



ThermoFisher
S C I E N T I F I C

Ion Reporter™ Software

Data Analysis Essentials

The world leader in serving science

Системы Ion GeneStudio S5 | Портфолио для решения широкого спектра задач

Ion GeneStudio™ S5



Быстрый.

Ion GeneStudio™ S5 Plus



Гибкий.

Ion GeneStudio™ S5 Prime



Мощный.



Чип Ion 510™

2–3 млн. прочтений
До 400 п.н.



Чип Ion 520™

3–6 млн. прочтений
До 600 п.н.



Чип Ion 530™

15–20 млн. прочтений
До 600 п.н.



Чип Ion 540™

60–80 млн. прочтений
До 200 п.н.



Чип Ion 550™

100–130 млн. прочтений
До 200 п.н.

For Research Use Only. Not for use in diagnostic procedures. * Throughputs based on 200bp sequencing

Платформа Ion Torrent NGS: 45 минут ручного труда

Система Ion Chef™



Подготовка библиотек, матрицы
и загрузка чипов

Простой запуск <30 мин.

Всего 2 стадии пипетирования

Протокол, не требующий присутствия оператора

Система Ion GeneStudio™ S5



Секвенирование,
анализ данных

Простой запуск <15 мин.

Готовые к использованию реактивы

Масштабируемая производительность: 2–130 млн.

For Research Use Only. Not for use in diagnostic procedures.

45 минут ручного труда от ДНК до результата | 2 стадии пипетирования / образец



For Research Use Only. Not for use in diagnostic procedures.

Ion Reporter Software

← → ↻ 🏠 <https://ionreporter.thermofisher.com/ir/> ☆ * 🌐 ☰

ion torrent
Sequencing for all™

INFORMATICS
WORKFLOW

DESIGN


SEQUENCE

REPORT

ION REPORTER™ SOFTWARE


A secure, hosted data analysis tool to simplify the informatics associated with routine assays around DNA variation.

[Sign In](#) or [Register new account](#)




ANALYZE

Automated analysis of 1, 2 or 3 samples.




ANNOTATE

Richly annotate your variants from a wide range of public or custom annotations.



FILTER

Rapidly filter variants to identify those that are biologically relevant.



REPORT & EXPORT

Simplified reporting and exporting of variants and data.

Agenda

Sequence

Import

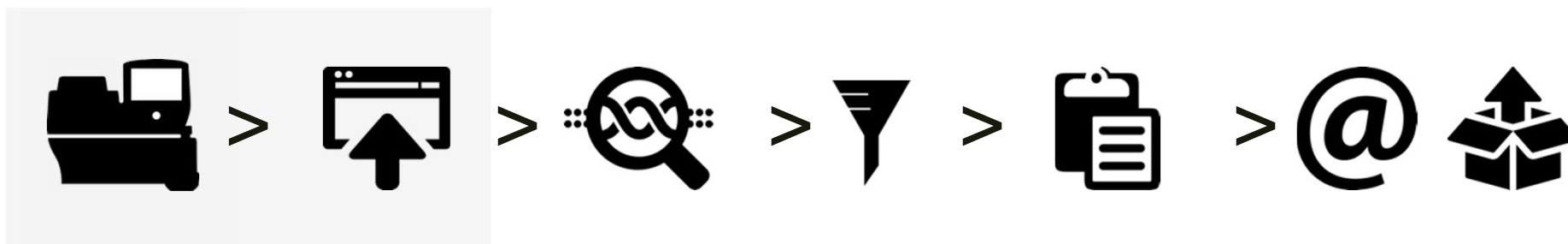
Analyze

Filter

Report

Share

Export



- Launch Analysis and Workflow Overview
- Variant Review, Filter, Classification, and Report
- Data Upload
- Download, Export and Share

Ion Reporter™ Software Overview

Ion Reporter

Hi, IR Admin0/1 TBHelpSign Out⚙

Home

Samples

Analyses

Workflows

Admin

OverviewLaunchMy Variants

Analysis Results

BackDownload ▾Selected Variants ▾Switch To ▾Generate Report

Analysis Name: AmpliSeq Exome 1

To learn more about reviewing your results, visit the [help guide](#).

SummaryFunctionalPopulationOntologiesPharmacogenomicsSomaticQC

⚙ Actions ▾

Search

<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	Locus	Genot...	Ref	Genes	Str...	Transcript	Coding	Amino Aci...	Variant Effect	PhyloP
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	chr1:871334	T/T	G	SAMD11	+	NM_152486.2				0.01
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	chr1:874820	C/T	C	SAMD11	+	NM_152486.2	c.686C>T	p.Pro229Leu	missense	-0.76
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	chr1:874821	T/C	T	SAMD11	+	NM_152486.2	c.687T>C	p.(=)	synonymous	-1.67
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	chr1:876499	G/G	A	SAMD11	+	NM_152486.2				0.09
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	chr1:877715	G/G	C	SAMD11	+	NM_152486.2				-1.08
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	chr1:878723	T/G	T	NOC2L ... (2)	+, -	NM_152486.2, NM_015658.3	c.1655T>G	p.Val552Gly	missense	2.07
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	chr1:880238	G/G	A	NOC2L ... (2)	+, -	NM_152486.2, NM_015658.3				-0.61
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	chr1:881627	A/A	G	NOC2L	-	NM_015658.3	c.1843C>T	p.(=)	synonymous	2.4

