

TruSeq Custom Amplicon Low Inputキットを用いた ターゲットリシーケンス – ドライ編 –

Mar 11, 2016



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テクニカルアプリケーションサイエンティスト

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本日の内容

▶ BaseSpace cloud app, TruSeq Ampliconを用いたデータ解析

- ーシーケンスデータの参照
- ー変異の解析結果の参照

▶ VariantStudioソフトウェアを用いたデータ解析

- ーデータのインポート
- ー変異の絞り込み
- ーアノテーション
- ーデータベースを利用した知見の入手

注：解析にはデモデータを使用しており、細かくは実際と異なる場合がございます。
データの参照と解析の流れをご案内する一例としてご参考下さい。

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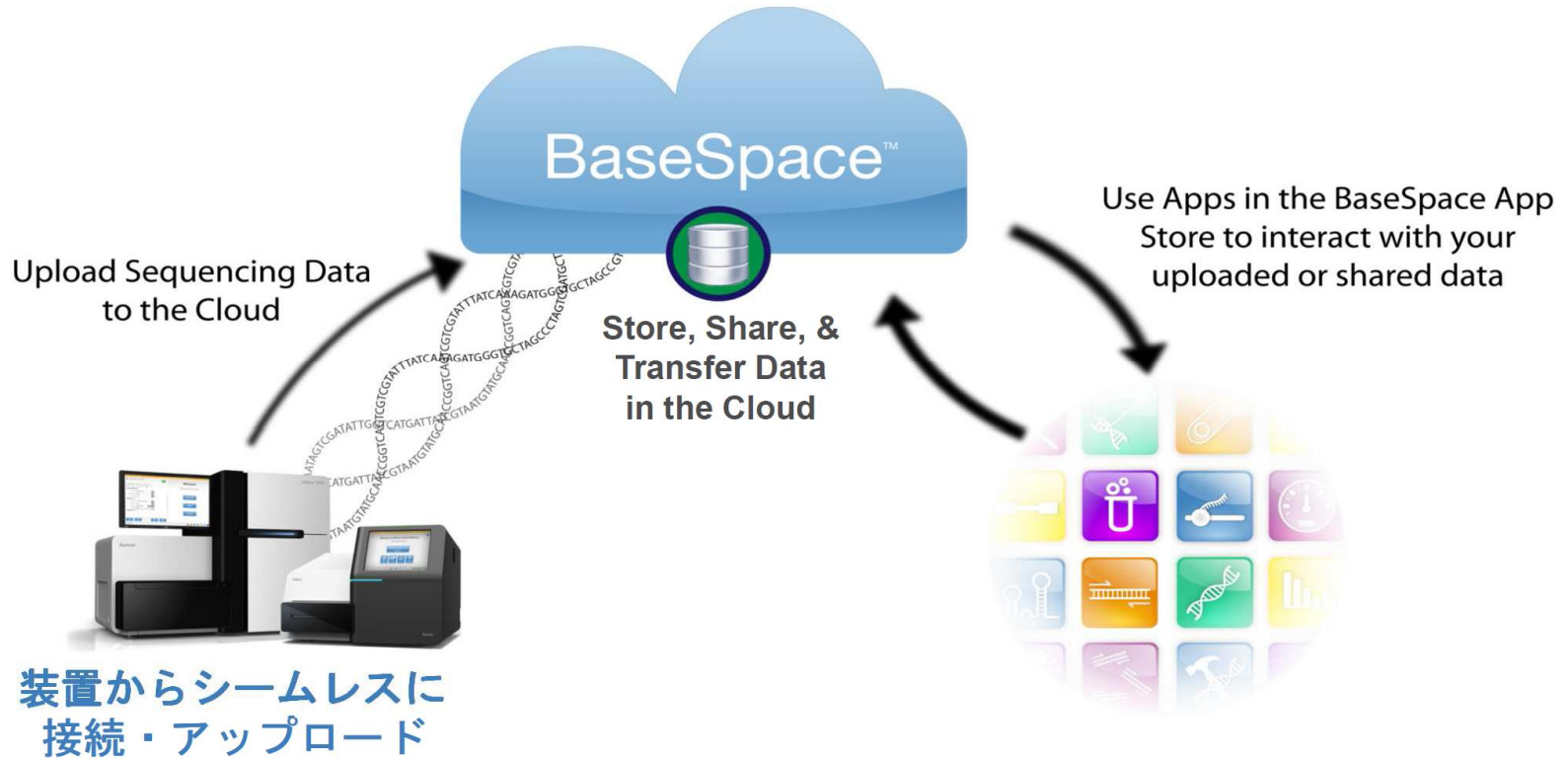
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BaseSpace cloudとは

Seamless upload, monitoring, storage, analysis, and collaboration

























The Future: Cloud Solutions will be everywhere and specifically valuable to researchers

BaseSpace appsとは

<イルミナコアアプリ>

 16S Metagenomics	 TopHat Alignment	 Cufflinks Assembly & DE	 RNA Express	 Variant Studio
 BWA Enrichment	 Isaac Enrichment	 Broad IGV	 TruSeq Amplicon	 Amplicon DS
 BWA WGS	 Isaac WGS	 Tumor Normal	 Long Read Assembly	 Long Read Phasing

<他社製アプリ>

 SPAdes	 Novo Align	 Advaita	 DNA Star
 AB SCIEX	 AB SCIEX	 AB SCIEX	 AB SCIEX
 SWATH Atlas	 MetaPhlan	 n of One	 My FLQ
 Lo Feq	 eGB	 Genomatix	 Genome Profiler
 OncoMD	 GeneTalk	 PathGEN Dx	 Melanoma Profiler
 TUTE	 DeepCheck HIV.HBV.HCV	 PEDANT	他

<イルミナラボアプリ>

 FastQC	 Kraken Metagenomics	 NextBio Annotates	 VCAT	 NextBio Transporter	 SRST2
 Fastq Toolkit	 Velvet Assembly	 Picard Space	 SRA Importer	 Prokka	他

TruSeq Amplicon app



TruSeq Amplicon
Illumina, Inc. [More info »](#)

Free

CATEGORIES Resequencing, Targeted Sequencing, Variant Analysis

Description

The TruSeq Amplicon workflow enables streamlined analysis of Illumina TruSeq Amplicon panels. Alignment is performed using the banded Smith-Waterman algorithm in the targeted regions (specified in a manifest file). For variant calling, users can select GATK 1.6, Isaac Variant Caller, or the Illumina-developed Somatic Variant Caller. Both Ensembl and Refseq are available for annotation of the output files.

This App supports TruSeq Custom Amplicon analysis via import of custom manifests.

FOR RESEARCH USE ONLY.

Current Limitations:

- This app supports Illumina TruSeq Custom Amplicon panels, and Illumina predesigned amplicon panels: the TruSeq Amplicon Cancer Panel, and the TruSight Myeloid Sequencing Panel.
- This app has been validated with Illumina MiSeq and NextSeq data only.

Technical Limitations:

- This app is designed for TruSeq Amplicon samples.
- Variants are only found in regions covered in the manifest.
- Reads must be at least 50 bases in length.
- No minimum number of reads, but use a reasonable input size to get your required coverage.
- App only supports running one manifest per analysis.
- Samples need to all be paired-end or all single-end; a warning is thrown if single-end.
- Samples all need to have the same read lengths

マニフェストファイルのダウンロード

MyIllumina / Custom Products

The screenshot shows the MyIllumina Custom Products page. On the left is a navigation menu with items: Catalog, Quick Order, Home, Account, Orders, Saved Carts, Custom Products (highlighted with a blue arrow), and Support Bulletins. The main content area is divided into three sections: DesignStudio, Assay Design Tool (ADT), and BlueGnome. DesignStudio includes buttons for Nextera Rapid Capture, TruSeq Targeted RNA, and TruSeq Custom Amplicon. Assay Design Tool (ADT) includes a button for Infinium iSelect Genotyping and a link for Estimate Project Cost (Internal Users Only). BlueGnome includes a button for BlueFish Probes and links for Configurator and BlueFish QC Reports. On the right, the 'Product Files' section is highlighted with a red border and contains a list of manifest files and a 'GET PRODUCT FILES' button. Below it is the 'Additional Resources' section with links to templates, ADT status codes, and custom design.

Product Files

- Manifest files for MiSeq Reporter analysis of TruSeq Amplicon results
- iSelect, OPA and VeraCode Manifest and LMS Definition files

[GET PRODUCT FILES](#)

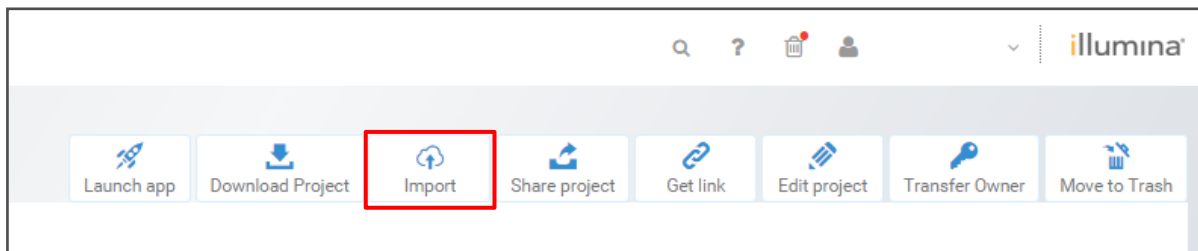
Additional Resources

- [Templates for iSelect](#)
- [ADT Status Codes](#)
- [Custom Infinium Genotyping Design](#)

シーケンスの際に使用するマニフェストファイルはイルミナホームページからダウンロードし、BaseSpaceプロジェクト内にインポートする必要がある

<https://my.illumina.com/Custom/Index>

BaseSpaceへのインポート



A screenshot of the BaseSpace 'PROJECTS' page. The navigation bar at the top includes 'DASHBOARD', 'PREP', 'RUNS', 'PROJECTS' (which is highlighted), 'APPS', and 'PUBLIC DATA'. Below the navigation bar, the text 'Projects: test_webinar : Import' is displayed. A red-bordered box contains the following text:

Due to browser limitations, imports have the following constraints:

- Maximum of **16 files** at a time
- Maximum of **25 Gb** at a time
- **One** sample at a time
- FASTQ Files must adhere to the [Illumina standard](#)

Below this box, the text 'Drag and drop or [select files](#)' is visible. At the bottom, there are three panels for file upload:

- Samples**: File type: .fastq.gz (with an 'FQ' icon)
- Analyses**: File type: .vcf, .vcf.gz (with a 'VCF' icon)
- Manifests**: File type: .txt (with a document icon)

TruSeq Amplicon appへのデータインプット



TruSeq Amplicon v2.0.0

illumina, Inc.

