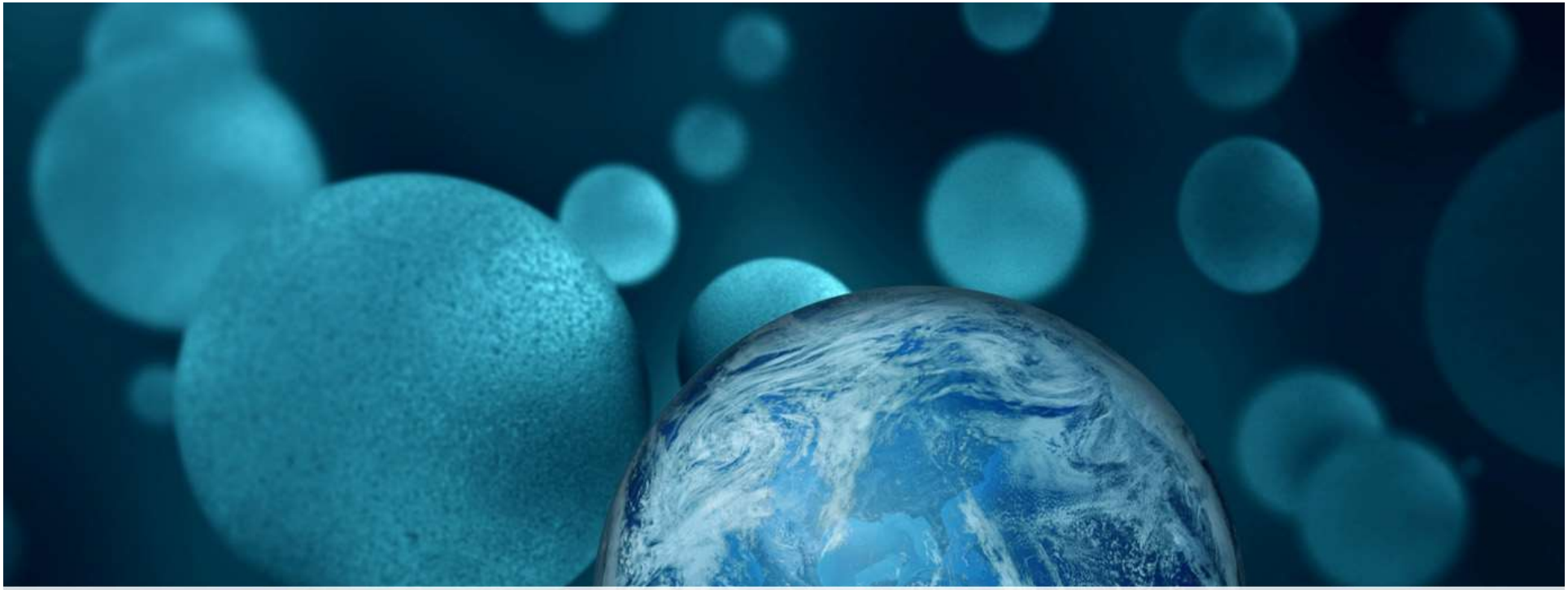

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Ion Reporter™ Software

Data Analysis Essentials

The world leader in serving science

Системы Ion GeneStudio S5 | Портфолио для решения широкого спектра задач

Ion GeneStudio™ S5	Ion GeneStudio™ S5 **Plus**	Ion GeneStudio™ S5 **Prime**
		
Быстрый.	**Гибкий.**	**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

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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 отДНК до результата | 2 стадии пипетирования / образец

Автоматизированная подготовка библиотек Ion AmpliSeq™

Добавить
Образец и пул праймеров в картридж для подготовки библиотек

Загрузить картридж
в систему Ion Chef

15 мин ручного труда

Автоматизированная подготовка матрицы

Добавить
Библиотеку в картридж для подготовки матрицы

Загрузить картридж
в систему Ion Chef

15 мин ручного труда

Секвенирование

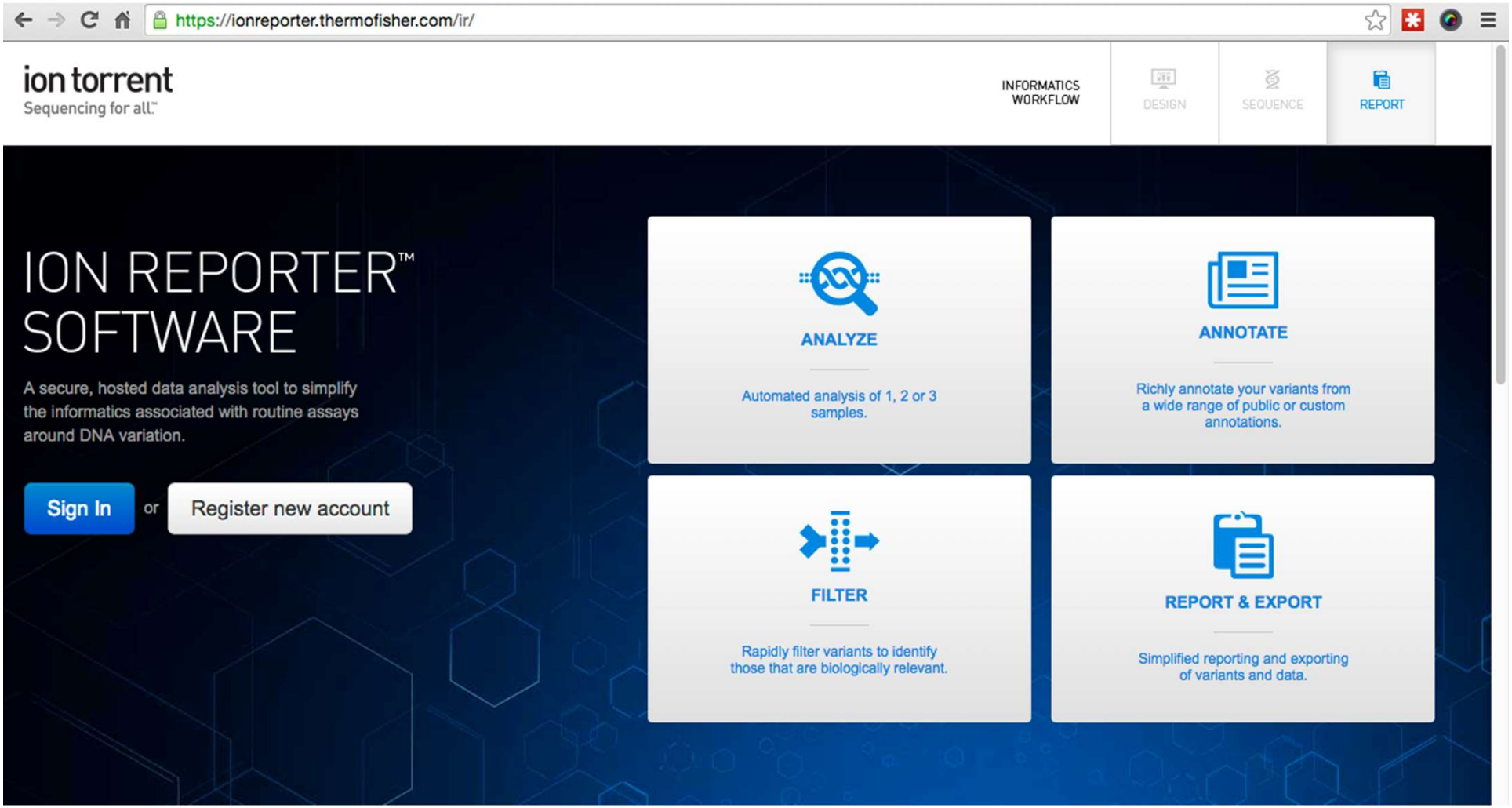
Загрузить реактивы
в систему Ion GeneStudio™ S5

