of the CNVs detected.

Robust detection of fusions

Cancer can arise from epigenetic changes, expression level changes, and gene fusions that are undetectable by standard sequencing.[14,15] The TruSight Oncology 500 assays detect and characterize fusions agnostic from the partner. To achieve comparable results with RNA analysis, 40 ng RNA is recommended for use with TruSight Oncology 500 while a range of 40–80 ng RNA is recommended for use with TruSight Oncology 500 High-Throughput. In cases where FFPE RNA yields from FFPE tissues are low, 40 ng RNA input can still be used to detect variants expressed at mid to high levels with TruSight Oncology 500 High-Throughput. However, when available, 80 ng RNA input helps maximize sensitivity for fusions present at low concentrations (Table 11).

Table 9: Sensitive detection of *BRCA* LR

BRCA 1/2 LR detected	Estimated LR VAF
BRCA1 loss exon 8	0.26
BRCA2 loss exon 21–24	0.44
BRCA1 loss exon 14–24	0.51
BRCA1 loss exon 21–24	0.85
BRCA1 loss exon 1–3	0.48
BRCA1 loss exon 1–23	0.70
BRCA2 gain exon 25–27	0.37
BRCA1 loss exon 1–3	0.86
BRCA1 gain exon 5–16	0.83
BRCA1 gain exon 17–18	0.51
BRCA1 gain exon 1–16	0.61
BRCA1 gain exon 13	0.69
BRCA2 gain exon 25	0.40
BRCA2 gain exon 11–27	0.54
BRCA2 gain exon 12–13	0.35
BRCA1 loss exon 22	0.92

These data were generated with DRAGEN TruSight Oncology 500 v2 software.

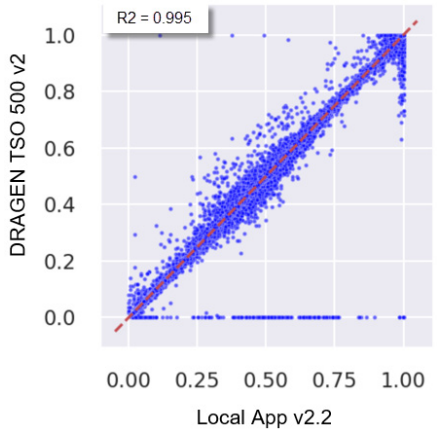

Figure 8: High concordance of VAF between TruSight Oncology 500 data analyzed using DRAGEN TruSight Oncology v2 software and TruSight Oncology 500 Local App v2.2.

Table 10: Highly sensitive DNA small variant detection

Gene	Mutation	DRAGEN TruSight Oncology 500 v2 (Mean VAF)
Variant type: single nucleotide variant (SNV)		
AKT1	E17K	5%
APC	R1450*	8%
BRAF	V600E	13%
CTNNB1	T41A	8%
EGFR	L858R	7%
EGFR	T790M	7%
FGFR3	S249C	6%
FOXL2	C134W	7%
GNAS	R201C	7%
IDH1	R132C	7%
KIT	D816V	8%
KRAS	G12D	7%
NOTCH1	P668S	5%
NRAS	Q61R	7%
PIK3CA	E545G	5%
RET	M918T	8%
TP53	R248Q	7%
Variant type: complex variant		
EGFR	L747_P753>Q	3%
Variant type: insertion		
APC	T1556Nfs*3	7%
ERBB2	A775-G776insYVMA	7%
Variant type: deletion		
FBXW7	FBXW7:G667fs	5%
PTEN	PTEN:K267fs*9	7%
TP53	TP53:C242fs*5	6%

The information in this table shows examples of concordance between data analyzed with DRAGEN TruSight Oncology 500 v2 software and the TruSight Oncology 500 Local App v2.2 and is not a comprehensive list of the SNVs and indels detected.

Table 11: Robust detection of fusions and splice variants

RNA fusion	RNA input amount			Tissue
	40 ng	60 ng	80 ng	
ALK-EML4	15	21	40	Lung
EGFR-RAB3IP	5	9	19	Brain
EGFR-METTL1	25	84	71	Brain
BRCA1-MPP2	25	28	29	Unknown
ALK-BRE	75	112	128	Sarcoma
CCDC170-ESR1	122	59	168	Kidney
MYC-MRPL13	27	35	52	Breast
MYC-STK3	11	39	28	Breast
ROS1;GOPC-ENC1	32	53	93	Lung
ROS1;GOPC-CD74	104	92	141	Lung
ANKUB1; RNF13-ETV5;DGKG	29	45	72	Uterus
NTRK3-SEMA6A	7	16	25	Skin
RET-NCOA4	74	78	154	Thyroid
EWSR1-ATF1	19	30	32	Sarcoma
EWSR1-CBY1	44	30	97	Sarcoma
BRCA2-NRXN3	33	60	84	Bone
FLT3-SMOX	50	72	54	Bone
FLT3-VWA8	29	51	69	Bone
FLT3-LCP1	12	32	47	Bone
Splice variant				
ARv7	26	38	46	Breast
EGFR v3	567	884	937	Brain
EGFR v3	1249	1614	2049	Brain

These data were generated with a Local App pipeline (not DRAGEN software).

