

Présentation générale



Version démo, release 10/21


Login

EIIA

DOCUMENTATION

LOGIN

LOGIN CHANGE PASSWORD

USERNAME 

PASSWORD

LOG IN

ANALYSIS NAME

COMMENT TEXT

DATE REQUESTED

HTS

SANGER

NORMAL

HIGH

URGENT

RESET FILTER

ANALYSES

VARIANTS

IMPORT

SEARCH

Recherche et
filtre des analyses

NOT READY 0

NO ANALYSES ARE NOT READY

Infobulle QC (regions
low coverage)

YOUR ANALYSES 1

2022-08-09 brca_sample_2.HBOC_v01

WARNING

HTS

1 • Interpretation • C. Collett • 2022-08-09 11:59
2 • Interpretation • C. Collett • 2022-08-09 12:00
3 • Interpretation • C. Collett • 2022-08-09 12:01
4 • Review (Ongoing) • H. Ibsen • 2022-08-10 11:38

Historique des
interprétations

Type de séquençage

INTERPRETATION 7

2022-08-09 brca_long_variants.HBOCUTV_v01

HTS

2022-08-09 brca_sample_1.HBOCUTV_v01

HTS

2022-08-09 brca_sample_1.HBOC_v01

WARNING

HTS

2022-08-09 brca_sample_2.HBOCUTV_v01

HTS

2022-08-09 brca_sample_3.HBOCUTV_v01

HTS

2022-08-09 brca_sample_allfiltered.HBOC_v01

WARNING

HTS

2022-08-09 brca_sample_master.HBOCUTV_v01

HTS

Overview

Équivalent du
dashboard

REVIEW 0

NO ANALYSES PENDING REVIEW.

MEDICAL REVIEW 0

NO ANALYSES PENDING MEDICAL REVIEW.

OTHERS' ANALYSES 0

OTHERS HAVE NO ANALYSES

FINALIZED 0

NO FINALIZED ANALYSIS

PREVIOUS 1 NEXT

Overview

Menu variations

OVERVIEW EIIA DOCUMENTATION H. IBSEN

VARIANTS (highlighted in red)

▶ SEARCH

▼ YOUR VARIANTS 0
YOU HAVE NO VARIANTS

▼ INTERPRETATION 6

2022-08-09	BRCA2 c.10G>T (p.Gly4Ter)	NEW	HBOC_v01
2022-08-09	BRCA2 c.51_52del (p.Arg18LeufsTer12)	NEW	HBOC_v01
2022-08-09	BRCA2 c.67*2T>A	NEW	HBOC_v01
2022-08-09	BRCA2 c.72A>T (p.Leu24Phe)	NEW	HBOC_v01
2022-08-09	BRCA2 c.97G>T (p.Glu33Ter)	NEW	HBOC_v01
2022-08-09	BRCA2 c.198A>G (p.Gln66=)	NEW	HBOC_v01

▼ PENDING REVIEW 0
NO VARIANTS PENDING REVIEW

▼ OTHERS' VARIANTS 0
OTHERS HAVE NO VARIANTS

▼ FINALIZED 0
NO FINALIZED VARIANTS

PREVIOUS 1 NEXT

Overview Import

OVERVIEW EIIA DOCUMENTATION H. IBSEN

ANALYSES

SEARCH

TYPE **VARIANTS** ANALYSES

HGVSc/GENOMIC e.g. c.123A>G or 13:100-300

GENE SEARCH GENE

USER SEARCH USER

ACTIVE IMPORTS 0

THERE ARE CURRENTLY NO ACTIVE IMPORTS.
TO CREATE A NEW ONE, USE 'NEW IMPORT' SECTION BELOW.

IMPORT HISTORY 0

PREVIOUS 1 NEXT

NEW IMPORT

IMPORT SOURCE **VARIANTS** SAMPLE REPOSITORY

Paste variant data in any of the following formats:

- Full HGVSc coordinates
- Genomic position
- VCF file
- SeqPilot export file

PASTE VARIANT DATA HERE

You can batch add multiple imports by using lines with the character "-" as separators. Note that all data in the same batch must be in the same format.

Batch example

+ PARSE DATA

▶ SEARCH

▼ NOT READY 0

NO ANALYSES ARE NOT READY

▼ YOUR ANALYSES 1

2022-08-09 brca_sample_2.HBOC_v01 WARNING HTS

1 • Interpretation • C. Collett • 2022-08-09 11:59
2 • Interpretation • C. Collett • 2022-08-09 12:00
3 • Interpretation • C. Collett • 2022-08-09 12:01
4 • Review (Ongoing) • H. Ibsen • 2022-08-10 11:38

▼ INTERPRETATION 7

2022-08-09 brca_long_variants.HBOCUTV_v01 HTS2022-08-09 brca_sample_1.HBOCUTV_v01 HTS2022-08-09 brca_sample_1.HBOC_v01 WARNING HTS2022-08-09 brca_sample_2.HBOCUTV_v01 HTS2022-08-09 brca_sample_3.HBOCUTV_v01 HTS2022-08-09 brca_sample_allfiltered.HBOC_v01 WARNING HTS2022-08-09 brca_sample_master.HBOCUTV_v01 HTS

Ouverture d'une analyse

▼ REVIEW 0

NO ANALYSES PENDING REVIEW.

▼ MEDICAL REVIEW 0

NO ANALYSES PENDING MEDICAL REVIEW.

▼ OTHERS' ANALYSES 0

OTHERS HAVE NO ANALYSES

▼ FINALIZED 0

NO FINALIZED ANALYSIS

INDICATIONS COMMENT

+ 0/0 FILTER DEFAULT FULL QUICK VISUAL

UNCLASSIFIED VARIANTS (8)

INH	GENE	HGVSc	CSQ	!	S	O	Q	R	CLASS
AD/AD;...	BRCA2	c.97delins(15)	frameshift						
AD/AD;...	BRCA2	c.292_305del	frameshift						
AD/AD;...	BRCA2	c.475+3_475...	intron						
AD/AD;...	BRCA2	c.583_595dup	frameshift						
AD/AD;...	BRCA2	c.682-2A>C	splice_acceptor						R
AD/AD;...	BRCA2	c.925dup	frameshift						R
AD/AD;...	BRCA2	c.1233dup	frameshift						R
AD/AD;...	BRCA2	c.1444del	frameshift						R

CLASSIFIED VARIANTS (0)

NOT RELEVANT VARIANTS (0)

TECHNICAL VARIANTS (0)

Liste de variations retenues

Détails de la variation c.97delins(15)

ANALYSIS SPECIFIC FOR VARIANT VERIFIED TECHNICAL NOT RELEVANT

ANALYSIS-SPECIFIC-COMMENTS

QUALITY

Proband **NEEDS VERIFICATION**

Filter: PASS
 Quality: 5000
 GQ: 99
 Depth: 187
 Ratio: 0.43
 ~ REF (G): 107
 ~ (15): 80

CLASSIFICATION SELECT CLASS

EVALUATION

REPORT

ACMG

SUGGESTED

SHOW REQ

ACMG SUGGESTED

PVS1 Null variant PM2 SUPPORTIVE Absent from controls

REGION

REGION-COMMENTS

VARDB SNV

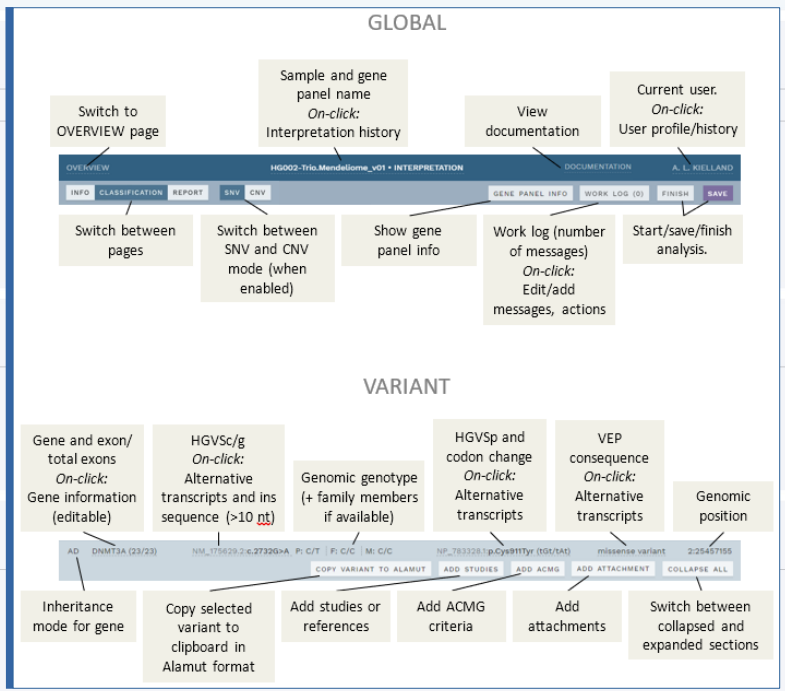
FREQUENCY

FREQUENCY-COMMENTS

GNOMAD EXONES
 GNOMAD GENOMES
 EAC
 INDB
 DBSNP

Écran d'ANALYSE

Équivaut à un mode read-only



Gène (exon/total)

