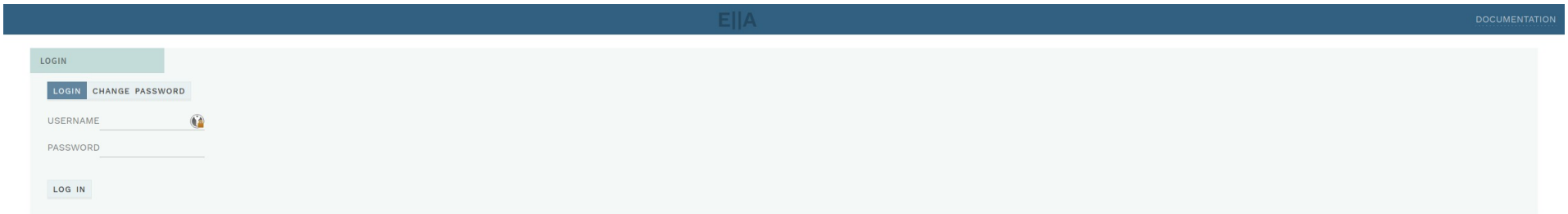
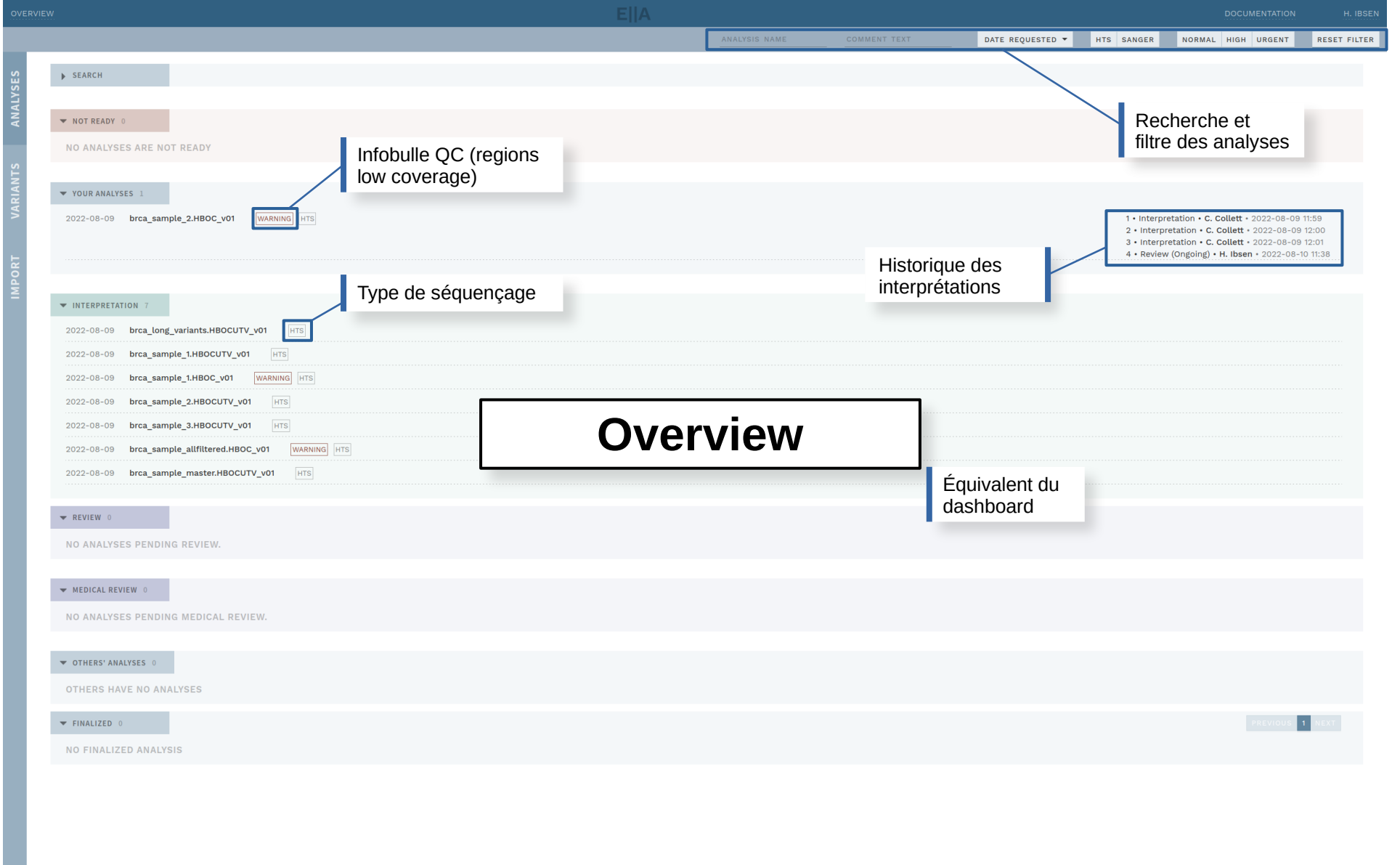
Présentation générale

