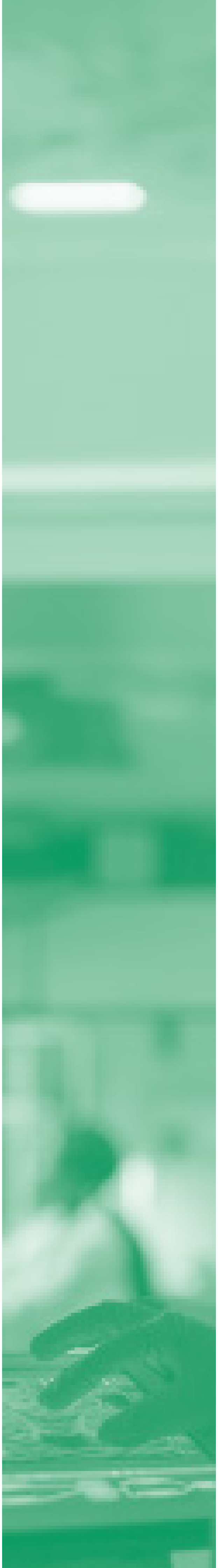


SEP 2023

strand
WE SOLVE PROBLEMS IN GENOMICS.

TECH NOTE

Showcasing Strand’s Expertise: **A Closer Look at StrandOmics**

StrandOmics is Strand’s cloud-based HIPAA-compliant platform for variant classification and interpretation.

It enables somatic variant visualization, prioritization, and reporting and is a customizable platform that has been used extensively in our engagements with key diagnostics customers.

In fact, StrandOmics and associated tools have been used to generate over 10,000 clinical reports.

Unveiling Key Features of StrandOmics

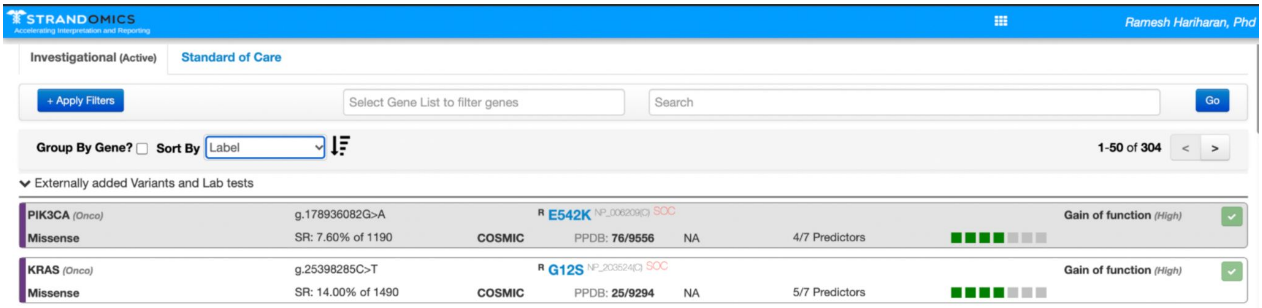
StrandOmics is driven by a range of key technology components and features, each enabling distinct functionalities. These functionalities are detailed below.

Variant call format file (VCF) input and parsing:

StrandOmics supports the parsing of VCF version 4.2 files that are input. It is capable of parsing SNPs, indels, structural variants (CNVs), and fusion variants (translocations). Additionally, it parses the phase from unphased VCF files. Furthermore, StrandOmics conducts parsing for all standard variant callers, including GATK, Illumina, built-in, and FreeBayes.

Variant prioritization engine:

StrandOmics is equipped with a variant prioritization engine that allows users to sort or report variants within a specimen based on parameters such as function, allele frequency, Pooled Population Database (PPDB) frequency, and more.



