

Hands-on Workshop

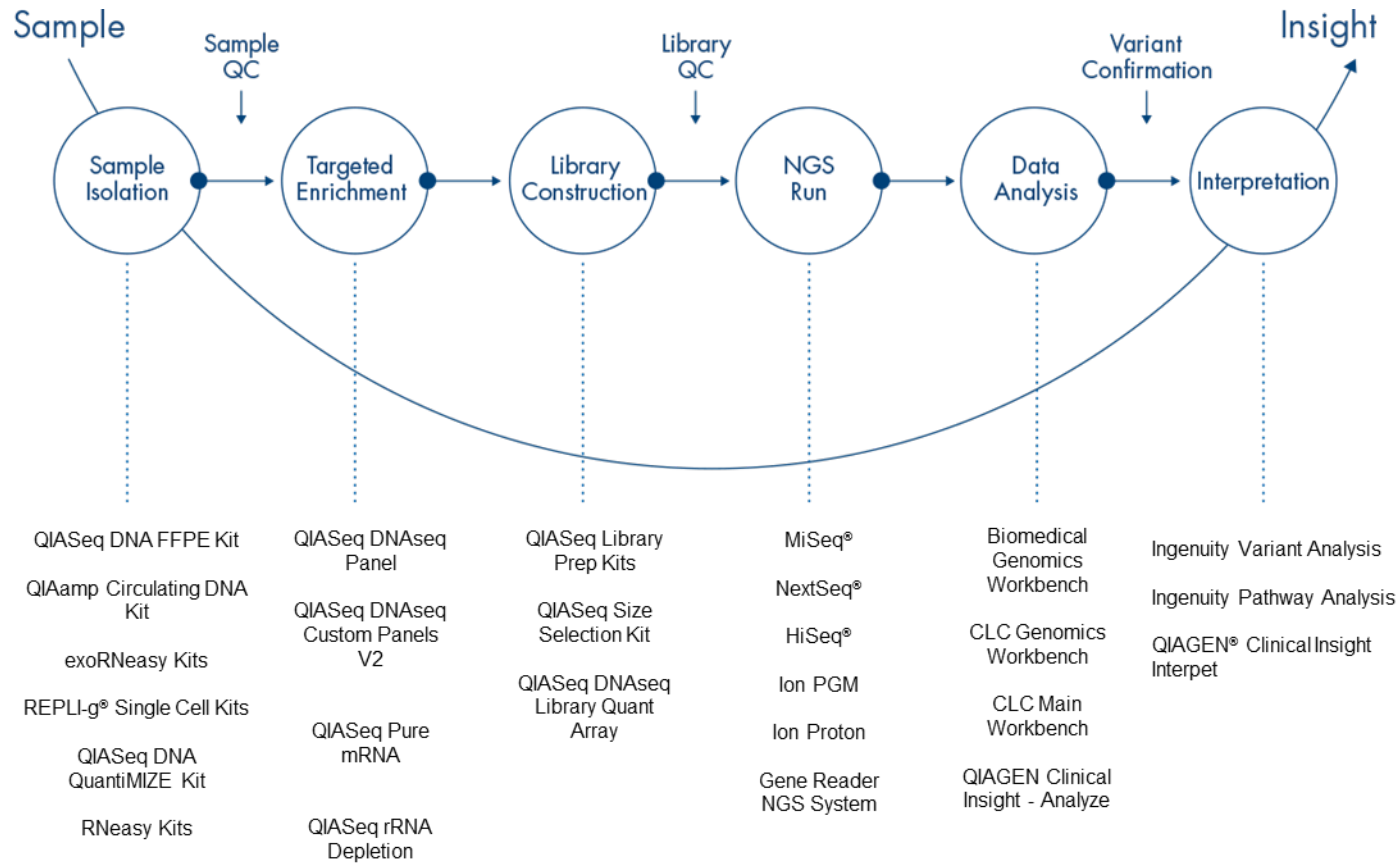
Clinical Genomics - Interpretation and Reporting with QIAGEN Bioinformatics

Ruth Burton PhD. – Clinical Applications Specialist (ruth.burton@qiagen.com)

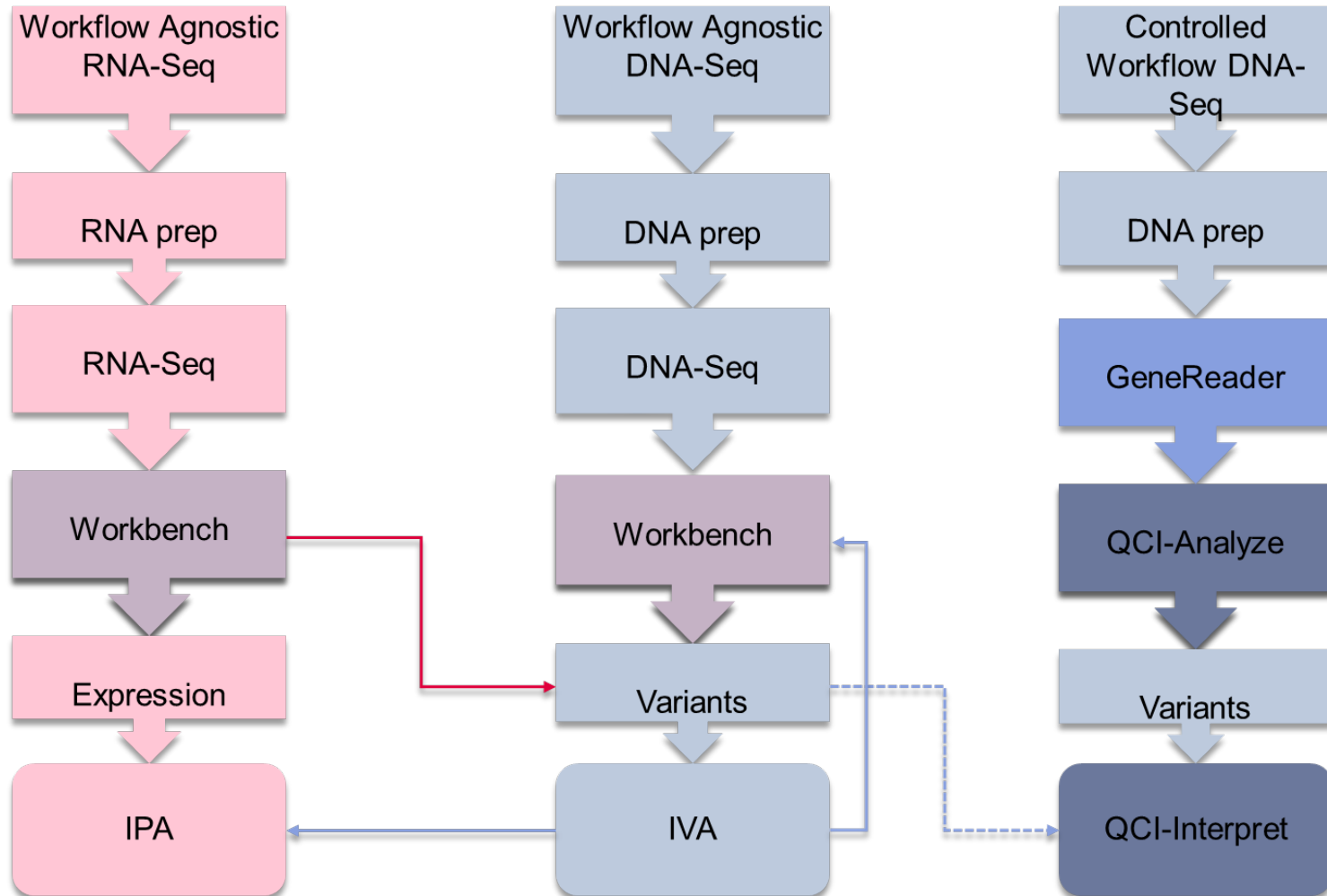
Agenda

- Introduction to the portfolio
- Getting started with IVA
- Filtering a whole exome sample
- QCI I demo
- Working with a trio
- Uploading and sharing samples
- Working with your own data

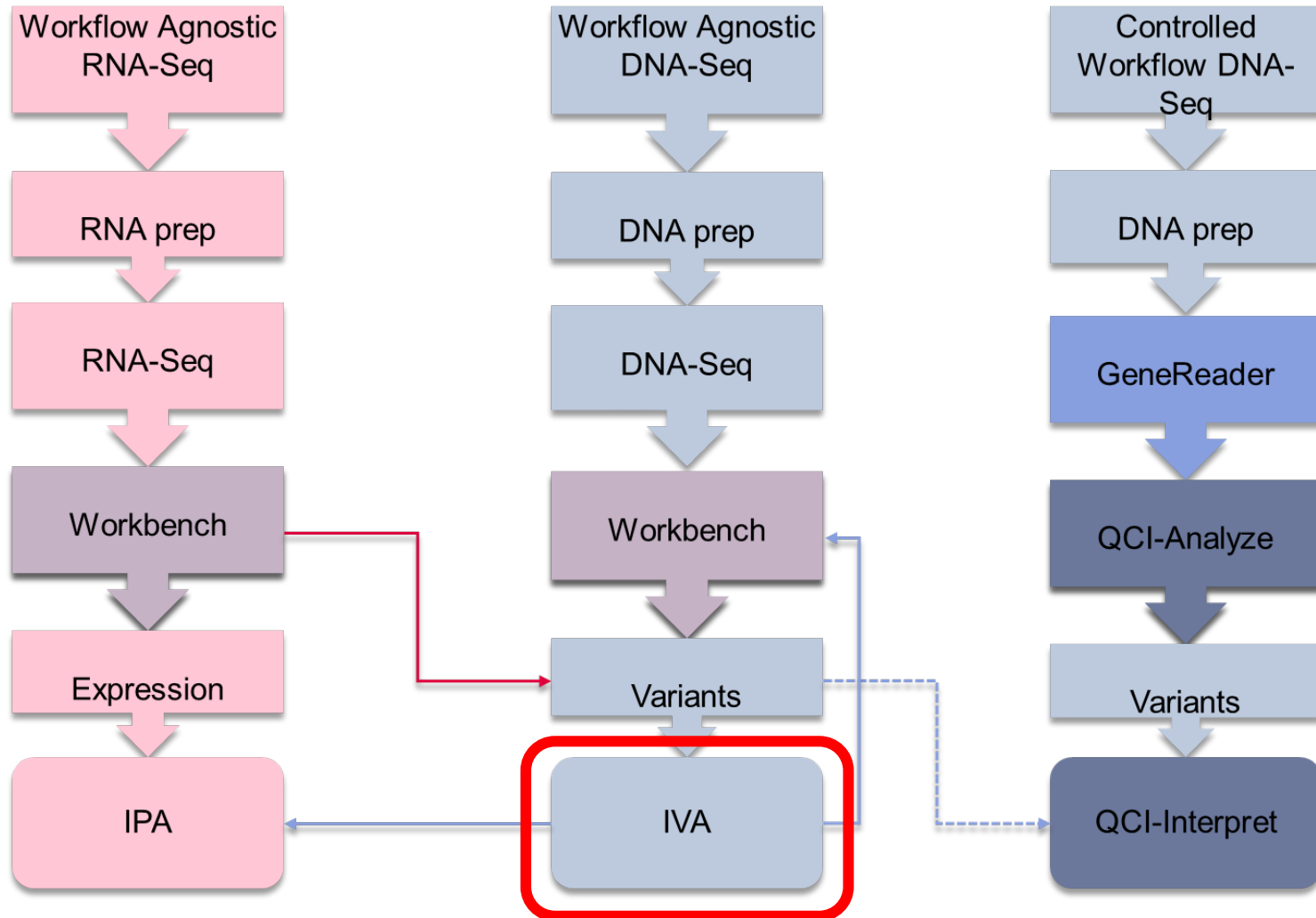
Introduction to the portfolio



Introduction to the portfolio



Introduction to the portfolio



Getting started with IVA

Log-on

<https://apps.ingenuity.com>

See your sheet for number

Login name:

variant_analysis1@ingenuity.com or vatraining1@ingenuity.com

Password: variant123

Logging on

Welcome! Please login

Email

Password

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Contact Customer Support

Customer Support
Phone: 650.381.5111
Hours: 6am - 5pm (PST)
Monday - Friday (excluding holidays)
AdvancedGenomicsSupport@qiagen.com

For Product and Sales related inquiries contact:
650.381.5056
bioinformaticssales@qiagen.com

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Welcome screen

What's new in build 4.4.20170525?