Filter Options

×

Variants

- Filtered In Variants (54402)
- Hidden Variants (0)
- Filtered Out Variants (0)

Samples

- Proband: Demo AmpliSeq Exome VCF
 - Gender : Female
 - Sample Type : DNA

Chromosome

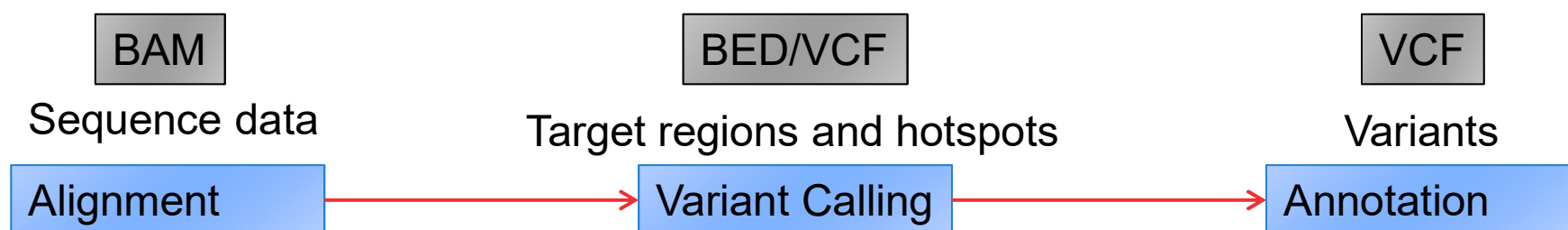
All ▾

Filter Chains

END FEEDBACK

Ion Reporter™ software workflows

- Workflow is a set of instructions for data analysis
- Key steps in a typical variant calling and annotation workflow

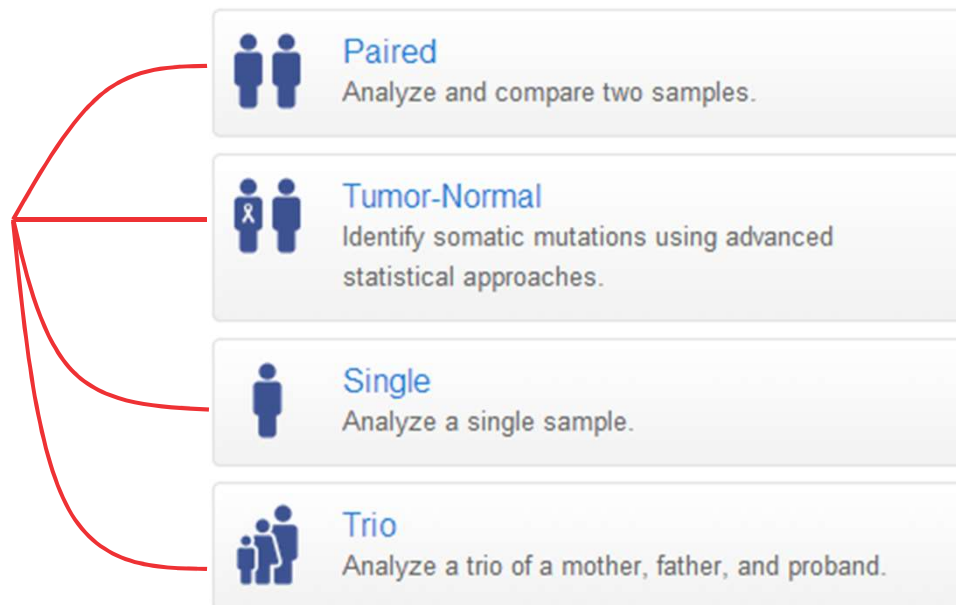
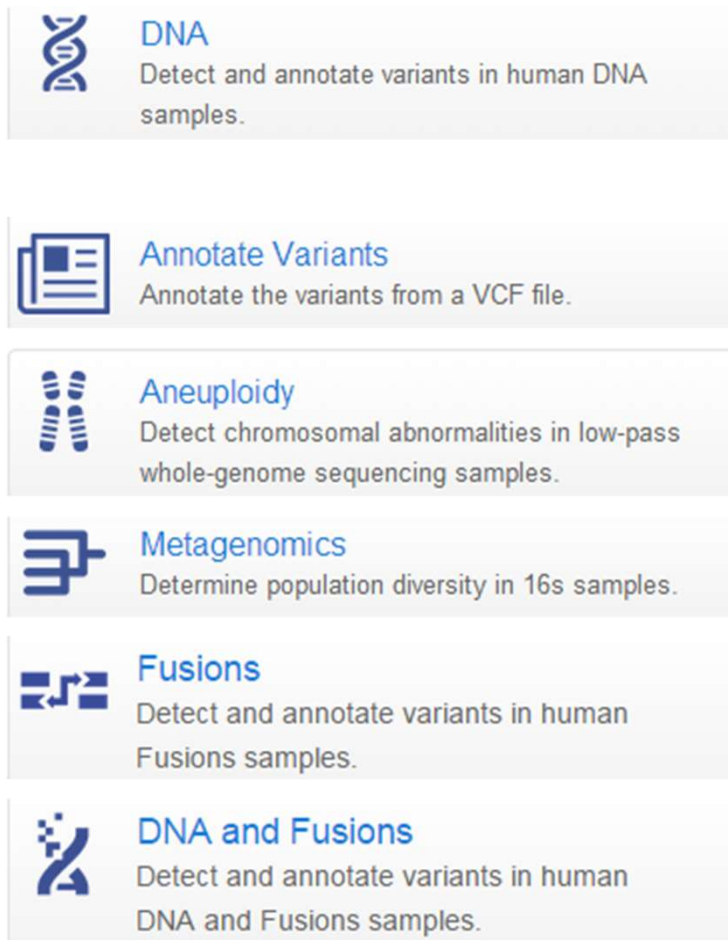