Protéine et codon touché

BRCA2 (3/27)

NM_000059.3:c.97delins(15) G/TACCCTTATAATGAC

NP_000050.2:p.Glu33TyrfsTer52 (Gaa/(15)aa)

GENE PANEL INFO

WORK LOG (0)

START INTERPRETATION

frameshift variant

13:32893243

COPY VARIANT TO ALAMUT

COLLAPSE ALL

AD/AD:AR/AD:SMu/AR

BRCA2 (3/27)

NM_000059.3:c.198A>G A/G

GENE INFO

LINKS

HGNC • ClinGen, HGMD, OMIM, PanelApp • ACMG Incidental

INHERITANCE

AD/AD:AR/AD:SMu/AR

PHENOTYPES

Fanconi anemia, complementation group D1 (AR)

Wilms tumor (AD:SMu)

Breast cancer, male, susceptibility to (AD)

Breast-ovarian cancer, familial, 2 (AD)

(Glioblastoma 3) (AR)

(Medulloblastoma) (AD:AR)

(Pancreatic cancer 2) (O)

(Prostate cancer) (AD)

BS1/BA1 CUTOFF

External: 0.0005/0.008 (overridden)

Internal: 0.0005/0.008 (overridden)

GENE COMMENTS

EDIT

NM_000059.3:c.51_52del AC/-

HGVSc

NM_000059.3(BRCA2):c.51_52del

NM_001136571.1(ZAR1L):

NM_001136571.2(ZAR1L):

PVS1 Null

NP_000050.2:p.Arg18LeufsTer12 (ACa/a)

PROTEIN

NP_000050.2(BRCA2):p.Arg18LeufsTer12

Transcrits alternatifs

GENE PANEL INFO

WORK LOG (1)

FINISH

SAVE

OVERVIEW OPTIONS

OVERVIEW COMMENT

PRIORITY: HIGH

EVENTS

ALL EVENTS

MESSAGES ONLY

SYSTEM 2022-08-09 11:28

PRIORITY SET TO NORMAL

HENRIK IBSEN 13:49

à faire en priorité !

HENRIK IBSEN 13:30

PRIORITY SET TO HIGH

Historique de l'analyse

NEW MESSAGE

ADD MESSAGE

GENE PANEL INFO

INFO:

Panel: HBOCUTV_v01

Genes: 7

Transcripts: 7

GENE LIST:

COPY ALL

COPY ALL WITH TRANSCRIPTS

FILTER

BRCA1

BRCA2

CDH1

PALB2

PTEN

STK11

TP53

Les analyses sur ELLA reposent sur des panels de gènes (quitte à inclure l'ensemble de l'exome)

5 MOST SIMILAR GENE PANELS:

NAME	ADDITIONAL	OVERLAPPING	MISSING
HBOC_v01	0	2	5

Showing BRCA1 to TP53

PREVIOUS

1

NEXT

FREQUENCY

FREQUENCY-COMMENTS

GNOMAD EXONES

GNOMAD GENOMES

EMAC

INDB

DBSNP

INFO CLASSIFICATION REPORT

AD/AD;AR/AD;SMu/AR BRCA2 (3/27) NM_000059.3:c.97delins(15) G/TACCCCTATAATGAC

INDICATIONS COMMENT

+ 0/1958 FILTER TRIO DEFAULT FULL QUICK VISUAL

UNCLASSIFIED VARIANTS (8)

INH	GENE	HGVSc	CSQ	!	S	O	Q	R	CLASS
AD/AD;...	BRCA2	c.97delins(15)	frameshift						
AD/AD;...	BRCA2	c.292_305del	frameshift						
AD/AD;...	BRCA2	c.475+3_475...	intron						
AD/AD;...	BRCA2	c.583_595dup	frameshift						
AD/AD;...	BRCA2	c.682-2A>C	splice_acceptor					R	
AD/AD;...	BRCA2	c.925dup	frameshift					R	
AD/AD;...	BRCA2	c.1233dup	frameshift					R	
AD/AD;...	BRCA2	c.1444del	frameshift					R	

CLASSIFIED VARIANTS (0)

NOT RELEVANT VARIANTS (0)

TECHNICAL VARIANTS (0)

SUC PVS1 Null variant PM2 SUPPORTIVE Absent from controls

REGION

REGION-COMMENTS

VARDB SHV

FREQUENCY

FREQUENCY-COMMENTS

GNOMAD EXONES
GNOMAD GENOMES
EAC
INDB
DBSNP

PREDICTION

ADD PREDICTION

PREDICTION-COMMENTS

CONSEQUENCE frameshift variant
SPLICEAI
OTHER

EXTERNAL

ADD EXTERNAL DB

EXTERNAL DB-COMMENTS

CLINVAR
HGMD PRO
OTHER

STUDIES & REFERENCES

STUDIES-COMMENTS

View (and optionally add back) filtered variants | Switch between filter configurations | Indications comment, mirrored on REPORT page

Inheritance (gene panel) | HGVS for default transcript | Consequence (VEP), LOF in red | Switch between modes of view

INDICATIONS COMMENT

+ 0/1958 FILTER TRIO DEFAULT FULL QUICK VISUAL

UNCLASSIFIED VARIANTS (11)

INH	GENE	HGVSc	CSQ	!	S	O	Q	R	CLASS
AD	CCND2	c.196-13A>C	intron		D				
AD	DNMT3A	c.2732G>A	missense		D				
XR	GRIA3	c.1181G>A	missense		X	O			
AD/AR ...	TTN TTN	c.34474C>A ...	missense in...		C				
AD/AR ...	TTN TTN	c.104251G>C ...	missense mi...		C			R	
AD	NKX2-5	c.61G>C	missense					R	
AD	VSKI	c.432C>G	missense					Q	R
AR	CBS	c.1105C>T	missense					R	
AR	GBE1	c.1134T>G	missense					R	
AR	SLC12A3	c.965C>T	missense,spl...					R	
AR;AD	SLC5A2	c.1961A>G	missense					R	

CLASSIFIED VARIANTS (6)

INH	GENE	HGVSc	CSQ	!	S	O	Q	R	CLASS
AD/AR	F11	c.403G>T	stop_gained					R	4* → 5
XR	AR	c.228_239dup	inframe_inse...		X	O	Q		→ 3
AD/AR	TMC1	c.2261-17_22...	intron		D		Q		→ 2
AR	ATM	c.295A>G	missense					R	3 → 2
AR	CFTR	c.3705T>G	missense					R	→ 2
XD	AMER1	c.1873A>G	missense		X	O		R	→ 2

NOT RELEVANT VARIANTS (1)

INH	GENE	HGVSc	CSQ	!	S	O	Q	R	CLASS
AR	ABCG5	c.293C>G	missense					R	

TECHNICAL VARIANTS (4)

INH	GENE	HGVSc	CSQ	!	S	O	Q	R	CLASS
AD/AR	GRHL2	c.1746C>A	stop_gained		I	D		T	
AD/AR	GRHL2	c.1754G>A	missense		I	D		T	
AD/AR	GRHL2	c.1755C>A	missense		I	D		T	
AD/AR	PMS2	c.1688G>A	missense		I			T	R

Currently viewed variant

Tags:
! = Variant warnings
F = Filtered variants (if added)
S = Segregation
O = Homozygous
Q = Quality issues
R = Reference available

Shading: >1 non-filtered variant in gene

Indicators for added ACMG criteria (show list with mouse-over)

Previously classified (outdated: *), new class S set in this analysis

New classification set, no existing class

Variants marked as not relevant

Variants marked as technical artefacts

Marked as reviewed/finalized

GENE PANEL INFO WORK LOG (0) START INTERPRETATION

frameshift variant 13:32893243

COPY VARIANT TO ALAMUT COLLAPSE ALL

Un clic ajoute l'URL de la db dans le presse-papier ou ouvre directement un onglet selon la configuration

EXTERNAL ADD EXTERNAL DB

EXTERNAL DB-COMMENTS

CLINVAR Review status: ★★☆☆ (reviewed by expert panel)
Submissions:
N/A - Pathogenic - Breast-ovarian cancer, familial 2 - BIC (BRCA2)
2016-09-08 - Pathogenic - not specified - ENIGMA
2015-10-02 - Pathogenic - not specified - CIMBA

HGMD PRO DM Breast cancer C101425

OTHER

INDICATIONS COMMENT

+ 0/0 FILTER DEFAULT **QUICK** VISUAL

UNCLASSIFIED VARIANTS (8)

INH	GENE	HGVSc	CSQ	!	S	O	Q	R	QUAL	DP	RATIO	HI.FREQ	HI.COUNT	EXTERNAL	CLASS
AD/AD;...	BRCA2	c.97delins(15)	frameshift						5000	187	0.43	-	-		T NR NP 2 BS1 BS2
AD/AD;...	BRCA2	c.292_305del	frameshift						5000	187	0.43	-	-		T NR NP 2 BS1 BS2
AD/AD;...	BRCA2	c.475+3_475...	intron						5000	187	0.43	-	-		T NR NP 2 BS1 BS2
AD/AD;...	BRCA2	c.583_595dup	frameshift						5000	187	0.43	-	-		T NR NP 2 BS1 BS2
AD/AD;...	BRCA2	c.682-2A>C	splice_acceptor					R	5000	187	0.43	-	-	HGMD Clinvar	T NR NP 2 BS1 BS2
AD/AD;...	BRCA2	c.925dup	frameshift					R	5000	187	0.43	-	-	HGMD Clinvar	T NR NP 2 BS1 BS2
AD/AD;...	BRCA2	c.1233dup	frameshift					R	5000	187	0.43	-	-	HGMD Clinvar	T NR NP 2 BS1 BS2
AD/AD;...	BRCA2	c.1444del	frameshift					R	5000	187	0.43	-	-	HGMD Clinvar	T NR NP 2 BS1 BS2