QIAGEN

BIOINFORMATICS POWERED BY INGENUITY  BIOBASE

Home > Products > Ingenuity Variant Analysis > Features

New features in the Spring 2017 Release

Improved Indel handling

Normalize indels in dbSNP to ensure that all short insertion/deletions are left-aligned, such that correctly formatted variants in user VCF data will be consistently annotated with the corresponding dbSNP record

Streamlined request to toggle pre-filtering

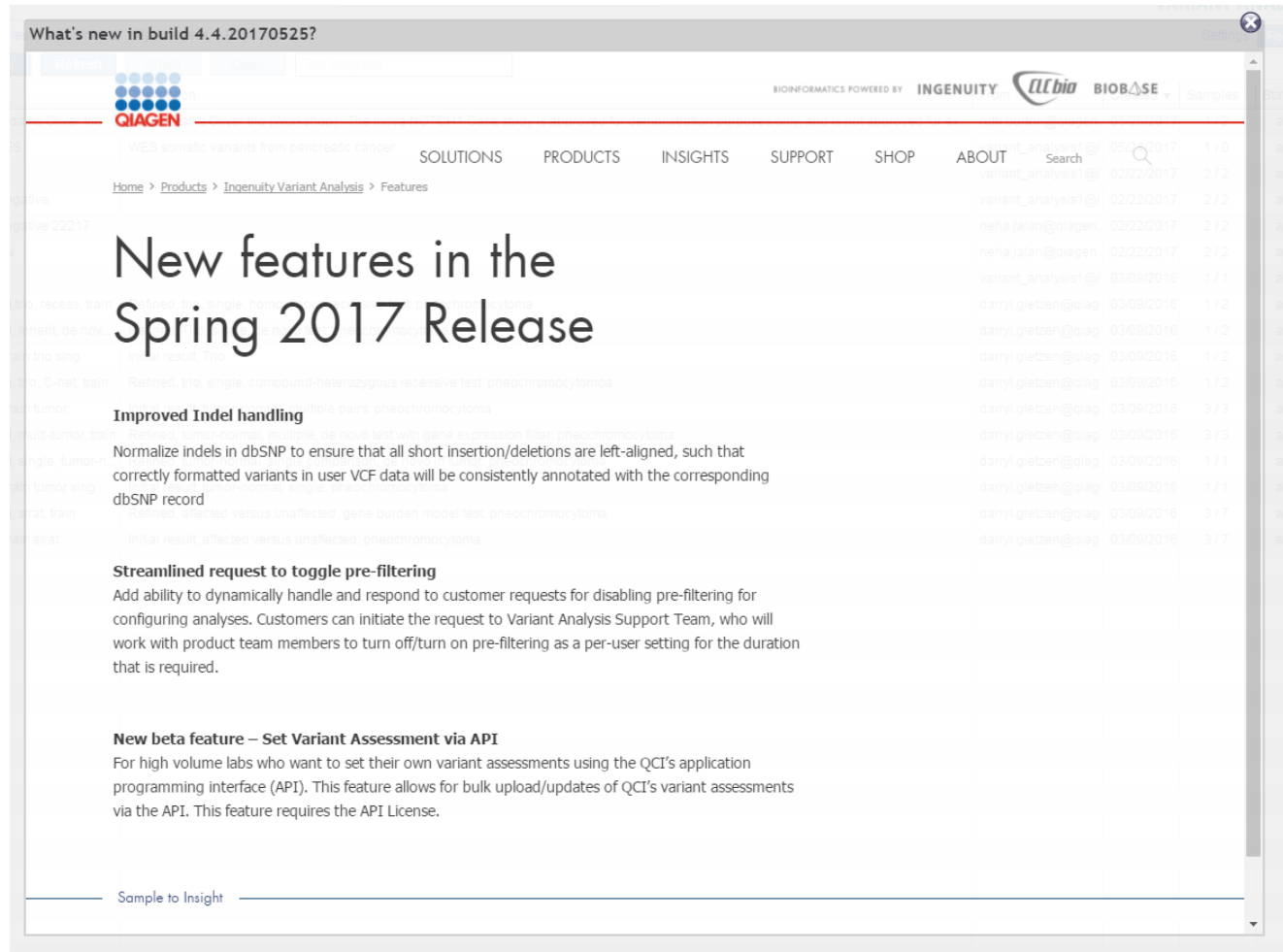
Add ability to dynamically handle and respond to customer requests for disabling pre-filtering for configuring analyses. Customers can initiate the request to Variant Analysis Support Team, who will work with product team members to turn off/turn on pre-filtering as a per-user setting for the duration that is required.

New beta feature – Set Variant Assessment via API

For high volume labs who want to set their own variant assessments using the QCI's application programming interface (API). This feature allows for bulk upload/updates of QCI's variant assessments via the API. This feature requires the API License.

Sample to Insight

Welcome screen, may see other info such as T&Cs or a sample is waiting to be imported...



What's new in build 4.4.20170525?

QIAGEN

BIOINFORMATICS POWERED BY INGENUITY *ILLUMINA* BIOBASE

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Sample to Insight

Find the demo sample, the default view is analysis – we need to find the sample

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Name	Description	From	Created	Samples	Status
Inova Adams Oliver trio	Inova Adams Oliver trio (Workshop). The Inova NOTCH1 Case study is a...	ruth.burton@qiagen.	05/25/2017	1 / 2	active
Test WES	WES somatic variants from pancreatic cancer	variant_analysis1@i	05/24/2017	1 / 0	active
hcc tn2		variant_analysis1@i	02/22/2017	2 / 2	active
triple negative		variant_analysis1@i	02/22/2017	2 / 2	active
triple negative 22217		neha.jalan@qiagen.	02/22/2017	2 / 2	active
HCC TN		neha.jalan@qiagen.	02/22/2017	2 / 2	active
Tutorial		variant_analysis1@i	03/09/2016	1 / 1	active
Refined, trio, recess, train	Refined, trio, single, homozygous recessive test: pheochromocytoma	darryl.gietzen@qiag	03/09/2016	1 / 2	active
Refined, Inherit, de nov...	Refined, Trio, single, de novo test: pheochromocytoma	darryl.gietzen@qiag	03/09/2016	1 / 2	active
Initial Train trio sing	Initial result, Trio	darryl.gietzen@qiag	03/09/2016	1 / 2	active
Refined, trio, C-het, train	Refined, trio, single, compound-heterozygous recessive test: pheochrom...	darryl.gietzen@qiag	03/09/2016	1 / 2	active
Initial Train tumor	Initial result, tumor-normal, multiple pairs: pheochromocytoma	darryl.gietzen@qiag	03/09/2016	3 / 3	active
Refined, multi-tumor, train	Refined, tumor-normal, multiple, de novo test with gene expression filter: ...	darryl.gietzen@qiag	03/09/2016	3 / 3	active
Refined, single, tumor-n...	Refined, tumor-normal, single comparison, de novo in tumor: pheochrom...	darryl.gietzen@qiag	03/09/2016	1 / 1	active
Initial Train tumor sing	Initial result, tumor-normal, single: pheochromocytoma	darryl.gietzen@qiag	03/09/2016	1 / 1	active
Refined, strat, train	Refined, affected versus unaffected, gene burden model test: pheochrom...	darryl.gietzen@qiag	03/09/2016	3 / 7	active
Initial Train strat	Initial result, affected versus unaffected: pheochromocytoma	darryl.gietzen@qiag	03/09/2016	3 / 7	active

Status: active

Name Test WES
Description WES somatic variants from pancreatic cancer
Created 05/24/2017 04:48 PM
From variant_analysis1@ingenuity.com
Genome GRCh37/HG19
Variant Count 2250
Filters
Confidence
Common Variants
Predicted Deleterious
Genetic Analysis
Cancer Driver Variants
Biological Context
Fields
Samples TCRBOA2-T-WEX Somatic Variants (case)

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Change to samples and search for “WEX”

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Showing 1 samples

ID	Barcode	Display Name	Description	Subject ID	From	Uploaded	Status
7837906	TCRBOA2-T-	TCRBOA2-T-WEX Som...	TCRBOA2-T-WEX Somatic Variants for demo		ruth.burton@qi	05/23/2017 04:	active

Highlight sample and click “Analyze”

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Showing 1 samples

ID	Barcode	Display Name	Description	Subject ID	From	Uploaded	Status
7837906	TCRBOA2-T-	TCRBOA2-T-WEX Som...	TCRBOA2-T-WEX Somatic Variants for d...		ruth.burton@qi	05/23/2017 04:	active

Status: active

Barcode TCRBOA2-T-WEX.read1 (paired) Read Ma

Display Name TCRBOA2-T-WEX Somatic Variants

Subject ID

Description TCRBOA2-T-WEX Somatic Variants for dem

Uploaded 05/23/2017 04:40 PM

From ruth.burton@qiagen.com

Genome GRCh37/HG19

Inferred Sex

Format vcf

Variant Count 2250
Ti/Tv Ratio: 0.9

Files TCRBOA2-T-WEX Somatic Variants.vcf
samplemetadata.txt

Sample ID 7837906

Type gene panel

ING dataPackageId DP_1879975951099517760290

[Edit](#) [Delete](#) [AFC Export](#)

A series of options follows, guiding you through the analysis

1 Cases and Controls
2 Focus the Analysis
3 Sample-specific options
4 Analyze

Select and drag samples to designate case/control status.
Drag samples from the left table to the desired list on the right. Drag to reorder samples. Their order here determines their order within the analysis views.

Load from prior analysis

Search samples by keyword

Name	Subject	Created	
103-00001-01	103-00001-01	05/25/17 09:58 AM	▲
103-00001-02	103-00001-02	05/25/17 09:58 AM	
103-00001-03	103-00001-03	05/25/17 09:58 AM	
TNBC4_small_Annotated_Va...	TNBC4_small Annotat...	02/22/17 04:34 PM	
23N_R1_001_sampled_Ann...	23N_R1_001 sampled...	02/22/17 04:34 PM	
23T_R1_001_sampled_Ann...	23T_R1_001 sampled...	02/22/17 04:34 PM	
HER2-2_small_Annotated_V...	HER2-2_small Annota...	02/22/17 04:34 PM	
HER2-1_small_Annotated_V...	HER2-1_small Annota...	02/22/17 04:20 PM	
TNBC3_small_Annotated_V...	TNBC3_small Annotat...	02/22/17 04:20 PM	
TNBC1_small_Annotated_V...	TNBC1_small Annotat...	02/22/17 04:20 PM	
HER2-3_small_Annotated_V...	HER2-3_small Annota...	02/22/17 04:20 PM	
HCC1187_Normal	HCC1187	02/22/17 04:20 PM	
HCC2218_Normal	HCC2218	02/22/17 04:20 PM	
HCC2218_Tumor	HCC2218	02/22/17 04:20 PM	
HCC1187_Tumor	HCC1187	02/22/17 04:20 PM	▼

1 Case (affected, tumor, responder, etc.)

TCRBOA2-TWEX Somatic Variants

0 Controls (unaffected, normal, nonresponder, etc.)

Use a library

Back
Next

A series of options follows, guiding you through the analysis

1 Cases and Controls
2 Focus the Analysis
3 Sample-specific options
4 Analyze

Please select the type of analysis you would like to start with.
This will set your starting filters to best practices. You can always change these settings later. If multiple options apply, start with one and start another analysis with a different selection later.

*** Analysis Design**

☐ Genetic disease
 Identify causal variants using trios, family analysis or case vs control models.

☒ Cancer
 Focus on somatic variants

☐ Stratification study Requires at least 2 cases and 2 controls
 Typically used to study complex disorders and with large numbers of samples (at least 50 recommended). Compare cases and controls statistically

☐ Other
 Use the most general filter settings.

☐ Settings from a previous analysis

Back
Next

A series of options follows, guiding you through the analysis

1 Cases and Controls
2 Focus the Analysis
3 Sample-specific options
4 Analyze

Select the cancer model for your analysis.
Optionally, provide biological terms so that you can easily find relevant literature and citations. Adding these terms will not constrain your results. You can always view the variants that are not annotated biologically as well as those that are.

This dataset concerns: Cancer

* Which cancer model is most applicable?

any cancer

Which biological terms describe this disease?

Enter relevant phenotypes, pathways, processes, or domains

pancreatic can

x pancreatic cancer [disease]

Back
Next

A series of options follows, guiding you through the analysis

1 Cases and Controls

2 Focus the Analysis

3 Sample-specific options

4 Analyze

✕

Your analysis is ready to begin
Some analysis take time to run. You will receive an email when the analysis is complete.