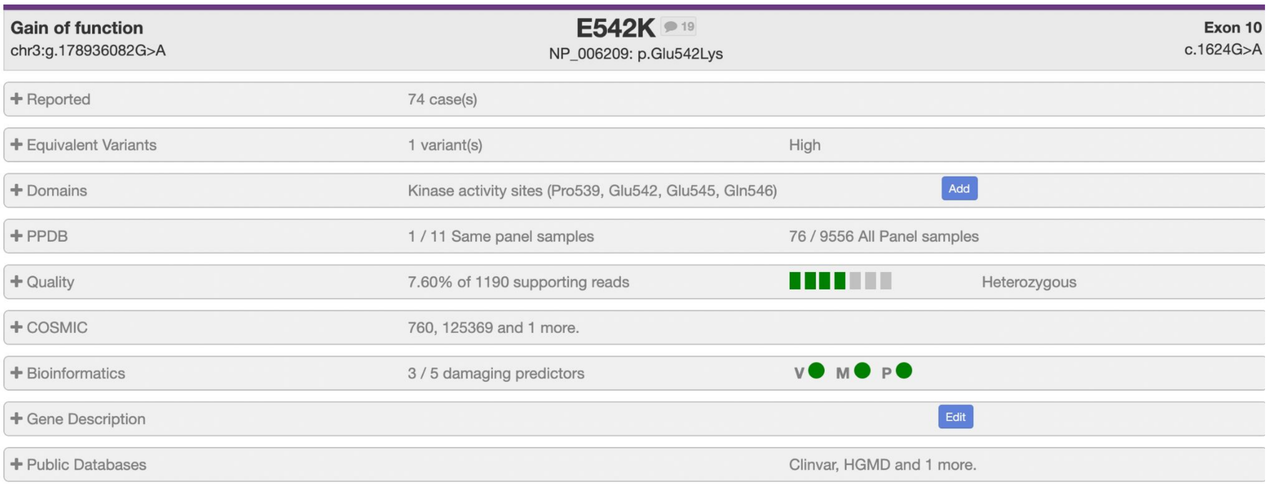
PIK3CA (Onco) g.178936082G>A R E542K NP_006209(C) SOC Gain of function (High) ✓
Missense SR: 7.60% of 1190 COSMIC PPDB: 76/9556 NA 4/7 Predictors

KRAS (Onco) g.25398285C>T R G12S NP_203524(C) SOC Gain of function (High) ✓
Missense SR: 14.00% of 1490 COSMIC PPDB: 25/9294 NA 5/7 Predictors

Figure 1: The top 2 variants in StrandOmics, as sorted by label, in descending order, for a colorectal cancer specimen.

Variant Card and Variant Quality:

Within StrandOmics are variant cards that contain specimen-level variant information, capturing all variant-level data in a single view. For example, a variant card for a *PIK3CA* mutation in a colorectal cancer specimen includes information such as the number of cases in which this variant was reported, equivalent *in vitro/in vivo* variants, domain information on kinase activity, variant quality encapsulated by various read-level statistics, COSMIC information, and additional data consolidated from various sources.



Gain of function
chr3:g.178936082G>A

E542K
NP_006209: p.Glu542Lys

Exon 10
c.1624G>A

+ Reported	74 case(s)		
+ Equivalent Variants	1 variant(s)	High	
+ Domains	Kinase activity sites (Pro539, Glu542, Glu545, Gln546)		
+ PPDB	1 / 11 Same panel samples	76 / 9556 All Panel samples	
+ Quality	7.60% of 1190 supporting reads		Heterozygous
+ COSMIC	760, 125369 and 1 more.		
+ Bioinformatics	3 / 5 damaging predictors		
+ Gene Description			
+ Public Databases		Clinvar, HGMD and 1 more.	

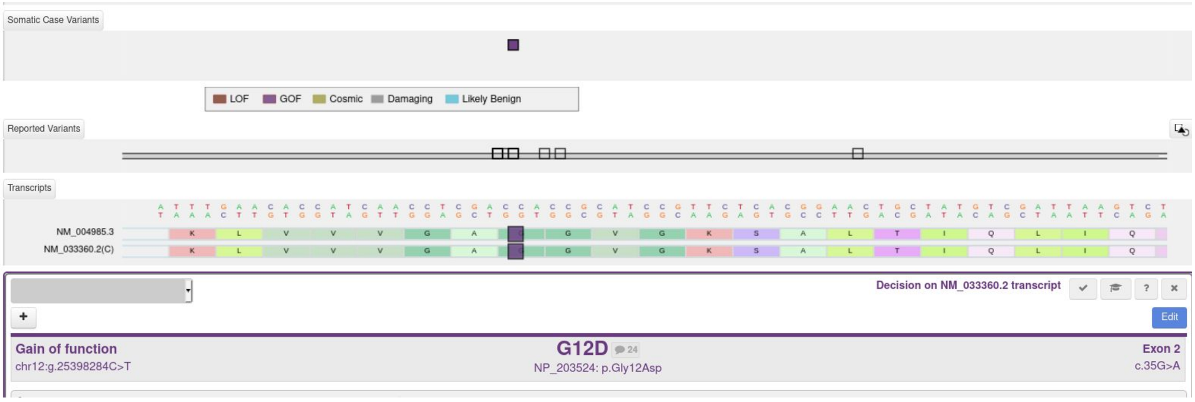
Figure 2: The variant card for the E542K variant in the PIK3CA gene of a CRC specimen