Перенести чип
в систему Ion GeneStudio™ S5 для секвенирования

15 мин ручного труда

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

Ion Reporter Software



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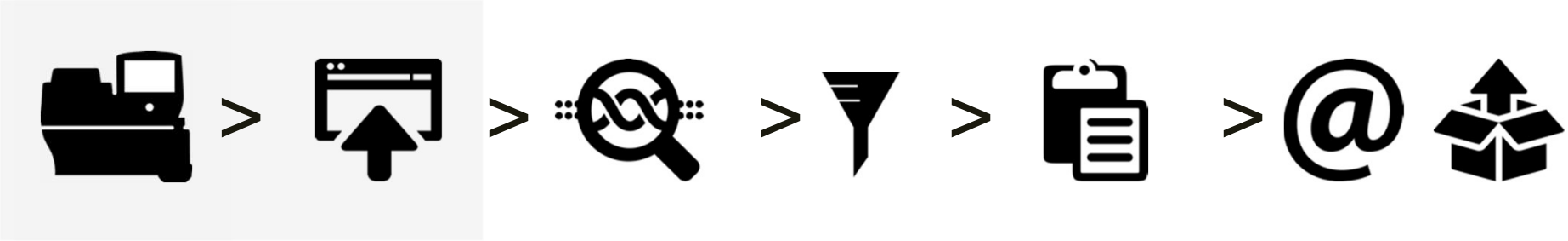
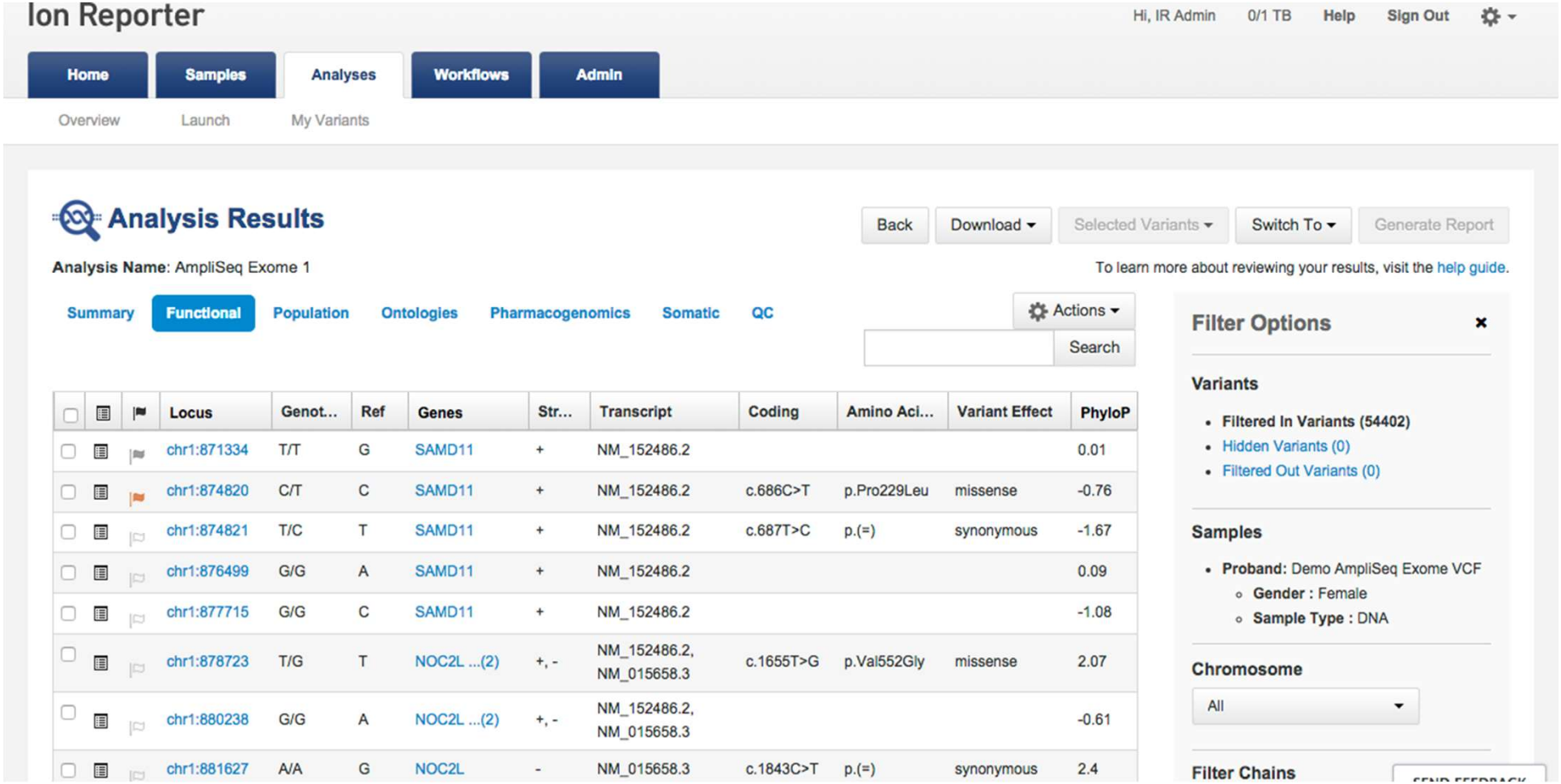
Agenda

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



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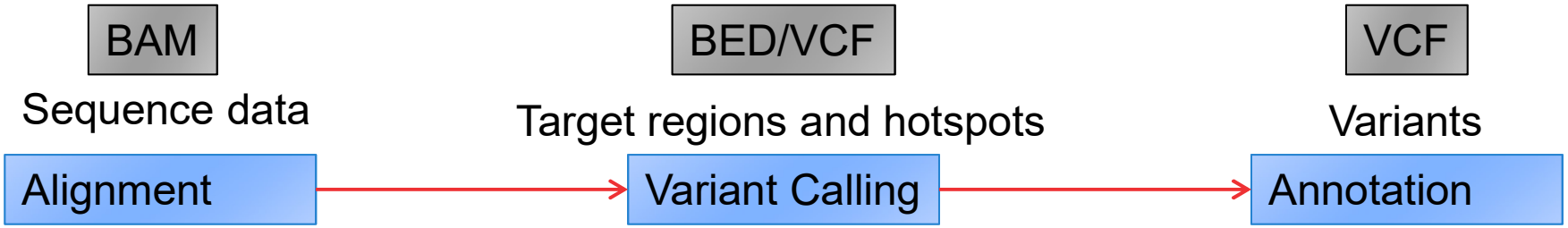
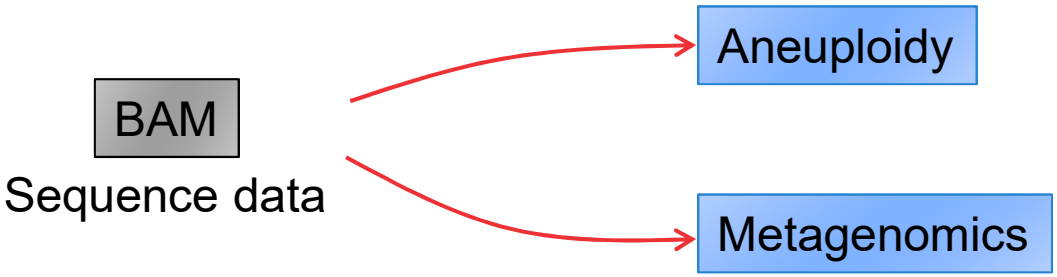
Ion Reporter™ software workflows

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

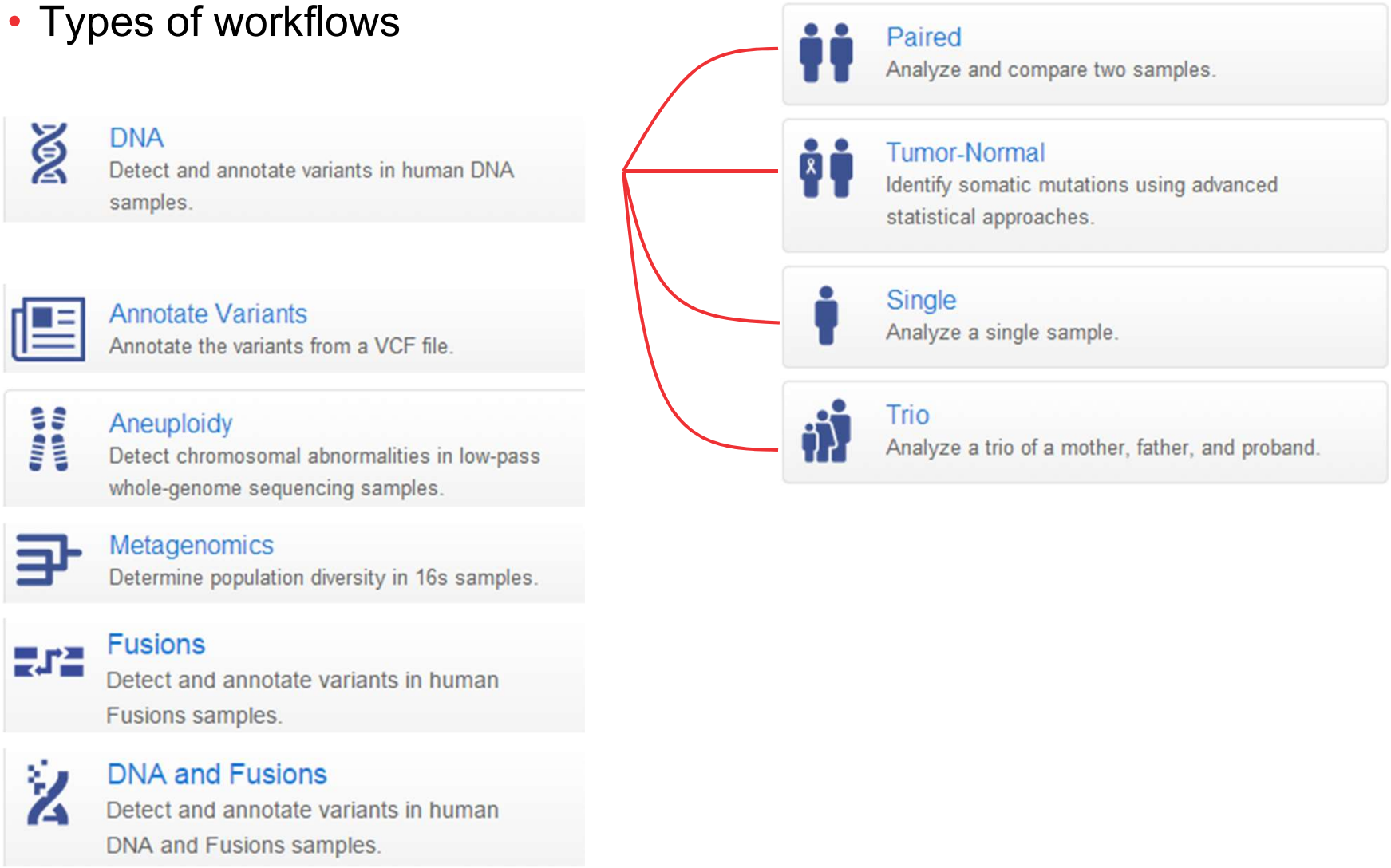


- Other workflows



Ion Reporter™ software workflows

- Types of workflows



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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