Summary
1 case sample(s)
0 control sample(s)
0 custom annotation(s)

Name your analysis

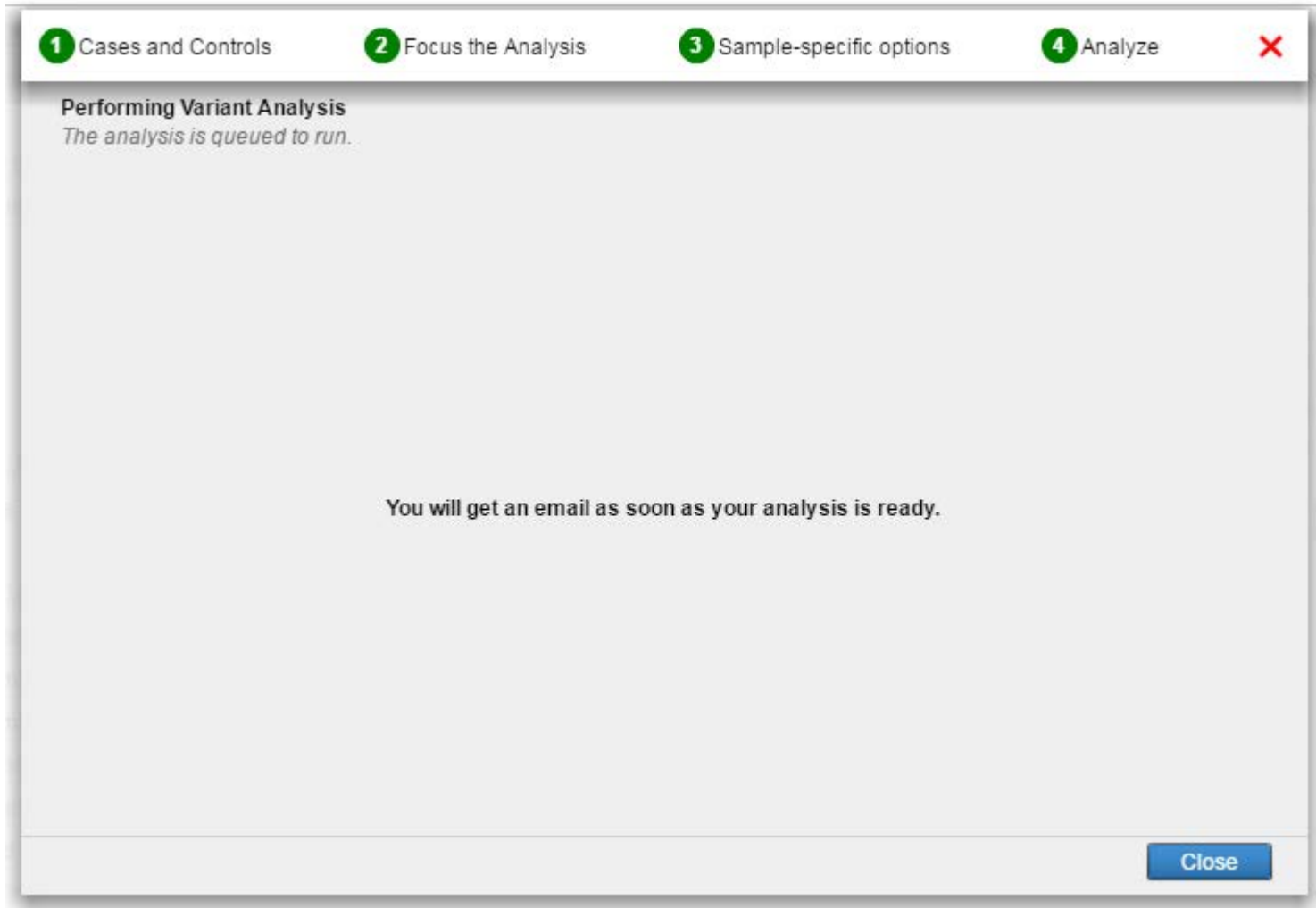
* Name

Description

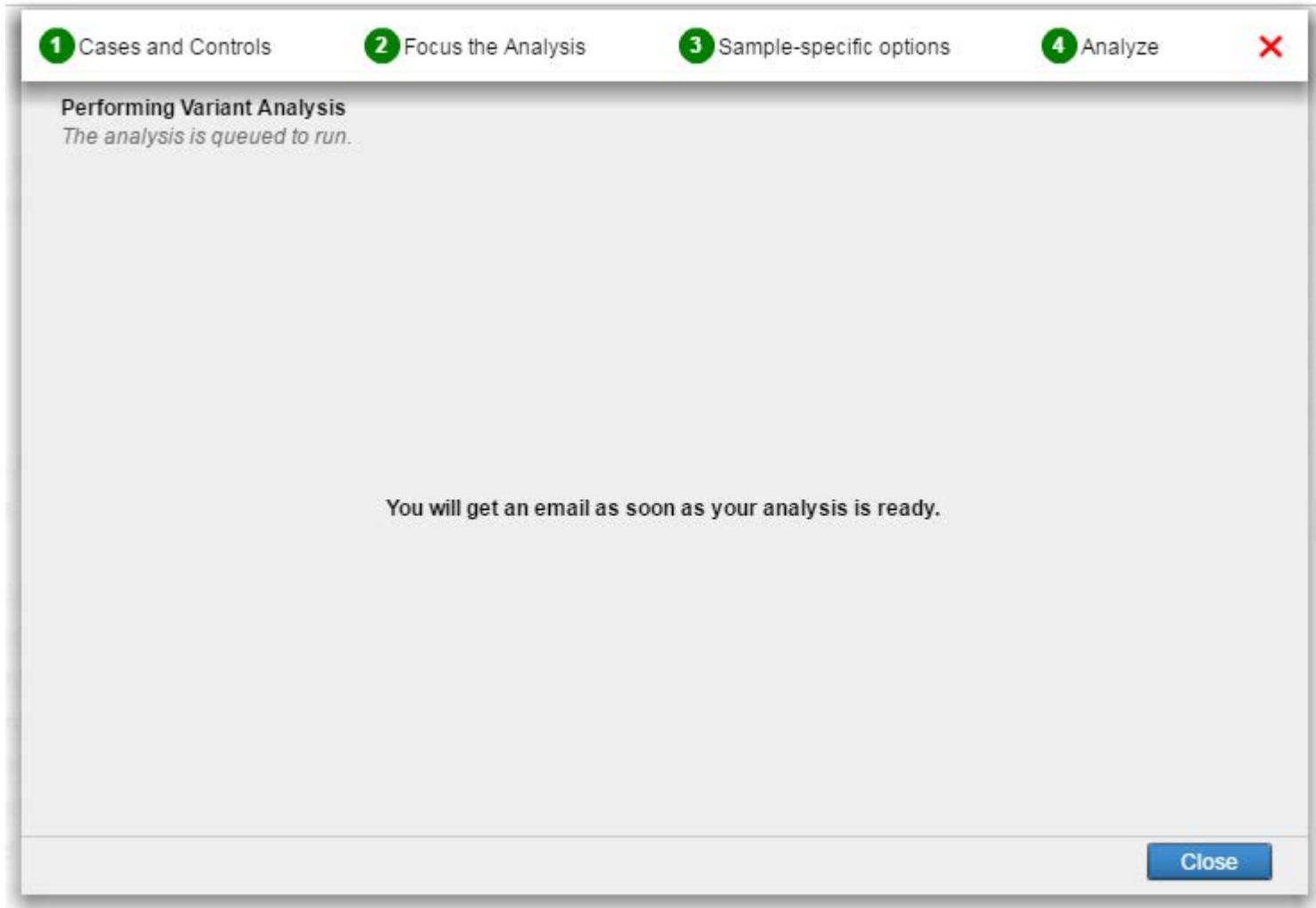
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Analyze

A series of options follows, guiding you through the analysis



A series of options follows, guiding you through the analysis



Can wait for the email or press “Refresh”

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Open

Name	Description	From	Created ▼	Samples	Status
Test WEX		variant_analysis1@i	05/31/2017	1 / 0	running
Inova Adams Oliver trio	Inova Adams Oliver trio (Workshop). The Inova NOTCH1 Case study is a...	ruth.burton@qiagen.	05/25/2017	1 / 2	active
Test WES	WES somatic variants from pancreatic cancer	variant_analysis1@i	05/24/2017	1 / 0	active
hcc tn2		variant_analysis1@i	02/22/2017	2 / 2	active
triple negative		variant_analysis1@i	02/22/2017	2 / 2	active
triple negative 22217		neha.jalan@qiagen.	02/22/2017	2 / 2	active
HCC TN		neha.jalan@qiagen.	02/22/2017	2 / 2	active
Tutorial		variant_analysis1@i	03/09/2016	1 / 1	active
Refined, trio, recess, train	Refined, trio, single, homozygous recessive test: pheochromocytoma	darryl.gietzen@qia	03/09/2016	1 / 2	active

When active can open analysis and review the filtering steps

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Name	Description	From	Created ▼	Samples	Status
Test WEX		variant_analysis1@i	05/31/2017	1 / 0	active
Inova Adams Oliver trio	Inova Adams Oliver trio (Workshop). The Inova NOTCH1 Case study is a...	ruth.burton@qiagen.	05/25/2017	1 / 2	active
Test WES	WES somatic variants from pancreatic cancer	variant_analysis1@i	05/24/2017	1 / 0	active
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triple negative 22217		neha.jalan@qiagen.	02/22/2017	2 / 2	active
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Refined, trio, recess, train	Refined, trio, single, homozygous recessive test: pheochromocytoma	darryl.gietzen@qia	03/09/2016	1 / 2	active
Refined, Inherit, de nov...	Refined, Trio, single, de novo test: pheochromocytoma	darryl.gietzen@qia	03/09/2016	1 / 2	active
Initial Train trio sing	Initial result, Trio	darryl.gietzen@qia	03/09/2016	1 / 2	active
Refined, trio, C-het, train	Refined, trio, single, compound-heterozygous recessive test: pheochrom...	darryl.gietzen@qia	03/09/2016	1 / 2	active
Initial Train tumor	Initial result, tumor-normal, multiple pairs: pheochromocytoma	darryl.gietzen@qia	03/09/2016	3 / 3	active
Refined, multi-tumor, train	Refined, tumor-normal, multiple, de novo test with gene expression filter: ...	darryl.gietzen@qia	03/09/2016	3 / 3	active
Refined, single, tumor-n...	Refined, tumor-normal, single comparison, de novo in tumor: pheochrom...	darryl.gietzen@qia	03/09/2016	1 / 1	active
Initial Train tumor sing	Initial result, tumor-normal, single: pheochromocytoma	darryl.gietzen@qia	03/09/2016	1 / 1	active

Status: active ×

Name Test WEX

Description

Created 05/31/2017 04:27 PM

From variant_analysis1@ingenuity.com

Genome GRCh37/HG19

Variant Count 2250

Filters

Confidence
Common Variants
Predicted Deleterious
Genetic Analysis
Cancer Driver Variants
Biological Context

Fields

Samples TCRBOA2-T-WEX Somatic Variants (case)

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When active can open analysis and review the filtering steps

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Name	Description	From	Created ▼	Samples	Status
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hcc tn2		variant_analysis1@i	02/22/2017	2 / 2	active
triple negative		variant_analysis1@i	02/22/2017	2 / 2	active
triple negative 22217		neha.jalan@qiagen.	02/22/2017	2 / 2	active
HCC TN		neha.jalan@qiagen.	02/22/2017	2 / 2	active
Tutorial		variant_analysis1@i	03/09/2016	1 / 1	active
Refined, trio, recess, train	Refined, trio, single, homozygous recessive test: pheochromocytoma	darryl.gietzen@qia	03/09/2016	1 / 2	active
Refined, Inherit, de nov...	Refined, Trio, single, de novo test: pheochromocytoma	darryl.gietzen@qia	03/09/2016	1 / 2	active
Initial Train trio sing	Initial result, Trio	darryl.gietzen@qia	03/09/2016	1 / 2	active
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Initial Train tumor	Initial result, tumor-normal, multiple pairs: pheochromocytoma	darryl.gietzen@qia	03/09/2016	3 / 3	active
Refined, multi-tumor, train	Refined, tumor-normal, multiple, de novo test with gene expression filter: ...	darryl.gietzen@qia	03/09/2016	3 / 3	active
Refined, single, tumor-n...	Refined, tumor-normal, single comparison, de novo in tumor: pheochrom...	darryl.gietzen@qia	03/09/2016	1 / 1	active
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Cancer Driver Variants
Biological Context

Fields

Samples TCRBOA2-T-WEX Somatic Variants (case)

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A filtering cascade has been applied... let's look at the filters in order

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Settings Feedback

Summary | Variants | Genes | Groups/Complexes | Pathways | Processes | Diseases | Overview

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135 variants

Filter Cascade

Variants	Genes
2250	1652
↓	
<p>× Confidence</p> <p>1928 1442</p>	
↓	
<p>× Common Variants</p> <p>1385 1084</p>	
↓	
<p>× Predicted Deleterious</p> <p>588 475</p>	
↓	
<p>× Genetic Analysis</p> <p>588 475</p>	
↓	
<p>× Cancer Driver Variants</p> <p>531 430</p>	
↓	
<p>× Biological Context</p> <p>135 111</p>	

☒ Recalculate when filters change

Add Filter

Chr...	Position	Gene Region	Gene Symbol	Protein Variant	Variant Findings	Case Samples	Translation Impact	SIFT Functio...	SIFT Sc...	PolyPhe...	Regulatory Site	Reg...
1	1960591	Exonic	GABRD	p.R245G			missense	Damaging	0.00	Probably		
1	34164470	Exonic	CSMD2	p.A1230S, p.A12			missense	Damaging	0.03	Benign		
1	109727724	Exonic, Intronic	KIAA1324	p.T238fs*21, p.T	2		frameshift					
1	160389105	Exonic	VANGL2	p.R169H			missense	Damaging	0.02	Probably		
1	180913650	Exonic	KIAA1614	p.G1095G			synonymous				Splice Site Loss	
1	201181728	Exonic	IGFN1	p.L2572fs*16	2		frameshift					
1	236156953	Exonic	NID1	p.T916M			missense	Tolerated	0.07	Benign		
2	29287826	Exonic	C2orf71	p.L1259P			missense	Tolerated	0.07	Probably		
2	71801337	Exonic	DYSF	p.A1048S, p.A10			missense	Tolerated	0.69	Benign		
2	95542415	Exonic	TEKT4	p.I222V, p.I404V			in-frame	Damaging	0.04	Possibly I		
2	95542418	Exonic	TEKT4	p.A223T, p.A405			in-frame	Tolerated	0.27	Benign		
2	97633356	Exonic	FAM178B	p.A212V			missense					
2	160193995	Exonic, Intronic	BAZ2B	p.S1861P			missense					
2	165551296	Exonic	COBLL1	p.L869fs*12, p.L	23		frameshift					
2	220344853	Exonic	SPEG	p.T1778I			missense	Damaging	0.00	Probably		
2	220471785	Exonic	STK11IP	p.R382H			missense	Damaging	0.00			
3	49723321	Exonic	MST1	p.W408G			missense	Damaging	0.00	Probably		
3	50273858	5'UTR, Exonic	GNAI2	p.A31T	27		missense	Tolerated	0.28	Benign	ENCODE TFBS	POL
3	124732449	Exonic	HEG1	p.S667_S672du	2		in-frame					
3	195508105	Exonic, Intronic	MUC4	p.A3449V			missense	Tolerated	0.10			
3	195508114	Exonic, Intronic	MUC4	p.T3446N			missense	Damaging	0.03			
3	195508921	Exonic, Intronic	MUC4	p.V3177A			missense	Activating	1.00			
3	195508930	Exonic, Intronic	MUC4	p.L3174P			missense	Tolerated	0.08			
4	5578113	Exonic	EVC2	p.Q1042del, p.Q			in-frame					
4	134072602	Exonic	PCDH10	p.R436P			missense	Tolerated	0.20	Possibly I		
4	164246874	Exonic	NPY1R	p.D246N	1		missense	Damaging	0.04	Probably		
4	187630956	Exonic	FAT1	p.L9P			missense	Damaging	0.00	Possibly I		
5	9197415	Exonic	SEMA5A	p.N312fs*13			frameshift					
5	112479044	Exonic	MCC	p.H252R, p.H62			missense			Possibly I		
5	120021716	Exonic	PRR16	p.K53R, p.K6R, I			missense	Tolerated	0.30	Probably		