Analysis Name:

Save Results To:

Sample(s):

Targeted Amplicons:

Custom Manifest File:

Variant Caller: GATK Starling Somatic

Somatic variant caller threshold (percentage):

Read Stitching: On Off

Annotation: RefSeq Ensembl

This app is free.


プロジェクト内にインポートしたマニフェストファイルを指定する

Analysis Infoタブ

Analysis Info

Inputs

Output Files


 Analysis Reports

- Summary
- A-StN-50ng-rep1
- A-StC-50ng-rep1
- A-ReN-50ng-rep1
- A-ReC-50ng-rep1
- A-LuN-50ng-rep1
- A-LuC-50ng-rep1

Analysis Info

NAME	TruSeq Amplicon (single strand analysis for pool A)
APPLICATION	TruSeq Amplicon Version: 2.0.0
DATE STARTED	Monday, January 11th 2016, 6:01:44 pm
DATE COMPLETED	Monday, January 11th 2016, 6:30:11 pm
DURATION	28 minutes 27 seconds
SESSION TYPE	Multi-Node
STATUS	Complete (1 Node Complete)

Logs (last checked 8:52:01am UTC)

 [Log Files](#)

Please view the Multi-Node details page to see logs for this analysis.

解析ログはこのページから取得可能。テクニカルサポートからお願いする場合がございます

Inputsタブ

Analysis Info

Inputs

Output Files

Analysis Reports

- Summary
- A-StN-50ng-rep1
- A-StC-50ng-rep1
- A-ReN-50ng-rep1
- A-ReC-50ng-rep1
- A-LuN-50ng-rep1
- A-LuC-50ng-rep1

Inputs

Analysis Name
TruSeq Amplicon

Save Results To

NAME
MiniSeq: TruSeq Custom Amplicon Low Input Dual Strand

Sample(s)

NAME
A-ReN-50ng-rep1
A-StC-50ng-rep1
A-LuN-50ng-rep1
A-StN-50ng-rep1
A-LuC-50ng-rep1
A-ReC-50ng-rep1

Targeted Amplicons
Custom Panel (select manifest file below) ▼

Custom Manifest File

NAME
TruSeq_CAT_Manifest_A.txt

Variant Caller

GATK Starling Somatic

Somatic variant caller threshold (percentage)

10

Read Stitching
















On Off

Annotation

RefSeq Ensembl

解析の条件を確認することができます

Output Filesタブ

Analysis Info	Output Files				
Inputs	NAME	ITEM(S) ANALYZED	DATE CREATED	TYPE	SIZE
Output Files	 A-StN-50ng-rep1	 A-StN-50ng-rep1	Jan 11 2016	App Result	57.05 MB
	A-StN-50ng-rep1_S4.bam		bam	56.21 MB	
	A-StN-50ng-rep1_S4.bam.bai		bai	439.16 KB	
	A-StN-50ng-rep1_S4.coverage.csv		csv	12.73 KB	
	A-StN-50ng-rep1_S4.genome.vcf.gz		gz	191.10 KB	
	A-StN-50ng-rep1_S4.genome.vcf.gz.tbi		tbi	1.65 KB	
	A-StN-50ng-rep1_S4.report.html		html	48.59 KB	
	A-StN-50ng-rep1_S4.report.pdf		pdf	100.64 KB	
	A-StN-50ng-rep1_S4.summary.csv		csv	2.12 KB	
	A-StN-50ng-rep1_S4.varianttable.txt		txt	2.95 KB	
	A-StN-50ng-rep1_S4.vcf.gz		gz	16.93 KB	
	A-StN-50ng-rep1_S4.vcf.gz.ant		ant	42.87 KB	
	A-StN-50ng-rep1_S4.vcf.gz.ant.idx		idx	18 B	
	A-StN-50ng-rep1_S4.vcf.gz.tbi		tbi	1.51 KB	

下流の解析に使用するファイルの一覧が取得できます。.vcfファイルは後に使用します



Output Filesタブ

Analysis Info

Inputs

Output Files

Output Files

NAME	ITEM(S) ANALYZED	DATE CREATED	TYPE	SIZE
 A-StN-50ng-rep1	 A-StN-50ng-rep1	Jan 11 2016	App Result	57.05 MB

Details

Sample name	A-StN-50ng-rep1
Date created	Jan 9, 2016
Genome	--
Paired end	Paired
Number of Clusters	695,727
Number of Reads	1,391,454
Read 1 length	151
Read 2 length	151
Size	108.38 MB

Origin

Originating runs:

- [MiniSeq: TruSeq Amplicon somatic panel](#)

Files

FASTQ Files

[TruSeq Amplicon \(single strand analysis for pool A\)](#)

[Amplicon DS](#)

Open App Session

Download Selected

<input type="checkbox"/>	NAME ▼	SIZE	TYPE	STATUS	DATE
<input type="checkbox"/>	A-StN-50ng-rep1_S1_L001_R2_001.fastq.gz	56.87 MB	.gz	UploadComplete	Jan 9, 2016
<input type="checkbox"/>	A-StN-50ng-rep1_S1_L001_R1_001.fastq.gz	51.5 MB	.gz	UploadComplete	Jan 9, 2016

サンプルの詳細（fastqファイルなど）情報を取得することができます

Analysis Reports - Amplicon Summary

Amplicon Summary ⁱ

Number of Amplicon Regions	Total Length of Amplicon Regions
125	18,423 bp

Read Level Statistics ⁱ

Read	Total Aligned Reads	Percent Aligned Reads
1	616,943	88.68%
2	606,566	87.18%

Base Level Statistics ⁱ

Read	Percent Q30	Total Aligned Bases	Percent Aligned Bases	Mismatch Rate
1	93.61%	92,277,982	92.14%	0.33%
2	89.10%	90,656,239	90.15%	0.34%

解析の結果はAnalysis Reportsタブから確認することができます
シーケンスとリファレンスゲノムへのマッピング結果
- リード数、マッピング率、クオリティスコアなど

Analysis Reports - Small Variant Summary

Small Variants Summary i

	SNVs	Insertions	Deletions
Total Passing	24	0	1
Percent Found in dbSNP	62.50%	-	100.00%
Het/Hom Ratio	3	-	-
Ts/Tv Ratio	3	-	-

Variants by Sequence Context i

	SNVs	Insertions	Deletions
Number in Genes	24	0	1
Number in Exons	13	0	0
Number in Coding Regions	11	0	0
Number in UTR Regions	2	0	0
Number in Splice Site Regions	1	0	0

Genes include exons, introns and UTR regions. Exons include coding and UTR regions. UTR regions include 5' and 3' UTR regions. Splice site regions include regions annotated as splice acceptor, splice donor, splice site or splice region.

SNV, insertion, deletionの結果

- 各変異の数
- ヘテロ／ホモ率

- Transition/Transversion率
- 変異の位置情報

Analysis Reports - Small Variant Summary

Variants by Consequence 

	SNVs	Insertions	Deletions
Frameshifts	-	0	0
Non-synonymous	6	0	0
Synonymous	5	-	-
Stop Gained	0	0	0
Stop Lost	0	0	0

Variation consequences are calculated following the guidelines at http://uswest.ensembl.org/info/genome/variation/predicted_data.html#consequences

SNV, insertion, deletionの結果

- 変異がもたらす結果のタイプ

Analysis Reports - Variants Table

Variants Table ⁱ

Export (CSV)

Chr	Pos	Ref	Alt	Type	Context	Consequence	dbSNP	COSMIC	ClinVar	Qual	Alt Freq
chr1	1152...	G	A	SNV	Intergeni...	downstream_g...				100	0.393
chr1	1152...	G	A	SNV	Intergeni...	downstream_g...				100	0.395
chr2	2944...	G	A	SNV	Intron	splice_region_...	rs46226...			100	0.511
chr4	1808...	A	T	SNV	Intron,Int...	intron_variant,...				100	0.141
chr4	5514...	A	G	SNV	Coding	synonymous_v...	rs18737...	COSM143...		100	0.998
chr4	5559...	T	C	SNV	Intron	intron_variant	rs10086...			100	1.000
chr7	5525...	C	T	SNV	Coding,In...	synonymous_v...	rs22290...	COSM858...		100	0.494
chr9	2197...	C	T	SNV	Coding,In...	missense_varia...	rs37312...			100	0.320
chr10	8965...	C	T	SNV	Coding	synonymous_v...	rs15065...	COSM5168		100	0.505
chr10	8972...	C	T	SNV	Coding	synonymous_v...				100	0.573
chr10	8972...	A	G	SNV	Coding	synonymous_v...				100	0.576
chr10	8972...	T	C	SNV	3P_UTR	3_prime_UTR_...				100	0.450
chr11	534197	C	T	SNV	Intron,Int...	intron_variant,...	rs41258...			100	0.553
chr17	7577...	A	C	SNV	Intron	intron_variant	rs12951...			100	0.472
chr17	7577...	G	A	SNV	Intron	intron_variant	rs12947...			100	0.480
chr17	7578...	C	T	SNV	Intron	intron_variant	rs29094...			100	1.000
chr17	7578...	G	C	SNV	Coding	missense_varia...	rs10425...	COSM459	non-path	100	0.993