- Other workflows



Ion Reporter™ software workflows

- Types of workflows



Annotations

- Information describing the variant type and location

Type	SNV
Coding (Nt change)	c.292A>G
Amino Acid Change	p.Thr98Ala
Genotype	A/G
Variant Effect	Missense
Gene	DDR2
Location	Exonic
Exon	5
Locus	Chr1:162724529
Transcript	NM_006182.3
Strands	+

Annotations

- Databases
 - Gene Ontology – controlled vocabulary for describing gene products
 - OMIM® – Online Mendelian Inheritance in Man™
 - PFAM – Database of protein families
 - ClinVar – Relationships among variation and human health
 - DrugBank – Drugs known to target the gene affected by the variant
 - COSMIC – Catalog of somatic mutations in cancer
 - dbSNP – Database of genomic variants

Annotations

- Annotation scores predicts
 - SIFT – whether an a.a. substitution affects protein function
 - Grantham – distance between two a.a. in evolutionary terms
 - PolyPhen-2 – possible impact on the structure and function of protein
 - Phylop – measures evolutionary conservation
- Other annotations
 - Population allele frequencies, 5000 exome, 1000 genome projects
 - Genetic category – for variant inheritance in trio samples
 - HotSpot information
 - Custom annotations

Filters

- Use filters to change which variants are displayed
 - 500Exomes African/European/Global
 - Allele Ratio
 - Allele Read-Count
 - ClinVar
 - CNV Confidence Range
 - COSMIC
 - dbSNP
 - DrugBank
 - Filtered Coverage
 - Functional Scores
 - Gene Ontology
 - Gene Symbol
 - HotSpot
 - Ingenuity Variant Analysis
 - Location
 - Minor Allele Frequency
 - My Variants
 - OMIM®
 - Pfam
 - PValue
 - UCSC common SNPS
 - Variant Effect
 - Variant Type
 - Zygotity

Launch Analysis and Workflow Overview



Ion Reporter Software

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Sequencing for all™

INFORMATICS
WORKFLOW

DESIGN


SEQUENCE

REPORT

ION REPORTER™ SOFTWARE


A secure, hosted data analysis tool to simplify the informatics associated with routine assays around DNA variation.

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ANALYZE

Automated analysis of 1, 2 or 3 samples.




ANNOTATE

Richly annotate your variants from a wide range of public or custom annotations.



FILTER

Rapidly filter variants to identify those that are biologically relevant.



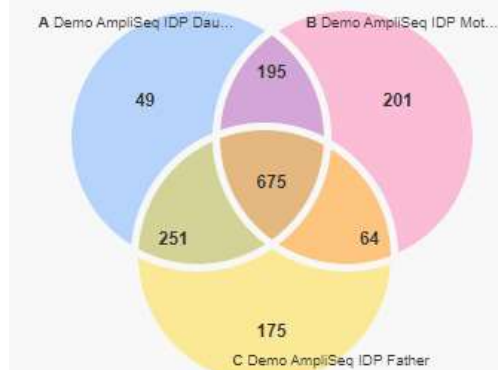
REPORT & EXPORT

Simplified reporting and exporting of variants and data.

Launch Analysis – Hands on Practice

1. Login into Ion Reporter > Analyses Tab
2. Launch Analysis > Manual

Samples



idp Go

Research Category Research Application Workflow Target Group Version Reference

ion	Research Category	Research Application	Workflow Name	Version	Reference	Sample Group	Modified On
ion	Inherited Disease	DNA	AmpliSeq IDP single sample	5.12	hg19	Single	Sep 13 2019 02:17 AM
ion	Inherited Disease	DNA	AmpliSeq IDP trio	5.12	hg19	Trio	Sep 13 2019 02:17 AM

idp Go

Samples

<input type="checkbox"/>	<input type="checkbox"/>	Sample	Gender	Sample Type
<input type="checkbox"/>	<input type="checkbox"/>	Demo AmpliSeq IDP Daughter	Female	DNA
<input checked="" type="checkbox"/>	<input type="checkbox"/>	Demo AmpliSeq IDP Mother	Female	DNA
<input checked="" type="checkbox"/>	<input type="checkbox"/>	Demo AmpliSeq IDP Father	Male	DNA

Launch Ion Reporter Analysis – Hands on Practice

1. Login into Ion Reporter > Analyses Tab
2. Launch Analysis > Manual
 - **Workflow** : “AmpliSeq IDP single sample” > Next
 - **Samples** : “Demo AmpliSeq IDP Mother, Demo AmpliSeq IDP Father” > Next
 - **Plugins** : Next
 - **Confirm & Launch** : Enter analysis name > Next
3. Launch Analysis

Launch Analysis

Workflow Samples Plugins **Confirm & Launch**

Analysis ready to launch!