Sample Legend [\[hide\]](#)

<p>Gene Function</p> <p>loss normal gain</p> <p>Identical to Reference Genome</p> <p>Heterozygous Variant</p> <p>Heterozygous/Ambiguous</p> <p>Homozygous Variant</p> <p>Copy Number Gain/Heterozygous</p> <p>Copy Number Gain/Homozygous</p> <p>Hemizygous</p> <p>Nullizygous</p> <p>Gene Fusion</p> <p>No genotype</p>	<p>Confident Call</p> <p>No Yes</p> <p>Loss normal gain</p>
--	---

A filtering cascade has been applied... let's look at the filters in order

Filter

Rename

Keep only

▼

variants which satisfy all of these criteria:

☒ Call quality is at least in any case or at least in any control

AND

☐ Variant passed upstream pipeline filtering

AND

☐ Read depth is at least in any case or at least in any control

AND

☐ Genotype quality is at least in any case or at least in any control

AND

☐ Allele fraction is at least in any case or at least in any control

AND

☒ Outside top % most exonically variable 100base windows in healthy public genomes

AND

☐ Outside top % [most exonically variable genes](#) in healthy public genomes (1000 Genomes)

Subsequent filters only treat a variant as present for samples that also satisfy the Keep criteria.

Apply

3

49723321

Exonic

MSI1

p.W408G

missense

A filtering cascade has been applied... let's look at the filters in order

Filter

Rename

Exclude

▼

variants that are observed in any of these populations with an allele frequency of

☐

at least

▼

3

▲▼

% of

all

▼

in the

Allele Frequency Community (includes ExAC and CGI)

Gain access to the community - [contribute your samples](#)

☒

at least

▼

0.5

▲▼

% in the

1000 Genomes Project

☒

at least

▼

0.5

▲▼

% of

all

▼

in the

ExAC

☒

at least

▼

0.5

▲▼

% of

all

▼

NHLBI ESP exomes

are present in

☐

dbSNP

or

☐

DGV

* The public Complete Genomics genomes are included in the AFC

Apply

A filtering cascade has been applied... let's look at the filters in order

Filter

Predicted Deleterious

Rename

Keep only

▼

variants that

are experimentally observed to be associated with a phenotype:

Disease-associated according to computed ACMG Guidelines classification

☒ Pathogenic
 ☒ Likely Pathogenic
 ☐ Uncertain Significance
 ☐ Likely Benign
 ☐ Benign

Listed in

☒ HGMD®
 ☐ ClinVar

OR

are associated with gain of function of a gene

☐ Established in the Literature
 ☐ Gene Fusion
 ☐ Inferred activating mutation by Ingenuity
 ☐ Predicted gain of function by BSIFT
 ☐ microRNA Binding Site
 ☐ Copy Number Gain

OR

are associated with loss of function of a gene

☒ Frameshift, in-frame indel, or start/stop codon change
 ☒ Missense unless predicted tolerated by ☐ SIFT or ☐ PolyPhen-2
 ☐ Predicted deleterious by having CADD score >
☐ Nullizygous
 ☒ Splice site loss up to bases into intron or ☒ as predicted by MaxEntScan
 ☐ Deleterious to a microRNA
 ☐ Copy Number Loss
 ☐ Promoter Loss ☐ with ENCODE TFBS
 ☐ Enhancer

Apply

A filtering cascade has been applied... let's look at the filters in order

Filter

Genetic Analysis

Rename

Use recommended settings for: (Custom) Inferred gain- or loss-of-function variants Set

☐ Pair/match samples from the same individual ⓘ
☐ Restrict to transmitted variants

☒ Case Samples

Keep only variants which are

☒ associated with gain of function
To control specific gain of function types, use the Predicted Deleterious filter

OR

☒ Homozygous
☒ Compound Heterozygous
☒ Haploinsufficient
☒ Hemizygous
☐ Nullizygous

☒ Het-ambiguous
☒ Heterozygous

AND

the genotypes selected above occur in
 at least 1 of the 1 case samples (100%)
 at variant level

☐ Control Samples

Exclude variants which are

☐ associated with gain of function
To control specific gain of function types, use the Predicted Deleterious filter

OR

☐ Homozygous
☐ Compound Heterozygous
☐ Haploinsufficient
☐ Hemizygous

☐ Het-ambiguous
☐ Heterozygous

AND

Apply

Sample to Insight

QIAGEN Sample to Insight | www.qiagenbioinformatics.com | www.qiagen.com

26

A filtering cascade has been applied... let's look at the filters in order

Filter

Cancer Driver Variants

Rename

Keep only

▼

variants that are found in

☒ Cancer-associated mouse knockout phenotypes
[View list of phenotypes](#)

☒ Cancer-associated cellular processes with

appropriate directionality

▼

[View list of processes](#)

☒ Cancer-associated pathways with

appropriate directionality

▼

[View list of pathways](#)

☒ Cancer therapeutic targets
[View list of drug targets](#)

☒ Published cancer literature

variant and gene level

▼

 findings

☐ Known or predicted cancer subnetwork regulatory sites
[View list of disease genes](#)

☐ COSMIC at a frequency

greater than or equal to

▼

0.01

▲▼

 %

☒ TCGA at a frequency

greater than or equal to

▼

0.01

▲▼

 %

AND

Involved in any of the diseases listed below

▼

any cancer

Apply

A filtering cascade has been applied... let's look at the filters in order

Filter

Biological Context

Rename

Keep only

▼

variants

Genes within

☐

1 hop

▼

downstream or

☐

1 hop

▼

upstream of the genes implicated in the biological terms entered

that are known or predicted to

Affect

▼

genes listed below or genes implicated in the following diseases, processes, pathways, phenotypes, domains, activities, or biomarkers

Enter and select term

× pancreatic cancer [disease]

Upload gene list file(s)...

☒

include diseases consistent with the phenotypes above

Apply

As well as filtering IVA provides a wealth of information...

Edit Columns Export Create List <input type="text" value="Search gene, chr, or db SNP"/> 135 variants									
Chr...	Position	Gene Region	Gene Symbol	Protein Variant	Variant Findings	Case Samples	Translation Impact	SIFT Functio...	SIFT
1	1960591	Exonic	GABRD	p.R245G			missense	Damaging	0.00
1	34164470	Exonic	CSMD2	p.A1230S, p.A12			missense	Damaging	0.03
1	109727724	Exonic, Intronic	KIAA1324	p.T238fs*21, p.T	2		frameshift		
1	160389105	Exonic	VANGL2	p.R169H			missense	Damaging	0.02
1	180913650	Exonic	KIAA1614	p.G1095G			synonymous		
1	201181728	Exonic	IGFN1	p.L2572fs*16	2		frameshift		
1	236156953	Exonic	NID1	p.T916M			missense	Tolerated	0.07
2	29287826	Exonic	C2orf71	p.L1259P			missense	Tolerated	0.07
2	71801337	Exonic	DYSF	p.A1048S, p.A10			missense	Tolerated	0.69
2	95542415	Exonic	TEKT4	p.I222V, p.I404V			in-frame	Damaging	0.04
2	95542418	Exonic	TEKT4	p.A223T, p.A405			in-frame	Tolerated	0.27
2	97633356	Exonic	FAM178B	p.A212V			missense		
2	160193995	Exonic, Intronic	BAZ2B	p.S1861P			missense		
2	165551296	Exonic	COBLL1	p.L869fs*12, p.L	23		frameshift		
2	220344853	Exonic	SPEG	p.T1778I			missense	Damaging	0.00
2	220471785	Exonic	STK11IP	p.R382H			missense	Damaging	0.00
3	49723321	Exonic	MST1	p.W408G			missense	Damaging	0.00
3	50273858	5'UTR, Exonic	GNAI2	p.A31T	27		missense	Tolerated	0.28
3	124732449	Exonic	HEG1	p.S667_S672du	2		in-frame		
3	195508105	Exonic, Intronic	MUC4	p.A3449V			missense	Tolerated	0.10
3	195508114	Exonic, Intronic	MUC4	p.T3446N			missense	Damaging	0.03
3	195508921	Exonic, Intronic	MUC4	p.V3177A			missense	Activating	1.00
3	195508930	Exonic, Intronic	MUC4	p.L3174P			missense	Tolerated	0.08
4	5578113	Exonic	EVC2	p.Q1042del, p.Q			in-frame		
4	134072602	Exonic	PCDH10	p.R436P			missense	Tolerated	0.20
4	164246874	Exonic	NPY1R	p.D246N	1		missense	Damaging	0.04
4	187630956	Exonic	FAT1	p.L9P			missense	Damaging	0.00
5	9197415	Exonic	SEMA5A	p.N312fs*13			frameshift		
5	112479044	Exonic	MCC	p.H252R, p.H62			missense		
5	120021716	Exonic	PRR16	p.K53R, p.K6R,			missense	Tolerated	0.30

Variant: chr2 | 165551296 | D... x

View : [More Details](#)
[Path to Phenotype](#)
[Variant Findings \(23\)](#)

Classification : [Uncertain Significance](#)
Red Activity: [More Details](#)
Gene Symbol : **COBLL1**
cordon-bleu WH2 rep
protein like 1

Cytoband : q24.3
Position : chr2:165551296 [IGV](#)

Gene Symbol : COBLL1
Position : 165551296
Cytoband : 2q24.3
Gene Region : Exonic
c.2606delT,
Transcript : c.2720delT,
Variant : c.2744delT,
c.2921delT
p.L869fs*12,
p.L907fs*12,
Protein Variant : p.L915fs*12,
p.L974fs*12

Translation
Impact : frameshift

CADD Score : 35.000
ExAC
Frequency : 0.269%
ExAC East
Asian : 0.216%
Frequency
ExAC South
Asian : 0.153%
Frequency
ExAC African
Frequency : 0.172%
ExAC
European : 0.338%
Frequency

As well as filtering IVA provides a wealth of information...

Variant: chr2 165551296 Deletion	
	Path to Phenotype
	Variant Findings (23)
Classification :	Uncertain Significance
Inferred Activity:	More Details
Gene Symbol :	COBLL1
	cordon-bleu WH2 repeat protein like 1
Cytoband :	q24.3
Position :	chr2:165551296 [IGV] [UCSC]
Gene Symbol :	COBLL1
Position :	165551296
Cytoband :	2q24.3
Gene Region :	Exonic
Transcript Variant :	c.2606delT, c.2720delT, c.2744delT, c.2921delT
Protein Variant :	p.L869fs*12, p.L907fs*12, p.L915fs*12, p.L974fs*12
Translation :	frameshift
Impact :	
CADD Score :	35.000
ExAC Frequency :	0.269%
ExAC East Asian Frequency :	0.216%
ExAC South Asian Frequency :	0.153%
ExAC African Frequency :	0.172%
ExAC European Frequency :	0.338%
ExAC Latino Frequency :	0.112%
ExAC Homozygous Count :	0
dbSNP ID :	772030741
COSMIC ID :	1400553
	4449589
Cosmic Frequency :	0.99, 1.19

As well as filtering IVA provides a wealth of information...

Variant: chr2 | 165551296 | Deletion (COBL1 p.L869fs*12, etc) - Computed Classification Explana... ✕

Computed classification based on [ACMG guidelines](#): **Uncertain Significance for Schizophrenia**

Evidence for pathogenicity

- PP3 - Multiple lines of computational evidence support a deleterious effect on the gene or gene product [CADD = 35.0] (Supporting)

Evidence against pathogenicity

- none

IPA Gene View: COBL1

Summary

Entrez Gene Name:	cordon-bleu WH2 repeat protein like 1
Synonym(s):	1810047P18RIK, Cobl-like 1, COBLR1, cordon-bleu WH2 repeat protein-like 1, D430044D16Rik, KIAA0977
Protein Functions / Functional Domains:	cadherin binding, protein binding
Subcellular Location:	cell-cell adherens junctions, Extracellular Space, plasma, vesicles
Canonical Pathway:	--

Top findings from Ingenuity Knowledge Base

regulates:	--
regulated by:	IL4, GMNN, SOX3, SOX2, MYC, KLF4, POU5F1, SOX1, Mycobacterium tuberculosis H37Rv, MAP3K8, FOS, Sos
binds:	PACSIN3, KIAA0368, PKM, ASMTL, DYNC1L1, JUP, PPP4R3A, KIAA1107, TGS1, RASSF1, ITGB3BP, DLG3, DYNC2H1, BRINP1, SORT1
role in cell:	--
disease:	non-insulin-dependent diabetes mellitus, gastric epithelial cancer, gastric carcinoma, colorectal carcinoma

Human Isoforms From RefSeq [More Info](#)

Loading

GO Annotations

Molecular Function:	actin monomer binding
Biological Process:	actin filament network formation; actin filament polymerization
Cellular Component:	cell-cell adherens junction; extracellular vesicular exosome

As well as filtering IVA provides a wealth of information...