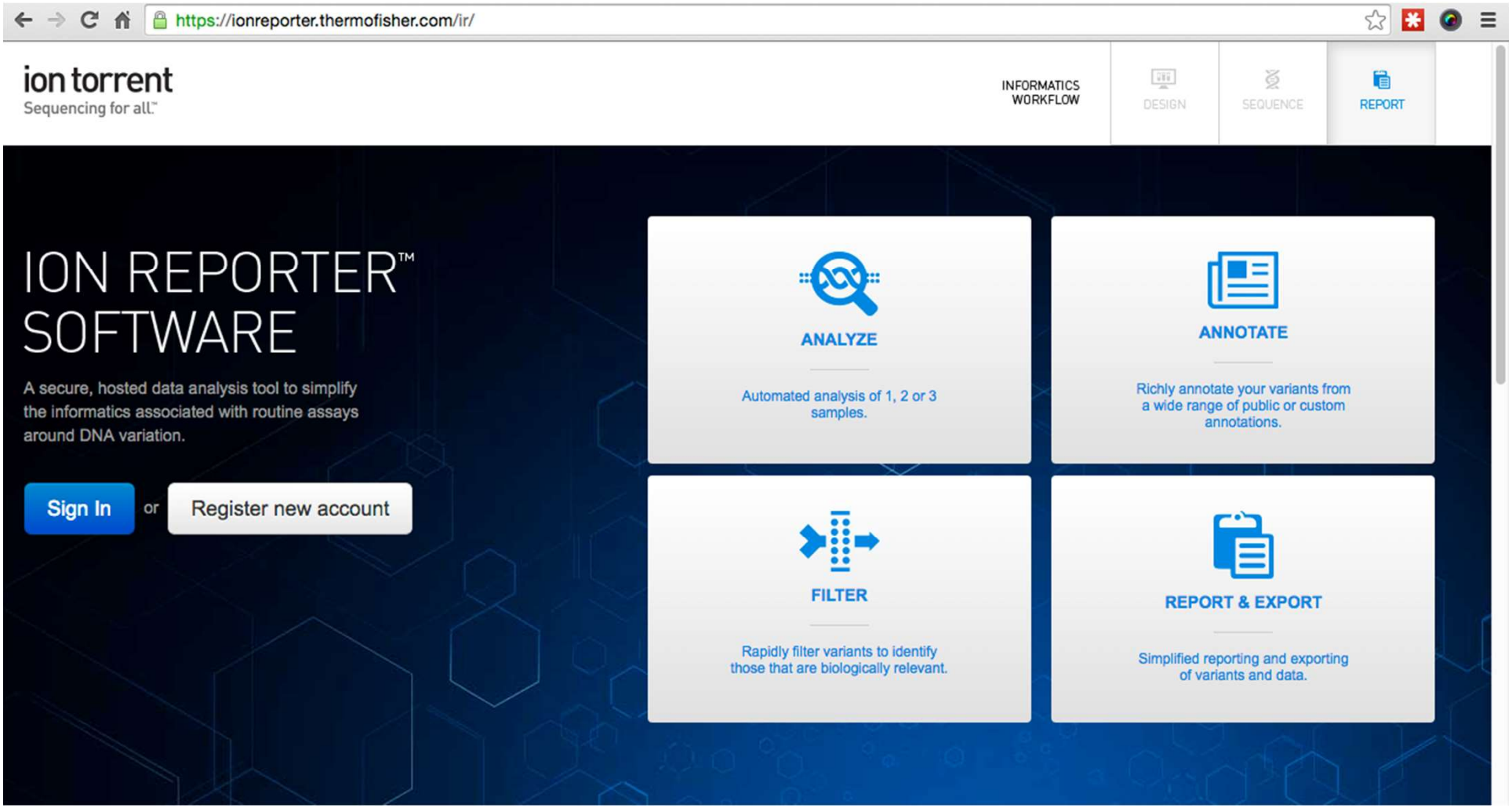
- Other annotations
  - Population allele frequencies, 5000 exome, 1000 genome projects
  - Genetic category – for variant inheritance in trio samples
  - HotSpot information
  - Custom annotations

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
  - Zygosity

Launch Analysis and Workflow Overview

Ion Reporter Software



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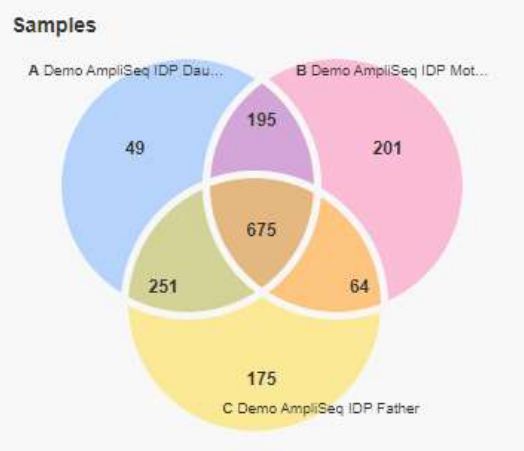
Launch Analysis – Hands on Practice

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



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

		Sample	Gender	Sample Type
☐		Demo AmpliSeq IDP Daughter	Female	DNA
☑		Demo AmpliSeq IDP Mother	Female	DNA
☑		Demo AmpliSeq IDP Father	Male	DNA

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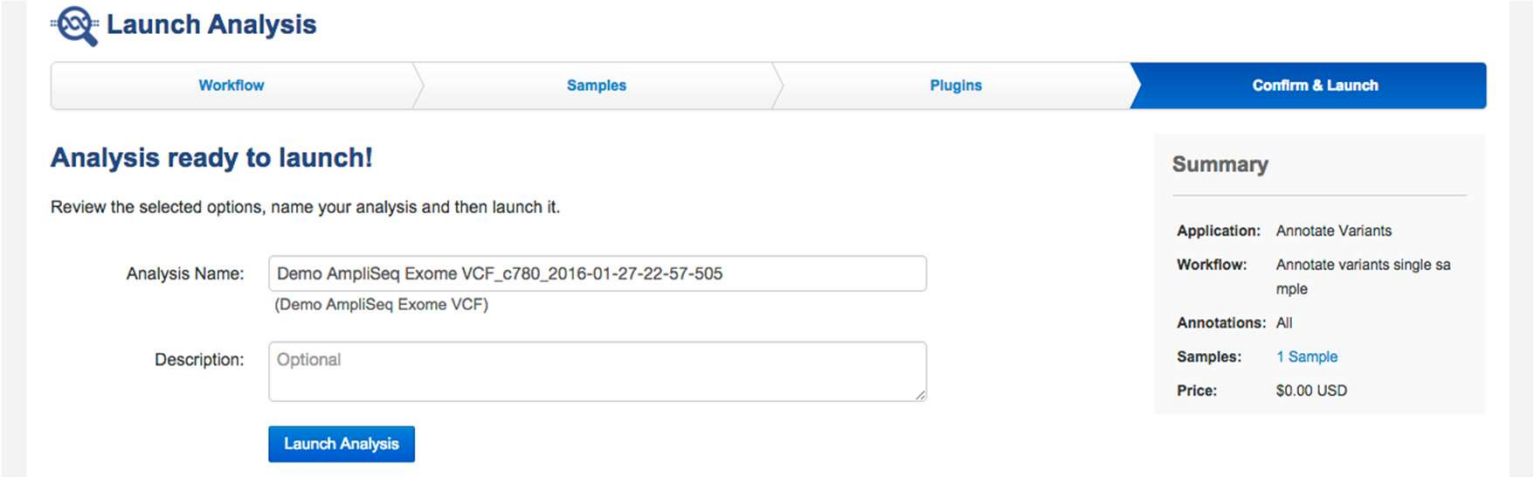
Launch Ion Reporter Analysis – Hands on Practice

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



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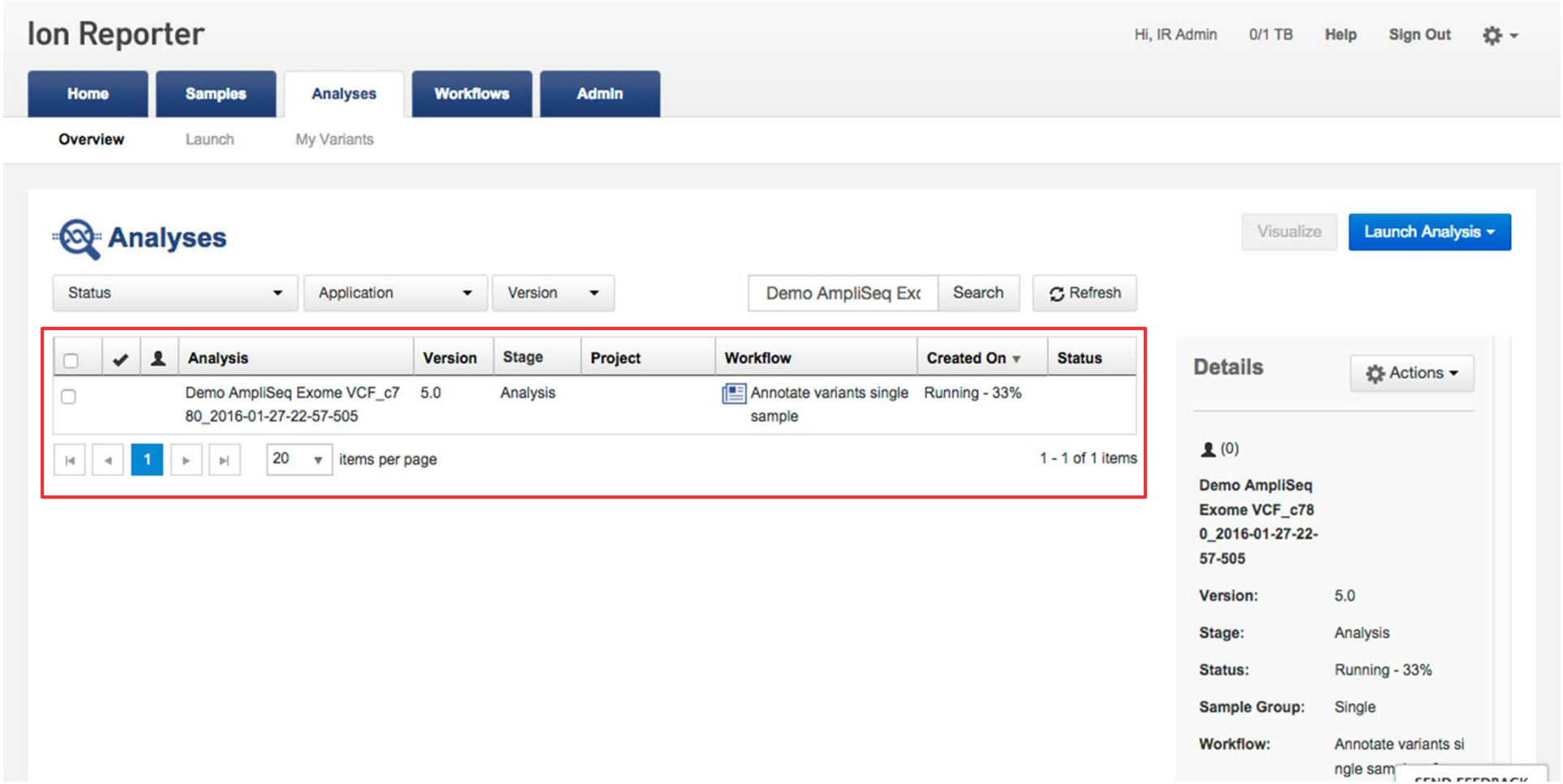
Launch Ion Reporter Analysis