Plan for the future

TruSight Oncology 500 and TruSight Oncology 500 High-Throughout integrate easily into labs currently using NGS, enabling them to offer CGP capabilities without exploring an entirely new technology. By consolidating multiple independent, single biomarker assays into one assay, labs can save sample, time, and money, while increasing the chances of identifying a positive biomarker. In addition, bringing tumor assays in house allows labs to keep sample and raw data.

Enhanced product attributes

Illumina offers high levels of service and support to ensure operational success for laboratories. To enable greater efficiency, TruSight Oncology 500 products‡ feature:

- Advanced change notification—Illumina notifies laboratories six months before any significant changes are made to a product in the TruSight Oncology 500 portfolio†
- Certificate of Analysis—Every TruSight Oncology 500 product† is issued with a certificate of analysis (CoA) by the Illumina Quality Assurance Department that ascertains the product has met its predetermined product release specifications and quality
- Extended shelf life—The minimum guaranteed shelf life for TruSight Oncology 500 reagents is extended to six months, reducing the risk of product expiration and enabling labs to use reagents according to current testing needs

Summary

TruSight Oncology 500 and TruSight Oncology 500 High-Throughput are NGS-based, hybrid-capture assays that enable CGP through analysis of key biomarkers present in guidelines and clinical trials, in a single assay using a small amount of sample. Combining DNA and RNA hybrid-capture with sophisticated informatics reduces errors and yields high-quality data, even from FFPE samples. With TruSight Oncology 500 High-Throughput, labs can increase their batch sizes and process more samples per week across a broad range of sequencing platforms. Leveraging the power of DRAGEN secondary analysis enables TruSight Oncology 500 to improve lab efficiency and produce meaningful results.

‡ For TruSight Oncology 500 bundles on the NextSeq 550Dx Instrument, enhanced features apply only to library preparation kits and not to core consumables.

Learn more

TruSight Oncology 500 and TruSight Oncology 500 High-Throughput

DRAGEN secondary analysis

Illumina Connected Analytics

Illumina Connected Insights

Ordering information: TruSight Oncology 500

	Sample type	Library prep		Velsera included
		Product	Catalog no.	
Manual	DNA	TruSight Oncology 500 DNA Kit[a] (16 indexes, 48 samples)	20028213	
		TruSight Oncology 500 DNA Kit, plus Velsera[a] (16 indexes, 48 samples)	20032624	✓
		TruSight Oncology 500 DNA Kit, for Use with NextSeq[b] (16 indexes, 48 samples)	20028214	
		TruSight Oncology 500 DNA Kit, for Use with NextSeq, plus Velsera[b] (16 indexes, 48 samples)	20032625	✓
	DNA/RNA	TruSight Oncology 500 DNA/RNA Bundle[a] (16 indexes, 24 samples)	20028215	
		TruSight Oncology 500 DNA/RNA Bundle, plus Velsera[a] (16 indexes, 24 samples)	20032626	✓
		TruSight Oncology 500 DNA/RNA Bundle, for Use with NextSeq[b] (16 indexes, 24 samples)	20028216	
		TruSight Oncology 500 DNA/RNA Bundle, for Use with NextSeq, plus Velsera[b] (16 indexes, 24 samples)	20032627	✓
Automated	DNA	TruSight Oncology 500 DNA Automation[a] Kit (16 indexes, 64 samples)	20045504	
		TruSight Oncology 500 DNA Automation Kit, plus Velsera[a] (16 indexes, 64 samples)	20045506	✓
		TruSight Oncology 500 DNA Automation Kit, for Use with NextSeq[b] (16 indexes, 64 samples)	20045505	
		TruSight Oncology 500 DNA Automation Kit, for Use with NextSeq, plus Velsera[b] (16 indexes, 64 samples)	20045507	✓
	DNA/RNA	TruSight Oncology 500 DNA/RNA[a] Automation Kit (16 indexes, 32 samples)	20045508	
		TruSight Oncology 500 DNA/RNA Automation Kit, plus Velsera[a] (16 indexes, 32 samples)	20045509	✓
		TruSight Oncology 500 DNA/RNA Automation Kit, for Use with NextSeq[b] (16 indexes, 32 samples)	20045990	
		TruSight Oncology 500 DNA/RNA Automation Kit, for Use with NextSeq, plus Velsera[b] (16 indexes, 32 samples)	20045991	✓

a. Includes library prep and enrichment reagents; does not include NextSeq 550 System sequencing reagents. NextSeq 550 System sequencing reagents are available separately. Visit illumina.com/products/by-type/sequencing-kits/cluster-gen-sequencing-reagents/nextseq-series-kits-v2-5.html.
b. Includes library prep and enrichment reagents and NextSeq 550 System sequencing reagent.