CLASSIFIED VARIANTS (0)

NOT RELEVANT VARIANTS (0)

TECHNICAL VARIANTS (0)

Lancer l'interprétation

INDICATIONS COMMENT

+ 0/0 FILTER DEFAULT **VISUAL**

UNCLASSIFIED VARIANTS (8)

INH	GENE	HGVSc	CSQ	!	S	O	Q	R	QUAL	RATIO	CLASS
AD/AD;...	BRCA2	c.97delins(15)	frameshift						5000	0.43	T
AD/AD;...	BRCA2	c.292_305del	frameshift						5000	0.43	T
AD/AD;...	BRCA2	c.475+3_475...	intron						5000	0.43	T
AD/AD;...	BRCA2	c.583_595dup	frameshift						5000	0.43	T
AD/AD;...	BRCA2	c.682-2A>C	splice_acceptor					R	5000	0.43	T
AD/AD;...	BRCA2	c.925dup	frameshift					R	5000	0.43	T
AD/AD;...	BRCA2	c.1233dup	frameshift					R	5000	0.43	T
AD/AD;...	BRCA2	c.1444del	frameshift					R	5000	0.43	T

CLASSIFIED VARIANTS (0)

NOT RELEVANT VARIANTS (0)

TECHNICAL VARIANTS (0)

TRACK SELECTION

PRESETS: DEFAULT TEST 1 TEST 2 TEST 3

GLOBAL TRACKS: REFGENE GENEPANEL CLASSIFICATIONS

GROUP TRACKS: VARIANTS

ANALYSIS TRACKS: VARIANTS

IGV hg19 13 13:32,893,219-32,893,267 Q 49 bp

Cursor Guide Center Line Track Labels Save SVG

32,893,220 bp 32,893,225 bp 32,893,230 bp 32,893,235 bp 32,893,240 bp 32,893,245 bp 32,893,250 bp 32,893,255 bp 32,893,260 bp 32,893,265 bp

G G A C C A A T T A A G T C T T A A T T G G T T T G A A G A A C T T T C T T C A G A A G C T C C A C

Variants

INDICATIONS COMMENT

+ 0/0 FILTER DEFAULT FULL QUICK VISUAL

UNCLASSIFIED VARIANTS (8)

INH	GENE	HGVSc	CSQ	! S O Q R	CLASS
AD/AD;...	BRCA2	c.97delins(15)	frameshift		
AD/AD;...	BRCA2	c.292_305del	frameshift		
AD/AD;...	BRCA2	c.475+3_475...	intron		
AD/AD;...	BRCA2	c.583_595dup	frameshift		
AD/AD;...	BRCA2	c.682-2A>C	splice_acceptor		[R]
AD/AD;...	BRCA2	c.925dup	frameshift		[R]
AD/AD;...	BRCA2	c.1233dup	frameshift		[R]
AD/AD;...	BRCA2	c.1444del	frameshift		[R]

CLASSIFIED VARIANTS (0)

NOT RELEVANT VARIANTS (0)

TECHNICAL VARIANTS (0)

ANALYSIS SPECIFIC FOR VARIANT VERIFIED TECHNICAL NOT RELEVANT

ANALYSIS-SPECIFIC-COMMENTS

Écran d'interprétation

Il est maintenant possible d'ajouter une classification, une publication,...

QUALITY

Proband

NEEDS VERIFICATION

Filter: PASS

Quality: 5000

GQ: 99

Depth: 187

Ratio: 0.43

~ REF (G): 107

~ (15): 80

CLASSIFICATION SELECT CLASS FINALIZE

EVALUATION

REPORT

ACMG (SUGGESTED CLASS:)

SUGGESTED

HIDE REQ

ACMG SUGGESTED

PVS1 Null variant ADD PM2 SUPPORTIVE Absent from controls ADD

ACMG REQ

GP - last exon not important Last exon not important GP - LOF missense LOF and missense = disease R - not in last exon Not in last exon + PVS1 R - nu

R - no freq Not in controls + PM2 + PM2 SUPPORTIVE

frameshift variant

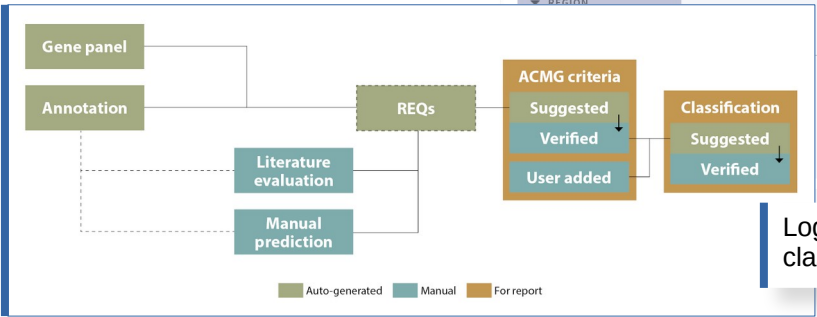
De novo (unconfirmed) ADD

Commentaire perso

B / U M A H1 H2 P Tx

SHOW: PATHOGENIC BENIGN OTHER

PVS1	Null variant
PS1	Known pathogenic aa
PS2	De novo (confirmed)
PS3	Functional damage
PS4	Increased prevalence in patients
PM1	Functional domain
PM2	Absent from controls
PM3	In trans pathogenic & AR
PM4	In-frame/stop-loss
PM5	Novel at known pathogenic aa
PM6	De novo (unconfirmed)
PP1	Cosegregation
PP2	Missense: important
PP3	Predicted pathogenic
PP4	Phenotype: single gene
PP5	Reported pathogenic, evidence unavailable



Logiciel focalisé sur la classification ACMG

INDICATIONS COMMENT

+ 0/0 FILTER DEFAULT FULL QUICK VISUAL

UNCLASSIFIED VARIANTS (7)

INH	GENE	HGVSc	CSQ	!	S	O	Q	R	CLASS
AD/AD;...	BRCA2	c.292_305del	frameshift						
AD/AD;...	BRCA2	c.475+3_475...	intron						
AD/AD;...	BRCA2	c.583_595dup	frameshift						
AD/AD;...	BRCA2	c.682-2A>C	splice_acceptor						R
AD/AD;...	BRCA2	c.925dup	frameshift						R
AD/AD;...	BRCA2	c.1233dup	frameshift						R
AD/AD;...	BRCA2	c.1444del	frameshift						R

CLASSIFIED VARIANTS (1)

INH	GENE	HGVSc	CSQ	!	S	O	Q	R	CLASS
AD/AD;...	BRCA2	c.97delins(15)	frameshift						→ 4

NOT RELEVANT VARIANTS (0)

TECHNICAL VARIANTS (0)

Classification appliquée

ANALYSIS SPECIFIC FOR VARIANT VERIFIED TECHNICAL NOT RELEVANT

ANALYSIS-SPECIFIC-COMMENTS

QUALITY

Proband

NEEDS VERIFICATION

Filter: PASS

Quality: 5000

GQ: 99

Depth: 187

Ratio: 0.43

~ REF (G): 107

~ (15): 80

Note pense-bête ou destinée à un relecteur

CLASSIFICATION CLASS 4 FINALIZE

Commentaire sur l'évaluation perso à inclure dans le rapport

Commentaire automatiquement ajouté au rapport

ACMG (SUGGESTED CLASS: 4)

PVS1 Null variant REMOVE - PM2 + Absent from controls REMOVE

PVS1-COMMENT PM2-COMMENT

SUGGESTED

HIDE REQ

ACMG SUGGESTED

PVS1 Null variant ADD PM2 SUPPORTIVE Absent from controls ADD

ACMG REQ

GP - last exon not important Last exon not important GP - LOF missense LOF and missense = disease R - not in last exon Not in last exon + PVS1 R - null variant Null variant + PVS1

R - no freq Not in controls + PM2 + PM2 SUPPORTIVE

Évaluation ACMG

REGION

REGION-COMMENTS

VARBB SNV

FREQUENCY

FREQUENCY-COMMENTS

OVERVIEW

BRCA2 (2/27) NM_000050.2:p.Glu33TyrfsTer52 (Gaa/15)aa frameshift variant

ADD PREDICTION

ORTHOLOG CONSERVATION: CONSERVED | NON-CONSERVED

PARALOG CONSERVATION: CONSERVED | NON-CONSERVED

DNA CONSERVATION: CONSERVED | NON-CONSERVED

DOMAIN: CRITICAL FUNCTIONAL DOMAIN | CRITICAL FUNCTIONAL AMINO ACID

REPEAT: REPEAT REGION | NON-REPEAT REGION

SPICE SITE EFFECT: SPICE SITE LOST | DE NOVO SPICE SITE | NO SPICE SITE EFFECT

DOCUMENTATION H. IBSEN

GENE PANEL INFO WORK LOG (0) START INTERPRETATION

NP_000050.2:p.Glu33TyrfsTer52 (Gaa/15)aa frameshift variant 13:32893243

COPY VARIANT TO ALAMUT COLLAPSE ALL

AD/AD/...	BRCA2	c.475+3_475...	intron	
AD/AD/...	BRCA2	c.583_595dup	frameshift	
AD/AD/...	BRCA2	c.682-2A>C	splice_acceptor	R
AD/AD/...	BRCA2	c.925dup	frameshift	R
AD/AD/...	BRCA2	c.1233dup	frameshift	R
AD/AD/...	BRCA2	c.1444del	frameshift	R