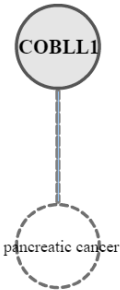
Coding Effects

Gene Symbol	Region	Transcript ID	Transcript Variant	Protein Variant	Translation Impact	SLF
COBL1	Exonic	NM_001278458.1	c.2921delT	p.L974fs*12	frameshift	
COBL1	Exonic	NM_001278460.1	c.2744delT	p.L915fs*12	frameshift	
COBL1	Exonic	NM_001278461.1	c.2606delT	p.L869fs*12	frameshift	
COBL1	Exonic	NM_014900.4	c.2720delT	p.L907fs*12	frameshift	

Variant: chr2 | 165551296 | q24.3 | Deletion | A (COBL1 p.L907fs*12, etc)

Details | Path to Phenotype | Variant Findings

Click on lines to see supporting citations.



Findings (2 citations)

COSMIC:4961732

- Mutant human **COBL1** gene (somatic substitution c.920>G>A) is observed with pancreatic ductal adenocarcinoma in human (COSMIC: observed in 2 of 704 samples).

COSMIC:3379821

- Mutant human **COBL1** gene (somatic substitution c.951>G>A) is observed with pancreatic ductal adenocarcinoma in human (COSMIC: observed in 2 of 704 samples).

As well as filtering IVA provides a wealth of information...

Variant: chr2 | 165551296 | q24.3 | Deletion | A (COBLL1 p.L907fs*12, etc)
✕

Details
Path to Phenotype
Variant Findings

Findings (9 citations)

[Genomic analysis of smoothened inhibitor resistance in basal cell carcinoma.](#) *Cancer Cell.* (2015)

- Mutant human **COBLL1** gene (somatic frameshift deletion c.2921delT translating to p.L974fs*12) is observed with basal cell carcinoma in human skin (COSMIC: observed in 1 of 49 samples).
- Mutant human **COBLL1** gene (somatic frameshift deletion c.2720delT translating to p.L907fs*12) is observed with basal cell carcinoma in human skin (COSMIC: observed in 1 of 49 samples).

[RNF43 is frequently mutated in colorectal and endometrial cancers.](#) *Nat Genet.* (2014)

- Mutant human **COBLL1** gene (somatic frameshift deletion c.2720delT translating to p.L907fs*12) is observed with adenocarcinoma in human colon (COSMIC: observed in 3 of 127 samples).
- Mutant human **COBLL1** gene (somatic frameshift deletion c.2921delT translating to p.L974fs*12) is observed with adenocarcinoma in human colon (COSMIC: observed in 3 of 127 samples).

[Mutational landscape of aggressive cutaneous squamous cell carcinoma.](#) *Clin Cancer Res.* (2014)

- Mutant human **COBLL1** gene (somatic frameshift heterozygous deletion c.2720delT translating to p.L907fs*12) is observed with squamous-cell carcinoma in skin from human head and neck (COSMIC: observed in 2 of 39 samples).
- Mutant human **COBLL1** gene (somatic frameshift heterozygous deletion c.2921delT translating to p.L974fs*12) is observed with squamous-cell carcinoma in skin from human head and neck (COSMIC: observed in 2 of 39 samples).

[The mutational burdens and evolutionary ages of early gastric cancers are comparable to those of advanced gastric cancers.](#) *J Pathol.* (2014)

- Mutant human **COBLL1** gene (somatic frameshift deletion c.2720delT translating to p.L907fs*12) is observed with mixed intestinal and diffuse adenocarcinoma-unclassifiable in human stomach (COSMIC: observed in 1 of 2 samples).
- Mutant human **COBLL1** gene (somatic frameshift deletion c.2921delT translating to p.L974fs*12) is observed with small intestine cancer in human stomach (COSMIC: observed in 1 of 13 samples).
- Mutant human **COBLL1** gene (somatic frameshift deletion c.2720delT translating to p.L907fs*12) is observed with small intestine cancer in human stomach (COSMIC: observed in 1 of 13 samples).
- Mutant human **COBLL1** gene (somatic frameshift deletion c.2921delT translating to p.L974fs*12) is observed with diffuse adenocarcinoma in human stomach (COSMIC: observed in 1 of 2 samples).
- Mutant human **COBLL1** gene (somatic frameshift deletion c.2720delT translating to p.L907fs*12) is observed with diffuse adenocarcinoma in human stomach (COSMIC: observed in 1 of 2 samples).
- Mutant human **COBLL1** gene (somatic frameshift deletion c.2921delT translating to p.L974fs*12) is observed with mixed intestinal and diffuse adenocarcinoma-unclassifiable in human stomach (COSMIC: observed in 1 of 2 samples).

[Whole-genome sequencing and comprehensive molecular profiling identify new driver mutations in gastric cancer.](#) *Nat Genet.* (2014)

- Mutant human **COBLL1** gene (somatic frameshift heterozygous deletion c.2720delT translating to p.L907fs*12) is observed with small intestine cancer in human stomach (COSMIC: observed in 6 of 57 samples).
- Mutant human **COBLL1** gene (somatic frameshift heterozygous deletion c.2921delT translating to p.L974fs*12) is observed with small intestine cancer in human stomach (COSMIC: observed in 6 of 57 samples).

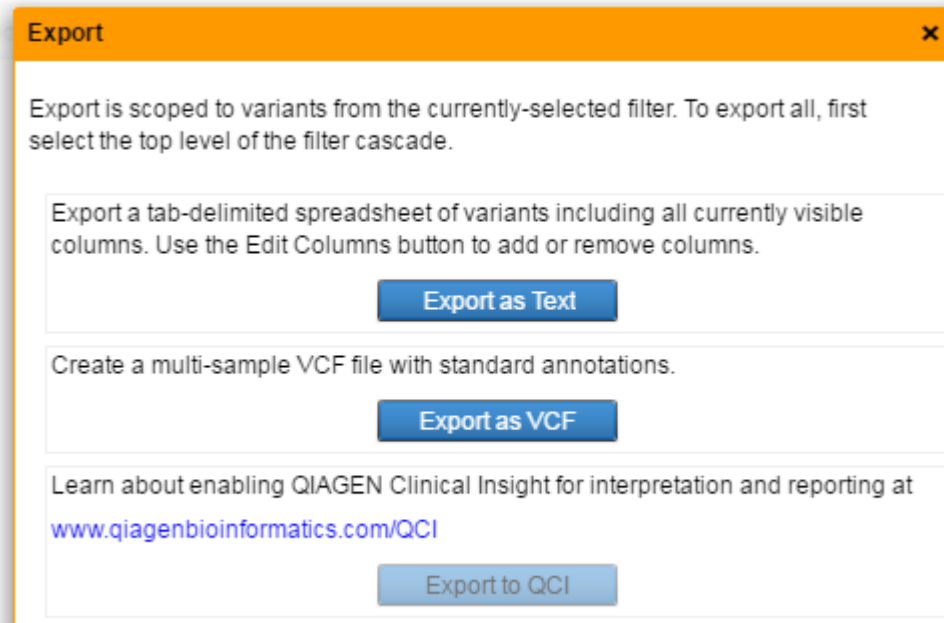
[Whole-exome sequencing of pancreatic neoplasms with acinar differentiation.](#) *J Pathol.* (2014)

- Mutant human **COBLL1** gene (somatic frameshift deletion c.2720delT translating to p.L907fs*12) is observed with acinar-cell carcinoma in human pancreas (COSMIC: observed in 1 of 17 samples).

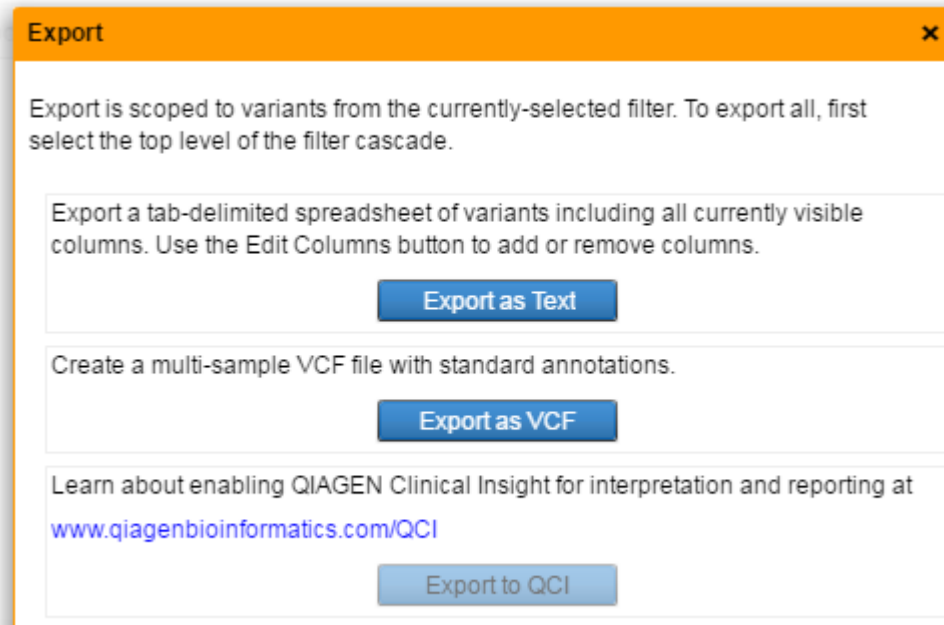
[The genomic landscape of oesophagogastric junctional adenocarcinoma.](#) *J Pathol.* (2013)

- Mutant human **COBLL1** gene (somatic frameshift deletion c.2720delT translating to p.L907fs*12) is observed with adenocarcinoma in human lower third of esophagus (COSMIC: observed in 1 of 8 samples).

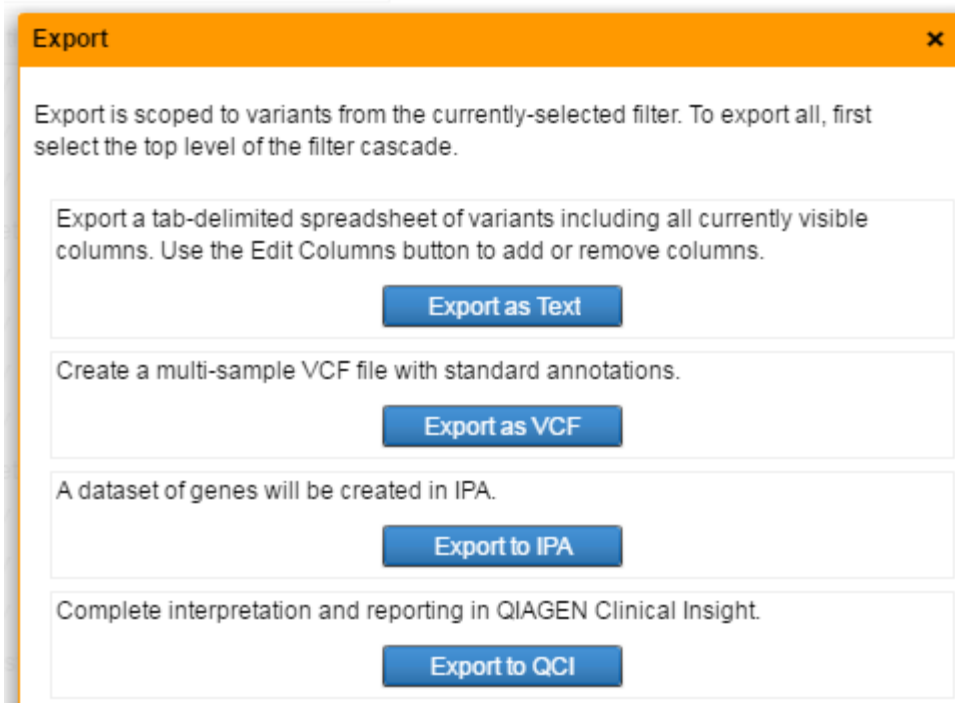
What can you do with your filtered list?



To export to QCI I you need a QCI I account



To export to QCI I you need a QCI I account



Choose your workflow

Which pipeline will you use?

Somatic

Germline/Hereditary
Including Hereditary Cancer

Add in patient meta data

Test Product Profile (optional)

Custom specification of default values that remain constant for a given test product (includes reporting method, report template, and more). Consult with our clinical science team to create one or more test product profile(s).

Test Product Code (required)

Your laboratory's unique identifier for the test that was ordered (usually a catalog # or SKU).

Accession ID (required)

Your laboratory's unique identifier for the sample.

Test Date (required)

Date that this accession entered your system.

Diagnosis (required)

This is used to match treatments and trials and for display on the report.

Diagnosis Stage (optional)

This is used to contextualize clinical trials to the selected stage. If left unselected, all clinical trials pertinent to the diagnosis will be displayed.

Primary Tumor Site (required)

This is used to display treatments and trials relevant to the selected tissue type when a match to the specific diagnosis cannot be made. Select "Unknown" only if no choice is relevant to this sample.