Ordering information: TruSight Oncology 500 and TruSight Oncology 500 High-Throughput

	Sample type	Library prep		Velsera included	Automation	
		Product	Catalog no.		Product	Catalog no.
Manual	DNA	TruSight Oncology 500 DNA High-Throughput Kit[a] (48 samples)	20040765		Beckman Coulter i-Series	Contact Illumina sales
		TruSight Oncology 500 DNA High-Throughput Kit, with Velsera[a] (48 samples)	20040769	✓	Hamilton Microlab STAR	Contact Illumina sales
		TruSight Oncology 500 DNA High-Throughput Kit[a] (144 samples)	20040767			
		TruSight Oncology 500 DNA High-Throughput, with Velsera[a] (144 samples)	20040771	✓		
	DNA/RNA	TruSight Oncology 500 DNA/RNA High-Throughput Kit[a] (24 samples)	20040764			
		TruSight Oncology 500 DNA/RNA High-Throughput Kit, with Velsera[a] (24 samples)	20040768	✓		
		TruSight Oncology 500 DNA/RNA High-Throughput Kit[a] (72 samples)	20040766			
		TruSight Oncology 500 DNA/RNA High-Throughput Kit, with Velsera[a] (72 samples)	20040770	✓		
Automated	DNA	TruSight Oncology 500 DNA High-Throughput Automation Kit[a] (64 samples)	20049283			
		TruSight Oncology 500 DNA High-Throughput Automation Kit[a] (64 samples) plus Velsera	20049277	✓		
		TruSight Oncology 500 DNA High-Throughput Automation Kit[a] (144 samples)	20049285			
		TruSight Oncology 500 DNA High-Throughput Automation Kit[a] (144 samples) plus Velsera	20049279	✓		
	DNA/RNA	TruSight Oncology 500 DNA/RNA High-Throughput Automation Kit[a] (32 samples)	20049282			
		TruSight Oncology 500 DNA/RNA High-Throughput Automation Kit[a] (32 samples) plus Velsera	20049276	✓		
		TruSight Oncology 500 DNA/RNA High-Throughput Automation Kit[a] (72 samples)	20049284			
		TruSight Oncology 500 DNA/RNA High-Throughput Automation Kit[a] (72 samples) plus Velsera	20049278	✓		

a. Includes library prep and enrichment reagents; does not include IDT for Illumina indexes or sequencing reagents for the NovaSeq 6000 System or NovaSeq X Series.

Ordering information: TruSight Oncology 500 High-Throughput, continued

Consumables		
Product		Catalog no.
Index kits		
Manual	IDT for Illumina UMI DNA/RNA UD Indexes Set A, Ligation (96 indexes, 96 samples)	20034701
Manual	IDT for Illumina UMI DNA/RNA UD Indexes Set B, Ligation (96 indexes, 96 samples)	20034702
Automated	IDT for Illumina UMI DNA/RNA UD Indexes for Automation Set A, Ligation (96 indexes, 96 samples)	20066404
Automated	IDT for Illumina UMI DNA/RNA UD Indexes for Automation Set B, Ligation (96 indexes, 96 samples)	20063213
NovaSeq 6000 sequencing reagent kits		
NovaSeq 6000 SP Reagent Kit v1.5 (200 cycles)		20040719
NovaSeq 6000 S1 Reagent Kit v1.5 (200 cycles)		20028318
NovaSeq 6000 S2 Reagent Kit v1.5 (200 cycles)		20028315
NovaSeq 6000 S4 Reagent Kit v1.5 (200 cycles)		20028313
NovaSeq X sequencing reagent kits		
NovaSeq X Series 1.5B Reagent Kit (200 cycles)		20104704
NovaSeq X Series 10B Reagent Kit (200 cycles)		20085595
NovaSeq X Series 25B Reagent Kit (300 cycles)		20104706

Ordering information: Analysis options

On-premises variant calling	
Product	Catalog no.
Illumina DRAGEN Server v3	20040619
Illumina DRAGEN Server v4	20051343
Illumina DRAGEN Server Advance Exchange Plan	20032797
Cloud-based variant calling	
Illumina Connected Analytics Basic Annual Subscription	20044874
Illumina Connected Analytics Professional Annual Subscription	20044876
Illumina Connected Analytics Enterprise Annual Subscription	20038994
Illumina Connected Analytics Enterprise Compliance Add-on (applies to Basic only)	20066830
Illumina Connected Analytics Training and Onboarding	20049422
Illumina Connected Analytics Data Storage: Illumina Analytics, 1 credit	20042038
Illumina Connected Analytics Data Storage: Illumina Analytics Starter Pack, 1000 credits	20042039
Illumina Connected Analytics Data Storage: Illumina Analytics, 5000 credits	20042040
Illumina Connected Analytics Data Storage: Illumina Analytics, 50,000 credits	20042041
Illumina Connected Analytics Data Storage: Illumina Analytics, 100,000 credits	20042042
Cloud-based variant reporting	
Illumina Connected Insights—Annual Subscription	20090137
Illumina Connected Insights—Oncology Genome Equivalent Sample-VCF	20090138
Illumina Connected Insights—Training and Onboarding	20092376
Informatics Professional Services	20071787

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