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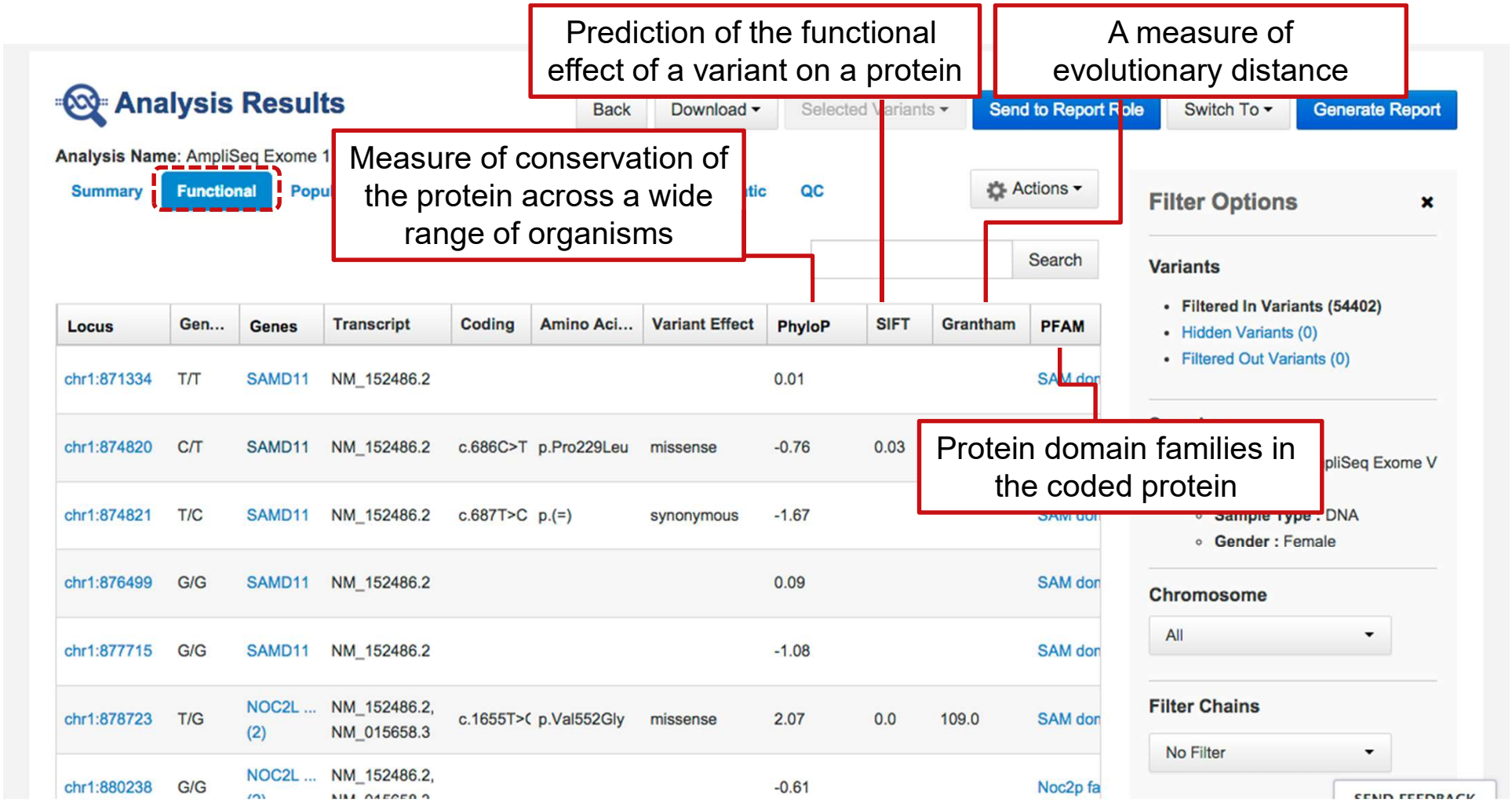
Variant Review, Filter, Classification, and Report

Analysis Results - Functional



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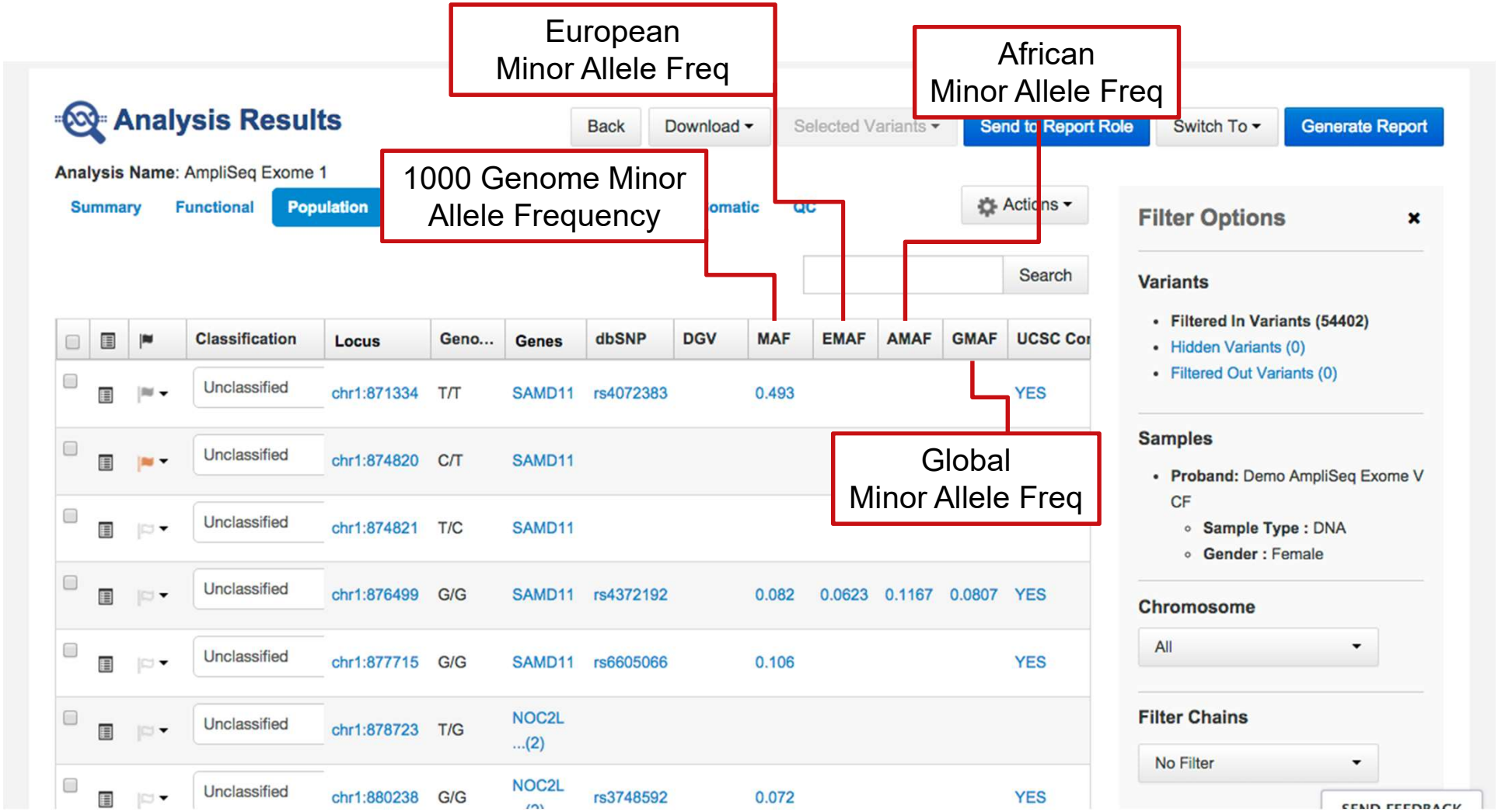
Analysis Results - Population