Add in patient meta data

Test Product Code (required)

Test

Your laboratory's unique identifier for the test that was ordered (usually a catalog # or SKU).

Accession ID (required)

TCRBOA2-T-WEX.

Your laboratory's unique identifier for the sample.

Test Date (required)

2017-05-31



Date that this accession entered your system.

Diagnosis (required)

Pancreatic cancer

✔ This diagnosis will be used to match treatments and trials and for display on the report.

Diagnosis Stage (optional)

Stage IV



This is used to contextualize clinical trials to the selected stage. If left unselected, all clinical trials pertinent to the diagnosis will be displayed.

Primary Tumor Site (required)

Pancreas



This is used to display treatments and trials relevant to the selected tissue type when a match to the specific diagnosis cannot be made. Select "Unknown" only if no choice is relevant to this sample.

Tumor content (optional)

40

Estimated tumor content of the sample. Enter a value between 1 and 100.

Interpretation and Reporting

Advanced



The software will “learn” from your classifications...

Interpretation and Reporting

Advanced



Reporting Method (optional)

Lab policy for reuse of variant reportability from prior tests.

☒ review all - default

☐ rereport all

☐ custom

Treatments Policy (optional)

Custom specification of default filters used for treatment matching. Consult with our clinical science team to create one.

Trials Policy (optional)

Custom specification of default filters used for clinical trials matching. Consult with our clinical science team to create one.

Further meta data...

Information about the patient.

Sex (optional)

☐ Male ☒ Female

This is used to limit to gender-appropriate clinical trials.

Ethnicity (optional)

Caucasian ▼

Age (recommended)

65

This is used to limit to age-appropriate clinical trials (e.g. pediatric for children).

Back

Continue

Further meta data...

The information below is used only on the final report. For validation samples, you can use the data that has been prefilled for you below, or hit Clear Data and enter your own information.

Patient Name (optional)

Michelle Doe

Client Name (optional)

General Hospital

Client ID (optional)

ABC123

Physician Name (optional)

Dr. E Smith

Pathologist Name (optional)

Dr. R Jones

Specimen Type (optional)

biopsy

Specimen ID (optional)

ABC123

Specimen Dissection (optional)

manual

Date of Birth (optional)

May ▼ 31 ▼ 2017 ▼
 Month Day Year



Report Date
Feb 10, 2016

Somatic Test

Patient Information	Client Information	Specimen
Patient Name: Michelle Doe	Client: General Hospital	Specimen Type: biopsy
Date of Birth:	Client ID: ABC123	Specimen ID: K23456
Ethnicity: Caucasian	Physician: Dr. E Smith	Collection Date: Feb 2, 2016
Sex: male	Pathologist: Dr. R Jones	Accession Date: Feb 2, 2016
Accession: 1234567		Primary Tumor Site: Lung
		Diagnosis: Lung Cancer

Interpretation

5 Clinically Significant Variants Reported 2 Approved Therapies 5 Potential Clinical Trials

3 alterations were identified that are associated with an available treatment. Clinical trials were identified that target the detected alterations. 2 alterations are associated with resistance to cetuximab/irinotecan, gefitinib, afatinib, erlotinib therapies. EGFR p.Q787Q is not likely to be a therapeutic target because there is no change in the amino acid at this position. Similar to other alterations in circulating cDNA, the monitoring of this variant may be reflective of disease progression or treatment, clinical correlation is advised. The functional consequences and clinical significance of EGFR p.Q787Q is not established. The relevance of therapies targeting this alteration is uncertain. Similar to other alterations in circulating cDNA, the monitoring of this variant may be reflective of disease progression or treatment, clinical correlation is advised. Additional Findings This is Hebrew

Variant Reported	FDA Approved Therapies for Indication	FDA Approved Therapies for Other Indications	Therapies Associated with Resistance	Potential Clinical Trials
TP53 p.S246fs*7	irinotecan			
KRAS p.G12D		regorafenib	panitumumab cetuximab	5 potential trials
APC p.Q2403H				
ROS1 p.Q2032R				

Page 1 of 7
QIAGEN 11730 Sequetur Blvd, Third Floor, Redwood City, CA 94063 qiagenbioinformatics.com | QIAGEN.com
QIAGEN Clinical Insight - Interpret software was used in sequence interpretation.

Option for customized report...

Specimen Collection Date - YYYY-MM-DD (optional)

2017-05-31



Report Template (required)

DemoReport

If you have a customized template available to you, enter its name. Otherwise, use "DemoReport".

Provide Access To: (optional)

valid@email.com; UserGroupName;

List of QCI account email addresses and/or groups separated by semi-colons.

Back

Submit

Go to results...

Your sample has been submitted successfully.

The results will be available in QIAGEN
Clinical Insight within a few minutes.



[Go To Results](#)

More samples to upload?

Upload and annotate another sample.




[Upload Another](#)

Learn how to configure and upload another
sample via API.



[API Explorer](#)

The list of variants...


Clinical Insight

[Variant List](#)
[Variant Detail](#)
[Review & Report](#)

[Ruth Burton](#) | [Test List](#) | [Contact Us](#) | [Logout](#)

Accession ID (Test Product Code)	Tumor Content	Age	Sex	Ethnicity	Diagnosis	Stage
TCRBOA2-T-WEX. (Test)	40%	65	Female	Caucasian	Pancreatic cancer	IV

Phenotype: pancreatic cancer
Age of Onset: 71 years
Gene Prevalence: 1.29%
Disease Prevalence: 1/7692

Gene: **LAMA3**
Transcript: NM_198129.2

Variant: c.5575C>T
p.Q1859* loss

Somatic Frequency: -
Population Frequency: 0% ExAC
Allele Fraction: 8.70% (of 23 reads)
Impact: Stop Gain

Computed Classification

Pathogenic

pancreatic cancer

Previous Assessment
Pathogenic
for pancreatic cancer
ruth.burton@qiagen.com
May 24, 2017

Open

< Previous

Next >

Use Classification

View Bibliography

Sort By: Classification
View: Grid Table

LAMA3 c.5575C>T p.Q1859*	MYH6 c.5565+1G>A	RHOBTB2 c.910G>T p.E304*	SPTLC2 c.865G>T p.E289*	SMAD3 c.3G>T p.M1I
ABCA7 c.2689A>G p.T897A	ABHD8 c.956C>A p.A319D	ADAMTS17 c.52_54delCTG p.L18del	ADNP c.3047delA p.K1016fs*11	AKAP1 c.-121G>T
ANAPC5 c.1301G>A p.R434H	ANGPT4 c.333C>A p.I111I	ANKRD30B c.1027A>G p.T343A	ANKRD30B c.1051T>G p.S351A	ANKRD30B c.1057_1058delCCin... p.P353I
ANKRD30B c.1062_1063delAAin... p.K355Q	ANKRD30B c.1072T>C p.S358P	ANKRD30B c.1077delG p.K360fs*11	ANKRD30B c.1102A>G p.T368A	AP3D1 c.2650G>C p.A884P
ARHGEF17 c.1463G>A p.R488Q	ARHGEF17 c.462C>T p.D154D	ARHGEF17 c.5912G>T p.G1971V	ARID1B c.96C>T p.A32A	ARMC12 c.372G>T p.V124V


Show
☒ To be assessed (201)
☒ Assessed

Actionability
☒ Therapies for this cancer 1
☒ Therapies for other cancers 2
☒ Resistance R
☒ Clinical Trials CT
☒ None

Classification
☒ Pathogenic ■
☒ Likely Pathogenic ■
☒ Uncertain Significance □

Origin
☒ Likely Somatic
☒ Likely Germline

Two views... a lot of information is already given about the variant


Clinical Insight

[Variant List](#)
[Variant Detail](#)
[Review & Report](#)

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Accession ID (Test Product Code)
TCRBOA2-T-WEX. (Test)

Tumor Content: 40% Age: 65 Sex: Female Ethnicity: Caucasian Diagnosis: Pancreatic cancer Stage: IV

Phenotype: pancreatic cancer Age of Onset: 71 years Gene Prevalence: 1.29% Disease Prevalence: 1/7692

Gene: **LAMA3**
Transcript: NM_198129.2

Variant: c.5575C>T
p.Q1859* **loss**



Somatic Frequency: -
Population Frequency: 0% ExAC
Allele Fraction: 8.70% (of 23 reads)
Impact: Stop Gain

Computed Classification

Pathogenic
pancreatic cancer

Previous Assessment
Pathogenic
for pancreatic cancer
ruth.burton@qiagen.com
May 24, 2017

[Open](#) [< Previous](#) [Next >](#) [Use Classification](#) [View Bibliography](#)

Sort By: Classification View:  

Gene	Alteration	Chr:Position	Function	Impact	Allele Fraction	Somatic Frequency for Diagnosis
LAMA3	c.5575C>T p.Q1859*	Chr18:21474984	loss	stop gain	8.70% (of 23 reads)	-
MYH6	c.5565+1G>A	Chr14:23853650	loss	-	5.41% (of 37 reads)	-
RHOBTB2	c.910G>T p.E304*	Chr8:22864668	loss	stop gain	12% (of 17 reads)	-
SPTLC2	c.865G>T p.E289*	Chr14:78023475	loss	stop gain	5.56% (of 36 reads)	-
SMAD3	c.3G>T p.M11	Chr15:67358495	loss	start loss	7.41% (of 27 reads)	-
ABCA7	c.2689A>G p.T897A	Chr19:1051158	loss	missense	5.00% (of 40 reads)	-
ABHD8	c.956C>A p.A319D	Chr19:17405290	normal	missense	6.90% (of 29 reads)	-
ADAMTS17	c.52_54delCTG p.L18del	Chr15:100882051	loss	in-frame	5.71% (of 35 reads)	-
ADNP	c.3047delA p.K1016fs*11	Chr20:49508204	loss	frameshift	5.41% (of 111 reads)	0.41%
AKAP1	c.-121G>T	Chr17:55163802	normal	-	13% (of 15 reads)	-
ANAPC5	c.1301G>A p.R434H	Chr12:121766122	normal	missense	5.36% (of 56 reads)	-

Show

☒ To be assessed (201)

☒ Assessed

Actionability

☒ Therapies for this cancer **1**

☒ Therapies for other cancers **2**

☒ Resistance **R**

☒ Clinical Trials **CT**

☒ None

Classification

☒ Pathogenic

☒ Likely Pathogenic

☒ Uncertain Significance

Origin

☒ Likely Somatic

☒ Likely Germline

ACMG Guidelines are used for classification

Computed classification explanation

Computed classification based on ACMG guidelines: **Pathogenic for pancreatic cancer**

Evidence for pathogenicity

- PVS1 - Null variant (nonsense, frameshift, canonical +/-1 or 2 splice sites, initiation codon) in a gene where loss of function (LOF) is a known mechanism of disease (Very Strong)
- PM2 - Absent from controls (or at extremely low frequency if recessive) in Exome Sequencing Project, 1000 Genomes Project, or ExAC [In these sources of population frequency data, this variant's frequency is 0% or $\leq 0.001\%$] (Moderate)
- PP3 - Multiple lines of computational evidence support a deleterious effect on the gene or gene product [CADD = 43.0] (Supporting)

Evidence against pathogenicity

- None

Variant details provide supporting evidence for classification

Accession ID (Test Product Code)	Tumor Content	Age	Sex	Ethnicity	Diagnosis	Stage
TCRBOA2-T-WEX. (Test)	40%	65	Female	Caucasian	Pancreatic cancer	IV

Phenotype: pancreatic cancer	Age of Onset 71 years <i>i</i>	Gene Prevalence 1.29% <i>i</i>	Disease Prevalence 1/7692 <i>i</i>
---	-----------------------------------	-----------------------------------	---------------------------------------

Gene LAMA3 Transcript NM_198129.2	Variant c.5575C>T p.Q1859* loss <i>i</i>	Somatic Frequency: - <i>i</i> Population Frequency: 0% ExAC Allele Fraction: 8.70% (of 23 reads) Impact: Stop Gain	Computed Classification <i>i</i> Pathogenic pancreatic cancer	Previous Assessment Pathogenic for pancreatic cancer ruth.burton@qiagen.com May 24, 2017
--	--	---	---	---

Variant List
< Previous
Next >
Use Classification
Open Assessment
View Bibliography



► Assessment


► Variant details

▼ Laboratory observations (2)

	Date	Accession ID	Assessment	Phenotype	Reportability	Genotype	Assessor
►	May 24, 2017	TCRBOA2-T-WEX.read1	Pathogenic	pancreatic cancer	Reportable	Het-Amb	ruth.burton@qiagen.com
►	May 11, 2017	TCRBOA2-T-WEX.v2	Pathogenic	pancreatic cancer	Reportable	Het-Amb	ruth.burton@qiagen.com