CLASSIFIED VARIANTS (0)

NOT RELEVANT VARIANTS (0)

TECHNICAL VARIANTS (0)

VARDDB SNV

FREQUENCY

FREQUENCY-COMMENTS

GNOMAD EXOMES GNOMAD GENOMES EAC INDB DBSNP

EXTERNAL

BRCA EXCHANGE Visit database

LOVD IARC HCl Visit database

LOVD - SHARED BRCA2 Visit database

UMD BRCASHARE Visit database

X - BIC (NOT IN USE) Visit database

X - LOVD GENOMED CHINA (NOT IN USE) Visit database

OTHER ENTER VALUE

Ajout manuel de pathogénicité estimée selon les databases

PREDICTION

ADD PREDICTION

PREDICTION-COMMENTS

CONSEQUENCE frameshift variant

SPLICEAI OTHER

ADD STUDIES

ADD STUDIES

SEARCH PUBMED MANUAL

SEARCH PHRASE brca2

Ajout de publications concernant la variation

EXTERNAL

ADD EXTERNAL DB

EXTERNAL DB-COMMENTS

CLINVAR HGMD PRO OTHER

STUDIES & REFERENCES

ADD STUDIES SHOW IGNORED (0)

STUDIES-COMMENTS

SEARCH RESULTS

BRCA2 mutation analysis of 87 Spanish breast/ovarian cancer families. Campos B et al. (2001), Ann. Oncol. 12(12), 1699-703. ADD

Detection of BRCA1 and BRCA2 mutations in breast cancer families by a comprehensive two-stage screening procedure. Spitzer C et al. (2000), Int. J. Cancer 88(4), 474-81. ADD

BRCA1 and BRCA2 mutation status and cancer family history of Danish women affected with multifocal or bilateral breast cancer at a young age. Berghthorsson JT et al. (2003), J. Med. Genet. 38(6), 361-8. ADD

BRCA1 and BRCA2 germline mutation spectrum and frequencies in Belgian breast/ovarian cancer families. Claes K et al. (2004), Br. J. Cancer 90(5), 1244-51. ADD

BRCA2 mutation analysis of 87 Spanish breast/ovarian cancer families. Campos B et al. (2001), Ann. Oncol. 12(12), 1699-703. ADD

Detection of BRCA1 and BRCA2 mutations in breast cancer families by a comprehensive two-stage screening procedure. Spitzer C et al. (2000), Int. J. Cancer 88(4), 474-81. ADD

An improved high-throughput heteroduplex mutation detection system for screening BRCA2 mutations fluorescent mutation detection (F-MD). Edwards SM et al. (2003), Hum. Mutat. 17(3), 220-32. ADD

BRCA1 and BRCA2 mutation status and cancer family history of Danish women affected with multifocal or bilateral breast cancer at a young age. Berghthorsson JT et al. (2003), J. Med. Genet. 38(6), 361-8. ADD

Two percent of men with early-onset prostate cancer harbor germline mutations in the BRCA2 gene. Adern C et al. (2003), Cancer 97(3), 1-11. ADD

BRCA2 germline mutations in Cypriot patients with familial breast/ovarian cancer. Hadjilavros A et al. (2003), Hum. Mutat. 21(2), 171. ADD

Analysis of BRCA1 and BRCA2 genes in Spanish breast/ovarian cancer patients: a high proportion of mutations unique to Spain and evidence of founder effects. Diaz O et al. (2003), Hum. Mutat. 22(4), 301-12. ADD

BRCA2 gene a candidate for clinical testing in familial colorectal cancer type X. Carru P et al. (2015), Clin. Genet. 87(6), 562-7. ADD

BRCA1 and BRCA2 germline mutation spectrum and frequencies in Belgian breast/ovarian cancer families. Claes K et al. (2004), Br. J. Cancer 90(5), 1244-51. ADD

Identification and evaluation of 55 genetic variations in the BRCA1 and the BRCA2 genes of patients from 59 Japanese breast cancer families. Kawahara M et al. (2004), J. Hum. Genet. 45(7), 391-5. ADD

Hereditary breast and ovarian cancer in Cyprus: identification of a founder BRCA2 mutation. Hadjilavros A et al. (2004), Cancer Genet. Cytogenet. 153(2), 150-6. ADD

Prevalence of BRCA2 mutations in a hospital based series of unselected breast cancer cases. Kim SY et al. (2005), J. Med. Genet. 42(1), 45. ADD

Complete allelic analysis of BRCA1 and BRCA2 variants in young Nigerian breast cancer patients. Puchner J et al. (2005), J. Med. Genet. 42(3), 176-82. ADD

High frequency of germ-line BRCA2 mutations among Hungarian male breast cancer patients without family history. Cookay B et al. (1999), Cancer Res 59(1), 995-8. ADD

SHOWING FIRST 19 SEARCH RESULTS ONLY

AD

BRCA2 (2/27)

NM_000059.3:c.10G>T

NP_000050.2:p.Gly4Ter (Gga/Tga)

stop gained

13:32890607

COPY VARIANT TO ALAMUT

ADD STUDIES

ADD ACMG

ADD ATTACHMENT

COLLAPSE ALL

WORK LOG (0)

FINISH

SAVE

X ADD STUDIES

CANCEL

SAVE

STUDIES

MODE
Add reference by searching, extracting from pubmed,
or add manually (published or unpublished study, e.g. in-house)

SEARCH | PUBMED | MANUAL

STATUS
Is the study published?

PUBLISHED | UNPUBLISHED

AUTHORS

Authors*

TITLE

Title*

JOURNAL / BOOK

Journal/book*

VOLUME

Volume

ISSUE

Issue

YEAR

Year*

PAGES

Pages

ABSTRACT

Abstract

+ ADD REFERENCE

EXTERNAL DB-COMMENTS

CLINVAR

Review status: ★★☆☆ (reviewed by expert panel)

Submissions:

2020-01-01 - Pathogenic - not provided - GeneKor MSA
2017-03-14 - Pathogenic - Breast and/or ovarian cancer - CHEO Genetics Diagnostic Laboratory,Children's Hospital of Eastern Ontario
2017-01-13 - Pathogenic - not provided - Clinical Genetics Karolinska University Hospital,Karolinska University Hospital
2016-11-23 - Pathogenic - Hereditary cancer-predisposing syndrome - Ambry Genetics
2016-10-18 - Pathogenic - not specified - ENIGMA
2015-10-02 - Pathogenic - not specified - CIMBA

HGMD PRO

DM
Breast and/or ovarian cancer
CM082514

Ajout possible d'articles en
attente de publication

STUDIES & REFERENCES

ADD STUDIES

SHOW IGNORED (0)

STUDIES-COMMENTS

PENDING

Greek BRCA1 and BRCA2 mutation spectrum: two BRCA1 mutations account for half the carriers found among high-risk breast/ovarian cancer patients.

Konstantopoulou I et al. (2008), Breast Cancer Res. Treat.: 107(3), 431-41. • HGMD CLINVAR

EVALUATE

IGNORE

Missing data for Pubmed ID 29446198 in database. Please add the reference manually.

Missing data for Pubmed ID 31159747 in database. Please add the reference manually.

✕ REFERENCE EVALUATION
Abstract
CLOSE

Spectrum of genetic variants of BRCA1 and BRCA2 in a German single center study.
Meisel C et al. (2017), Arch. Gynecol. Obstet.: 295(3), 1227-1238.