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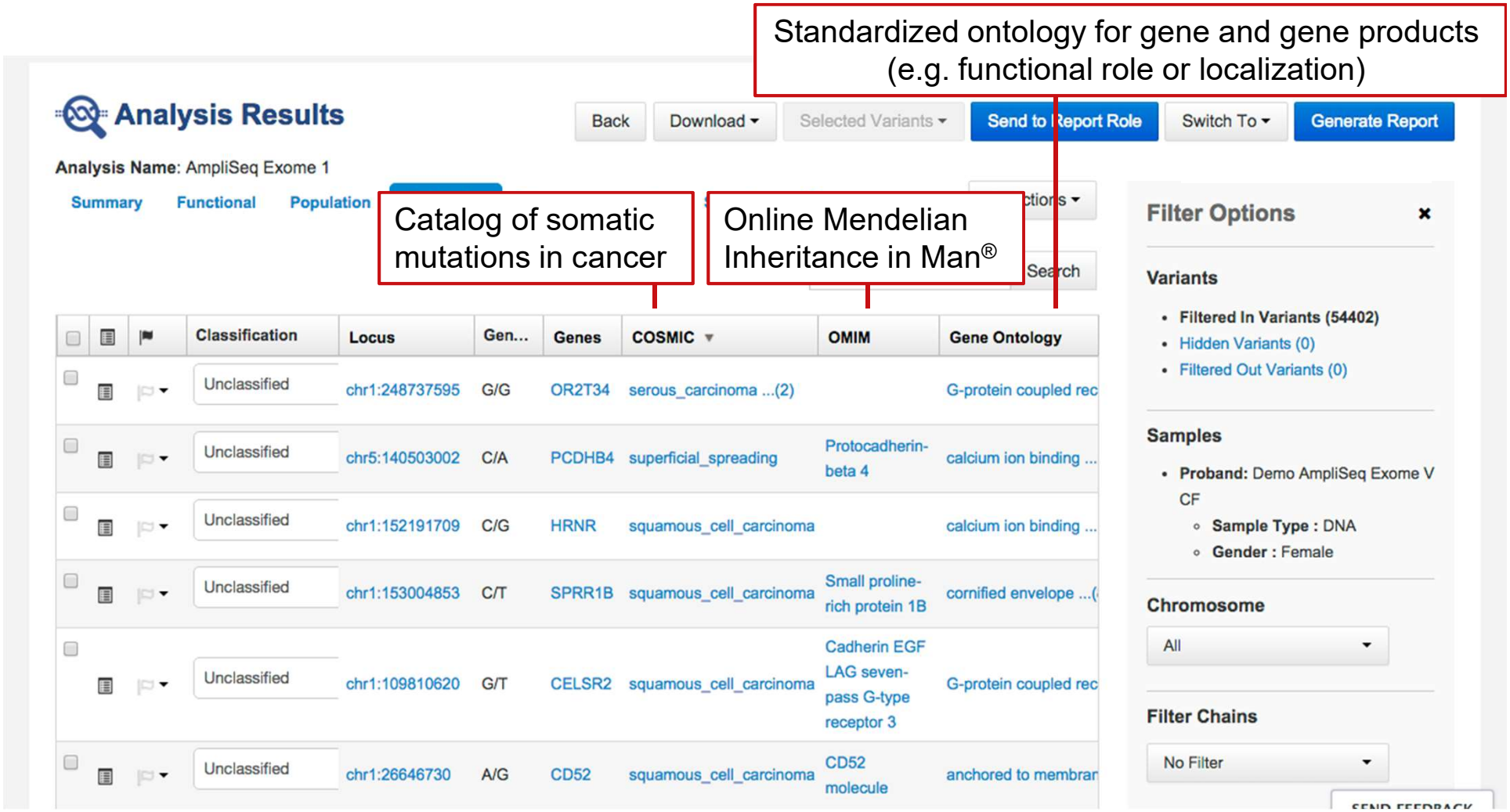
Analysis Results - Ontologies



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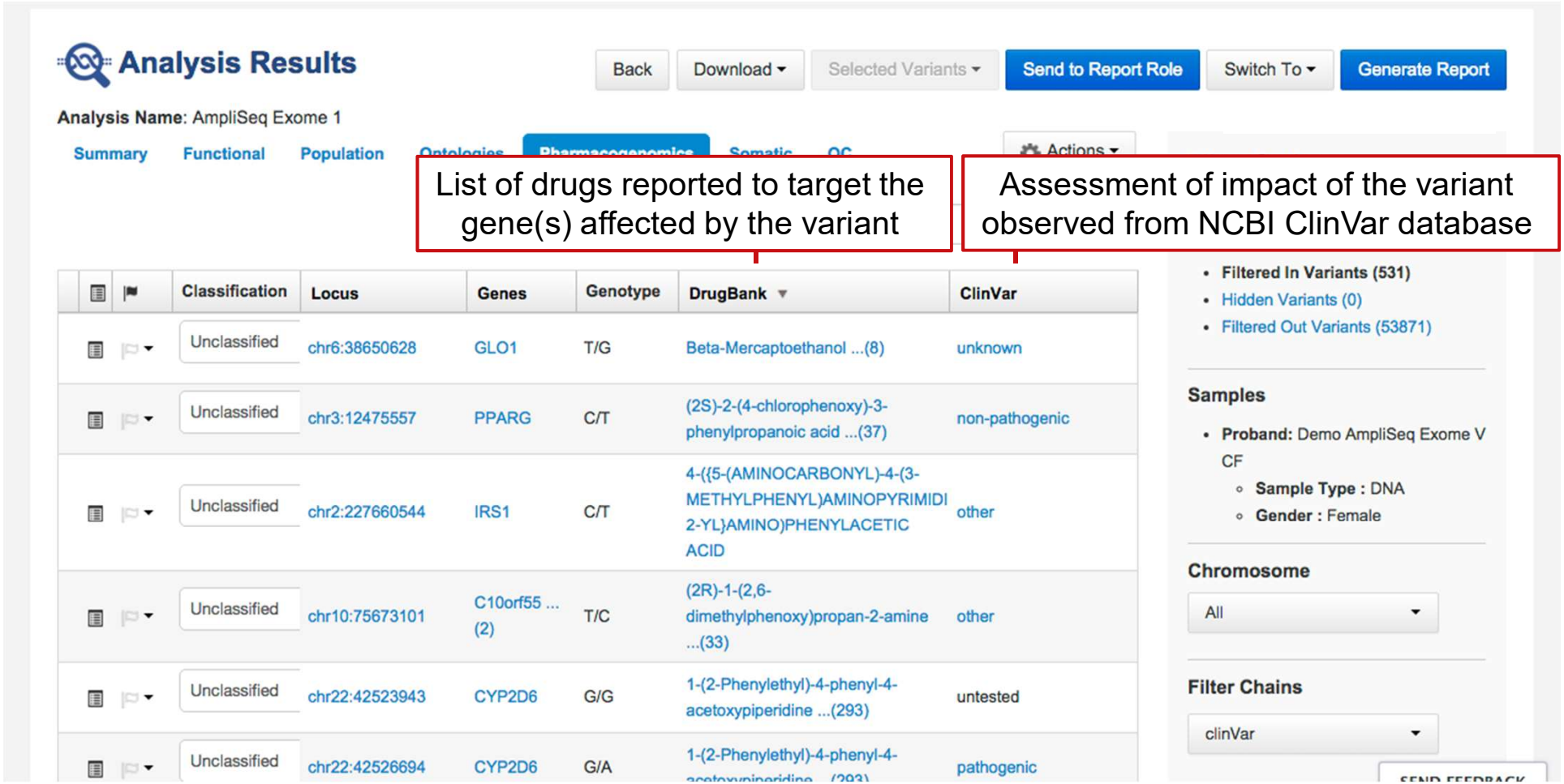
Analysis Results - Pharmacogenomics