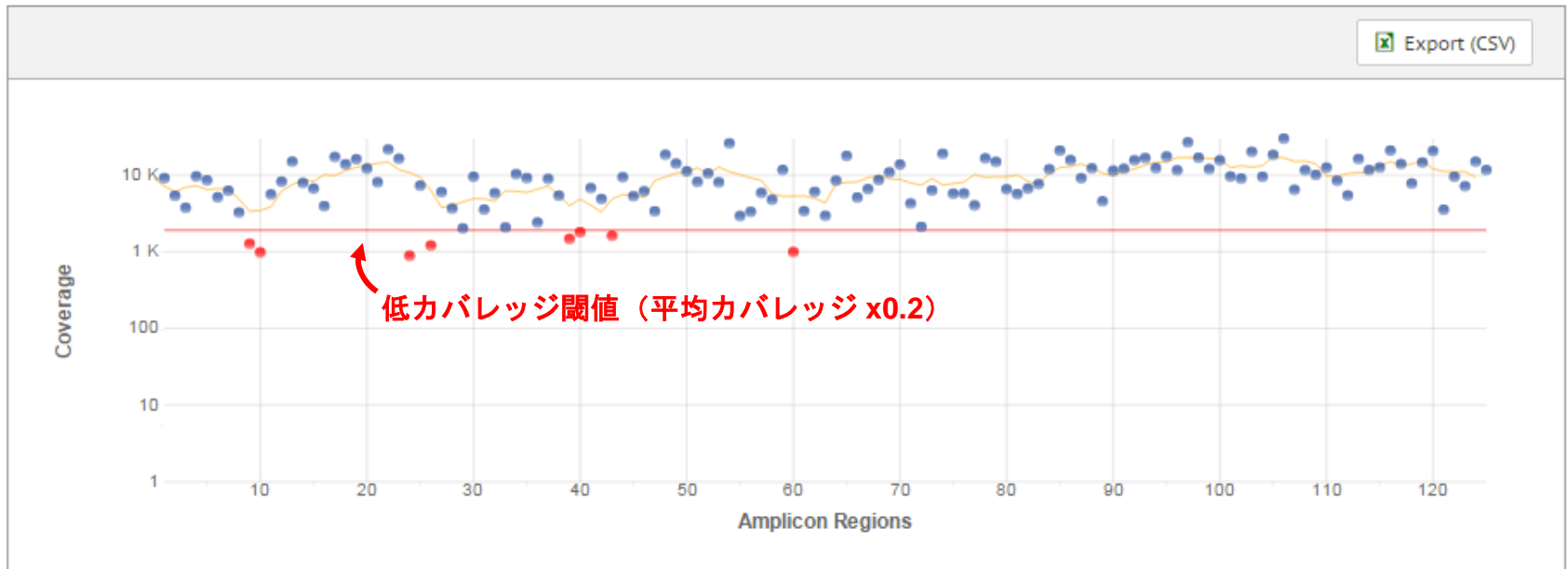
変異の詳細を確認でき、結果をダウンロードすることができます

Analysis Reports - Coverage Summary

Coverage Summary *i*

Amplicon Mean Coverage	Uniformity of Coverage
9568.7	93.60% 80%がスペック

Coverage by Amplicon Region *i*



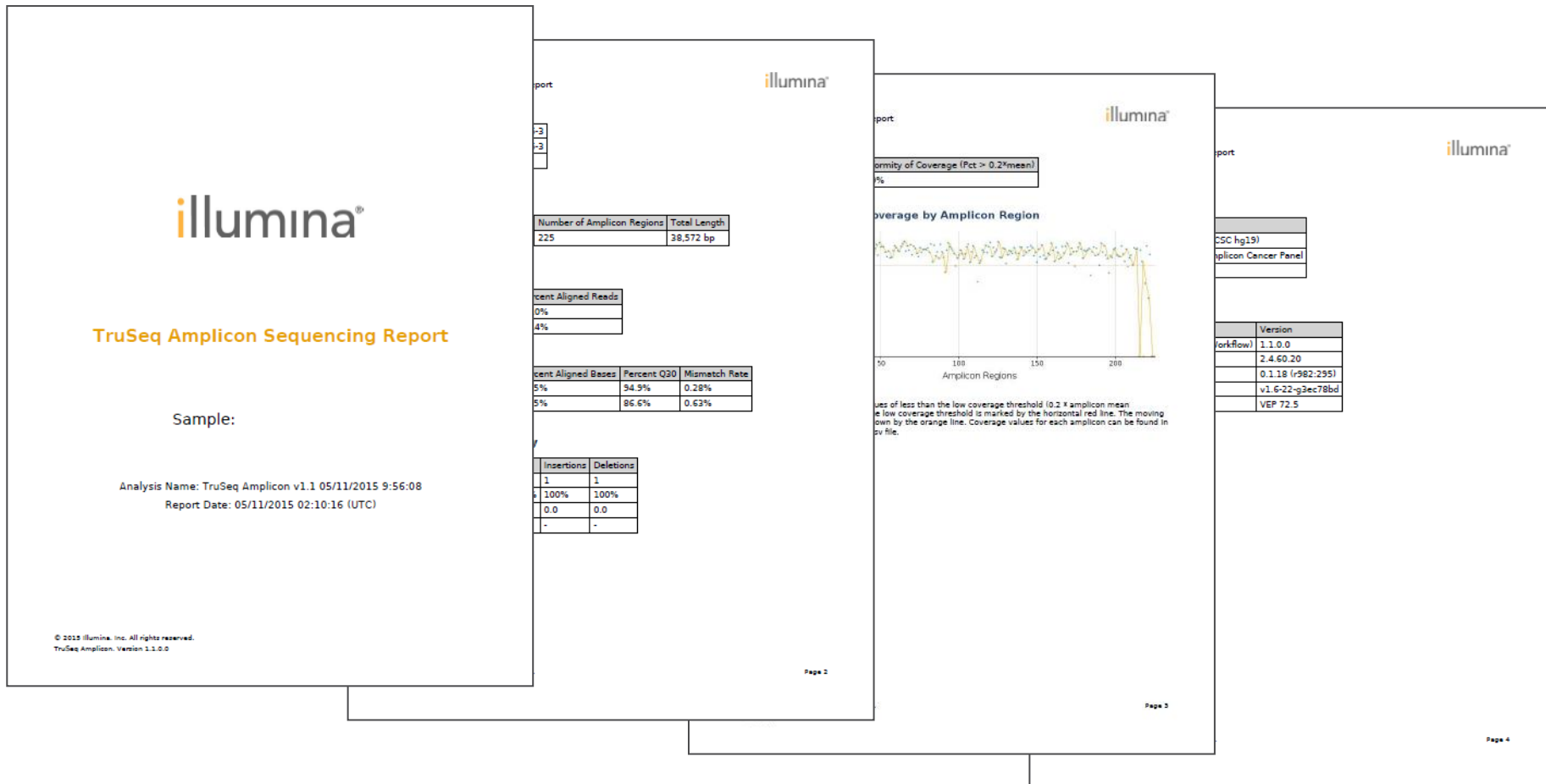
カバレッジ情報

- 平均カバレッジ、低カバレッジ変異
- マウスオンでどの変異か、個別のカバレッジが確認可能

Analysis Reports - PDF Summary Report

Results for Sample

 PDF Summary Report



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TruSeq Amplicon Sequencing Report

Sample:

Analysis Name: TruSeq Amplicon v1.1 05/11/2015 9:56:08
Report Date: 05/11/2015 02:10:16 (UTC)

© 2015 Illumina, Inc. All rights reserved.
TruSeq Amplicon, Version 1.1.0.0

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Number of Amplicon Regions	Total Length
225	38,572 bp

Percent Aligned Reads
0%
4%

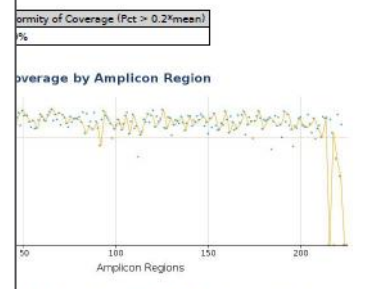
Percent Aligned Bases	Percent Q30	Mismatch Rate
5%	34.5%	0.28%
5%	86.6%	0.63%

Insertions	Deletions
1	1
100%	100%
0.0	0.0
-	-

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Uniformity of Coverage (Pct > 0.2 * mean)

Coverage by Amplicon Region



Percent of amplicon regions with coverage of less than the low coverage threshold (0.2 * amplicon mean). The low coverage threshold is marked by the horizontal red line. The moving average is shown by the orange line. Coverage values for each amplicon can be found in the .cov file.