E||A

Version démo, release 10/21

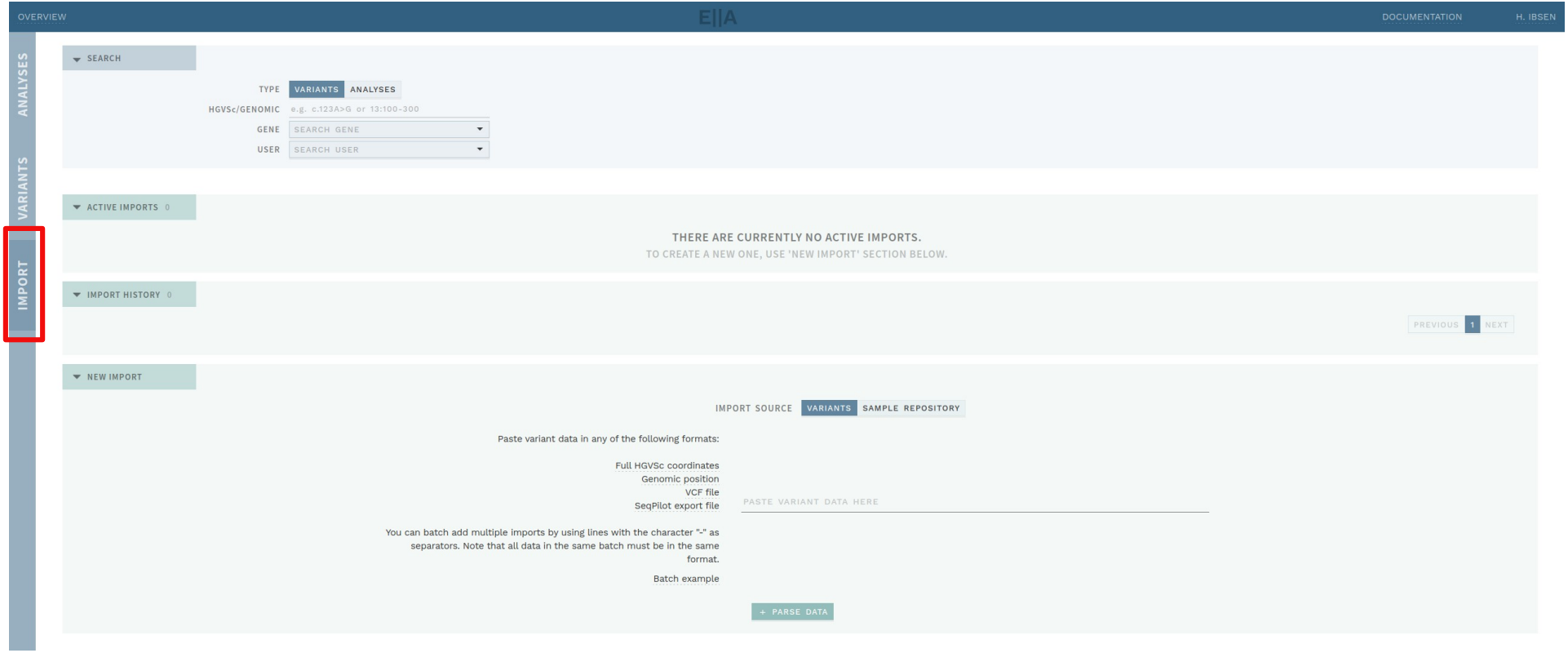
Login

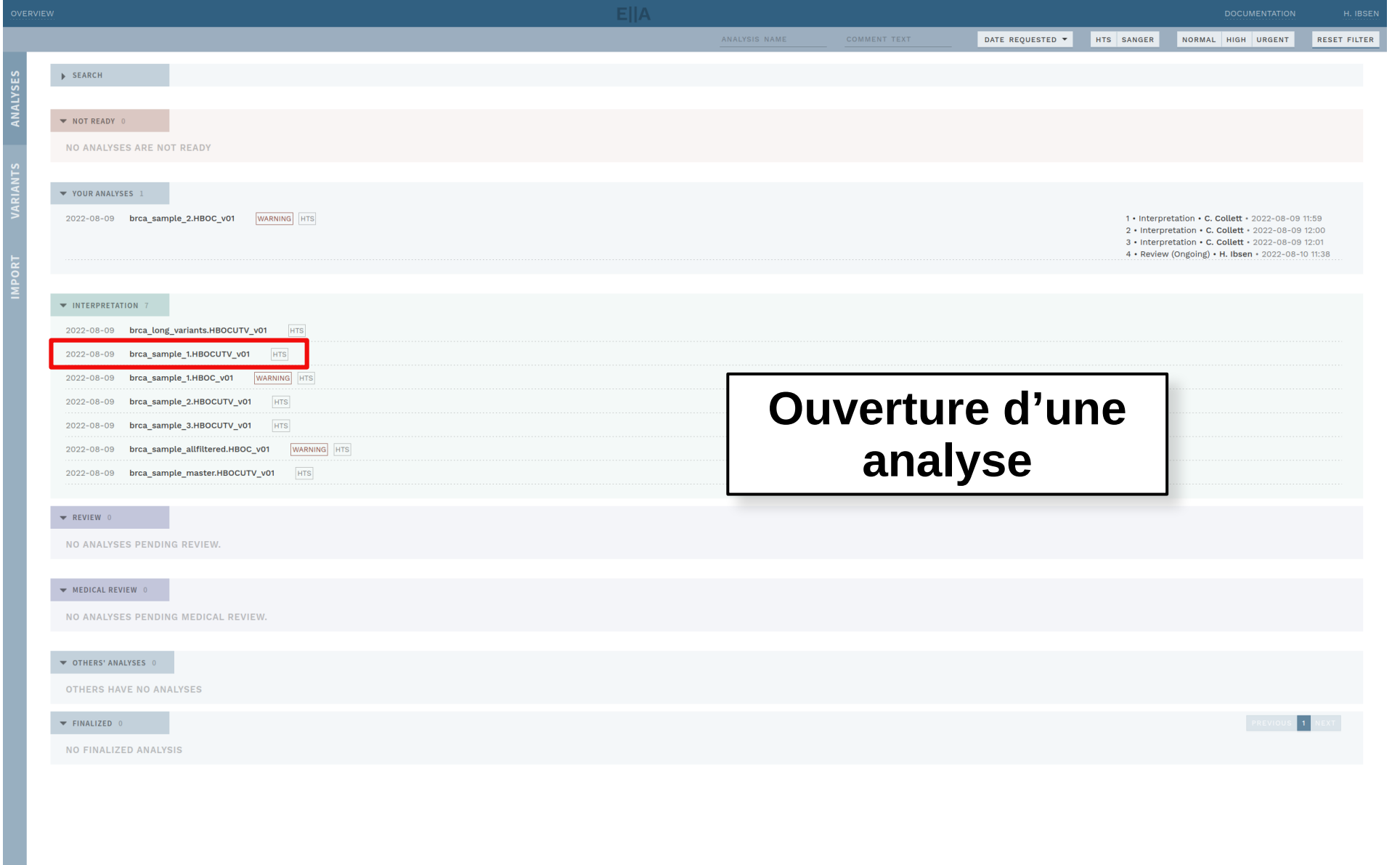

E|A
DOCUMENTATION
LOGIN
LOGIN
CHANGE PASSWORD
USERNAME
PASSWORD
LOG IN


OVERVIEW
E||A
DOCUMENTATION
H. IBSEN
ANALYSIS NAME
COMMENT TEXT
DATE REQUESTED
HTS
SANGER
NORMAL
HIGH
URGENT
RESET FILTER
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
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
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
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
Recherche et filtre des analyses
Infobulle QC (regions low coverage)
Historique des interprétations
Type de séquençage
Overview
Équivalent du dashboard