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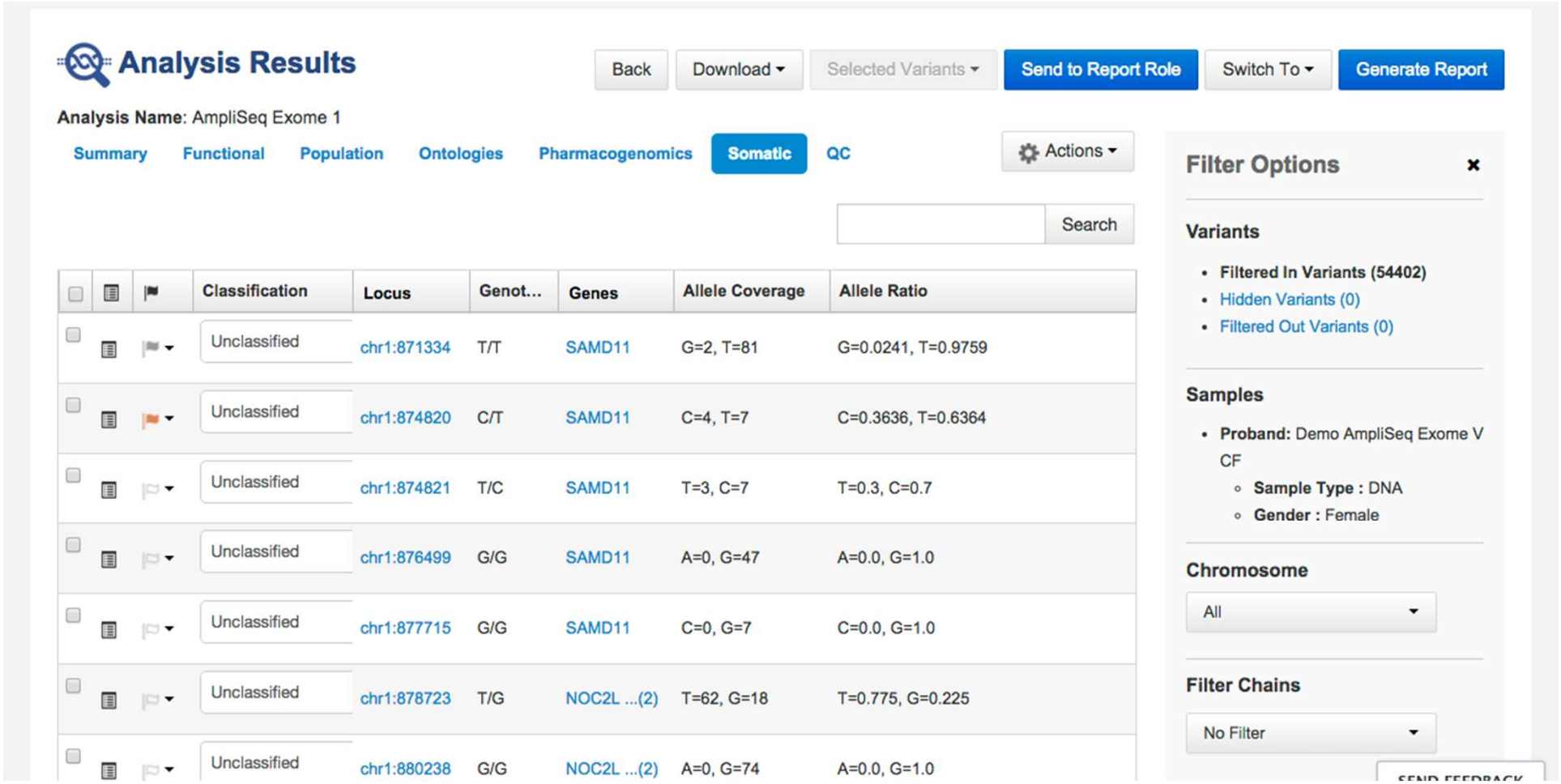
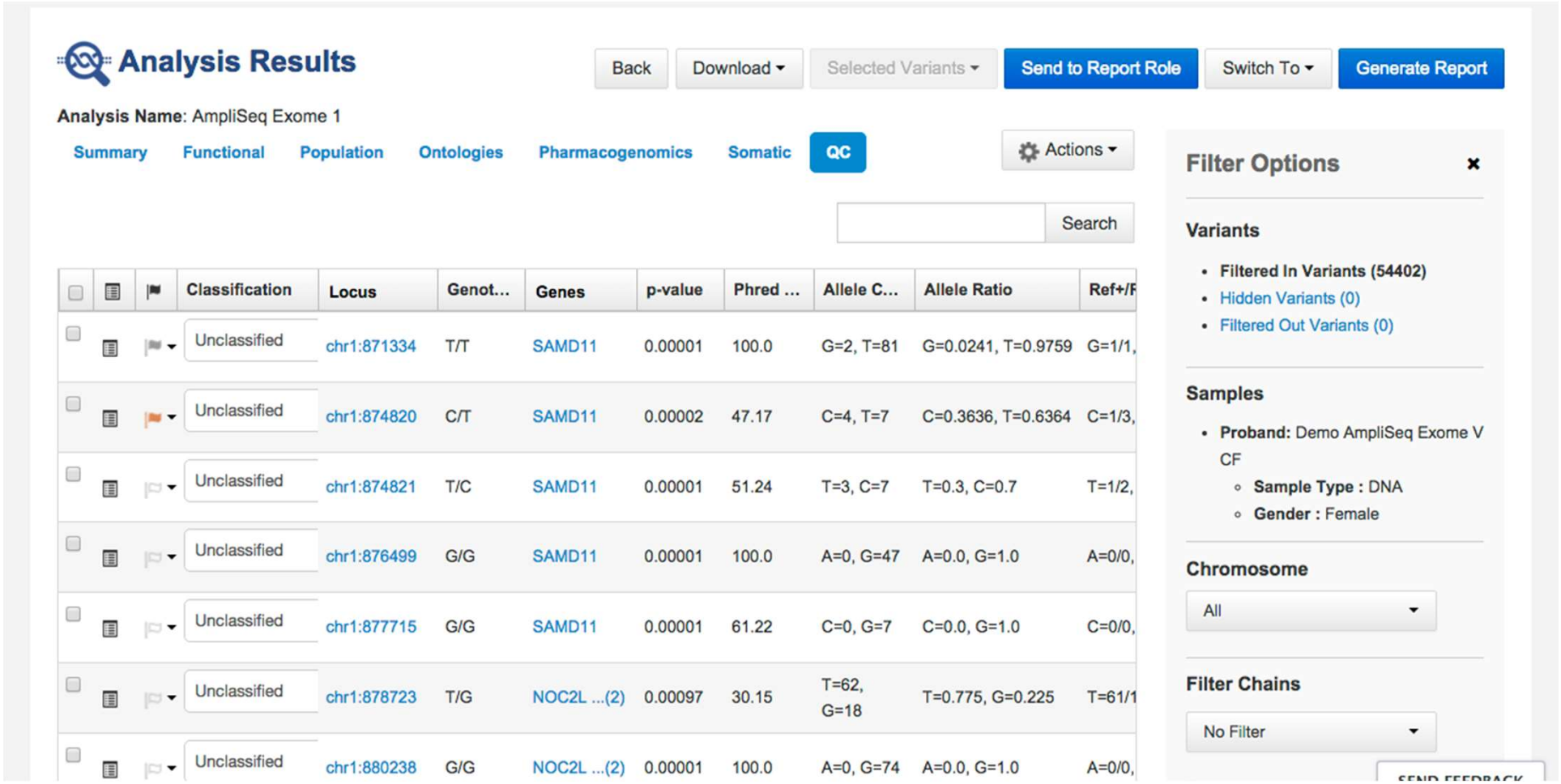
Analysis Results - Somatic



ThermoFisher SCIENTIFIC

Analysis Results - QC



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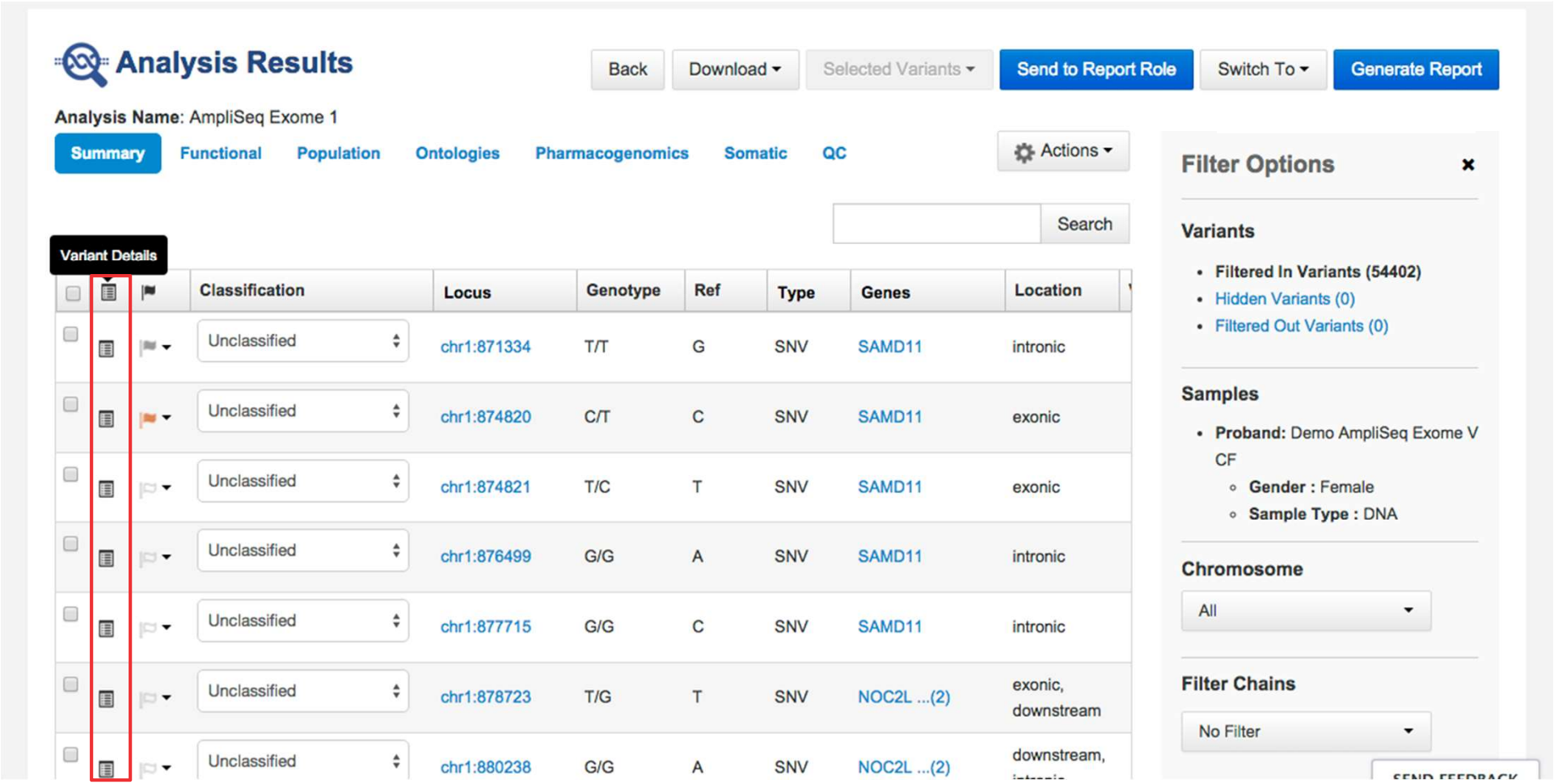
Analysis Results - Variant Details