Version
1.1.0.0
2.4.60.20
0.1.18 (r982:295)
v1.6-22-g3ec78bd
VEP 72.5

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Page 3

Page 4

シーケンス結果はリンクからPDFで取得することができます

本日の内容

▶ BaseSpace cloud app, TruSeq Ampliconを用いたデータ解析

ーシーケンスデータの参照

ー変異の解析結果の参照

▶ VariantStudioソフトウェアを用いたデータ解析

ーデータのインポート

ー変異の絞り込み

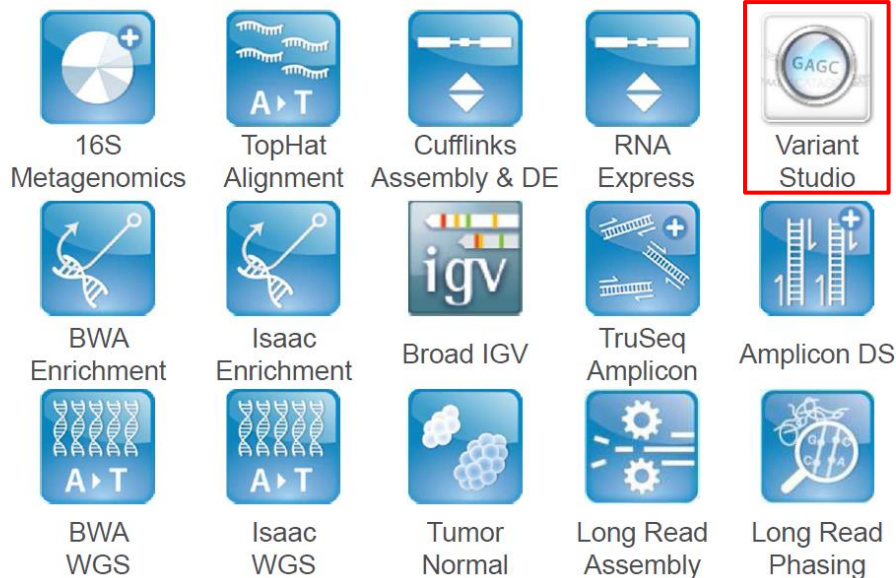
ーアノテーション

ーデータベースを利用した知見の入手

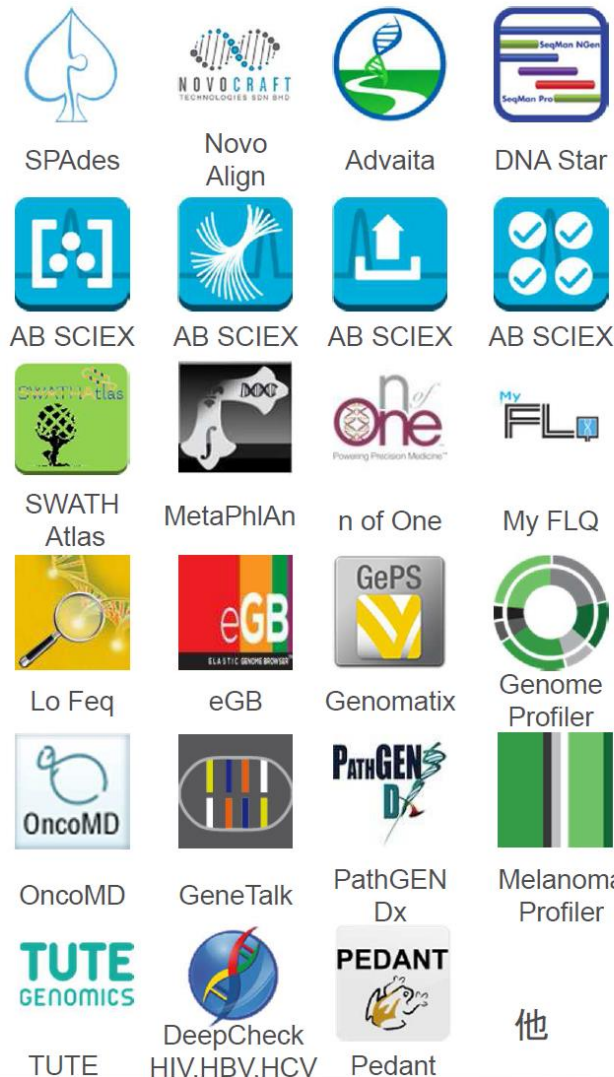
注：解析にはデモデータを使用しており、細かくは実際と異なる場合がございます。
データの参照と解析の流れをご案内する一例としてご参考下さい。

BaseSpace app : VariantStudioを用いたデータ解析

<イルミナコアアプリ>



<他社製アプリ>



<イルミナラボアプリ>



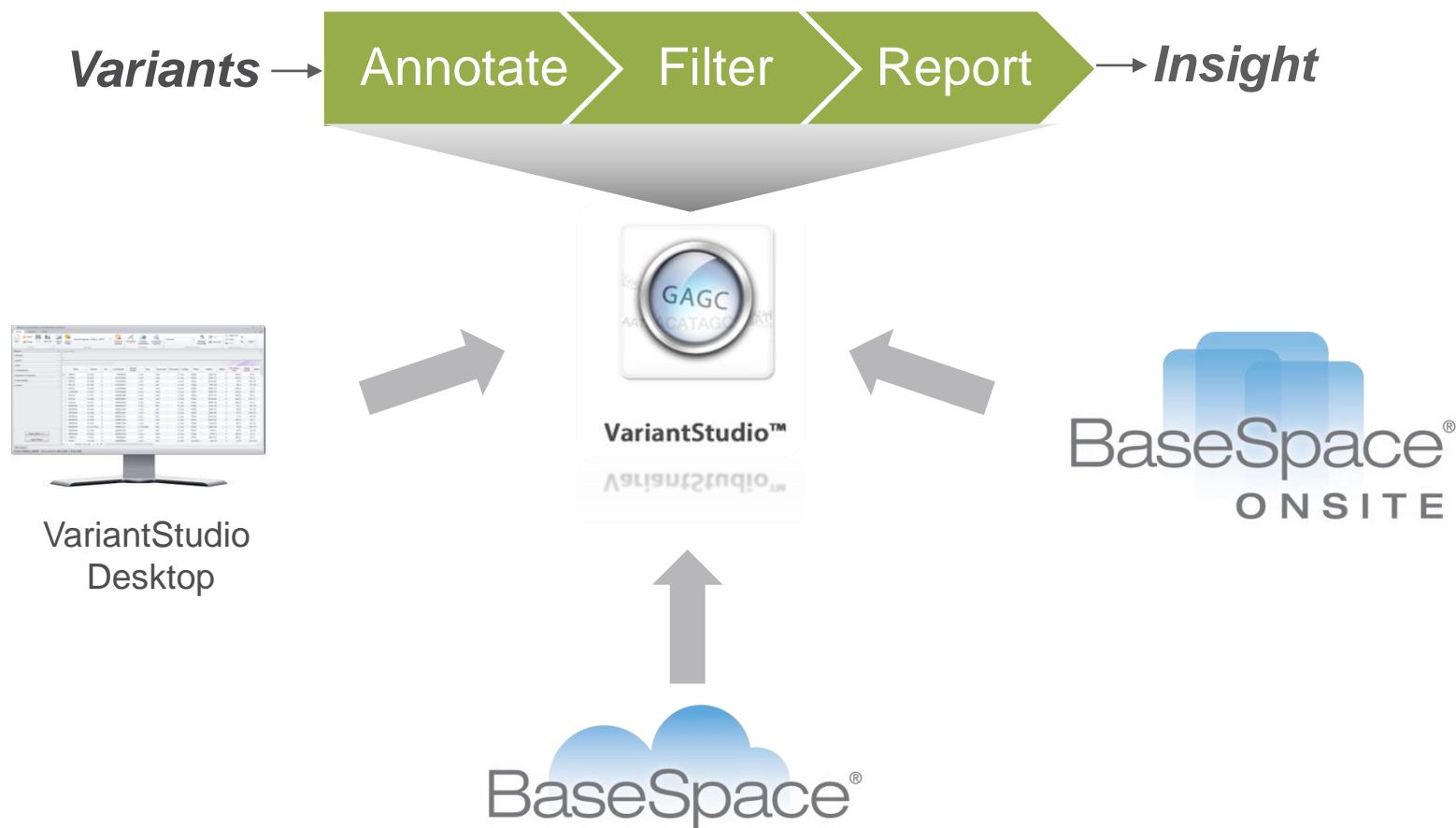
他

他

他

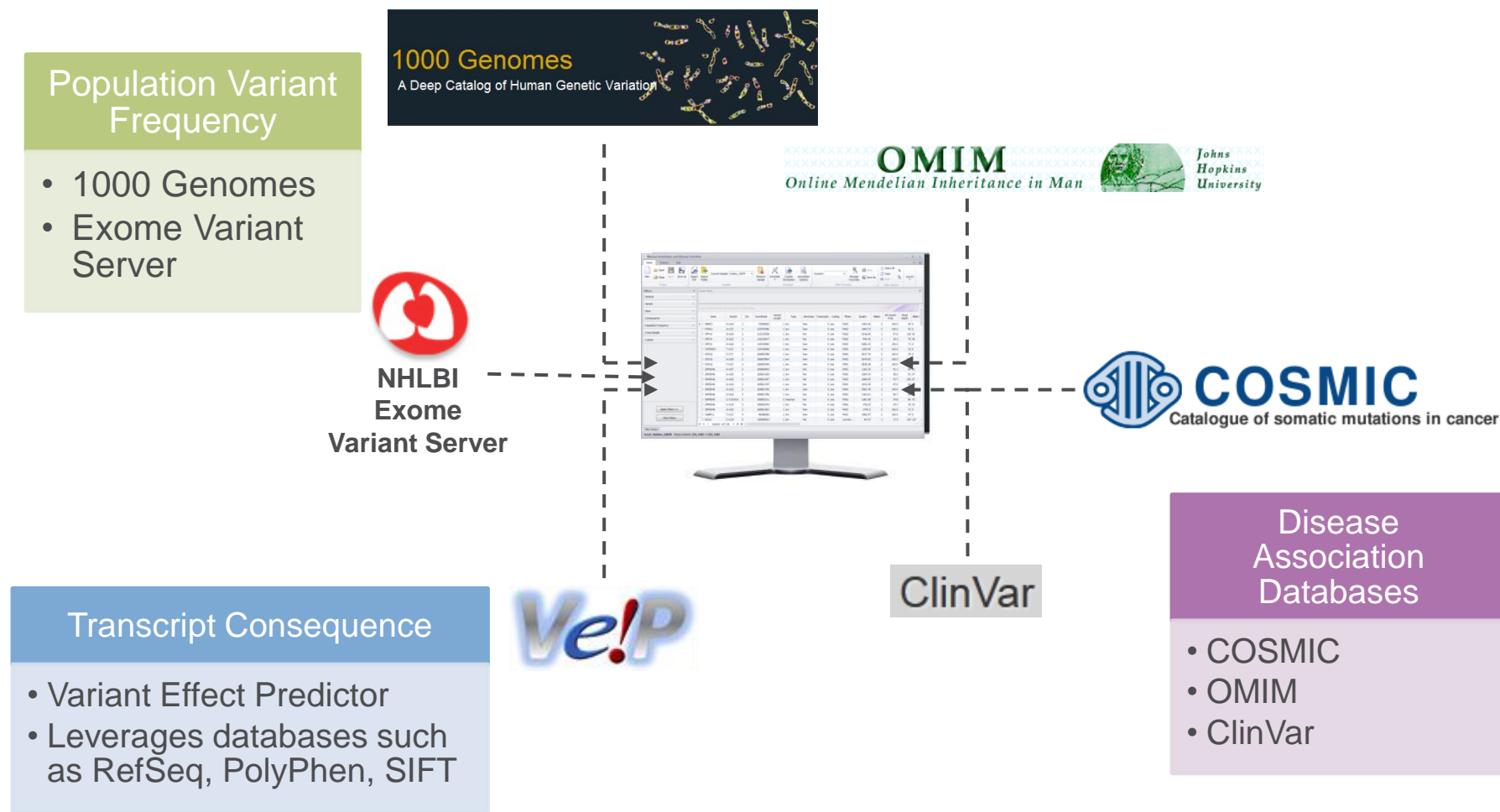
VariantStudioとは

- ▶ 変異解析のためのイルミナソフトウェア。
- ▶ リシーケンスappsで生成されるvcfファイルを読み込ませることで、コールした変異をアノテーション・フィルターし、解析レポートの形にするまでアシストします。



VariantStudioとは

- ▶ インターネット上の各種データベースの情報を簡便に統合し、知見を得ることができます



VariantStudioへのvcfファイルのインポート

ここから.vcfファイルを選択できます

Gene	Variant	Chr	Coordinate	Classification	Type	Genotype	Exonic	Filters	Quality	GQX	Inherited From	Alt Variant Freq	Read Depth	Alt Read Depth	Allelic Depths	C

Sample: STN-4-10-2015-3_S1 Genes, Variants: (24, 41) -> (24, 41)