Embracing the Visual Realm

StrandOmics seamlessly merges variant information with visual insights:

Genome Browser:

The Genome Browser offers an intuitive visualization of CNVs, SNVs, and fusions within their immediate context. This interactive view presents variants based on nature, read depth, frequency, and phase.



Decision on NM_033360.2 transcript

Gain of function
chr12:g.25398284C>T

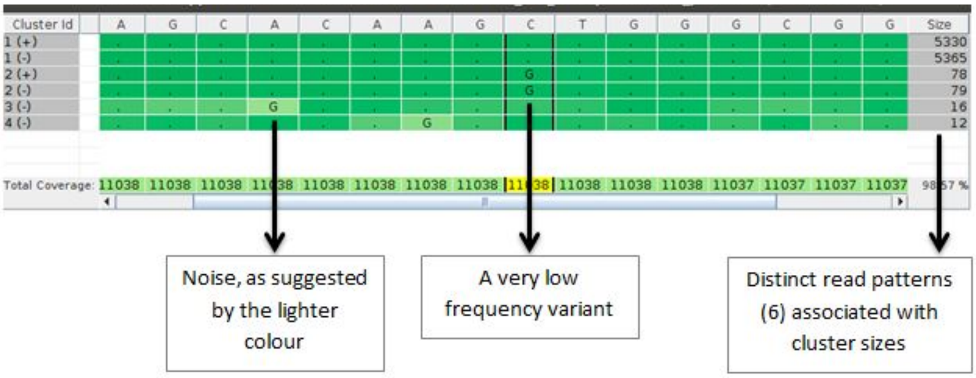
G12D 24
NP_203524: p.Gly12Asp

Exon 2
c.35G>A

Figure 3: An example of how an SNV G12D in chr12 is visualized in the Genome Browser

Variant Support View:

The VSV provides the user with the ability to view variants by nature, read depth, frequency, and phase. It does so by clustering reads and presenting all the read patterns about the variant. It is worth highlighting that the VSV enables instant verification of the variants by color coding the clusters based on average base quality/mapping quality.



Cluster Id	A	G	C	A	C	A	A	G	C	T	G	G	G	C	G	G	Size
1 (+)	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	5330
1 (-)	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	5365
2 (+)	.	.	.	.	.	.	.	.	G	.	.	.	.	.	.	.	78
2 (-)	.	.	.	.	.	.	.	.	G	.	.	.	.	.	.	.	79
3 (-)	.	.	.	G	.	.	.	.	.	.	.	.	.	.	.	.	16
4 (-)	.	.	.	.	.	.	G	.	.	.	.	.	.	.	.	.	12
Total Coverage:	11038	11038	11038	11[illegible]38	11038	11038	11038	11038	11[illegible]38	11038	11038	11038	11037	11037	11037	11037	98[illegible]57 %