Review the selected options, name your analysis and then launch it.

Analysis Name:
(Demo AmpliSeq Exome VCF)

Description:

Launch Analysis

Summary

Application: Annotate Variants
Workflow: Annotate variants single sample
Annotations: All
Samples: 1 Sample
Price: \$0.00 USD

Launch Ion Reporter Analysis

Ion Reporter

Hi, IR Admin0/1 TBHelpSign Out⚙

HomeSamplesAnalysesWorkflowsAdmin

OverviewLaunchMy Variants

Analyses

StatusApplicationVersionDemo AmpliSeq ExSearchRefresh

<input type="checkbox"/>	<input checked="" type="checkbox"/>	<input type="checkbox"/>	Analysis	Version	Stage	Project	Workflow	Created On	Status
<input type="checkbox"/>			Demo AmpliSeq Exome VCF_c780_2016-01-27-22-57-505	5.0	Analysis		Annotate variants single sample	Running - 33%	

20 items per page

1 - 1 of 1 items

Details

⚙ Actions

👤 (0)

Demo AmpliSeq Exome VCF_c780_2016-01-27-22-57-505

Version: 5.0

Stage: Analysis

Status: Running - 33%

Sample Group: Single

Workflow: Annotate variants single sam

Variant Review, Filter, Classification, and Report



Analysis Results - Functional

Analysis Results

Analysis Name: AmpliSeq Exome 1

Summary **Functional** Popu

Back Download Selected Variants Send to Report Role Switch To Generate Report

Actions Search

Filter Options

Variants

- Filtered In Variants (54402)
- Hidden Variants (0)
- Filtered Out Variants (0)

Sample Type : DNA

Gender : Female

Chromosome

All

Filter Chains

No Filter

SEND FEEDBACK

Prediction of the functional effect of a variant on a protein

A measure of evolutionary distance

Measure of conservation of the protein across a wide range of organisms

Protein domain families in the coded protein

Locus	Gen...	Genes	Transcript	Coding	Amino Aci...	Variant Effect	PhyloP	SIFT	Grantham	PFAM
chr1:871334	T/T	SAMD11	NM_152486.2				0.01			SAM don
chr1:874820	C/T	SAMD11	NM_152486.2	c.686C>T	p.Pro229Leu	missense	-0.76	0.03		
chr1:874821	T/C	SAMD11	NM_152486.2	c.687T>C	p.(=)	synonymous	-1.67			SAM don
chr1:876499	G/G	SAMD11	NM_152486.2				0.09			SAM don
chr1:877715	G/G	SAMD11	NM_152486.2				-1.08			SAM don
chr1:878723	T/G	NOC2L ... (2)	NM_152486.2, NM_015658.3	c.1655T>C	p.Val552Gly	missense	2.07	0.0	109.0	SAM don
chr1:880238	G/G	NOC2L ... (2)	NM_152486.2, NM_015658.3				-0.61			Noc2p fa

Analysis Results - Population

Analysis Results

Analysis Name: AmpliSeq Exome 1

Summary Functional **Population**

Back Download Selected Variants Send to Report Role Switch To Generate Report

1000 Genome Minor Allele Frequency

European Minor Allele Freq

African Minor Allele Freq

Global Minor Allele Freq

	Classification	Locus	Geno...	Genes	dbSNP	DGV	MAF	EMAF	AMAF	GMAF	UCSC Cor
	Unclassified	chr1:871334	T/T	SAMD11	rs4072383		0.493				YES
	Unclassified	chr1:874820	C/T	SAMD11							
	Unclassified	chr1:874821	T/C	SAMD11							
	Unclassified	chr1:876499	G/G	SAMD11	rs4372192		0.082	0.0623	0.1167	0.0807	YES
	Unclassified	chr1:877715	G/G	SAMD11	rs6605066		0.106				YES
	Unclassified	chr1:878723	T/G	NOC2L ...(2)							
	Unclassified	chr1:880238	G/G	NOC2L ...	rs3748592		0.072				YES

Filter Options

Variants

- Filtered In Variants (54402)
- Hidden Variants (0)
- Filtered Out Variants (0)

Samples

- Proband: Demo AmpliSeq Exome V CF
 - Sample Type : DNA
 - Gender : Female

Chromosome

All

Filter Chains

No Filter

SEND FEEDBACK

Analysis Results - Ontologies

Standardized ontology for gene and gene products
(e.g. functional role or localization)

Catalog of somatic mutations in cancer

Online Mendelian Inheritance in Man®

Analysis Results

Analysis Name: AmpliSeq Exome 1

Summary Functional Population

Back Download Selected Variants Send to Report Role Switch To Generate Report

	Classification	Locus	Gen...	Genes	COSMIC	OMIM	Gene Ontology
	Unclassified	chr1:248737595	G/G	OR2T34	serous_carcinoma ... (2)		G-protein coupled rec
	Unclassified	chr5:140503002	C/A	PCDHB4	superficial_spreading	Protocadherin-beta 4	calcium ion binding ...
	Unclassified	chr1:152191709	C/G	HRNR	squamous_cell_carcinoma		calcium ion binding ...
	Unclassified	chr1:153004853	C/T	SPRR1B	squamous_cell_carcinoma	Small proline-rich protein 1B	cornified envelope ... (
	Unclassified	chr1:109810620	G/T	CELSR2	squamous_cell_carcinoma	Cadherin EGF LAG seven-pass G-type receptor 3	G-protein coupled rec
	Unclassified	chr1:26646730	A/G	CD52	squamous_cell_carcinoma	CD52 molecule	anchored to membrar