VariantStudioへのvcfファイルのインポート

The screenshot shows the Illumina VariantStudio interface. The top menu bar includes Home, Annotation & Classification, Reports, and Help. Below the menu is a toolbar with icons for New, Open, Close, Save, Save As, Import VCF, Add Variants to Sample, Import Folder, Remove Sample, Manage Favorites, Save, Select All, Copy, Column Order, Smaller, Larger, Save As Default, Apply Default, and Restore Default. The main window is divided into several sections:

- Filters:** A sidebar on the left with expandable sections for General, Variant, Gene, Consequence, Population Frequency, Cross Sample Subtraction, Family Based, Custom, and Classification. At the bottom of this sidebar are buttons for "Apply Filters =>" and "Clear Filters".
- Gene View:** A horizontal track at the top of the main area showing a genomic region on Chromosome 2 (Chr 2) with the ERBB4 gene. The coordinates range from 213,240,442 to 213,403,352. This section is highlighted with a red border.
- Variant Table:** A table listing detected variants. The columns are Gene, Variant, Chr, Coordinate, Classification, Type, Genotype, Exonic, Filters, Quality, GQX, Inherited From, Alt Variant Freq, Read Depth, Alt Read Depth, and Allelic Depths. The first row shows a deletion in the ERBB4 gene.
- Variant Navigation:** At the bottom of the table, it shows "Variant 1 of 41" and navigation arrows.
- Display Options:** Checkboxes for "Show Population Frequencies", "Show Transcript Info", "Show Custom Annotations", "Show ClinVar", and "Show Cosmic".
- Filter History:** A section at the bottom left showing the current filter history.
- Sample Information:** At the bottom of the window, it displays "Sample: STN-4-10-2015-3_S1 Genes, Variants: (24, 41) -> (24, 41)".

A red text overlay in the center of the variant table reads: **.vcfファイルがインポートされました** (VCF file imported).

Gene	Variant	Chr	Coordinate	Classification	Type	Genotype	Exonic	Filters	Quality	GQX	Inherited From	Alt Variant Freq	Read Depth	Alt Read Depth	Allelic Depths
ERBB4	TA>TA/T	2	212578379	...	deletion	het	yes	PASS	5970.44	99		26.6	1811	354	979,354
VHL	C>C/A	3	10191577	...	snv	het	yes	LowQD	1006.82	99		12.8	4999	623	4259,...
PIK3CA	C>C/T	3	178916931	...	snv	het	yes	LowQD	3332.57	99		13.4	2221	297	1924,...
PIK3CA	C>C/A	3	178921639	...	snv	het	no	PASS	37479.86	99		42.2	4426	1862	2555,...
FGFR3	G>A/A	4	1807894	...	snv	hom	yes	PASS	152252.8	99		99.6	4999	4972	19,4972
FGFR3	C>C/T	4	1808422	...	snv	het	yes	PASS	3448.42	99		18.8	1416	266	1149,...
PDGFRA	A>G/G	4	55141055	...	snv	hom	yes	PASS	156252.3	99		100	5000	4994	1,4994
KIT	A>A/C	4	55593712	...	snv	het	yes	LowQD	5851.06	99		15.4	4532	688	3776,...
KIT	T>C/C	4	55599436	...	snv	hom	no	PASS	182513.1	99		99.6	5000	4862	19,4862
KDR	A>G/G	4	55946...	...	snv	hom	yes	PASS	5715...	99		99.8	4999	4985	8,4985
APC	G>A/A	5	7171...	...	snv	het	yes	PASS	5715...	99		99.9	5000	4995	4,4995
SMO	A>A/G	7	128846469	...	snv	het	no	PASS	50895.7	99		35.4	5000	1764	3214,...
CDKN2A	C>C/A	9	21971281	...	snv	het	no	LowQD	359.25	99		9.9	4028	397	3596,...
GNAQ	G>G/GA	9	80343587	...	insertion	het	yes	PASS	38973.66	99		60.2	5000	1947	1287,...
GNAQ	A>G/G	9	80409345	...	snv	hom	no	PASS	156405	99		99.8	5000	4986	8,4986
GNAQ	C>C/T	9	80409528	...	snv	het	yes	LowQD	559.8	99		7.6	3519	269	3250,...
ABL1	C>C/T	9	133738328	...	snv	het	yes	PASS	2743.03	99		14.2	1114	158	956,158
ABL1	C>C/T	9	133738438	...	snv	het	yes	LowQD	195.7	99		7.5	2086	156	1926,...

VariantStudioへのvcfファイルのインポート

The screenshot shows the Illumina VariantStudio interface. The 'Table Options' menu is open, and the 'Column Order' option is highlighted with a red box and the text 'ここをクリック' (Click here). The main window displays a table of variant data for sample STN-4-10-2015-3_51.

Gene	Variant	Chr	Coordinate	Classification	Type	Genotype	Exonic	Filters	Quality	GQX	Inherited From	Alt Variant Freq	Read Depth	Alt Read Depth	Allelic Depths
ERBB4	TA>TA/T	2	212578379	...	deletion	het	yes	PASS	5970.44	99		26.6	1811	354	979,354
VHL	C>C/A	3	10191577	...	snv	het	yes	LowQD	1006.82	99		12.8	4999	623	4259,...
PIK3CA	C>C/T	3	178916931	...	snv	het	yes	LowQD	3332.57	99		13.4	2221	297	1924,...
PIK3CA	C>C/A	3	178921639	...	snv	het	no	PASS	37479.86	99		42.2	4426	1862	2555,...
FGFR3	G>A/A	4	1807894	...	snv	hom	yes	PASS	152252.8	99		99.6	4999	4972	19,4972
FGFR3	C>C/T	4	1808422	...	snv	het	yes	PASS	3448.42	99		18.8	1416	266	1149,...
PDGFRA	A>G/G	4	55141055	...	snv	hom	yes	PASS	156252.3	99		100	5000	4994	1,4994
KIT	A>A/C	4	55593712	...	snv	het	yes	LowQD	5851.06	99		15.4	4532	688	3776,...
KIT	T>C/C	4	55599436	...	snv	hom	no	PASS	182513.1	99		99.6	5000	4862	19,4862
KDR	A>G/G	4	55946081	...	snv	hom	yes	PASS	157163.2	99		99.8	4999	4985	8,4985
APC	G>A/A	5	112175770	...	snv	hom	yes	PASS	155164.6	99		99.9	5000	4995	4,4995
SMO	A>A/G	7	128846469	...	snv	het	no	PASS	50895.7	99		35.4	5000	1764	3214,...
CDKN2A	C>C/A	9	21971281	...	snv	het	no	LowQD	359.25	99		9.9	4028	397	3596,...
GNAQ	G>G/GA	9	80343587	...	insertion	het	yes	PASS	38973.66	99		60.2	5000	1947	1287,...
GNAQ	A>G/G	9	80409345	...	snv	hom	no	PASS	156405	99		99.8	5000	4986	8,4986
GNAQ	C>C/T	9	80409528	...	snv	het	yes	LowQD	559.8	99		7.6	3519	269	3250,...
ABL1	C>C/T	9	133738328	...	snv	het	yes	PASS	2743.03	99		14.2	1114	158	956,158
ABL1	C>C/T	9	133738438	...	snv	het	yes	LowQD	195.7	99		7.5	2086	156	1926,...

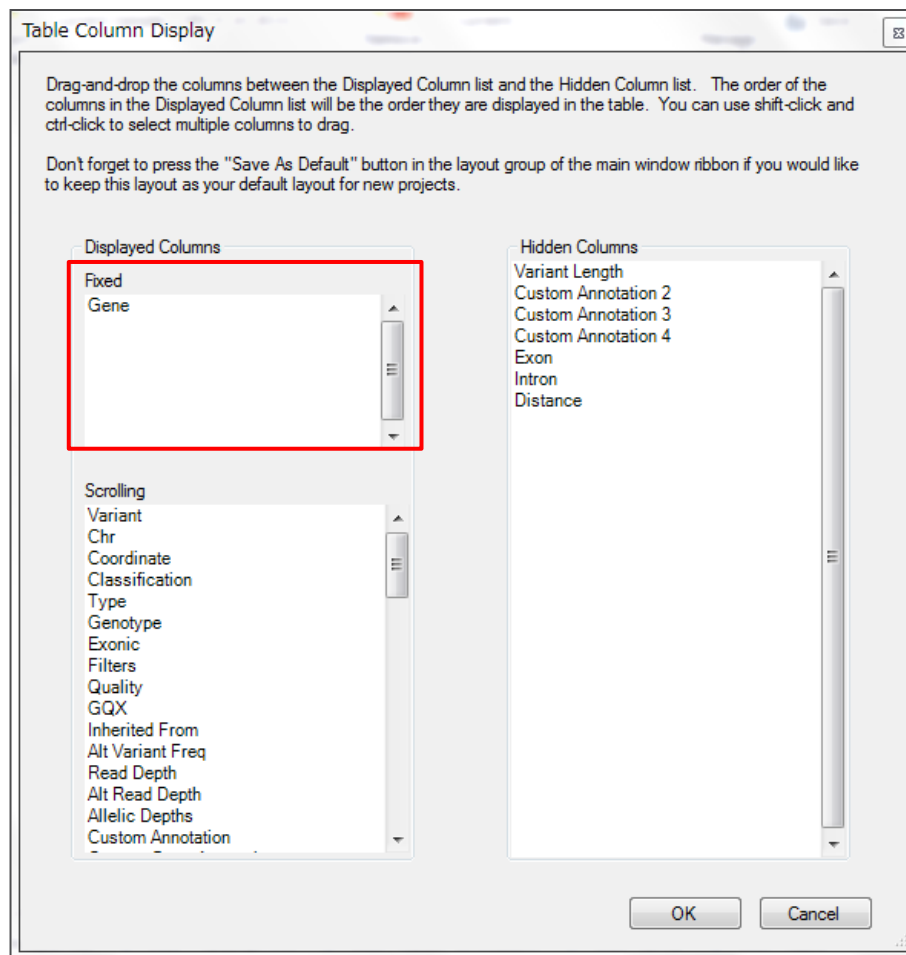
Variant 1 of 41

Show Population Frequencies
 Show Transcript Info
 Show Custom Annotations
 Show ClinVar
 Show Cosmic

Filter History

Sample: STN-4-10-2015-3_51 Genes, Variants: (24, 41) -> (24, 41)

VariantStudioへのvcfファイルのインポート



ドラッグで固定するカラムを選択することができます
‘Gene’の固定をお勧めします

Annotate: 変異のアノテーション

Annotation Options
View and modify the current annotation options

Gene	Num Transcripts	Transcript	Consequence	cDNA Position	CDS Position	Protein Position	Amino Acids	Codons	HGNC	Transcript HGNC	Canoni...	Sift	Polyf
ERBB4			n	n	n	n	n	n	n	n	n	n	n
VHL			n	n	n	n	n	n	n	n	n	n	n
PIK3CA	0												
PIK3CA	0												
FGFR3	0												
FGFR3	0												
PDGFRA	0												
KIT	0												
KIT	0												
KDR	0												
APC	0												
SMO	0												
CDKN2A	0												
GNAQ	0												
GNAQ	0												
GNAQ	0												
ABL1	0												
ABL1	0												

Geneのラムが固定されました

Variant 1 of 4

Show Population Frequencies Show Transcript Info Show Custom Annotations Show ClinVar Show Cosmic

Variants Genes No-Call Regions

Filter History

Sample: S11-4-10-2015-3_S1 Genes, Variants: (24, 41) -> (24, 41)

Annotate: 変異のアノテーション

The screenshot shows the Illumina VariantStudio software interface. The 'Annotate' menu item is highlighted with a red box and the text 'ここをクリック' (Click here). The main window displays a table of variant annotations with columns for Gene, Num Transcripts, Transcript, Consequence, cDNA Position, CDS Position, Protein Position, Amino Acids, Codons, HGNC, Transcript HGNC, Canoni..., Sift, and Polyf. The table lists 18 genes, all with 0 transcripts. The status bar at the bottom indicates 'Sample: S11-4-10-2015-3_S1 Genes, Variants: (24, 41) -> (24, 41)'.