Source: HGMD (Additional phenotype: Breast and/or ovarian cancer)
Source: CLINVAR

<p>RELEVANCE Is the reference relevant?</p> <p>CONCLUSION Author variant classification</p> <p>FAMILY Variant segregates with disease? <input type="checkbox"/></p> <p>Variant confirmed/unconfirmed de novo in patient?</p> <p>Variant cis/trans with pathogenic?</p> <p>POPULATION Observed in UNRELATED affecteds? <input type="checkbox"/></p> <p>Observed in healthy individual/population?</p> <p>PROTEIN Abnormal protein function? <input type="checkbox"/></p> <p>RNA Abnormal splicing/protein expression? <input type="checkbox"/></p> <p>IN SILICO Results of prediction tools? <input type="checkbox"/></p> <p>OVERALL QUALITY</p>	<p>YES INDIRECTLY NO IGNORE</p> <p>PATHOGENIC VUS NEUTRAL NOT CLASSIFIED</p> <p>YES NO CHOOSE QUALITY ▾</p> <p>CONFIRMED UNCONFIRMED</p> <p>CIS TRANS</p> <p>>=4 AFFECTED 3 AFFECTED 1-2 AFFECTED</p> <p>YES NO</p> <p>YES NO CHOOSE QUALITY ▾</p> <p>YES NO CHOOSE QUALITY ▾</p> <p>PATHOGENIC VUS NEUTRAL Enter tools...</p> <p>EXCELLENT GOOD PASSABLE LACKING POOR</p>
---	---

COMMENTS

Évaluation de la pertinence et conclusion des publications liées

STUDIES & REFERENCES
ADD STUDIES
SHOW IGNORED (0)

STUDIES-COMMENTS

EVALUATED	<p>Published studies</p> <p>BRCA1 and BRCA2 germline mutation spectrum and frequencies in Belgian breast/ovarian cancer families. Claes K et al. (2004), Br. J. Cancer: 90(6), 1244-51. • User</p> <p>EVALUATION</p>	<p>RE-EVALUATE</p> <p>IGNORE</p>
PENDING	<p>BRCA1 and BRCA2 mutation status and cancer family history of Danish women affected with multifocal or bilateral breast cancer at a youn... Bergthorsson JT et al. (2001), J. Med. Genet.: 38(6), 361-8. • User</p> <p>Detection of BRCA1 and BRCA2 mutations in breast cancer families by a comprehensive two-stage screening procedure. Spitzer E et al. (2000), Int. J. Cancer: 85(4), 474-81. • User</p>	<p>EVALUATE</p> <p>IGNORE</p>

AD/AD;AR/AD;SMu/AR

BRCA2 (3/27)

NM_000059.3:c.97delins(15) G/TACCCTTATAATGAC

NP_000050.2:p.Glu33TyrfsTer52 (Gaa/(15)aa)

frameshift variant

13:32893243

COPY VARIANT TO ALAMUT ADD STUDIES ADD ACMG ADD ATTACHMENT COLLAPSE ALL

INDICATIONS COMMENT

+ 0/0 FILTER DEFAULT FULL QUICK VISUAL

UNCLASSIFIED VARIANTS (8)

INH GENE HGVS CSQ ! S O Q R CLASS

Attachment
Filename: ELLA_114721.png
File size: 26.18 kB
Uploaded: 10-08-2022 11:47 by Henrik Ibsen

IGV hg19 13 13:32,893,219-32,893,267 49 bp
G G A C C A A T A A G T C T T A A T T G A A G A C T T C T T C A G A G C T C C A C

Upload de tous les formats possible

ANALYSIS SPECIFIC FOR VARIANT VERIFIED TECHNICAL NOT RELEVANT

ANALYSIS-SPECIFIC-COMMENTS

ACMG (SUGGESTED CLASS: 4)
PVS1 Null variant REMOVE
PM2 Absent from controls REMOVE
SUGGESTED SHOW REQ
ACMG SUGGESTED
PVS1 Null variant ADD
PS4 Increased prevalence in patients ADD
PP1 MODERATE Cosegregation ADD
PM2 SUPPORTIVE Absent from controls ADD
EXISTING Class 4 HISTORY
Classification made by Henrik Ibsen (testgroup01) on 2022-08-10 for HBOCUTV_v01
Report changed by Henrik Ibsen (testgroup01) on 2022-08-10

REGION

REGION-COMMENTS

SNV

UNCLASSIFIED VARIANTS (7)

INH	GENE	HGVSc	CSQ	!	S	O	Q	R	CLASS
AD/AD...	BRCA2	c.292_305del	frameshift						
AD/AD...	BRCA2	c.475+3_475...	intron						
AD/AD...	BRCA2	c.583_595dup	frameshift						
AD/AD...	BRCA2	c.682-2A>C	splice_acceptor						R
AD/AD...	BRCA2	c.925dup	frameshift						R
AD/AD...	BRCA2	c.1233dup	frameshift						R
AD/AD...	BRCA2	c.1444del	frameshift						R

CLASSIFIED VARIANTS (1)

INH	GENE	HGVSc	CSQ	!	S	O	Q	R	CLASS
AD/AD...	BRCA2	c.97delins(15)	frameshift						R 4

NOT RELEVANT VARIANTS (0)

TECHNICAL VARIANTS (0)

REPORT

Indications recommandées

Commentaire sur l'interprétation

CLINICAL REPORT

NM_000059.3(BRCA2):c.[1];[1] Likely pathogenic variant c.97delins(15) p.Glu33Tyrfs1er52 à inclure dans le rapport

Commentaire ajouté à la variation lors de l'interprétation

Commentaire libre

Reporting

SIFIED VARIANTS (7)

GENE SELECT NEXT ROUND

SEND FROM/TO: INTERPRETATION → INTERPRETATION

NOT READY → **INTERPRETATION** → REVIEW → MEDICAL REVIEW → FINALIZE

Choix de la prochaine étape à valider

FINISH

a inclure dans le rapport

ANALYSES

VARIANTS

IMPORT

SEARCH

NOT READY 0

NO ANALYSES ARE NOT READY

YOUR ANALYSES 1

2022-08-09 brca_sample_2.HBOC_v01 [WARNING] [HTS]

- 1 • Interpretation • C. Collett • 2022-08-09 11:59
- 2 • Interpretation • C. Collett • 2022-08-09 12:00
- 3 • Interpretation • C. Collett • 2022-08-09 12:01
- 4 • Review (Ongoing) • H. Ibsen • 2022-08-10 11:38

INTERPRETATION 6

2022-08-09 brca_sample_1.HBOCUTV_v01 [HTS]

2022-08-09 brca_sample_1.HBOC_v01 [WARNING] [HTS]

2022-08-09 brca_sample_2.HBOCUTV_v01 [HTS]

2022-08-09 brca_sample_3.HBOCUTV_v01 [HTS]

2022-08-09 brca_sample_allfiltered.HBOC_v01 [WARNING] [HTS]

2022-08-09 brca_sample_master.HBOCUTV_v01 [HTS]

REVIEW 1

2022-08-09 brca_long_variants.HBOCUTV_v01 [HTS]

- 1 • Interpretation • H. Ibsen • 2022-08-10 11:49
- 2 • Review

MEDICAL REVIEW 0

NO ANALYSES PENDING MEDICAL REVIEW.

OTHERS' ANALYSES 0

OTHERS HAVE NO ANALYSES

FINALIZED 0

NO FINALIZED ANALYSIS

Revue

Équivalent d'une seconde interprétation pour confirmer l'interprétation initiale

OVERVIEW brca_long_variants.HBOCUTV_v01 - REVIEW DOCUMENTATION H. IBSEN

INFO CLASSIFICATION REPORT GENE PANEL INFO WORK LOG (1) FINISH SAVE

UNCLASSIFIED VARIANTS (1)

INH	GENE	HGVSc	CSQ	!	S	O	Q	R	CLASS
AD/AD;...	BRC2	c.925dup	frameshift						R

CLASSIFIED VARIANTS (3)

INH	GENE	HGVSc	CSQ	!	S	O	Q	R	CLASS
AD/AD;...	BRC2	c.97delins(15)	frameshift						V R 4
AD/AD;...	BRC2	c.1233dup	frameshift						R 3
AD/AD;...	BRC2	c.1444del	frameshift						R → 2

NOT RELEVANT VARIANTS (4)

INH	GENE	HGVSc	CSQ	!	S	O	Q	R	CLASS
AD/AD;...	BRC2	c.292_305del	frameshift						
AD/AD;...	BRC2	c.475+3_475...	intron						
AD/AD;...	BRC2	c.583_595dup	frameshift						
AD/AD;...	BRC2	c.682-2A>C	splice_acceptor						R

TECHNICAL VARIANTS (0)

REPORT

Indications recommandées

Commentaire sur l'interprétation

→ Le commentaire peut être modifié lors de la revue

CLINICAL REPORT

NM_000059.3(BRCA2):c.[.];[.] Likely pathogenic variant c.97delins(15) p.Glu33TyrfsTer52 à inclure dans le rapport

NM_000059.3(BRCA2):c.[.];[.] Variant of uncertain significance c.1233dup p.Pro412ThrfsTer9

Tri possible en variations non pertinentes

Choix des variations à inclure

Les commentaires et classifications peuvent être modifiés par le reviewer

Possibilité d'imposer des exigences pour le rendu

FINALIZED VARIANTS (1)

SEND FROM/TO: REVIEW → FINALIZED

NOT READY → INTERPRETATION → REVIEW → MEDICAL REVIEW → FINALIZE

FINALIZE NOT POSSIBLE:

- SOME VARIANTS ARE MARKED AS NOT RELEVANT, WHILE THIS IS DISALLOWED IN CONFIGURATION.
- SOME VARIANTS ARE MISSING CLASSIFICATIONS: BRCA2 C.925DUP (P.SER309PHEFSTER6)
- SOME VARIANTS HAVE CLASSIFICATIONS THAT ARE NOT FINALIZED: BRCA2 C.1444DEL (P.ALA483GLNFSTER2)

GENE	HGVSc	CSQ	!	S	O	Q	R	CLASS
BRCA2	c.292_305del	frameshift						

YOUR PROFILE

Username: testuser1
Full name: Henrik Ibsen
Password expiry: 2027-03-30 17:03
E-mail: testuser1@foo.bar

YOUR USER GROUP

Group: testgroup01
Other users in group:
Bjørnstjerne Bjørnson
Camilla Collett

GENEPANELS AND INFO

Number of analysis worked on: 2
Number of variants worked on: 1
Genepanels:
HBOCUTV v01
HBOC v01



Gestion des droits

ANALYSES

SEARCH

NOT READY 0

NO ANALYSES ARE NOT READY

VARIANTS

YOUR ANALYSES 0

YOU HAVE NO ONGOING ANALYSES

IMPORT

INTERPRETATION 2

2022-08-09 HG002-Trio.Mendeliome_v01 HTS

2022-08-09 NA12878.Ciliopati_v03 HTS

REVIEW 0

NO ANALYSES PENDING REVIEW.