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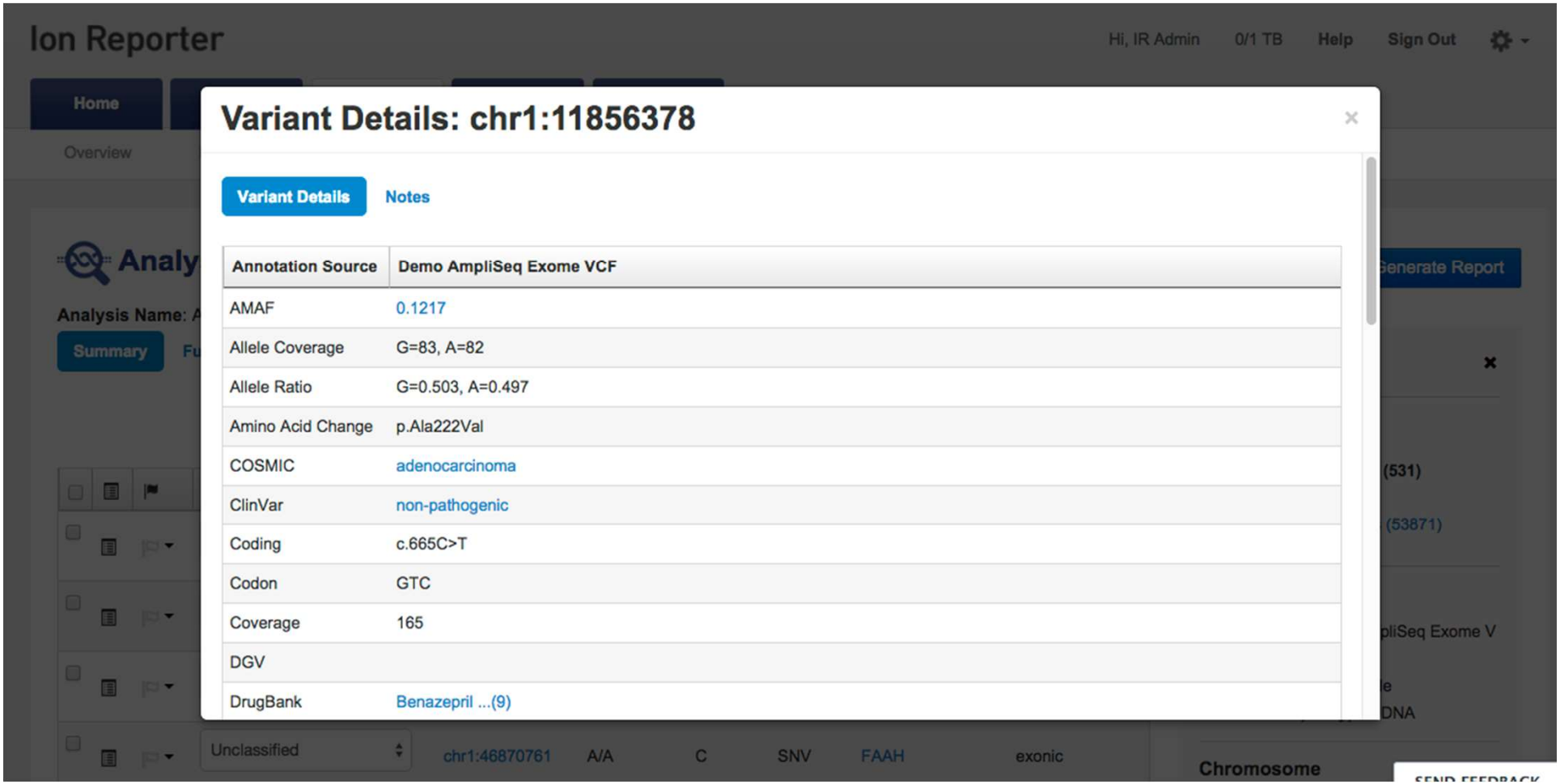
Analysis Results - Variant Details



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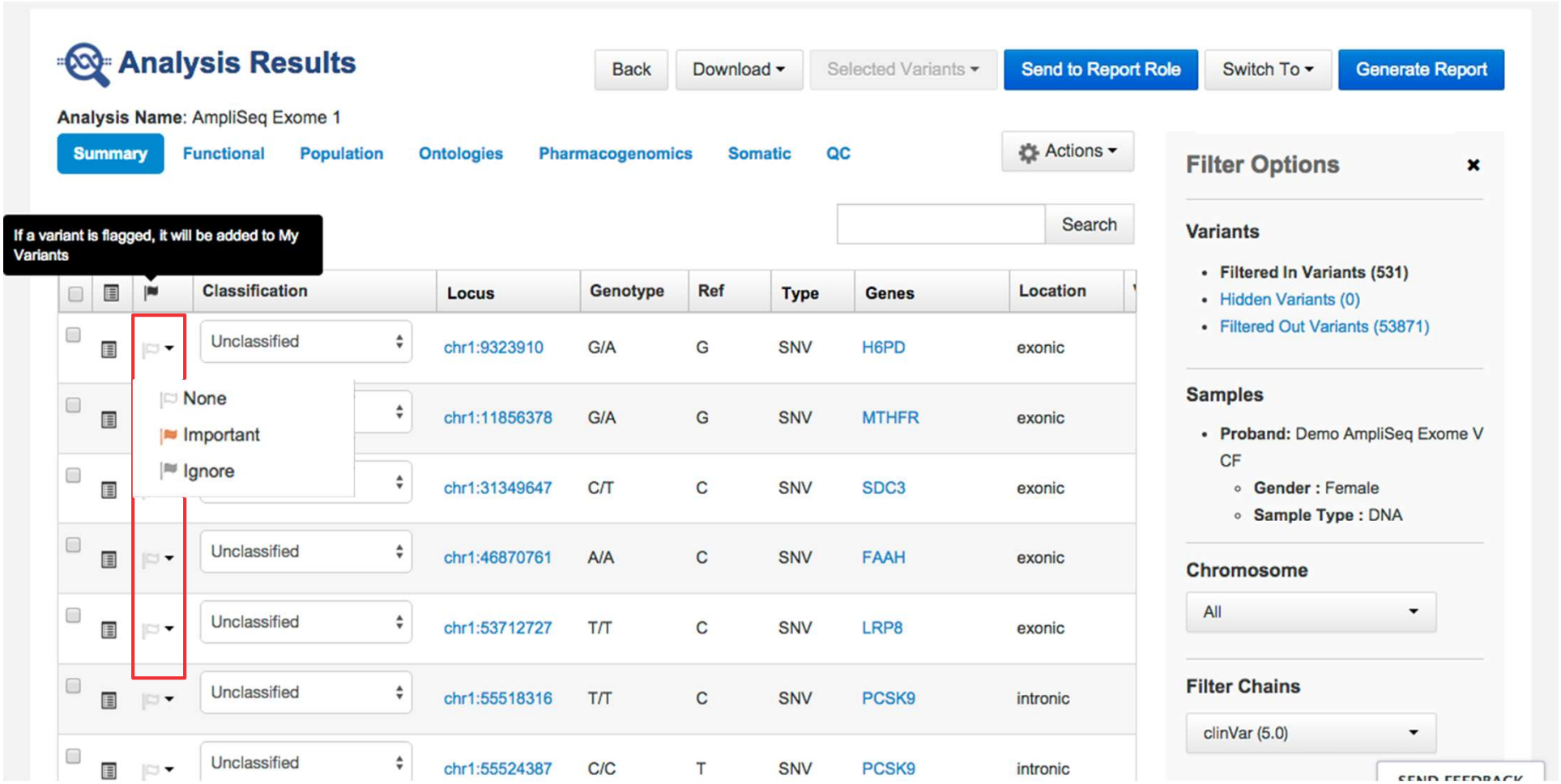
My Variants



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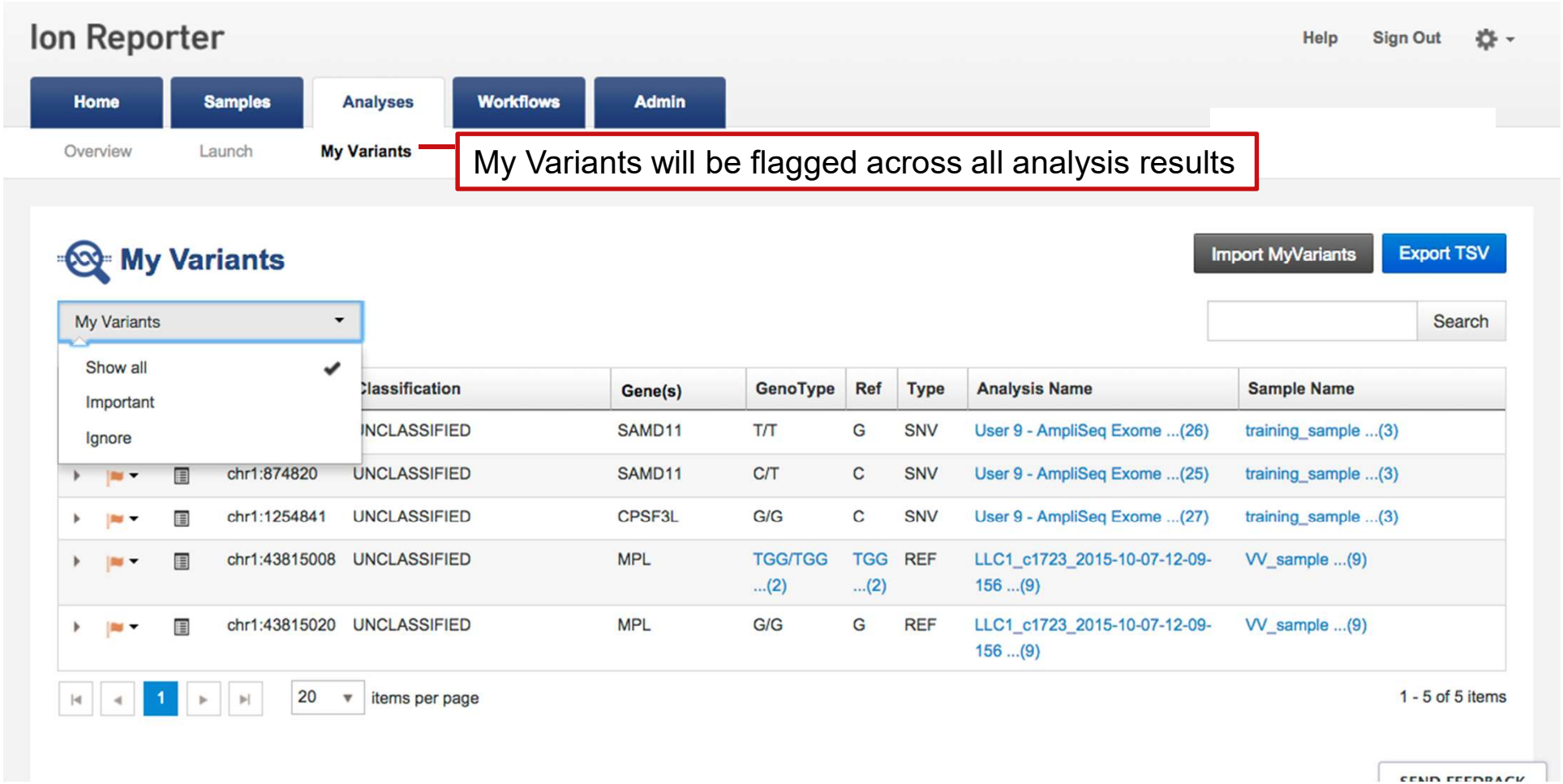
My Variants



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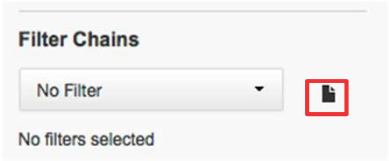
Filter Chain