Annotation Options
View and modify the current annotation options

Gene	Num Transcripts	Transcript	Consequence	cDNA Position	CDS Position	Protein Position	Amino Acids	Codons	HGNC	Transcript HGNC	Canoni...	Sift	Polyf
ERBB4	0												
VHL	0												
PIK3CA	0												
PIK3CA	0												
FGFR3	0												
FGFR3	0												
PDGFRA	0												
KIT	0												
KIT	0												
KDR	0												
APC	0												
SMO	0												
CDKN2A	0												
GNAQ	0												
GNAQ	0												
GNAQ	0												
ABL1	0												
ABL1	0												

Variant 1 of 41

Show Population Frequencies Show Transcript Info Show Custom Annotations Show ClinVar Show Cosmic

Filter History

Sample: S11-4-10-2015-3_S1 Genes, Variants: (24, 41) -> (24, 41)

Annotate: 変異のアノテーション

The screenshot displays the Illumina VariantStudio interface. The top menu includes Home, Annotation & Classification, Reports, and Help. The main toolbar contains icons for Annotate, Custom Annotation, Custom Gene Annotation, Annotation Options, Set Default Transcripts, Apply Classifications from Database, View Classification Database, and Classification Settings.

On the left, there is a 'Filters' panel with expandable sections for General, Variant, Gene, Consequence, Population Frequency, Cross Sample Subtraction, Family Based, Custom, and Classification. Below the filters are 'Apply Filters =>' and 'Clear Filters' buttons.

The main area is titled 'Gene View' and contains a table of variant annotations. The table has the following columns: Gene, Num Transcripts, Transcript, Consequence, cDNA Position, CDS Position, Protein Position, Amino Acids, Codons, HGNC, Transcript HGNC, Canoni..., Sift, and Polyf. The table lists various genes such as ERBB4, VHL, PIK3CA, FGFR3, PDGFRA, KIT, KDR, APC, SMO, CDKN2A, GNAQ, and ABL1, along with their respective transcript IDs and variant consequences.

A red text overlay is positioned over the table, reading: 各変異についてアノテーション情報が取得されました (Annotation information was obtained for each variant).

At the bottom of the interface, there are navigation controls for 'Variant 1 of 41', checkboxes for 'Show Population Frequencies', 'Show Transcript Info', 'Show Custom Annotations', 'Show ClinVar', and 'Show Cosmic', and tabs for 'Variants', 'Genes', and 'No-Call Regions'. The status bar at the very bottom indicates 'Sample: S11-4-10-2015-3_S1 Genes, Variants: (24, 41) -> (24, 41)'.

Filter: 変異の絞り込み（目的のキーワードがある場合）

The screenshot shows the Illumina VariantStudio interface. The 'Filters' panel on the left is expanded to 'General'. The 'Gene View' table shows a list of variants. A red box highlights the 'Gene' column header, and a red text overlay says 'ここに遺伝子名をタイプします' (Type the gene name here). Below the table, a red text overlay says 'リストされる遺伝子が絞られました' (The listed genes are filtered). The filter expression at the bottom is 'Contains([Gene], 'erbb')'. The status bar at the bottom shows 'Sample: STN-4-10-2015-3_S1 Genes, Variants: (24, 41) -> (24, 41)'.

Gene	Variant	Chr	Coordinate	Classification	Type	Genotype	Exonic	Filters	Quality	GQX	Inherited From	Alt Variant Freq	Read Depth	Alt Read Depth	Allelic Depths	...
ERBB4	TA>TA/T	2	212578379	...	deletion	het	yes	PASS	5970.44	99		26.6	1811	354	979,354	...
ERBB2	C>C/T	17	37881010	...	snv	het	yes	LowQD	1495.15	99		10.2	2058	209	1841,...	...
ERBB2	G>G/T	17	37881030	...	snv	het	yes	LowQD	2144.04	99		12	2058	247	1808,...	...

ここに遺伝子名をタイプします

リストされる遺伝子が絞られました

Contains([Gene], 'erbb')

Sample: STN-4-10-2015-3_S1 Genes, Variants: (24, 41) -> (24, 41)

Filter: 変異の絞り込み（条件に合うものを探したい場合）

The screenshot shows the Illumina VariantStudio interface. The 'Filters' panel on the left is highlighted with a red box and contains the following settings:

- Variant
- Pass Filter
- Quality > 0
- Read Depth > 0
- Alt Variant Freq > 0
- Show only variants:
 - Inside genes
 - In conserved regions
- Only variants without dbSNP ID
- Only variants with Cosmic annotation
 - where matches mutant allele
 - where not matches mutant allele
- Only variants with ClinVar annotation
 - where matches mutant allele
 - where not matches mutant allele
- Buttons: Apply Filters =>, Clear Filters

The main 'Gene View' table displays the following data:

Gene	Variant	Chr	Coordinate	Classification	Type	Genotype	Exonic	Filters	Quality	GQX	Inherited From	Alt Variant Freq	Read Depth	Alt Read Depth	Allelic Depths
ERBB4	TA>TA/T	2	212578379	...	deletion	het	yes	PASS	5970.44	99		26.6	1811	354	979,354
VHL	C>C/A	3	10191577	...	snv	het	yes	LowQD	1006.82	99		12.8	4999	623	4259,...
PIK3CA	C>C/T	3	178916931	...	snv	het	yes	LowQD	3332.57	99		13.4	2221	297	1924,...
PIK3CA	C>C/A	3	178921639	...	snv	het	no	PASS	37479.86	99		42.2	4426	1862	2555,...
FGFR3	G>A/A	4	1807894	...	snv	hom	yes	PASS	152252.8	99		99.6	4999	4972	19,4972
FGFR3	C>C/T	4	1808422	...	snv	het	yes	PASS	3448.42	99		18.8	1416	266	1149,...
PDGFRA	A>G/G	4	55141055	...	snv	hom	yes	PASS	156252.3	99		100	5000	4994	1,4994
KIT	A>A/C	4	55593712	...	snv	het	yes	LowQD	5851.06	99		15.4	4532	688	3776,...
KIT	T>C/C	4	55599436	...	snv	hom	no	PASS	182513.1	99		99.6	5000	4862	19,4862
KDR	A>G/G	4	55946081	...	snv	hom	yes	PASS	157163.2	99		99.8	4999	4985	8,4985
APC	G>A/A	5	112175770	...	snv	hom	yes	PASS	155164.6	99		99.9	5000	4995	4,4995
SMO	A>A/G	7	128846469	...	snv	het	no	PASS	50895.7	99		35.4	5000	1764	3214,...
CDKN2A	C>C/A	9	21971281	...	snv	het	no	LowQD	359.25	99		9.9	4028	397	3596,...
GNAQ	G>G/GA	9	80343587	...	insertion	het	yes	PASS	38973.66	99		60.2	5000	1947	1287,...
GNAQ	A>G/G	9	80409345	...	snv	hom	no	PASS	156405	99		99.8	5000	4986	8,4986
GNAQ	C>C/T	9	80409528	...	snv	het	yes	LowQD	559.8	99		7.6	3519	269	3250,...
ABL1	C>C/T	9	133738328	...	snv	het	yes	PASS	2743.03	99		14.2	1114	158	956,158
ABL1	C>C/T	9	133738438	...	snv	het	yes	LowQD	195.7	99		7.5	2086	156	1926,...

At the bottom of the interface, there are checkboxes for 'Show Population Frequencies', 'Show Transcript Info', 'Show Custom Annotations', 'Show ClinVar', and 'Show Cosmic', all of which are checked. Below the table, there are tabs for 'Variants', 'Genes', and 'No-Call Regions', with 'Variants' selected.

ここに表示条件を入力していきます

Filter: 変異の絞り込み（条件に合うものを探したい場合）

The image shows a variant filtering interface with four main panels:

- General:** Includes sections for Genotype (Heterozygote, Homozygote, Hemizygote), Variant Type (SNVs, Insertions, Deletions, Reference), and Chromosome (All Chromosomes, Autosomal, Chromosome: dropdown). A "Use Advanced Filter" checkbox is at the bottom.
- Variant:** Includes a "Pass Filter" section with input fields for Quality, Read Depth, and Alt Variant Freq. Below is a "Show only variants:" section with checkboxes for "Inside genes" and "In conserved regions". Further down are checkboxes for "Only variants without dbSNP ID", "Only variants with Cosmic annotation" (with sub-options for "where matches mutant allele" and "where not matches mutant allele"), and "Only variants with ClinVar annotation" (with sub-options for "where matches mutant allele" and "where not matches mutant allele").
- Consequence:** Starts with a checked "Show only variants that are:" checkbox. Below is a list of consequence types: Missense (checked), Polyphen "damaging" (checked), SIFT "deleterious" (checked), Frameshift (checked), Stop gained (checked), Stop lost (checked), Initiator codon (checked), In-frame insertion (checked), In-frame deletion (checked), and Splice (checked). A "Select All" button is at the bottom.
- Population Frequency:** Includes checkboxes for Global Frequency, American Pop Frequency, Asian Pop Frequency, African Pop Frequency, European Pop Frequency, and EVS Frequency, each with a frequency input field set to 1. A "Set all to:" section has a dropdown set to 1 and a "Set All" button.