MEDICAL REVIEW 0

NO ANALYSES PENDING MEDICAL REVIEW.

OTHERS' ANALYSES 0

OTHERS HAVE NO ANALYSES

FINALIZED 0

NO FINALIZED ANALYSIS

ANALYSIS INFO

Requested:
2022-08-09
Imported:
2022-08-09 11:28

SAMPLES

PROBAND	MOTHER	FATHER
Sample name: HG002	Sample name: HG004	Sample name: HG003
Imported: 2022-08-09 11:28	Imported: 2022-08-09 11:28	Imported: 2022-08-09 11:28
Family: TestFam	Family: TestFam	Family: TestFam
Sex: Male	Sex: Female	Sex: Male
Technology: HTS	Technology: HTS	Technology: HTS

ATTACHMENTS

ATTACHMENTS

Cas d'une analyse en trio

PIPELINE REPORT

Gene list for genes having below 100% coverage:

Gene	Transcript	Phenotype	Inheritance	Coverage (% bp) (2)
MSH2	NM_000251.2	Colorectal cancer, hereditary nonpolyposis, type 1	AD	2869 99.9%
MSH6	NM_000179.2	Colorectal cancer, hereditary nonpolyposis, type 5	AD	4123 99.2%
PMS2	NM_000535.5	Colorectal cancer, hereditary nonpolyposis, type 4	AD	2649 98.6%

(1) bp = basepair; + 4 bp = -2 og + 2 bp in Intron region to cover conserved splice site (based on Refseqs from UCSC refGene table, March 2015, GRCh37/hg19)

(2) Percentage of region covered at least 40 times

Regions covered by less than 40 reads

Start position (HGVSg)	End position (HGVSg)	Gene	Transcript	Exon	x covered
chr2:g.47630540N>N	chr2:g.47630543N>N	MSH2	NM_000251.2	exon1	36
chr2:g.48010497N>N	chr2:g.48010531N>N	MSH6	NM_000179.2	exon1	13
chr7:g.6013138N>N	chr7:g.6013175N>N	PMS2	NM_000535.5	exon15	11

AD DNMT3A (23/23)

NM_175629.2:c.2732G>A P: C/T | F: C/C | M: C/C

NP_783328.1:p.Cys911Tyr (tGt/tAt)

missense variant

2:25457155

COPY VARIANT TO ALAMUT COLLAPSE ALL

INDICATIONS COMMENT

+ 0/654 FILTER TRIODEFAULT FULL QUICK VISUAL

UNCLASSIFIED VARIANTS (20)

INH	GENE	HGVSc	CSQ	!	S	O	Q	R	CLASS
AD	DNMT3A	c.2732G>A	missense						MD
AR	ARSB	c.312_312+1...	frameshift, s...						ID
AR	PRKRA	c.785-2_785...	splice_acceptor						ID R
AR	PRKRA	c.785-5_785...	splice_region...						ID Q
AR	FCGR3A	c.512T>A	missense						IA O QR
XD	AMER1	c.1873A>G	missense						X O QR
XR	GRI3	c.1181G>A	missense						X O Q
AD/AR ...	TTN TTN	c.34474C>A ...	missense, int...						C
AD/AR ...	TTN TTN	c.104251G>C...	missense						C
AR	FMN2	c.3138A>T	synonymous						C Q
AR	FMN2	c.3495T>G	synonymous						C Q
AD	BRAF	c.2128-16_21...	splice_region...						IM Q
AD/AR	MSH2	c.942+2del	splice_donor						M R
AR	ARSB	c.312+3A>C	splice_region...						IM
AR	ARSB	c.312+2T>C	splice_donor						IM
AR	SERPINB8	c.197T>C	missense						M
AD	BRAF	c.2128-5dup	splice_region...						I O Q
AD/AR	F11	c.403G>T	stop_gained						M R
AD/AR	HTT	c.96_110dup	inframe_inse...						I Q
AR	STRC	c.3307-26_33...	intron						I Q

CLASSIFIED VARIANTS (0)

NOT RELEVANT VARIANTS (0)

TECHNICAL VARIANTS (0)

ANALYSIS SPECIFIC FOR VARIANT VERIFIED TECHNICAL NOT RELEVANT

ANALYSIS-SPECIFIC-COMMENTS

QUALITY	Proband (Male)	Father	Mother
P(de novo):	0.80	Filter: PASS	Filter: PASS
Filter:	PASS	Quality: 43	Quality: 43
Quality:	43	GQ: 83	GQ: 98
GQ:	47	Depth: 37	Depth: 36
Depth:	41	Ratio: 0.04	Ratio: 0.00
Ratio:	0.44	- REF (C): 45	- REF (C): 41
- REF (C):	23	- T: 2	- T: 0
- T:	18		

CLASSIFICATION SELECT CLASS

EVALUATION

REPORT

ACMG

SUGGESTED SHOW REQ ACMG SUGGESTED

REGION

REGION-COMMENTS

VARBB SNV

FREQUENCY

FREQUENCY-COMMENTS

GNOMAD EXOMES	GNOMAD GENOMES	EXAC	FINDB	POP	COUNT	NUM	HOM	FREQ	DBSNP
				OUSWES:	1	12840	0	1.000e-4	rs906113912
				OUSWGS:					
				OUSWES indications:					
				- PU:		1			

PREDICTION

ADD PREDICTION

OVERVIEW | INFO | CLASSIFICATION | REF | ACTIONS

AD DNMT3A (c.2732G>A)

INDICATIONS COMMENT

+ 0/654 FILTER

UNCLASSIFIED VARIANTS (20)

INH	GENE	HGVSc	CSQ	! S O Q R	QUAL	DP	RATIO	HI. FREQ	HI. COUNT	EXTERNAL	CLASS
AD	DNMT3A	c.2732G>A									
AR	ARSB	c.312_312+1									
AR	PRKRA	c.785-2_785-									
AR	PRKRA	c.785-5_785-									
AR	FCGR3	c.512T>A									
XD	AMER1	c.1873A>G									
XR	GRIA3	c.1181G>A									
AD/AR	TTN	c.34474C>A									
AD/AR	TTN	c.104251G>C									
AR	FMN2	c.3138A>T									
AR	FMN2	c.3495T>G									
AD	BRAF	c.2128-16_21									
AD/AR	MSH2	c.942+2del									
AR	ARSB	c.312+3A>C									
AR	ARSB	c.312+2T>C									
AR	SERPIN8	c.197T>C									
AD	BRAF	c.2128-5dup									
AD/AR	F11	c.403G>T									
AD/AR	HTT	c.96_110dup									
AR	STRC	c.3307-26_33									

CLASSIFIED VARIANT (1)

NOT RELEVANT VARIANTS (1)

TECHNICAL VARIANTS (0)

ADD FILTERED VARIANTS

INCLUDED VARIANTS (0)

ALL (654) | FREQUENCY (473) | REGION (22) | CONSEQUENCE (40) | PPY (9) | QUALITY (8) | SEGREGATION (102) | ALL GENES

654 VARIANTS FROM CURRENT FILTER SETTINGS

INH	GENE	HGVSc	CSQ	! S O Q R	QUAL	DP	RATIO	HI. FREQ	HI. COUNT	EXTERNAL	CLASS
AD	A2M	c.387C>G	synonymous		121	49	0.37	0.010904	1773	Clinvar	
AD/AR	AARS1	c.2900A>T	missense		125	40	0.40	0.016002	2444	Clinvar	
AR	ABAT	c.540+32_54...	intron		96	33	0.39	0.139500	7754	Clinvar	
AR	ABCG5	c.293C>G	missense		127	32	0.53	0.003226	447	HGMD Clinvar	
AR	ACAD8	c.1128+25_1...	splice_region...		154	23	0.52	0.007600	97		
AD/AR	ACAN	c.1051+9C>T	intron		129	29	0.52	0.039600	5531		
AD/AR	ACAN	c.3294C>T	synonymous		68	6	1.00	0.601200	18779		
AD/AR	ACAN	c.3351C>T	synonymous		10	3	1.00	0.113500	1983	Clinvar	
AD/AR	ACAN	c.3637G>A	missense		76	3	0.67	0.307500	4933		
AD/AR	ACAN	c.4170_4226del	inframe_dele...		40	26	0.24	0.032100	3302		
AD/AR	ACAN	c.3408C>T	synonymous		46	1	1.00	-	-		
AD/AR	ACAN	c.4290T>C	synonymous		115	27	0.30	0.000733	105		
AR	AD/AR	CARMIL...	g.67693916G...	synonymous...	128	33	0.45	0.002996	585	Clinvar	
AR	ACE	c.1586+14_1...	intron		33	16	0.31	1.000e-4	1		
AD	ACTC1	c.809-16_809...	intron		231	33	0.92	0.167300	3573	Clinvar	
AD	ACTC1	c.809-28_809...	intron		231	33	0.95	0.169800	3424	Clinvar	
AD	ACTN1	c.2663C>T	missense		126	32	0.41	0.035900	5028		
AR	ADA	c.390G>A	synonymous		129	33	0.55	0.062100	5176	Clinvar	
AR	ADAM17	c.1695T>C	synonymous		128	43	0.53	0.020000	3347	Clinvar	
AR	ADAM17	c.958-13dup	intron		102	24	0.43	0.072400	878		