Overview
Menu variations

Overview
Import



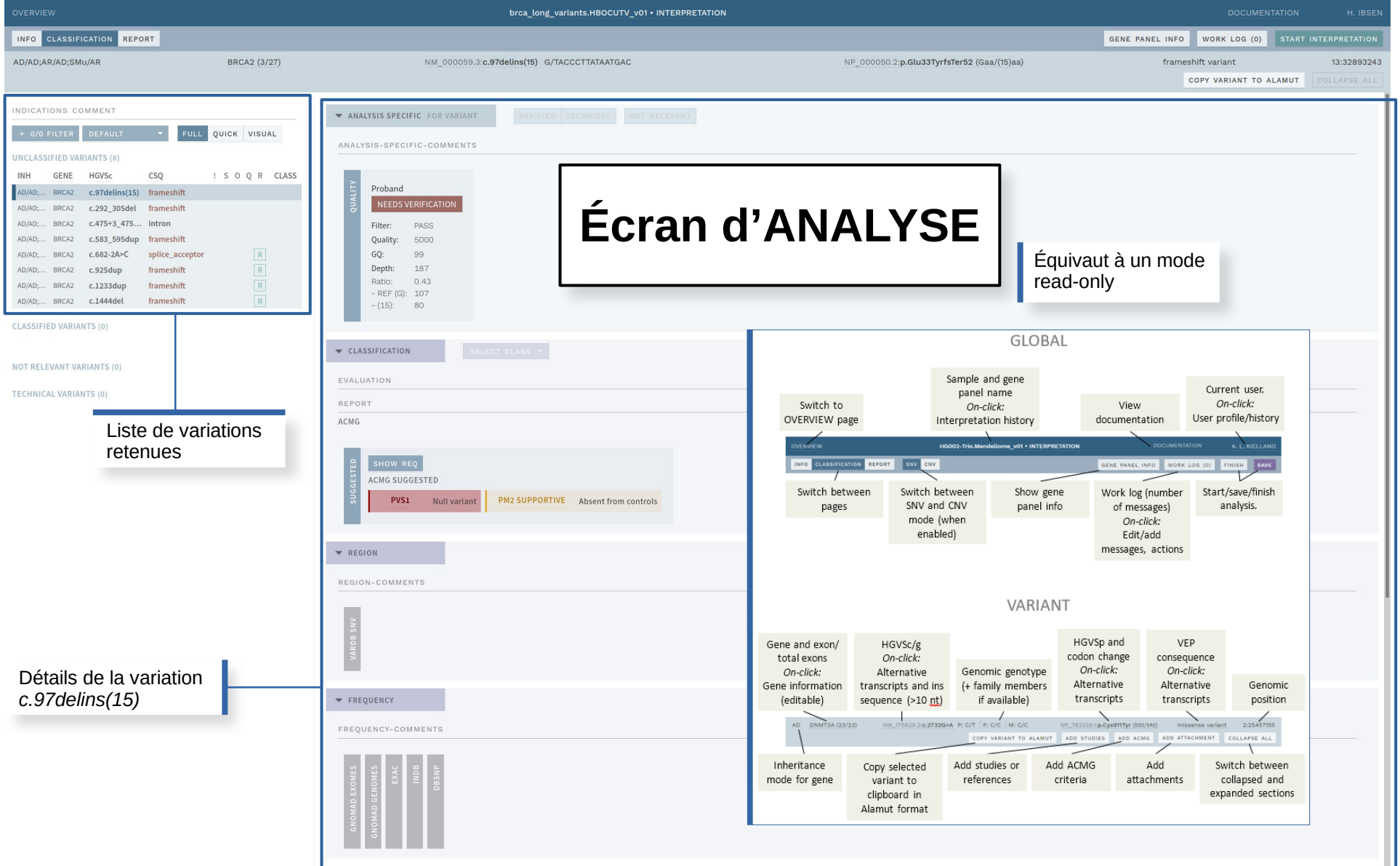

OVERVIEW
E||A
DOCUMENTATION
H. IBSEN
ANALYSIS NAME
COMMENT TEXT
DATE REQUESTED
HTS
SANGER
NORMAL
HIGH
URGENT
RESET FILTER
ANALYSES
VARIANTS
IMPORT
SEARCH
NOT READY 0
NO ANALYSES ARE NOT READY
YOUR ANALYSES 1
2022-08-09 brca_sample_2.HBOC_v01 WARNING HTS
1 • Interpretation • C. Collett • 2022-08-09 11:59
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 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
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
Ouverture d'une analyse