変異コールの情報、アノテーション情報から、任意の条件で変異を絞り込むことができます
条件設定が終わったら、'Apply Filter'をクリック

Filter: 変異の絞り込み（条件に合うものを探したい場合）

The screenshot shows the Illumina VariantStudio interface. The top menu bar includes Home, Annotation & Classification, Reports, and Help. Below the menu is a toolbar with various icons for file operations and sample management. The main window is divided into a left sidebar with filter categories and a central table view.

Filters:

- General
- Variant
- Gene
- Consequence
- Population Frequency
- Cross Sample Subtraction
- Family Based
- Custom
- Classification

Gene View Table:

Gene	Variant	Chr	Coordinate	Classification	Type	Genotype	Exonic	Filters	Quality	GQX	Inherited From	Alt Variant Freq	Read Depth	Alt Read Depth	Allelic Depths	Custom Annotation	Custom Gene Annotation	Custom Gene Annotation 2	Nu Trans
ZIC3	C>A/A	X	136649654	...	snv	hom	yes	PASS	375	69	mother	100	24	24	0,24				

Text overlay: リストされる遺伝子が絞られました (The gene listed is filtered)

Bottom status bar: Sample: NA12882_Synthetic Genes, Variants: (2703, 7846) -> (1, 1)

Filter: 変異の絞り込み（条件に合うものを探したい場合）

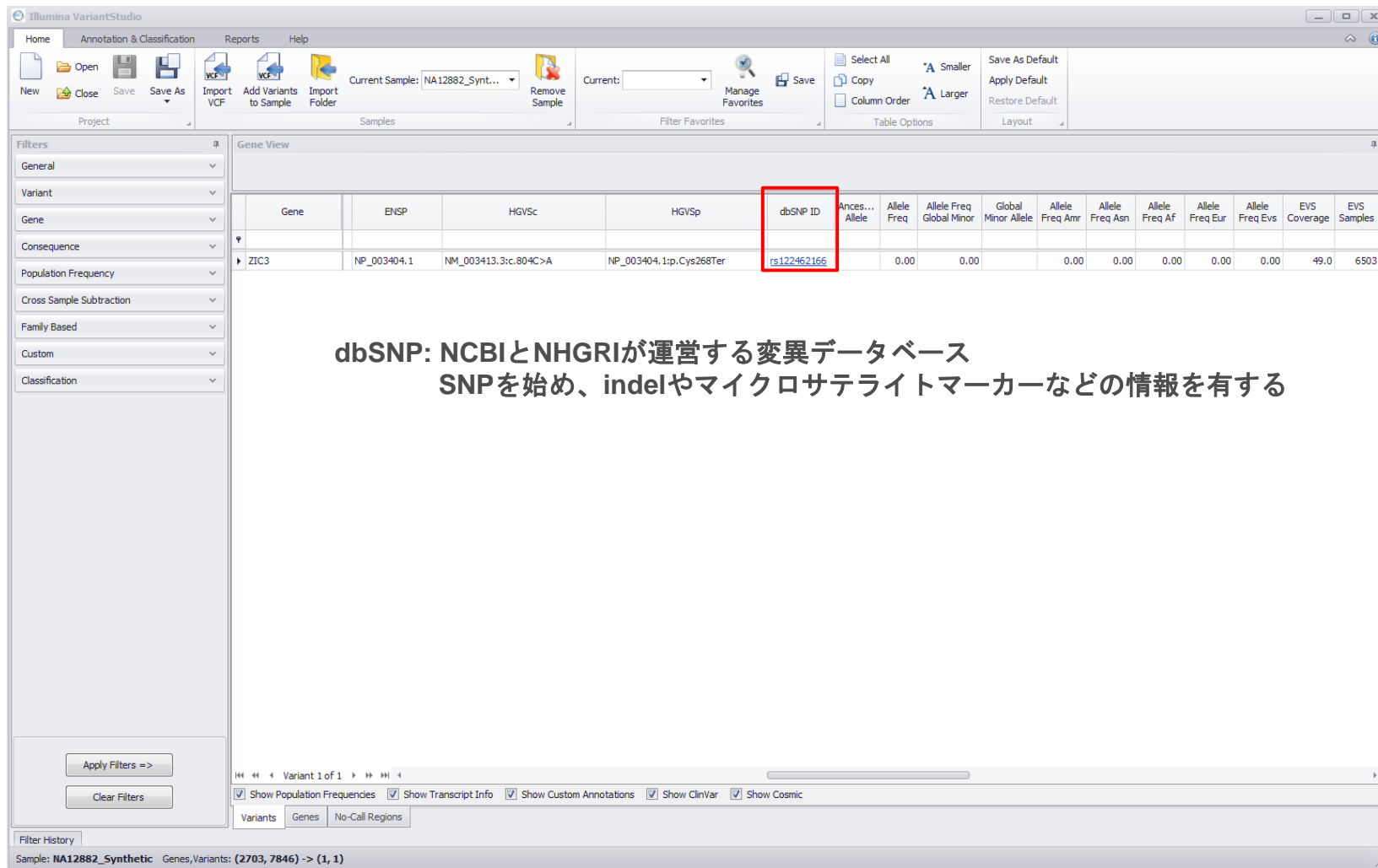
The screenshot shows the Illumina VariantStudio interface. The 'Save' button in the top toolbar is highlighted with a red box. A red text overlay reads: **ここをクリックすると、フィルター条件を保存できます** (Clicking here allows you to save filter conditions).

The main window displays a 'Gene View' table with the following data:

Gene	Variant	Chr	Coordinate	Classification	Type	Genotype	Exonic	Filters	Quality	GQX	Inherited From	Alt Variant Freq	Read Depth	Alt Read Depth	Allelic Depths	Custom Annotation	Custom Gene Annotation	Custom Gene Annotation 2	Nu Trans
ZIC3	C>A/A	X	136649654	...	snv	hom	yes	PASS	375	69	mother	100	24	24	0,24				

At the bottom of the interface, the status bar shows: **Sample: NA12882_Synthetic** Genes, Variants: (2703, 7846) -> (1, 1)

変異の意義を知る: dbSNP



The screenshot shows the Illumina VariantStudio interface. The 'Gene View' table displays the following data:

Gene	ENSP	HGVSc	HGVSp	dbSNP ID	Ances... Allele	Allele Freq	Allele Freq Global Minor	Global Minor Allele	Allele Freq Amr	Allele Freq Asn	Allele Freq Af	Allele Freq Eur	Allele Freq Evs	EVS Coverage	EVS Samples
ZIC3	NP_003404.1	NM_003413.3:c.804C>A	NP_003404.1:p.Cys268Ter	rs122462166		0.00	0.00		0.00	0.00	0.00	0.00	0.00	49.0	6503

Text overlay: dbSNP: NCBIとNHGRIが運営する変異データベース
SNPを始め、indelやマイクロサテライトマーカなどの情報を有する

Sample: NA12882_Synthetic Genes, Variants: (2703, 7846) -> (1, 1)

変異の意義を知る: dbSNP

NCBI dbSNP Short Genetic Variations

Search small variations in dbSNP or large structural variations in dbVar

Reference SNP (refSNP) Cluster Report: rs122462166 **** With Pathogenic allele ****

RefSNP	Allele	HGVS Names
Organism: human (Homo sapiens)	SNV: single nucleotide variation	NC_000023.10:g.136649654C>A
Molecule Type: Genomic	Variation Class:	NC_000023.11:g.137567495C>A
Created/Updated in build: 133/146	RefSNP Alleles: A/C (FWD)	NG_008115.1:g.6309C>A
Map to Genome Build: 107/Weight	Allele Origin: A:germline C:germline	NM_003413.3:c.804C>A
Validation Status:	Ancestral Allele: C	NP_003404.1:p.Cys268Ter
	Variation Viewer: VarView OMIM	
	Clinical Significance: With Pathogenic allele [ClinVar]	
	MAF/Minor Allele Count: NA	
	MAF Source:	

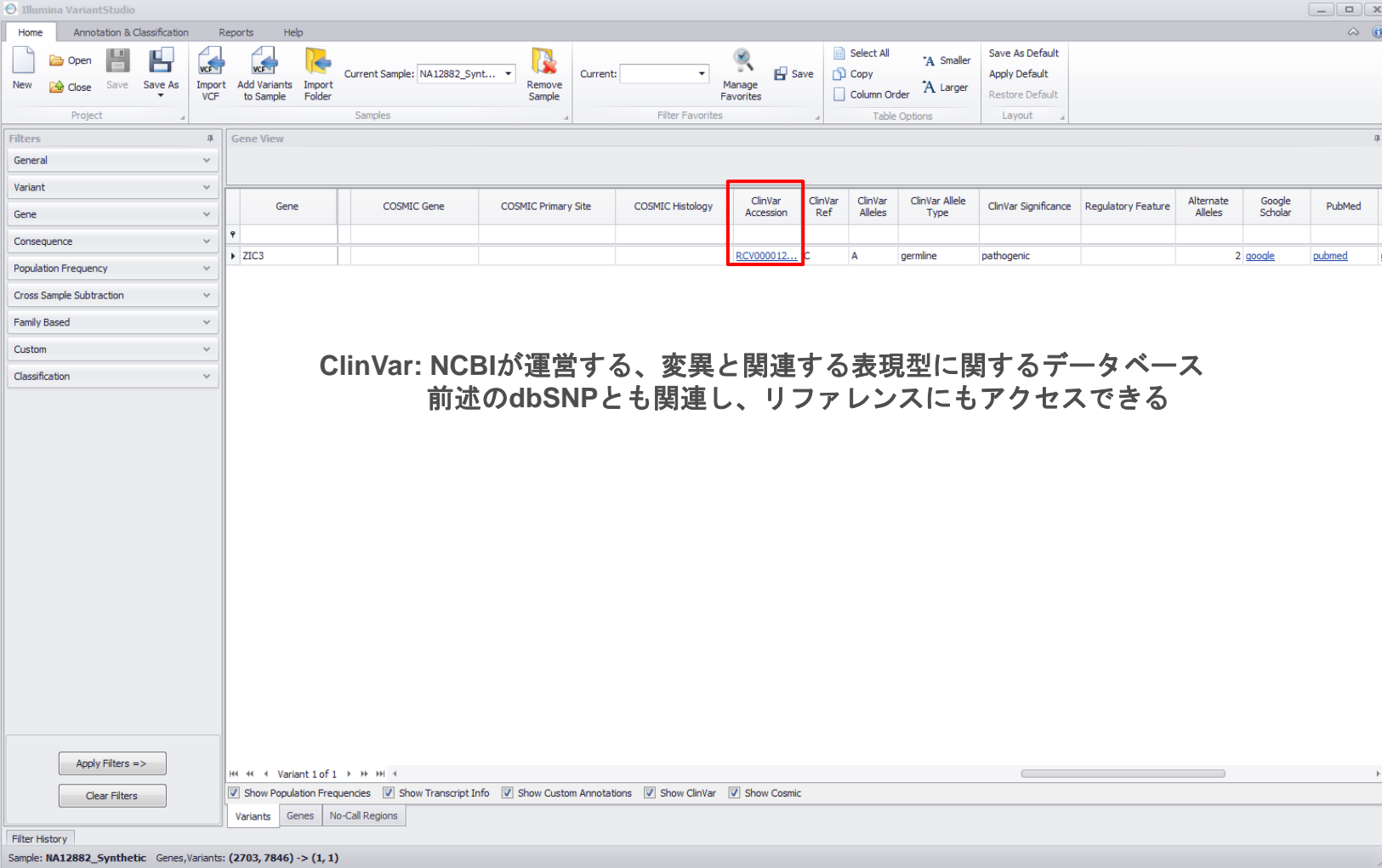
SNP Details are organized in the following sections:
[GeneView](#) [Map](#) [Submission](#) [Fasta](#) [Resource](#) [Diversity](#) [Validation](#)