Filter Options

Variants

- Filtered In Variants (54402)
- Hidden Variants (0)
- Filtered Out Variants (0)

Samples

- Proband: Demo AmpliSeq Exome V CF
 - Sample Type : DNA
 - Gender : Female

Chromosome

All

Filter Chains

No Filter

SEND FEEDBACK

Analysis Results - Pharmacogenomics

Analysis Results

Analysis Name: AmpliSeq Exome 1

Summary Functional Population Ontologies Pharmacogenomics Somatic OC

List of drugs reported to target the gene(s) affected by the variant

Assessment of impact of the variant observed from NCBI ClinVar database

	Classification	Locus	Genes	Genotype	DrugBank ▼	ClinVar
[Icon] [Dropdown]	Unclassified	chr6:38650628	GLO1	T/G	Beta-Mercaptoethanol ... (8)	unknown
[Icon] [Dropdown]	Unclassified	chr3:12475557	PPARG	C/T	(2S)-2-(4-chlorophenoxy)-3-phenylpropanoic acid ... (37)	non-pathogenic
[Icon] [Dropdown]	Unclassified	chr2:227660544	IRS1	C/T	4-({5-(AMINOCARBONYL)-4-(3-METHYLPHENYL)AMINOPYRIMIDI 2-YL}AMINO)PHENYLACETIC ACID	other
[Icon] [Dropdown]	Unclassified	chr10:75673101	C10orf55 ... (2)	T/C	(2R)-1-(2,6-dimethylphenoxy)propan-2-amine ... (33)	other
[Icon] [Dropdown]	Unclassified	chr22:42523943	CYP2D6	G/G	1-(2-Phenylethyl)-4-phenyl-4-acetoxypiperidine ... (293)	untested
[Icon] [Dropdown]	Unclassified	chr22:42526694	CYP2D6	G/A	1-(2-Phenylethyl)-4-phenyl-4-acetoxypiperidine ... (293)	pathogenic

- Filtered In Variants (531)
- Hidden Variants (0)
- Filtered Out Variants (53871)

Samples

- Proband: Demo AmpliSeq Exome V CF
 - Sample Type : DNA
 - Gender : Female

Chromosome


All ▼

Filter Chains

clinVar ▼

SEND FEEDBACK

Analysis Results - Somatic

 **Analysis Results**

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Analysis Name: AmpliSeq Exome 1

[Summary](#) [Functional](#) [Population](#) [Ontologies](#) [Pharmacogenomics](#) **[Somatic](#)** [QC](#) [Actions](#)

[Search](#)

		Classification	Locus	Genot...	Genes	Allele Coverage	Allele Ratio
<input type="checkbox"/>		Unclassified	chr1:871334	T/T	SAMD11	G=2, T=81	G=0.0241, T=0.9759
<input type="checkbox"/>		Unclassified	chr1:874820	C/T	SAMD11	C=4, T=7	C=0.3636, T=0.6364
<input type="checkbox"/>		Unclassified	chr1:874821	T/C	SAMD11	T=3, C=7	T=0.3, C=0.7
<input type="checkbox"/>		Unclassified	chr1:876499	G/G	SAMD11	A=0, G=47	A=0.0, G=1.0
<input type="checkbox"/>		Unclassified	chr1:877715	G/G	SAMD11	C=0, G=7	C=0.0, G=1.0
<input type="checkbox"/>		Unclassified	chr1:878723	T/G	NOC2L ... (2)	T=62, G=18	T=0.775, G=0.225
<input type="checkbox"/>		Unclassified	chr1:880238	G/G	NOC2L ... (2)	A=0, G=74	A=0.0, G=1.0

Filter Options ×

Variants

- **Filtered In Variants (54402)**
- [Hidden Variants \(0\)](#)
- [Filtered Out Variants \(0\)](#)

Samples

- **Proband: Demo AmpliSeq Exome V**
CF
 - **Sample Type** : DNA
 - **Gender** : Female

Chromosome


All

Filter Chains

No Filter

[SEND FEEDBACK](#)


Analysis Results - QC

 **Analysis Results**

















[Back](#) [Download](#) [Selected Variants](#) [Send to Report Role](#) [Switch To](#) [Generate Report](#)


Analysis Name: AmpliSeq Exome 1

[Summary](#) [Functional](#) [Population](#) [Ontologies](#) [Pharmacogenomics](#) [Somatic](#) **QC**

 Actions

[Search](#)