6 VARIANTS FROM CURRENT FILTER SETTINGS

INH	GENE	HGVSc	CSQ	! S O Q R	QUAL	DP	RATIO	HI. FREQ	HI. COUNT	EXTERNAL	CLASS
AD	CACNA1C	c.3049-10C>T	intron		217	33	0.39	0.003101	296	Clinvar	
AD	CHD7	c.4645-9T>C	intron		129	42	0.50	0.000189	32	Clinvar	
AD/AR	ITGA2B	c.671-13C>T	intron		121	31	0.35	0.000301	74	Clinvar	

ALL (654) | FREQUENCY (473) | REGION (22) | CONSEQUENCE (40) | PPY (9) | QUALITY (8) | SEGREGATION (102) | ALL GENES

6 VARIANTS FROM CURRENT FILTER SETTINGS

INH	GENE	HGVSc	CSQ	! S O Q R	QUAL	DP	RATIO	HI. FREQ	HI. COUNT	EXTERNAL	CLASS
AR	IVD	c.560-18C>T	intron		125	50	0.42	0.007117	851	Clinvar	
AD	MAP2K1	c.292-3C>T	splice_region...		127	42	0.43	0.000130	32	Clinvar	
AD	MSR1	c.1034-11del	intron		216	21	0.92	0.007000	90		
AD	RAB11B	c.431-8T>C	splice_region...		126	38	0.50	0.002472	564		
AR	RTTN	c.4303-12_43...	intron		172	15	0.80	-	-		
AD	TBX1	c.513-16C>T	intron		128	43	0.51	0.000552	106		

PREDICTION | ADD PREDICTION

AD DNMT3A (c.2732G>A) | INFO | CLASSIFICATION | REF | ACTIONS

INDICATIONS COMMENT

UNCLASSIFIED VARIANTS (20)

CLASSIFIED VARIANT (1)

NOT RELEVANT VARIANTS (1)

TECHNICAL VARIANTS (0)

AD DNMT3A (c.2732G>A) | INFO | CLASSIFICATION | REF | ACTIONS

INDICATIONS COMMENT

UNCLASSIFIED VARIANTS (20)

CLASSIFIED VARIANT (1)

NOT RELEVANT VARIANTS (1)

TECHNICAL VARIANTS (0)

AD DNMT3A (c.2732G>A) | INFO | CLASSIFICATION | REF | ACTIONS

INDICATIONS COMMENT

UNCLASSIFIED VARIANTS (20)

CLASSIFIED VARIANT (1)

NOT RELEVANT VARIANTS (1)

TECHNICAL VARIANTS (0)

INDICATIONS COMMENT
 + 3/654 FILTER TRIODEFAULT FULL QUICK VISUAL

UNCLASSIFIED VARIANTS (23)

INH	GENE	HGVSc	CSQ	! F S O	CLASS	QUAL	DP	RATIO	HI.FREQ	HI.COUNT	EXTERNAL	CLASS
AD	DNMT3A	c.2732G>A	missense	[M] [D]	Clinvar	43	41	0.44	1.000e-4	1		T NR NP 2 BS1 BS2
AR	ARSB	c.312_312+11...	frameshift, s...	[I] [D]	Clinvar	41	27	0.37	-	-		T NR NP 2 BS1 BS2
AR	PRKRA	c.785-2_785...	splice_acceptor	[I] [D]	Clinvar	41	27	0.37	-	-		T NR NP 2 BS1 BS2
AR	PRKRA	c.785-5_785...	splice_region...	[I] [D]	Clinvar	41	27	0.37	-	-		T NR NP 2 BS1 BS2
AR	FCGR3A	c.512T>A	missense	[I] [A] [O]	Clinvar	116	30	0.50	0.000547	85		T NR NP 2 BS1 BS2
XD	AMER1	c.1873A>G	missense	[X] [O]	Clinvar	121	31	0.35	0.000301	74		T NR NP 2 BS1 BS2
XR	GRI3A	c.1181G>A	missense	[X] [O]	Clinvar	232	45	1.00	0.013900	178		T NR NP 2 BS1 BS2
AD/AR	TTN TTN	c.34474C>A ...	missense, int...	[C]	Clinvar	128	28	0.54	0.005752	296		T NR NP 2 BS1 BS2
AD/AR	TTN TTN	c.104251G>C > ...	missense	[C]	Clinvar	129	25	0.48	0.002561	394		T NR NP 2 BS1 BS2
AR	FMN2	c.3138A>T	synonymous	[C] [Q]	Clinvar	40	8	0.50	0.000628	34		T NR NP 2 BS1 BS2
AR	FMN2	c.3495T>G	synonymous	[C] [Q]	Clinvar	23	3	0.33	2.318e-5	1		T NR NP 2 BS1 BS2
AD	BRAF	c.2128-16_21...	splice_region...	[I] [M] [Q]	Clinvar	15	9	0.44	0.003876	509		T NR NP 2 BS1 BS2
AD/AR	MSH2	c.942+2del	splice_donor	[M] [R]	HGMD Clinvar	15	25	0.40	-	-		T NR NP 2 BS1 BS2
AR	ARSB	c.312+3A>C	splice_region...	[I] [M]	Clinvar	48	26	0.46	-	-		T NR NP 2 BS1 BS2
AR	ARSB	c.312+2T>C	splice_donor	[I] [M]	Clinvar	48	25	0.48	-	-		T NR NP 2 BS1 BS2
AR	SERPINB8	c.197T>C	missense	[M]	Clinvar	116	30	0.50	0.000547	85		T NR NP 2 BS1 BS2
AD	BRAF	c.2128-5dup	splice_region...	[I] [O] [Q]	Clinvar	33	6	0.67	0.010500	135		T NR NP 2 BS1 BS2
AD	CACNA1C	c.3049-10C>T	intron	[I]	Clinvar	217	33	0.39	0.003101	296		T NR NP 2 BS1 BS2
AD	CHD7	c.4645-9T>C	intron	[I]	Clinvar	129	42	0.50	0.000189	32		T NR NP 2 BS1 BS2
AD/AR	F11	c.403G>T	stop_gained	[R]	HGMD Clinvar	119	37	0.35	0.001824	233		T NR NP 2 BS1 BS2
AD/AR	HTT	c.96_110dup	inframe_inse...	[I] [Q]	Clinvar	315	16	1.00	0.021200	272		T NR NP 2 BS1 BS2
AD/AR	ITGA2B	c.671-13C>T	intron	[I]	Clinvar	121	31	0.35	0.000301	74		T NR NP 2 BS1 BS2
AR	STRC	c.3307-26_33...	intron	[I] [Q]	Clinvar	232	45	1.00	0.013900	178		T NR NP 2 BS1 BS2

CLASSIFIED VARIANTS (0)

Classification rapide

Classification bénigne/non pertinente en un clic

INDICATIONS COMMENT
 + 3/654 FILTER TRIODEFAULT FULL QUICK VISUAL

UNCLASSIFIED VARIANTS (23)