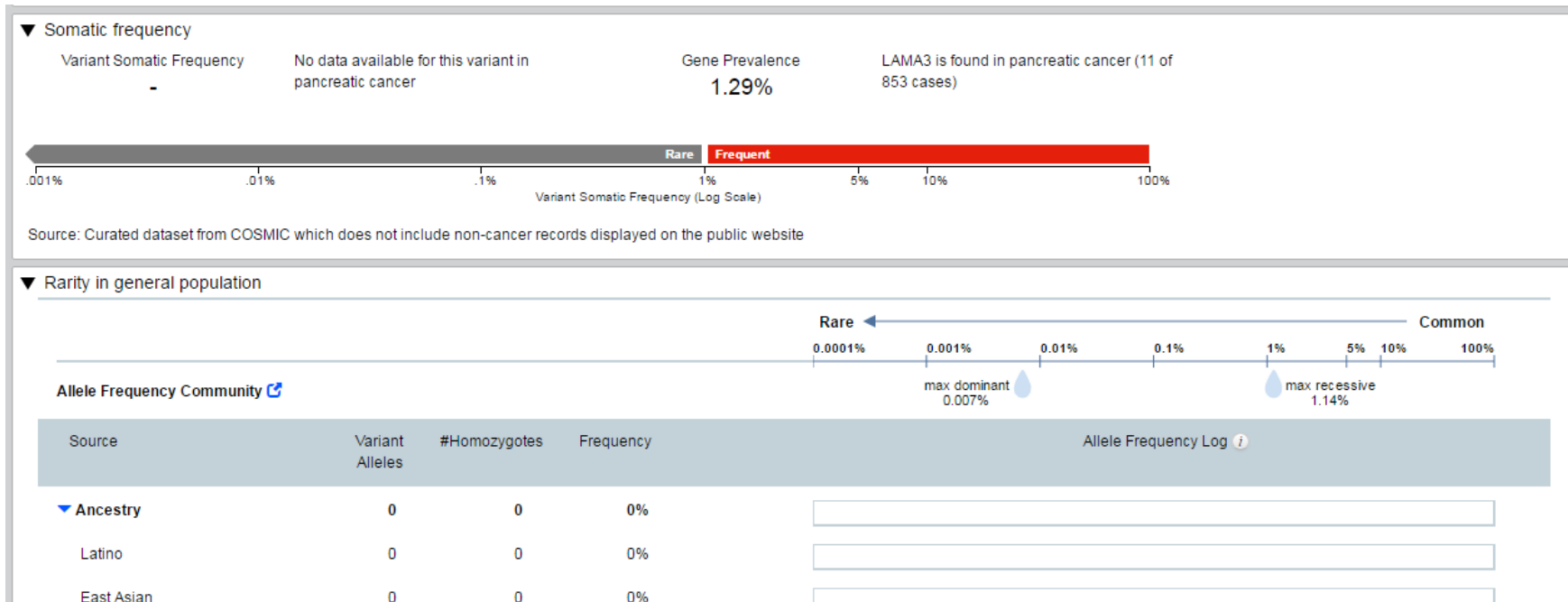
▼ Effect on Protein


 CHR 18: 21474984 c.5575C>T Exon 44 
 p.Q1859* Stop Gain SNV











NM_198129.2 ▼


Loss/Normal/Gain Fn
 Benign/Likely Benign
 Pathogenic/Likely Pathogenic
 Found in This Patient


Variant details provide supporting evidence for classification



Variant details provide supporting evidence for classification

▼ Predicted Biochemical Impact on NM_198129.2		
CADD 	 Deleterious 	 Predicts biochemical impact
PolyPhen	No Prediction	 Predicts no biochemical impact
SIFT	No Prediction 	 Predicts loss of function
Mutation Taster	View Prediction	 Predicts gain of function
BLOSUM	No Prediction	
PhyloP	 Highly Conserved	
MaxEntScan	No Prediction	
Gene Splicer	No Prediction	
B-SIFT	No Prediction	
QCI Inferred Activation 	No Prediction	

Key step is creating a report


Clinical Insight

[Variant List](#)
[Variant Detail](#)
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Accession ID (Test Product Code)	Tumor Content	Age	Sex	Ethnicity	Diagnosis	Stage
TCRBOA2-T-WEX. (Test)	40%	65	Female	Caucasian	Pancreatic cancer	IV

0 Days
System rec'd May 31, 2017

In Review
Current state

Change State

Sign Out

Preview Report

Marked Reportable

1
Variants

0
Clinical Trials

0
References

200
Unassessed Variants

Overall Interpretation

Positive
Presumed Positive
Inconclusive
Presumed Negative
Negative
Omit Interpretation

Add overall comment


Report Comment:

Pathogenic variants detected. NGS process passed QC

Edit

Reportable Variants

pancreatic cancer

Gene	Variant	Allele Fraction	Function	Assessment	References
 LAMA3	c.5575C>T p.Q1859*	8.70%	loss	Pathogenic	0

Key step is creating a report

Edit Comment for LAMA3 c.5575C>T p.Q1859* pancreatic cancer

No previous comments

Report comment:

Evidence for Pathogenicity

- PVS1 - Null variant (nonsense, frameshift, canonical +/-1 or 2 splice sites, initiation codon) in a gene where loss of function (LOF) is a known mechanism of disease (Very Strong)
- PM2 - Absent from controls (or at extremely low frequency if recessive) in Exome Sequencing Project, 1000 Genomes Project, or ExAC [In these sources of population frequency data, this variant's frequency is 0% or <= 0.001%] (Moderate)
- PP3 - Multiple lines of computational evidence support a deleterious effect on the gene or gene product [CADD = 43.0] (Supporting)

☐ Reuse this comment for other tests with
☐ any LAMA3 variant
☒ this variant

(maximum 32K characters)

Add Assessment
Cancel
Continue

Key step is creating a report



Somatic Test

Report Date May 31, 2017
Report Status -

Patient Information		Client Information		Specimen	
Patient Name	Michelle Doe	Client	General Hospital	Specimen Type	biopsy
Date of Birth	May 31, 2017	Client ID	ABC123	Specimen ID	ABC123
Ethnicity	Caucasian	Physician	Dr. E Smith	Collection Date	May 31, 2017
Sex	female	Pathologist	Dr. R Jones	Accession Date	May 31, 2017
Accession	TCRBOA2-T-WEX.			Primary Tumor Site	Pancreas
				Diagnosis	Pancreatic cancer
				Diagnosis Stage	IV

Interpretation

1 Clinically Significant Variant Reported 0 Approved Therapy 0 Potential Clinical Trials

Pathogenic variants detected. NGS process passed QC

Summary of Clinically Significant Variants

Variants Reported	Allele Fraction	Approved Therapies for Same Cancer	Approved Therapies for Other Cancers	Therapies Associated with Resistance	Potential Clinical Trials
LAMA3 p.Q1859*	8.7% (of 23 reads)				

Variant Details

Gene	Exon #	Nucleotide Change	Amino Acid Change	Effect on Protein
LAMA3	44	NM_198129.2: c.5575C>T	p.Q1859*	loss of function

Evidence for Pathogenicity

- PVS1 - Null variant (nonsense, frameshift, canonical +/-1 or 2 splice sites, initiation codon) in a gene where loss of function (LOF) is a known mechanism of disease (Very Strong)
- PM2 - Absent from controls (or at extremely low frequency if recessive) in Exome Sequencing Project, 1000 Genomes Project, or ExAC [In these sources of population frequency data, this variant's frequency is 0% or <= 0.001%] (Moderate)
- PP3 - Multiple lines of computational evidence support a deleterious effect on the gene or gene product [CADD = 43.0] (Supporting)

Genes Tested

KRAS NRAS KIT BRAF PDGFRA ALK EGFR ERBB2 PIK3CA ERBB3 ESR1 RAF1

Methods and Limitations

EXAMPLE Statement including sample type (FFPE, etc), method of extraction, amplification reactions, panel targeted regions, sequencing technology, etc. Additionally, a description of the data analysis software(s), genome of reference and the sensitivity of the methods should be described.

QIAGEN Clinical Insight - Interpret software was used in sequence analysis and interpretation. The application was internally designed and developed by QIAGEN. All analyses were based on: QIAGEN Clinical Insight-Interpret (4.4.20170525), Ingenuity Knowledge Base (Lorien 170520.000), CADD (v1.3), EVS (ESP6500SI-V2), Allele Frequency Community (2017-01-31), JASPAR (2013-11), Vista Enhancer hg18 (2012-07), Vista Enhancer hg19 (2012-07), Clinical Trials (Lorien 170520.000), BSIFT (2016-02-23), TCGA (2013-09-05), PolyPhen-2 (v2.2.2), 1000 Genome Frequency (phase3v5b), Clinvar (2017-01-04), DGV (2016-05-15), COSMIC (v79), ExAC (0.3.1), HGMD (2016.4), PhyloP hg18 (2009-11), PhyloP hg19 (2009-11), DbSNP (149), TargetScan (6.2), SIFT4G (2016-02-23) Weekly updates to Ingenuity Knowledge Base for clinical trials recruitment status and new findings from recent articles. Variants are reported according to HGVS nomenclature and were classified following ACMG guidelines. Information on therapeutic agents and clinical trials were obtained from publicly available information. Variants, therapies, and trials listed in this report are not ranked in order of potential clinical significance or predicted efficacy for this patient.

Trio Analysis, highlight and click “Open”

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 [My Libraries](#) |
 [My Analyses](#) |
 [Publications](#) |
 [Test WEX](#) [x]

[Create](#)
[Refresh](#)
[Share](#)
[Open](#)

Name	Description	From	Created ▼	Samples	Status
Test WEX		variant_analysis1@i	05/31/2017	1 / 0	active
Inova Adams Oliver trio	Inova Adams Oliver trio (Workshop). The Inova NOTCH1 Case study is a...	ruth.burton@qiagen.	05/25/2017	1 / 2	active
Test WES	WES somatic variants from pancreatic cancer	variant_analysis1@i	05/24/2017	1 / 0	active
hcc tn2		variant_analysis1@i	02/22/2017	2 / 2	active
triple negative		variant_analysis1@i	02/22/2017	2 / 2	active
triple negative 22217		neha.jalan@qiagen.	02/22/2017	2 / 2	active
HCC TN		neha.jalan@qiagen.	02/22/2017	2 / 2	active
Tutorial		variant_analysis1@i	03/09/2016	1 / 1	active
Refined.trio. recess. train	Refined. trio. single. homozygous recessive test: pheochromocytoma	darryl.aetzen@qiaa	03/09/2016	1 / 2	active

Here we have a different set up and filters, click on the top of the filter cascade...

My Samples | My Libraries | My Analyses | Publications | Inova Adams Oliver trio [x] Settings Feedback

Filter Cascade + ? 1 Summary | Variants | Genes | Groups/Complexes | Pathways | Processes | Diseases | Overview Copy Share Publish

Starting variant set limited by pre-filtering [show details](#)

Filter	Count	Genes
Confidence	44445	13631
Common Variants	3554	2920
Predicted Deleterious	1040	881
Genetic Analysis	52	36
Phenotype-Driven Ranking	12	6

☒ Recalculate when filters change Add Filter

Sample Legend [hide]

Gene Function: loss, normal, gain

Confident Call: No, Yes

- Identical to Reference Genome
- Heterozygous Variant
- Heterozygous/Ambiguous
- Homozygous Variant
- Copy Number Gain/Heterozygous
- Copy Number Gain/Homozygous
- Hemizygous
- Nullizygous
- Gene Fusion
- No genotype

Chr...	Position	Gene Region	Gene Symbol	Protein Variant	Variant Findings	Case Samples	Control Samples	Translation Impact	SIFT Functio...	SIFT Sc...	PolyPhe...	Rec
1	69081	Promoter	OR4F17 (inclu			-	-					
1	69270	Exonic	OR4F17 (inclu	p.S60S		-	-	synonymous				
1	69511	Exonic	OR4F17 (inclu	p.T141A	1	-	-	missense	Activating	0.68	Benign	
1	69675	Exonic	OR4F17 (inclu	p.N195K		-	-	missense	Damaging	0.01	Probably	
1	69761	Exonic	OR4F17 (inclu	p.D224V		-	-	missense	Tolerated	0.12	Benign	
1	69847	Exonic	OR4F17 (inclu	p.W253R		-	-	missense	Tolerated	0.49	Probably	
1	69897	Exonic	OR4F17 (inclu	p.S269S		-	-	synonymous				
1	865694	Exonic	SAMD11	p.H78Y		-	-	missense			Possibly I	
1	871215	Exonic	SAMD11	p.P123P		-	-	synonymous				
1	877782	Intronic	SAMD11			-	-					
1	877831	Exonic	SAMD11	p.R343R		-	-	synonymous				
1	878314	Exonic	SAMD11	p.G480G		-	-	synonymous				
1	881627	Exonic	NOC2L	p.L615L		-	-	synonymous				
1	883625	Intronic	NOC2L			-	-					
1	887801	Exonic	NOC2L	p.T394T		-	-	synonymous				
1	888639	Exonic	NOC2L	p.E306E		-	-	synonymous				
1	888659	Exonic	NOC2L	p.V300V	2	-	-	synonymous				
1	889158	Intronic	NOC2L			-	-					
1	889159	Intronic	NOC2L			-	-					
1	897325	Exonic	KLHL17	p.A203A		-	-	synonymous				
1	899928	Intronic	KLHL17			-	-					
1	902128	Exonic	PLEKHN1	p.A43V	17	-	-	missense	Damaging	0.03	Benign	EN
1	906272	Exonic	PLEKHN1	p.A166A		-	-	synonymous				
1	909238	Exonic	PLEKHN1	p.R452P, p.R487		-	-	missense	Tolerated	0.28	Benign	
1	909242	Exonic	PLEKHN1	p.G453G, p.G48		-	-	synonymous				
1	911595	Exonic	PERM1	p.V683A, p.V777		-	-	missense	Tolerated	0.06		
1	914333	Exonic	PERM1	p.E599Q, p.E69		-	-	missense	Tolerated	0.58		
1	914852	Exonic	PERM1	p.Q426E, p.Q52		-	-	missense	Activating	1.00		
1	914876	Exonic	PERM1	p.S418G, p.S512	1	-	-	missense	Damaging	0.05		
1	914940	Exonic	PERM1	p.A396A, p.A490		-	-	synonymous				

While the same principles are used as with the single sample here we can use the parental samples to detect *de novo* variants in the child...