<input type="checkbox"/>			Classification	Locus	Genot...	Genes	p-value	Phred ...	Allele C...	Allele Ratio	Ref+/F
<input type="checkbox"/>			Unclassified	chr1:871334	T/T	SAMD11	0.00001	100.0	G=2, T=81	G=0.0241, T=0.9759	G=1/1,
<input type="checkbox"/>			Unclassified	chr1:874820	C/T	SAMD11	0.00002	47.17	C=4, T=7	C=0.3636, T=0.6364	C=1/3,
<input type="checkbox"/>			Unclassified	chr1:874821	T/C	SAMD11	0.00001	51.24	T=3, C=7	T=0.3, C=0.7	T=1/2,
<input type="checkbox"/>			Unclassified	chr1:876499	G/G	SAMD11	0.00001	100.0	A=0, G=47	A=0.0, G=1.0	A=0/0,
<input type="checkbox"/>			Unclassified	chr1:877715	G/G	SAMD11	0.00001	61.22	C=0, G=7	C=0.0, G=1.0	C=0/0,
<input type="checkbox"/>			Unclassified	chr1:878723	T/G	NOC2L ... (2)	0.00097	30.15	T=62, G=18	T=0.775, G=0.225	T=61/1
<input type="checkbox"/>			Unclassified	chr1:880238	G/G	NOC2L ... (2)	0.00001	100.0	A=0, G=74	A=0.0, G=1.0	A=0/0,

Filter Options 

Variants

- **Filtered In Variants (54402)**
- [Hidden Variants \(0\)](#)
- [Filtered Out Variants \(0\)](#)

Samples

- **Proband: Demo AmpliSeq Exome V CF**
 - **Sample Type : DNA**
 - **Gender : Female**

Chromosome


All

Filter Chains

No Filter

[SEND FEEDBACK](#)

Analysis Results - Variant Details

 **Analysis Results**

[Back](#) [Download](#) [Selected Variants](#) [Send to Report Role](#) [Switch To](#) [Generate Report](#)

Analysis Name: AmpliSeq Exome 1

[Summary](#) [Functional](#) [Population](#) [Ontologies](#) [Pharmacogenomics](#) [Somatic](#) [QC](#) [Actions](#)

Variant Details

		Classification	Locus	Genotype	Ref	Type	Genes	Location
<input type="checkbox"/>		Unclassified	chr1:871334	T/T	G	SNV	SAMD11	intronic
<input type="checkbox"/>		Unclassified	chr1:874820	C/T	C	SNV	SAMD11	exonic
<input type="checkbox"/>		Unclassified	chr1:874821	T/C	T	SNV	SAMD11	exonic
<input type="checkbox"/>		Unclassified	chr1:876499	G/G	A	SNV	SAMD11	intronic
<input type="checkbox"/>		Unclassified	chr1:877715	G/G	C	SNV	SAMD11	intronic
<input type="checkbox"/>		Unclassified	chr1:878723	T/G	T	SNV	NOC2L ... (2)	exonic, downstream
<input type="checkbox"/>		Unclassified	chr1:880238	G/G	A	SNV	NOC2L ... (2)	downstream, intronic

Filter Options ×

Variants

- **Filtered In Variants** (54402)
- [Hidden Variants](#) (0)
- [Filtered Out Variants](#) (0)

Samples

- **Proband:** Demo AmpliSeq Exome V CF
 - **Gender :** Female
 - **Sample Type :** DNA

Chromosome

All

Filter Chains

No Filter

[SEND FEEDBACK](#)

Analysis Results - Variant Details

Ion Reporter

Hi, IR Admin 0/1 TB Help Sign Out

Home Overview

Analysis Name: A

Summary

Variant Details: chr1:11856378

Variant Details Notes

Annotation Source	Demo AmpliSeq Exome VCF
AMAF	0.1217
Allele Coverage	G=83, A=82
Allele Ratio	G=0.503, A=0.497
Amino Acid Change	p.Ala222Val
COSMIC	adenocarcinoma
ClinVar	non-pathogenic
Coding	c.665C>T
Codon	GTC
Coverage	165
DGV	
DrugBank	Benazepril ... (9)

Unclassified chr1:46870761 A/A C SNV FAAH exonic Chromosome

Generate Report

SEND FEEDBACK

My Variants

Analysis Results

Analysis Name: AmpliSeq Exome 1

Summary Functional Population Ontologies Pharmacogenomics Somatic QC

Back Download Selected Variants Send to Report Role Switch To Generate Report

Actions

If a variant is flagged, it will be added to My Variants

	Classification	Locus	Genotype	Ref	Type	Genes	Location
	Unclassified	chr1:9323910	G/A	G	SNV	H6PD	exonic
	None	chr1:11856378	G/A	G	SNV	MTHFR	exonic
	Important	chr1:31349647	C/T	C	SNV	SDC3	exonic
	Ignore						
	Unclassified	chr1:46870761	A/A	C	SNV	FAAH	exonic
	Unclassified	chr1:53712727	T/T	C	SNV	LRP8	exonic
	Unclassified	chr1:55518316	T/T	C	SNV	PCSK9	intronic
	Unclassified	chr1:55524387	C/C	T	SNV	PCSK9	intronic

Filter Options

Variants

- Filtered In Variants (531)
- Hidden Variants (0)
- Filtered Out Variants (53871)

Samples

- Proband: Demo AmpliSeq Exome V CF
 - Gender : Female
 - Sample Type : DNA

Chromosome

All

Filter Chains

clinVar (5.0)