INH	GENE	HGVSc	CSQ	! F S O Q R	QUAL	DP	RATIO	HI.FREQ	HI.COUNT	EXTERNAL	CLASS
AD	DNMT3A	c.2732G>A	missense	[M] [D]	43	41	0.44	1.000e-4	1		T NR NP 2 BS1 BS2
AR	ARSB	c.312_312+11...	frameshift, s...	[I] [D]	41	27	0.37	-	-		T NR NP 2 BS1 BS2
AR	PRKRA	c.785-2_785...	splice_acceptor	[I] [D]	41	27	0.37	-	-		T NR NP 2 BS1 BS2
AR	PRKRA	c.785-5_785...	splice_region...	[I] [D]	41	27	0.37	-	-		T NR NP 2 BS1 BS2
AR	FCGR3A	c.512T>A	missense	[I] [A] [O]	116	30	0.50	0.000547	85		T NR NP 2 BS1 BS2
XD	AMER1	c.1873A>G	missense	[X] [O]	121	31	0.35	0.000301	74		T NR NP 2 BS1 BS2
XR	GRI3A	c.1181G>A	missense	[X] [O]	232	45	1.00	0.013900	178		T NR NP 2 BS1 BS2
AD/AR	TTN TTN	c.34474C>A ...	missense, int...	[C]	128	28	0.54	0.005752	296		T NR NP 2 BS1 BS2
AD/AR	TTN TTN	c.104251G>C > ...	missense	[C]	129	25	0.48	0.002561	394		T NR NP 2 BS1 BS2
AR	FMN2	c.3138A>T	synonymous	[C] [Q]	40	8	0.50	0.000628	34		T NR NP 2 BS1 BS2
AR	FMN2	c.3495T>G	synonymous	[C] [Q]	23	3	0.33	2.318e-5	1		T NR NP 2 BS1 BS2
AD	BRAF	c.2128-16_21...	splice_region...	[I] [M] [Q]	15	9	0.44	0.003876	509		T NR NP 2 BS1 BS2
AD/AR	MSH2	c.942+2del	splice_donor	[M] [R]	15	25	0.40	-	-		T NR NP 2 BS1 BS2
AR	ARSB	c.312+3A>C	splice_region...	[I] [M]	48	26	0.46	-	-		T NR NP 2 BS1 BS2
AR	ARSB	c.312+2T>C	splice_donor	[I] [M]	48	25	0.48	-	-		T NR NP 2 BS1 BS2
AR	SERPINB8	c.197T>C	missense	[M]	116	30	0.50	0.000547	85		T NR NP 2 BS1 BS2
AD	BRAF	c.2128-5dup	splice_region...	[I] [O] [Q]	33	6	0.67	0.010500	135		T NR NP 2 BS1 BS2
AD	CACNA1C	c.3049-10C>T	intron	[I]	217	33	0.39	0.003101	296		T NR NP 2 BS1 BS2
AD	CHD7	c.4645-9T>C	intron	[I]	129	42	0.50	0.000189	32		T NR NP 2 BS1 BS2
AD/AR	F11	c.403G>T	stop_gained	[R]	119	37	0.35	0.001824	233		T NR NP 2 BS1 BS2
AD/AR	HTT	c.96_110dup	inframe_inse...	[I] [Q]	315	16	1.00	0.021200	272		T NR NP 2 BS1 BS2
AD/AR	ITGA2B	c.671-13C>T	intron	[I]	121	31	0.35	0.000301	74		T NR NP 2 BS1 BS2
AR	STRC	c.3307-26_33...	intron	[I] [Q]	232	45	1.00	0.013900	178		T NR NP 2 BS1 BS2

CLASSIFIED VARIANTS (0)

AR ARSB () NM_000046.3:c.312_312+1ins(67) P: -/GCGGGGGCGGCGGGGGGGGGCGGGGGGGGGGGCGGGCGGGCGGGGGGGCGG NP_000037.2:p.Ile105ArgfsTer44 (-)/(67) frameshift variantsplice region variant 5:78280759-78280760

COPY VARIANT TO ALAMUT ADD STUDIES ADD ACMG ADD ATTACHMENT COLLAPSE ALL

INDICATIONS COMMENT

+ 3/684 FILTER TRIODEFAULT FULL QUICK VISUAL

UNCLASSIFIED VARIANTS (22)

INH	GENE	HGVSc	CSQ	!	F	S	O	Q	R	CLASS
AR	ARSB	c.312_312+1i...	frameshift, s...	I	D					
AR	PRKRA	c.785-2_785-...	splice_acceptor	I	D				R	
AR	PRKRA	c.785-5_785-...	splice_region...	I	D			Q		
AR	FCG3A	c.512T>A	missense	I	A	O	Q	R		
XD	AMER1	c.1873A>G	missense		X	O	Q	R		
XR	GRIA3	c.1181G>A	missense		X	O	Q			
AD/AR ...	TTN TTN	c.34474C>A ...	missense, int...		C					
AD/AR ...	TTN TTN	c.104251G>C ...	missense		C					
AR	FMN2	c.3138A>T	synonymous		C			Q		
AR	FMN2	c.3495T>G	synonymous		C			Q		
AD	BRAF	c.2128-16_21...	splice_region...	I	M			Q		
AD/AR	MSH2	c.942+2del	splice_donor		M			R		
AR	ARSB	c.312+3A>C	splice_region...	I	M					
AR	ARSB	c.312+2T>C	splice_donor	I	M					
AR	SERPIN8	c.197T>C	missense	I	M					

WARNING 1

- Another variant is within 2 bp of this variant

ANALYSIS SPECIFIC FOR VARIANT VERIFIED TECHNICAL NOT RELEVANT

ANALYSIS-SPECIFIC-COMMENTS

QUALITY	Proband (Male)	Father	Mother
	NEEDS VERIFICATION	NEEDS VERIFICATION	NEEDS VERIFICATION
P(de novo):	0.39	Filter: PASS	Filter: PASS
Filter:	PASS	Quality: 41	Quality: 41
Quality:	41	GQ: 99	GQ: 91
GQ:	45	Depth: 53	Depth: 36
Depth:	27	Ratio: 0.00	Ratio: 0.00
Ratio:	0.37	- REF (C): 53	- REF (C): 41
- REF (C):	17	- (68): 0	- (68): 0
- (68):	10		

POTENTIAL CONFLICT

This variant is currently being worked on by Marie Spångberg in variant workflow.

WARNING 1

- This variant's existing classification was performed by a different user group: testgroup03.

INFORMATION

A NEW EVALUATION WAS LOADED FOR: BRCA2 C.97G>T

Système d'alertes et notifications

XD	AMER1	c.1873A>G	missense	X	O	Q	R
XR	GRIA3	c.1181G>A	missense	X	O	Q	
AD/AR ...	TTN TTN	c.34474C>A ...	missense, int...		C		
AR	FMN2	c.3138A>T	synonymous		C		Q
AR	FMN2	c.3495T>G	synonymous		C		Q
AD	BRAF	c.2128-16_21...	splice_region...	I	M		Q
AD/AR	MSH2	c.942+2del	splice_donor		M		R
AR	ARSB	c.312+3A>C	splice_region...	I	M		
AR	ARSB	c.312+2T>C	splice_donor	I	M		
AR	SERPIN8	c.197T>C	missense	I	M		
AD	BRAF	c.2128-5dup	splice_region...	I	M		O Q
AD	CACNA1C	c.3049-10C>T	intron		I		
AD	CHD7	c.4645-9T>C	intron		I		
AD/AR	F11	c.403G>T	stop_gained				R
AD/AR	HTT	c.96_110dup	inframe_inse...	I			Q
AD/AR	ITGA2B	c.671-13C>T	intron		I		
AR	STRC	c.3307-26_33...	intron	I			Q

FREQUENCY

FREQUENCY-COMMENTS

GNOMAD GENOMES	FILTER: RF	POP	COUNT	NUM	HOM	HEMI	FREQ
		AFR:	19	12358	0		0.001537
		AMR:	15	19408	0		0.000773
		ASJ:	15	6472	0		0.002318
		EA:	51	13158	0		0.003876
		E(F):	54	17066	0		0.003164
		E(NF):	298	89060	1		0.003346
		OTH:	15	3696	0		0.004058
		SA:	42	22694	0		0.001851
		TOT:	509	183912	1		0.002768

GNOMAD GENOMES	FILTER: RF	POP	COUNT	NUM	HOM	HEMI	FREQ
		AFR:	1	5734	0		0.000174
		AMR:	0	406	0		0
		ASJ:	0	228	0		0
		EA:	0	1300	0		0
		E(F):	0	728	0		0
		E(NF):	0	11276	0		0
		OTH:	0	556	0		0
		SA:					
		TOT:	1	20228	0		4.944e-5

EXAC	INDB	POP	COUNT	NUM	HOM	FREQ	DBSNP
		OUSWES:	33	12840	3	0.002600	rs766844227
		OUSWGS:	1	804	0	0.001200	
		OUSWGS indications:					
		- Mendel:		1			

Configuration

La configuration des comportements de l'IU, des panels, des annotations, des filtres, des groupes d'utilisateurs se fait directement en yml ou json ou via le CLI.

Possibilité d'analyse automatique lors du dépôt d'un sample dans un dossier pré-défini et via des regex servant à appliquer une whitelist/blacklist.

Un panel de gènes par défaut peut être appliqué automatiquement selon le usergroup destinataire.

```
{
  "name": "frequency",
  "config": {
    "groups": {
      "external": {
        "GNOMAD_GENOMES": ["G"],
        "GNOMAD_EXOMES": ["G"]
      },
      "internal": {
        "inDB": ["OUSWES"]
      }
    },
    "num_thresholds": {
      "GNOMAD_GENOMES": {
        "G": 5000
      },
      "GNOMAD_EXOMES": {
        "G": 5000
      }
    },
    "thresholds": {
      "AD": {
        "external": 0.005,
        "internal": 0.05
      },
      "default": {
        "external": 0.01,
        "internal": 0.05
      }
    }
  }
}
```

```
{
  "name": "quality",
  "config": {
    "qual": 100,
    "allele_ratio": 0.25,
    "filter_status": {
      "pattern": "PASS",
      "inverse": true,
    }
  }
}
```

```
{
  "name": "segregation",
  "config": {
    "denovo": {
      "enable": true,
      "gq_threshold": {
        "proband": 20,
        "mother": 20,
        "father": 20
      }
    },
    "compound_heterozygous": { "enable": true },
    "recessive_homozygous": { "enable": true },
    "no_coverage_parents": { "enable": true },
    "parental_mosaicism": { "enable": false }
  }
}
```