Noise, as suggested by the lighter colour

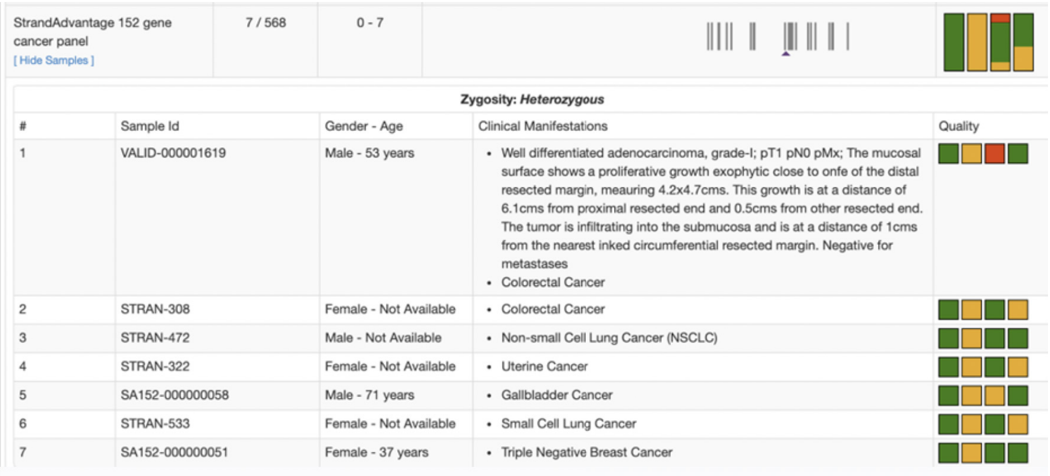
A very low frequency variant

Distinct read patterns (6) associated with cluster sizes

Figure 4: VSV showing clusters of haplotypes in a particular region, along with read support. The bases are colored by mean base quality, with light green representing lower base quality.

Historical annotations provided by the pooled patient database (PPDB):

Leveraging the pooled patient database (PPDB), StrandOmics captures variant instances across panels and specimens. Query-based on panel type and tumor type to access a detailed record of variant occurrences, complete with quality parameters.



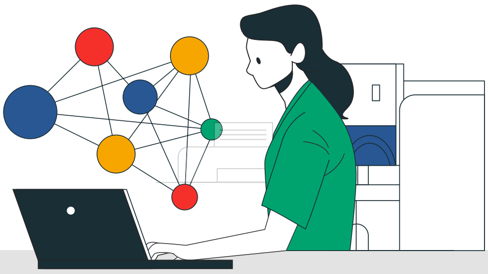
StrandAdvantage 152 gene cancer panel
[Hide Samples]
7 / 568
0 - 7

Zygosity: *Heterozygous*

#	Sample Id	Gender - Age	Clinical Manifestations	Quality
1	VALID-000001619	Male - 53 years	• Well differentiated adenocarcinoma, grade-I; pT1 pN0 pMx; The mucosal surface shows a proliferative growth exophytic close to onfe of the distal resected margin, meauring 4.2x4.7cms. This growth is at a distance of 6.1cms from proximal resected end and 0.5cms from other resected end. The tumor is infiltrating into the submucosa and is at a distance of 1cms from the nearest inked circumferential resected margin. Negative for metastases • Colorectal Cancer	🟩🟨🟥🟩
2	STRAN-308	Female - Not Available	• Colorectal Cancer	🟩🟨🟩🟨
3	STRAN-472	Male - Not Available	• Non-small Cell Lung Cancer (NSCLC)	🟩🟨🟩🟩
4	STRAN-322	Female - Not Available	• Uterine Cancer	🟩🟨🟩🟨
5	SA152-000000058	Male - 71 years	• Gallbladder Cancer	🟩🟨🟨🟩
6	STRAN-533	Female - Not Available	• Small Cell Lung Cancer	🟩🟨🟩🟨
7	SA152-000000051	Female - 37 years	• Triple Negative Breast Cancer	🟩🟨🟩🟩

Figure 5: PPDB provides a detailed view of the frequencies of PIK3CA E542K mutation in a 152 gene cancer panel. B, M, S and T stand for base quality, mapping quality, strand bias, and tail distance bias.

Lastly, this list of features would not be complete without mentioning the key pillars of StrandOmics— the knowledge bases (KBs). For more details on how StrandOmics is shaping the landscape of somatic and germline variant classification, please refer to our tech note.



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Our Genomics Solutions

We help you speed up the reporting of clinical NGS tests, both somatic and germline.

We enable pharmaceutical companies with clinical data and bioinformatics solutions for target discovery, translational research, and clinical research.

We help you build complex genomic software for managing and analyzing data and metadata from the entire gamut of omics modalities. We can help you accelerate releases and scale software operations.