Filter

Genetic Analysis

Rename

Use recommended settings for: (Custom) Dominant variants Set

☐ Pair/match samples from the same family ⓘ
☐ Restrict to transmitted variants

☒ Case Samples

Keep only variants which are

☒ associated with gain of function
To control specific gain of function types, use the Predicted Deleterious filter

OR

☒ Homozygous
☒ Compound Heterozygous
☒ Haploinsufficient
☒ Hemizygous
☐ Nullizygous

☒ Het-ambiguous
☒ Heterozygous

AND

the genotypes selected above occur in
 at least 1 of the 1 case samples (100%)
 at variant level

☒ Control Samples

Exclude variants which are

☒ associated with gain of function
To control specific gain of function types, use the Predicted Deleterious filter

OR

☒ Homozygous
☒ Compound Heterozygous
☒ Haploinsufficient
☒ Hemizygous

☒ Het-ambiguous
☒ Heterozygous

AND

the genotypes selected above occur in
 at least 1 of the 2 control samples (50%)
 at variant level

Apply

After the genetic analysis filter only variants unique to the proband remain

My Samples | My Libraries | My Analyses | Publications | Inova Adams Oliver trio [x]

Summary | Variants | Genes | Groups/Complexes | Pathways | Processes | Diseases | Overview Copy

Filter Cascade show details

Starting variant set limited by pre-filtering

Filter	Count
Starting variant set	45986
Confidence	44445
Common Variants	3554
Predicted Deleterious	1040
Genetic Analysis	52
Phenotype-Driven Ranking	12

☒ Recalculate when filters change Add Filter

Sample Legend hide

Gene Function: loss, normal, gain

Confident Call: No, Yes

- Identical to Reference Genome
- Heterozygous Variant
- Heterozygous/Ambiguous
- Homozygous Variant
- Copy Number Gain/Heterozygous
- Copy Number Gain/Homozygous
- Hemizygous
- Nullizygous
- Gene Fusion
- No genotype

Chr...	Position	Gene Region	Gene Symbol	Protein Variant	Variant Findings	Case Samples	Control Samples	Translation Impact	SIFT Functio...	SIFT :
1	17085590	Exonic, ncRNA	LOC10272456	p.Q376_R377de		1	--	in-frame		
1	248637199	Exonic	OR2T3/OR2T3	p.F183S	1	1	--	missense	Damaging	0.00
2	20867122	Exonic	GDF7	p.G45_G50dup		1	--	in-frame		
2	47641559	Splice Site	MSH2		4	1	--			
2	179301046	Exonic, Intronic	LOC10192702			1	--			
3	75786743	Exonic, Intronic	ZNF717			1	--			
3	75787198	Exonic, Intronic	ZNF717			1	--			
3	75787219	Exonic, Intronic	ZNF717			1	--			
3	121351218	Exonic	HCLS1			1	--			
3	195506136	Exonic, Intronic	MUC4			1	--			
3	195509006	Exonic, Intronic	MUC4			1	--			
3	195511369	Exonic, Intronic	MUC4			1	--			
3	195512534	Exonic, Intronic	MUC4			1	--			
3	195514715	Exonic, Intronic	MUC4			1	--			
4	15004878	Exonic	CPEB2	p.P201dup	25	1	--	in-frame		
5	140563921	Exonic	PCDHB16	p.S596*	2	1	--	stop gain		
5	173035295	3'UTR, Exonic	BOD1			1	--			
6	136582252	Exonic, Intronic	BCLAF1			1	--			
6	136599906	Exonic, Intronic	BCLAF1			1	--			
6	168376961	Exonic	HGC6.3	p.A125fs*4		1	--	frameshift		
7	100550614	Exonic, Intronic	MUC3A			1	--			
7	100550694	Exonic, Misma	MUC3A			1	--			
7	100550862	Exonic	MUC3A			1	--			
7	100550891	Exonic	MUC3A			1	--			
7	100550916	Exonic	MUC3A			1	--			
7	100551020	Exonic	MUC3A			1	--			
7	117188841	Exonic	CFTR	p.L454del	9	1	--	in-frame		
8	144940774	Exonic	EPPK1			1	--			
9	139399861	Exonic	NOTCH1	p.C1496Y	5	1	--	missense	Damaging	0.00
10	135438960	Exonic	FRG2/FRG2B	p.R160fs*5	2	1	--	frameshift		

Refine this further using phenotype information...

Filter

Phenotype-Driven Ranking

Rename

Ranks candidate genes by computing semantic similarity between supplied phenotypes and known disease-gene associations.

Enter and select term

- × syndactyly
- × brachydactyly
- × oligodactyly
- × aplasia cutis congenita
- × megalencephaly-cutis marmorata telangiectatica congenita (megalencephaly-capillary malformati...

[Upload file with HPO IDs...](#)

Apply

The result is a list of variants prioritized by the relationship with the patients phenotype

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Filter Cascade

Starting variant set limited by pre-filtering
[show details](#)

Variants	Genes
45986	13736

↓

× Confidence
44445
13631

↓

× Common Variants
3554
2920

↓

× Predicted Deleterious
1040
881

↓

× Genetic Analysis
52
36

↓

× Phenotype-Driven Ranking
12
6

☒ Recalculate when filters change

[Add Filter](#)

[Summary](#) | [Variants](#) | [Genes](#) | [Groups/Complexes](#) | [Pathways](#) | [Processes](#) | [Diseases](#) | [Overview](#)

[Add Filter](#)

Top 36 results

Disease	Gene	Caus	Transcript Variant	Classification	MOI	Case Samp	Control Samples	Score	Score Breakdown by Phenotype
Adams-Oliver syndrome type 5	NOTCH1	Yes	c.4487G>A	Uncertain Sig	domin	■	--	1.96	
Adams-Oliver syndrome	NOTCH1	Yes	c.4487G>A	Likely Pathog		■	--	1.85	
Adams-Oliver syndrome type 1	NOTCH1	No	c.4487G>A	Uncertain Sig	domin	■	--	1.85	
hypertrichotic osteochondrodys	MUC3A	No	unknown	Uncertain Sig	domin	■	--	1.69	
hypertrichotic osteochondrodys	MUC3A	No	unknown	Uncertain Sig	domin	■	--	1.69	
hypertrichotic osteochondrodys	MUC3A	No	unknown	Uncertain Sig	domin	■	--	1.69	
Fanconi anemia	AR	No	c.234_239delGCGAGCA	Uncertain Sig	recess	■	--	1.69	
hypertrichotic osteochondrodys	MUC3A	No	unknown	Uncertain Sig	domin	■	--	1.69	
hypertrichotic osteochondrodys	MUC3A	No	unknown	Uncertain Sig	domin	■	--	1.69	
Fanconi anemia	AR	No	c.1409_1420delGCGG	Likely Benigr	recess	■	--	1.69	
Down syndrome	NOTCH1	No	c.4487G>A	Uncertain Sig		■	--	1.68	
geleophysic dysplasia	LTBP3	No	c.103_105dupCTG; c.-;	Benign		■	--	1.42	
conotruncal heart malformation:	NOTCH1	No	c.4487G>A	Uncertain Sig		■	--	1.35	
Marfan syndrome	NOTCH1	No	c.4487G>A	Uncertain Sig	domin	■	--	1.05	
Turner syndrome	AR	No	c.234_239delGCGAGCA	Uncertain Sig		■	--	0.99	
Turner syndrome	AR	No	c.1409_1420delGCGG	Uncertain Sig		■	--	0.99	
tetralogy of Fallot	NOTCH1	No	c.4487G>A	Uncertain Sig		■	--	0.86	
acromicric dysplasia	LTBP3	No	c.103_105dupCTG; c.-;	Benign	domin	■	--	0.86	
amyloidosis	AR	No	c.1409_1420delGCGG	Uncertain Sig		■	--	0.85	
amyloidosis	NOTCH1	No	c.4487G>A	Uncertain Sig		■	--	0.85	
amyloidosis	AR	No	c.234_239delGCGAGCA	Uncertain Sig		■	--	0.85	
aortic valve disease type 1	NOTCH1	Yes	c.4487G>A	Uncertain Sig		■	--	0.56	
hypoplastic left heart syndrome	NOTCH1	No	c.4487G>A	Uncertain Sig		■	--	0.56	
Turcot syndrome	MSH2	Yes	c.744+2delT; c.942+2d	Pathogenic	recess	■	--	0.35	
cryptorchidism	AR	No	c.234_239delGCGAGCA	Uncertain Sig		■	--	0.35	

Sample Legend [hide]

Gene Function

loss normal gain

Identical to Reference Genome

Confident Call

No Yes

■ ■

Can view the key genes and variants...

Filter Cascade

Starting variant set limited by pre-filtering
[show details](#)

Variants	Genes
45986	13736

↓

× Confidence

Variants	Genes
44445	13631

↓

× Common Variants

Variants	Genes
3554	2920

↓

× Predicted Deleterious

Variants	Genes
1040	881

↓

× Genetic Analysis

Variants	Genes
52	36

↓

× Phenotype-Driven Ranking

Variants	Genes
12	6

☒ Recalculate when filters change

Add Filter

Summary | Variants | **Genes** | Groups/Complexes | Pathways | Processes | Diseases | Overview
Copy **Share** **Publis**

Edit Columns **Export** **Create List**

Search gene, chr, or db SNP



12 variants

Chr...	Position	Gene Region	Gene Symbol	Protein Variant	Variant Findings	Case Samples	Control Samples	Translation Impact	SIFT Functio...	SIFT Sc...	PolyPhe...	Re
2	47641559	Splice Site	MSH2		4	■	--					Sp
7	100550614	Exonic, Intron	MUC3A			■	--					
7	100550694	Exonic, Misma	MUC3A			■	--					
7	100550862	Exonic	MUC3A			■	--					
7	100550891	Exonic	MUC3A			■	--					
7	100550916	Exonic	MUC3A			■	--					
7	100551020	Exonic	MUC3A			■	--					
7	117188841	Exonic	CFTR	p.L454del	9	■	--	in-frame				
9	139399861	Exonic	NOTCH1	p.C1496Y	5	■	--	missense	Damaging	0.00	Probably	
11	65325325	5'UTR, Exonic	LTBP3	p.L35dup		■	--	in-frame				
X	66765159	Exonic	AR	p.Q79_Q80del	4	■	--	in-frame				
X	66766357	Exonic	AR	p.G470_G473de	1	■	--	in-frame				

Loading your own data...

My Samples | My Libraries | My Analyses | Publications | Inova Adams Oliver trio [×]

Summary | Variants | Genes | Groups/Complexes | Pathway

Filter Cascade  

Starting variant set limited by pre-filtering
[show details](#)

Variants	Genes
45986	13736

↓

× Confidence	
44445	13631

⌋

Chr...	Position	Gene Region	Gene Symbol	Protein
2	47641559	Splice Site	MSH2	
7	100550614	Exonic, Intronic	MUC3A	
7	100550694	Exonic, Mismatch	MUC3A	
7	100550862	Exonic	MUC3A	
7	100550891	Exonic	MUC3A	

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There are two options for large files the datastream is better...

1 Upload Sample

2 Saved

×

Upload file(s) for one sample. ⓘ

Supported formats: [VCF](#), [GVF](#), [Complete Genomics](#)

Barcode *

Display Name *

Subject ID

Description

Files * [Select sample file\(s\)...](#)

Have many samples to upload?
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Save

Just drag and drop your files...

Drag or select your data files and wait until all uploads are complete.

Having trouble? [Click here](#) to use previous version.

- Supported file formats: [VCF](#), [GVF](#), [Complete Genomics](#)
- If sample annotations should be applied in batch along with sample upload (e.g. familial relationships, clinical traits), please upload a manifest file along with sample VCFs. Download and edit our manifest template file [here](#)
- If a single sample draws from multiple VCF files, please bundle all VCF and manifest files into an archive file (zip, bz, bz2, gz) before uploading
- If there are any data format problems, QIAGEN Support will contact you to resolve them.


Upload files
Drag a file or click to browse

Sharing samples, ideal if you want to create your own account

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[Open](#)

Name	Description	From	Created ▼	Samples	Status
Test WEX		variant_analysis1@i	05/31/2017	1 / 0	active
Inova Adams Oliver trio	Inova Adams Oliver trio (Workshop). The Inova NOTCH1 Case study is a...	ruth.burton@qiagen.	05/25/2017	1 / 2	active
Test WES	WES somatic variants from pancreatic cancer	variant_analysis1@i	05/24/2017	1 / 0	active
hcc tn2		variant_analysis1@i	02/22/2017	2 / 2	active
triple negative		variant_analysis1@i	02/22/2017	2 / 2	active

Sharing samples, ideal if you want to create your own account

1
Select Recipients

2
Shared

X

Who are the recipients?
Sharing allows colleagues to copy analyses into their private workspace. Once shared, the data can not be recalled. Enter email addresses for recipients.

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Message

Analyses

Name ▲	Description	Samples
Test WEX		1 / 0

Any questions?

Come and visit us at our stand
Join us at our presentation on Wednesday

ruth_burton@qiagen.com

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