SEND FEEDBACK

My Variants

Ion Reporter

Help Sign Out ⚙

Home Samples Analyses Workflows Admin

Overview Launch My Variants

My Variants will be flagged across all analysis results

My Variants

Import MyVariants Export TSV

Search

My Variants

- Show all ✓
- Important
- Ignore

		Classification	Gene(s)	GenoType	Ref	Type	Analysis Name	Sample Name
		UNCLASSIFIED	SAMD11	T/T	G	SNV	User 9 - AmpliSeq Exome ... (26)	training_sample ... (3)
▶	🚩	chr1:874820 UNCLASSIFIED	SAMD11	C/T	C	SNV	User 9 - AmpliSeq Exome ... (25)	training_sample ... (3)
▶	🚩	chr1:1254841 UNCLASSIFIED	CPSF3L	G/G	C	SNV	User 9 - AmpliSeq Exome ... (27)	training_sample ... (3)
▶	🚩	chr1:43815008 UNCLASSIFIED	MPL	TGG/TGG ... (2)	TGG ... (2)	REF	LLC1_c1723_2015-10-07-12-09-156 ... (9)	VV_sample ... (9)
▶	🚩	chr1:43815020 UNCLASSIFIED	MPL	G/G	G	REF	LLC1_c1723_2015-10-07-12-09-156 ... (9)	VV_sample ... (9)

◀ ◁ 1 ▷ ▶ 20 items per page

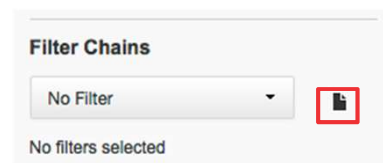
1 - 5 of 5 items

SEND FEEDBACK

Filter Chain

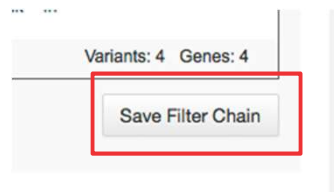
1. Click on New Filter Chain

- Enter a filter name
- Create the following filters



Variant Effect	all but unknown, synonymous
Location	Intronic, Exonic
Pvalue	0 to 0.0001
ClinVar	All pathogenic
MAF	<0.005
Disease research area	Phenylketonuria

- Scroll Down > Apply





2. Click “Save Filter Chain”

- Filter is now available for other IR Report

FilterChain Query

Minor Allele Frequency AND Variant Effect AND __ClinVar(20180729)__1 AND Location AND PValue

Selected Filters

Name	Value	
Minor Allele Frequency	0.0 <= Minor Allele Frequency <= 0.05	+ 
Variant Effect	Variant Effect in refAllele, missense, nonframeshiftInsertion, nonframeshiftDeletion, nonframeshiftBlockSubstitution, nonsense, stoploss, frameshiftInsertion, frameshiftDeletion, frameshiftBlockSubstitution	
__ClinVar(20180729)__1	__ClinVar(20180729)__1 in Pathogenic, Likely pathogenic, Conflicting interpretations of pathogenicity, Pathogenic/Likely pathogenic, Pathogenic, other, Uncertain significance, Likely pathogenic, Pathogenic, Conflicting interpretations of pathogenicity, Conflicting interpretations of pathogenicity, risk factor, Pathogenic, Uncertain significance, Pathogenic, Likely pathogenic, Conflicting interpretations of pathogenicity, Likely pathogenic, Pathogenic, Benign, Conflicting interpretations of pathogenicity, other, other, Pathogenic, other, other, Conflicting interpretations of pathogenicity, Conflicting interpretations of pathogenicity, other, Pathogenic, Pathogenic/Likely pathogenic, Pathogenic, Benign/Likely benign, Pathogenic/Likely pathogenic, drug response, Likely pathogenic, drug response	
Location	Location in intronic, exonic	
PValue	0.0 <= PValue <= 0.0001	

Cancel

Apply

Summary

Functional

Population

Ontologies

Pharmacogenomics

QC

Search

G

	Classification	Locus	Genotype	Ref	Type	N	Cones	DrugBank	ClinVar
	Unclassified	chr12:103234252	T/C	T	SNV	PAH	Beta(2-Thienyl)Alanine ... (10)	Pathogenic	
	Unclassified	chr1:94512565	C/T	C	SNV	ABCA4		Conflicting interpretations of pathogenicity, risk factor	
	Unclassified	chr17:17700037	AAGG/A	AAGG	INDEL	RAI1		Conflicting interpretations of pathogenicity	
	Unclassified	chr11:71146837	C/T	C	SNV	DHCR7	NADH	Conflicting interpretations of pathogenicity	
	Unclassified	chr10:55892642	T/C	T	SNV	PCDH15		Conflicting interpretations of pathogenicity	
	Unclassified	chr7:150644428	C/A	C	SNV	KCNH2	Alfuzosin ... (48)	Conflicting interpretations of pathogenicity	
	Unclassified	chr7:91694743	A/G	A	SNV	AKAP9		Conflicting interpretations of pathogenicity	
	Unclassified	chr2:220284876						Conflicting interpretations of pathogenicity	
	Unclassified	chr1:201331068						Conflicting interpretations of pathogenicity	

NM_000277.3(PAH):c.1241A>G (p.Tyr414Cys)

Cite this record

Interpretation:

Pathogenic

Review status:

★★★★☆ reviewed by expert panel FDA RECOGNIZED DATABASE

Submissions:

15 (Most recent: Mar 28, 2019)

Last evaluated:

Jul 27, 2018

Accession:

VCV000000593.3

Variation ID:

593

Description:

single nucleotide variant

Variant details

Conditions

Gene(s)

Aggregate interpretations per condition

Interpreted condition	Interpretation	Number of submissions	Review status	Last evaluated	Variation/condition record
Phenylketonuria	Pathogenic	9	reviewed by expert panel	Jul 27, 2018	RCV000150074.10

- По какой линии бабушка болела ФКУ?