Integrated Maps (Hint: click on 'Chr Pos' to see variant in the new NCBI variation viewer)

Assembly	Annotation Release	Chr	Chr Pos	Contig	Contig Pos	SNP to Chr	Contig allele	Contig to Chr	Neighbor SNP	Map Method
GRCh38.p2	107	X	137567495	NT_011786.17	20971929	Fwd	C	Fwd	view	mapup
GRCh37.p13	105	X	136649654	NT_011786.16	20917364	Fwd	C	Fwd	view	blast

データベース内を参照すると、変異の発病性などについて知ることができる

変異の意義を知る: ClinVar



The screenshot shows the Illumina VariantStudio interface. The main window displays a table of variant data for the gene ZIC3. The table has the following columns: Gene, COSMIC Gene, COSMIC Primary Site, COSMIC Histology, ClinVar Accession, ClinVar Ref, ClinVar Alleles, ClinVar Allele Type, ClinVar Significance, Regulatory Feature, Alternate Alleles, Google Scholar, and PubMed. The ClinVar Accession column is highlighted with a red box, showing the value RCV000012... for the variant. Below the table, there is a large text overlay in Japanese.

ClinVar: NCBIが運営する、変異と関連する表現型に関するデータベース
前述のdbSNPとも関連し、リファレンスにもアクセスできる

Variant 1 of 1

Show Population Frequencies Show Transcript Info Show Custom Annotations Show ClinVar Show Cosmic

Filter History

Sample: NA12882_Synthetic Genes, Variants: (2703, 7846) -> (1, 1)

変異の意義を知る: ClinVar

NCBI Resources How To Sign in to NCBI

ClinVar Search ClinVar for gene symbols, HGVS expressions, conditions, and more Search Help

Home About Access Using the website How to submit Statistics FTP site

NM_003413.3(ZIC3):c.804C>A (p.Cys268Ter)

Variation ID: 11434
Review status: (0/4) no assertion criteria provided

Interpretation

Clinical significance: [Pathogenic](#)
Last evaluated: Nov 1, 1997
Number of submission(s): 1
Condition(s): Heterotaxy, visceral, X-linked [\[MedGen - OMIM\]](#)
[See supporting ClinVar records](#)

Allele(s)

NM_003413.3(ZIC3):c.804C>A (p.Cys268Ter)

Allele ID: 26473
Variant type: single nucleotide variant
Cytogenetic location: Xq26
Genomic location:

- ChrX: 137567495 (on Assembly GRCh38)
- ChrX: 136649654 (on Assembly GRCh37)

Protein change: C270*, C268*
HGVS:

- NG_008115.1:g.6309C>A
- NM_003413.3:c.804C>A
- NC_000023.11:g.137567495C>A (GRCh38)
- NP_003404.1:p.Cys268Ter
- NC_000023.10:g.136649654C>A (GRCh37)

Links:

- OMIM: [300265.0002](#)
- dbSNP: [122462166](#)

1 Affected gene

Zic family member 3 (ZIC3) [Gene - OMIM - Variation Viewer]
Haploinsufficiency - *Sufficient evidence for dosage pathogenicity* (Apr 26, 2012)
Triplosensitivity - *No evidence available* (Apr 26, 2012)
[Search ClinVar for variants within ZIC3](#)
[Search ClinVar for variants including ZIC3](#)

Variant frequency in dbGaP

NM_003413.3(ZIC3):c.804C>A (p.Cys268Ter)
GRCh37 ChrX:136649654

	Called variants	Potential variants
Sample count	no data	0 of 39618

Called variants are **samples** submitted to dbGaP that have the variant allele.
Potential variants are **SRA runs** that display the allele in at least 30% of the reads covering the position, and have 10 or more passing reads covering the position.

Browser views

[RefSeqGene](#)
[Variation Viewer \[GRCh38 - GRCh37\]](#)
[UCSC \[GRCh38/hg38 - GRCh37/hg19\]](#)

Related information

[dbSNP](#)
[Gene](#)
[MedGen](#)

変異の発病性や、表現型について情報を得ることができる

変異の意義を知る: OMIM

The screenshot shows the Illumina VariantStudio interface. The main window displays a table of variants. The 'ClinVar OMIM' column for the variant ZIC3 is highlighted with a red box, showing the value 306955. Below the table, there is a text box with the following content:

OMIM: Online Mendelian Inheritance in Man
NCBIが提供する、変異と遺伝性疾患との関連性に特化したデータベース
リファレンスなどにもアクセス可能

The interface also includes a sidebar with filters, a top menu bar, and a status bar at the bottom indicating the current sample and variant details.

Gene	Regulatory Feature	Alternate Alleles	Google Scholar	PubMed	UCSC Browser	ClinVar RS	ClinVar Disease Name	ClinVar MedGen	ClinVar OMIM	ClinVar Orphanet	ClinVar GeneReviews	ClinVar SnoMedCt ID
ZIC3		2	google	pubmed	ucsc	rs122462166	Heterotaxy...		306955	116718		

変異の意義を知る: OMIM

#306955

ICD+

HETEROTAXY, VISCERAL, 1, X-LINKED; HTX1

Alternative titles; symbols

DEXTROCARDIA WITH OTHER CARDIAC MALFORMATIONS

LATERALITY, X-LINKED

SITUS INVERSUS, COMPLEX CARDIAC DEFECTS, AND SPLENIC DEFECTS, X-LINKED

Other entities represented in this entry:

CONGENITAL HEART DEFECTS, MULTIPLE TYPES, 1, X-LINKED, INCLUDED; CHTD1, INCLUDED

Phenotype-Gene Relationships

Location	Phenotype	Phenotype MIM number	Inheritance (<i>in progress</i>)	Phenotype mapping key	Gene/Locus	Gene/Locus MIM number
Xq26.3	Heterotaxy, visceral, 1, X-linked	306955	XLR	3	ZIC3	300265
Xq26.3	Congenital heart defects, nonsyndromic, 1, X-linked	306955	XLR	3	ZIC3	300265

Clinical Synopsis

Phenotypic Series

TEXT

A number sign (#) is used with this entry because X-linked heterotaxy-1 (HTX1) and multiple types of congenital heart defects-1 (CHTD1) are caused by mutation in the ZIC3 gene (300265) on chromosome Xq26.

Mutation in the ZIC3 gene can also cause VACTERL with or without hydrocephalus (VACTERLX; 314390), a disorder with overlapping features.

変異が関与する遺伝性疾患について知ることができる

Public dataの取得

The image shows two overlapping screenshots of the BaseSpace web interface. The top screenshot displays the 'Public Data' page with a search bar containing 'truseq custom amplicon'. Three project entries are listed, with the first one highlighted by a red box. The bottom screenshot shows the 'PROJECTS' page for the selected project, featuring a table of analyses with the first row highlighted by a red box.

Public Data Page:

- Search: truseq custom amplicon
- Categories: Exome, Resequencing, Small RNA, Targeted Sequencing
- Project 1: [MiniSeq: TruSeq Custom Amplicon Low Input Dual Strand](#) (targeted-sequencing, cancer-research)
- Project 2: [MiniSeq: TruSeq Custom Amplicon Low Input \(replicates of Horizon samples and NA12878\)](#) (targeted-sequencing, cancer-research)
- Project 3: [MiSeq v3: TruSeq Custom Amplicon Low Input \(HDx, 10ng samples\)](#) (resequencing)

Project Page:

Projects: MiniSeq: TruSeq Custom Amplicon Low Input Dual Strand

Analyses (5)

	NAME	LAST MODIFIED	STATUS	APPLICATION	SIZE
<input type="checkbox"/>	TruSeq Amplicon (single strand analysis for pool A)	Jan 12, 2016	Complete	TruSeq Amplicon	0 B
<input type="checkbox"/>	Amplicon DS	Jan 11, 2016	Complete	Amplicon DS	1 GB
<input type="checkbox"/>	TruSeq_CAT_Manifest_B	Jan 10, 2016	Complete	Imported	85 KB
<input type="checkbox"/>	TruSeq_CAT_Manifest_A	Jan 10, 2016	Complete	Imported	85 KB
<input type="checkbox"/>	FASTQ Generation 2016-01-08 23:48:23Z	Jan 09, 2016	Complete	FASTQ Generation	4 GB

本日の内容

▶ BaseSpace cloud app, TruSeq Ampliconを用いたデータ解析

- ーシーケンスデータの参照
- ー変異の解析結果の参照

▶ VariantStudioソフトウェアを用いたデータ解析

- ーデータのインポート
- ー変異の絞り込み
- ーアノテーション
- ーデータベースを利用した知見の入手

注：解析にはデモデータを使用しており、細かくは実際と異なる場合がございます。
データの参照と解析の流れをご案内する一例としてご参考下さい。