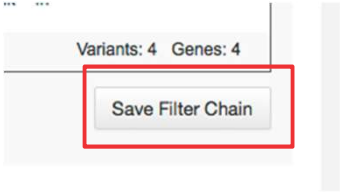
1. Click on New Filter Chain



   - 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

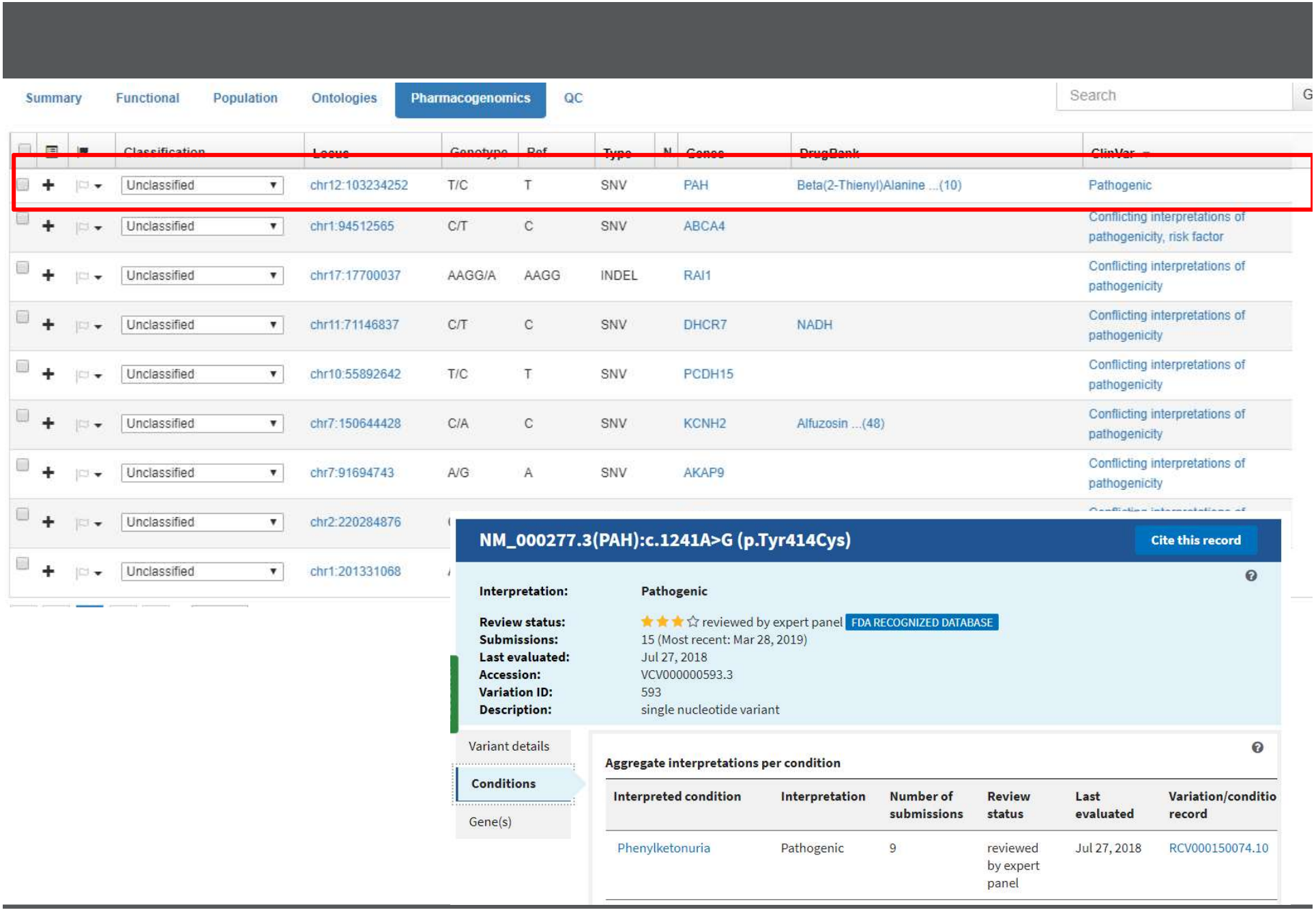
FilterChain Query

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



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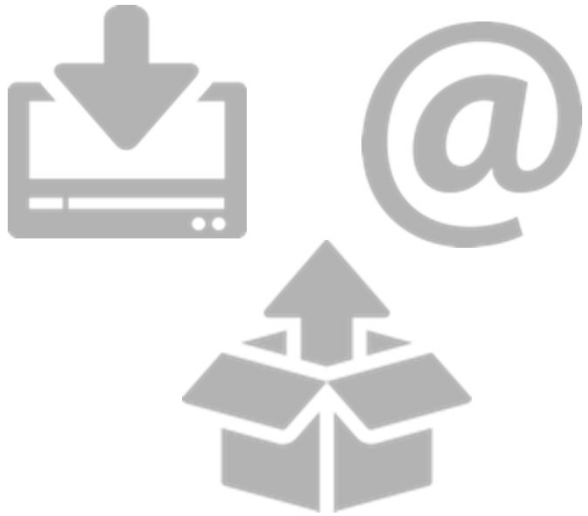
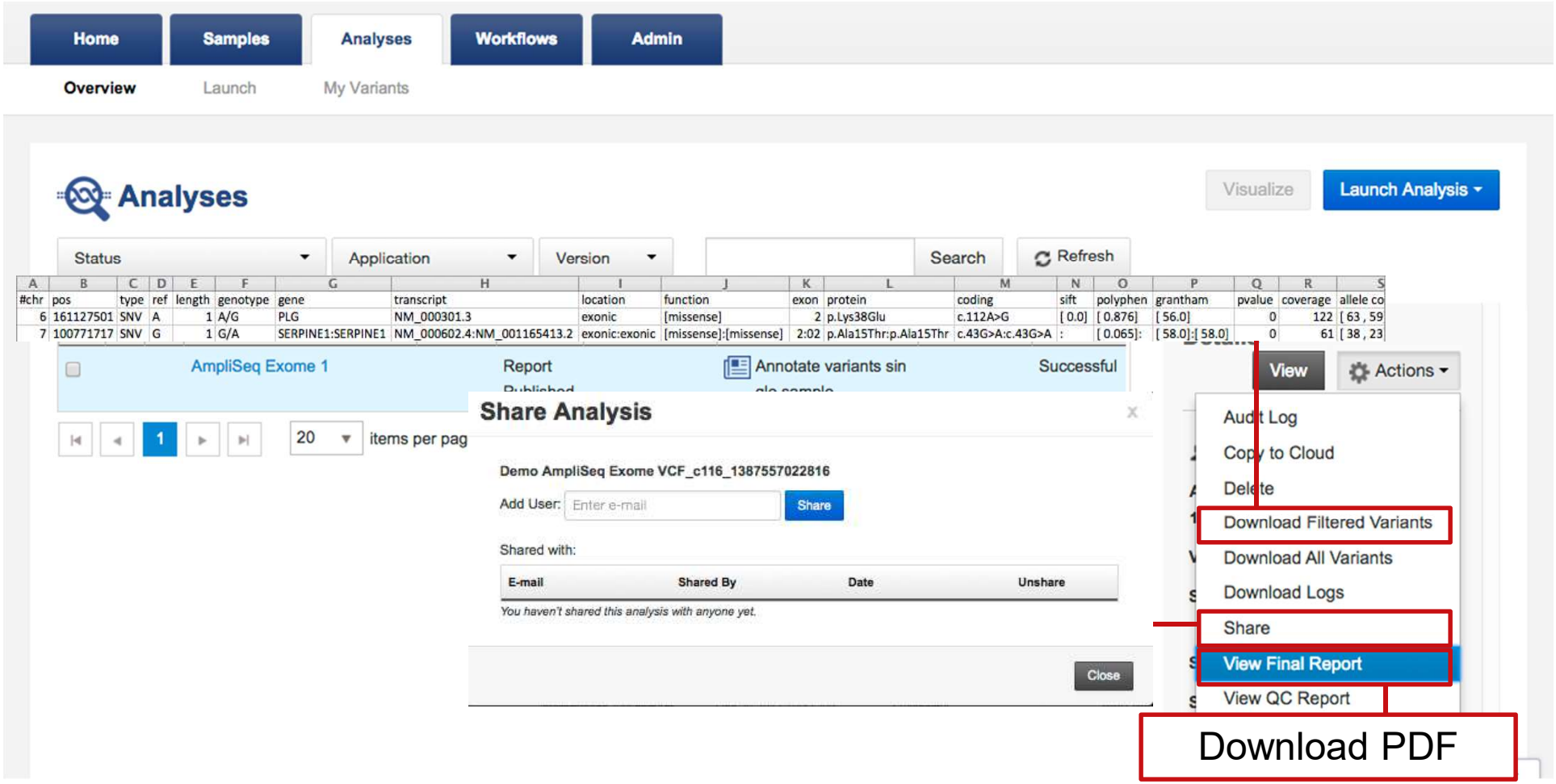
- По какой линии бабушка болела ФКУ?

Download, Export and Share



Download Report



#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

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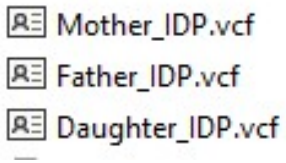
Семья с гипертрофической кардиомиопатией

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

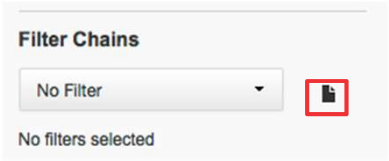
Мать, 45 лет - здорова

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



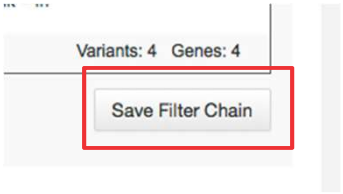
Filter Chain

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	Fibronectin type III domain containing
	I-set domain containing
	Myosin binding proteins
HCOP	Orthology Predictions for MYBPC3

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]