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A	B	C	D	E	F	G	H	I	J	K	L	M	N	O	P	Q	R	S
#chr	pos	type	ref	length	genotype	gene	transcript	location	function	exon	protein	coding	sift	polyphen	grantham	pvalue	coverage	allele co
6	161127501	SNV	A	1	A/G	PLG	NM_000301.3	exonic	[missense]	2	p.Lys38Glu	c.112A>G	[0.0]	[0.876]	[56.0]	0	122	[63, 59]
7	100771717	SNV	G	1	G/A	SERPINE1:SERPINE1	NM_000602.4:NM_001165413.2	exonic:exonic	[missense]:[missense]	2:02	p.Ala15Thr:p.Ala15Thr	c.43G>A:c.43G>A	:	[0.065]:	[58.0]:[58.0]	0	61	[38, 23]

AmpliSeq Exome 1

Report

Annotate variants sin

Successful

Share Analysis

1

20

items per pag

Demo AmpliSeq Exome VCF_c116_1387557022816

Add User:

Shared with:

E-mail	Shared By	Date	Unshare
You haven't shared this analysis with anyone yet.			

Close

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Download All Variants

Download Logs

Share

View Final Report

View QC Report

Download PDF

Семья с гипертрофической кардиомиопатией

Дочь, 20 лет - признаки синдрома малого выброса - кардиалгия, приступы стенокардии, головокружения, выраженная одышка. Обмороки на высоте нагрузки.


Проявления левожелудочковой сердечной недостаточности. Нарушения ритма сердца — желудочковые экстрасистолы.


Признаки увеличения левого желудочка и левого предсердия. Увеличение правого желудочка

Мать, 45 лет - здорова

Отец, 46 лет - неявные признаки синдрома малого выброса. Гипертрофия межжелудочковой перегородки.

 Mother_IDP.vcf

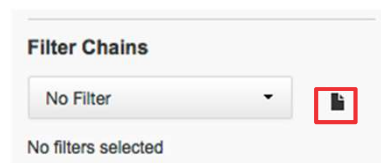
 Father_IDP.vcf

 Daughter_IDP.vcf

Filter Chain

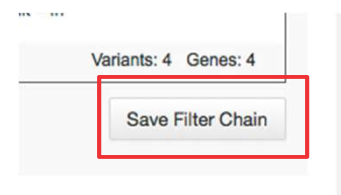
1. Click on New Filter Chain

- Enter a filter name
- Create the following filters



Variant Effect	all but unknown, synonymous
Location	Intronic, Exonic
Pvalue	0 to 0.0001
ClinVar	All pathogenic
MAF	<0.005
OMIM	all cardio (card)

- Scroll Down > Apply



2. Click “Save Filter Chain”

- Filter is now available for other IR Report

Гены с вариантами причастными к кардиомипатиям

Мать:

Locus	Genotype	Ref	Type	No Call Reason	Genes
chr1:201331068	A/G	A	SNV		TNNT2

Отец:

Locus	Genotype	Ref	Type	No Call Reason	Genes
chr11:47364234	T/T	C	SNV		MYBPC3

Дочь:

	Locus	Genotype	Ref	Type	No Call Reason	Genes
▼	chr1:201331068	A/G	A	SNV		TNNT2
▼	chr11:47364234	C/T	C	SNV		MYBPC3

Семья с гипертрофической кардиомиопатией

Symbol Report: MYBPC3 ⓘ

APPROVED SYMBOL ⓘ	MYBPC3
APPROVED NAME ⓘ	myosin binding protein C, cardiac
HGNC ID ⓘ	HGNC:7551
PREVIOUS SYMBOLS & NAMES ⓘ	CMH4, "myosin-binding protein C, cardiac"
SYNONYMS ⓘ	FHC, MYBP-C
LOCUS TYPE ⓘ	gene with protein product
CHROMOSOMAL LOCATION ⓘ	11p11.2
GENE FAMILY ⓘ	<u>Fibronectin type III domain containing</u> <u>I-set domain containing</u> <u>Myosin binding proteins</u>
HCOP ⓘ	<u>Orthology Predictions for MYBPC3</u>

Gene Family: Troponin complex subunits (TNN)

Troponin: Troponin is a complex of three regulatory proteins (troponin C, troponin I, and troponin T) that is integral to muscle contraction in skeletal muscle and cardiac muscle, but not smooth muscle. Discussions of troponin often pertain to its functional characteristics and/or to its usefulness as a diagnostic marker or therapeutic target for various heart disorders in particular as a highly specific marker for myocardial infarction or heart muscle cell death. [Source: [Wikipedia](#)]