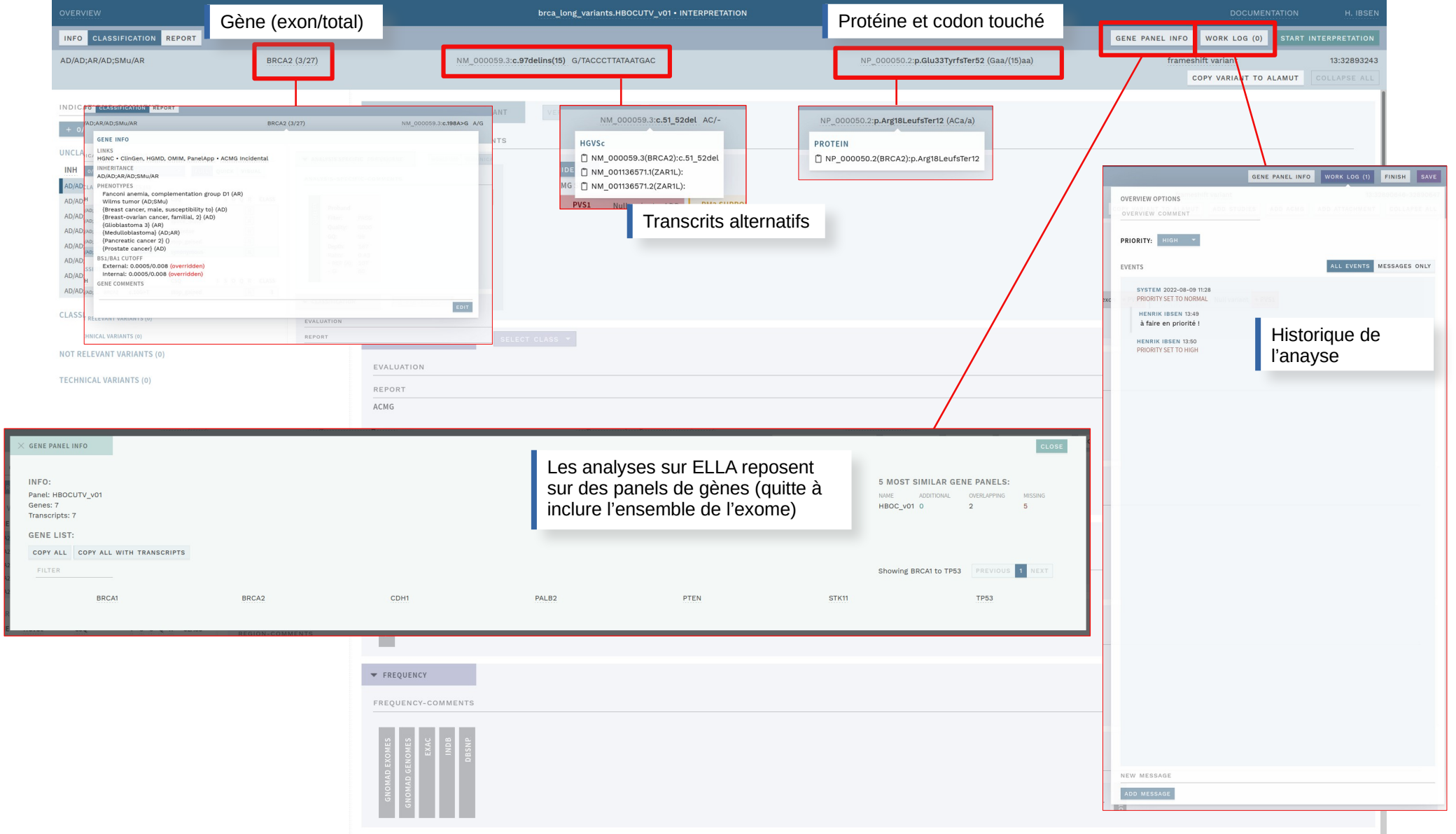
Écran d'ANALYSE

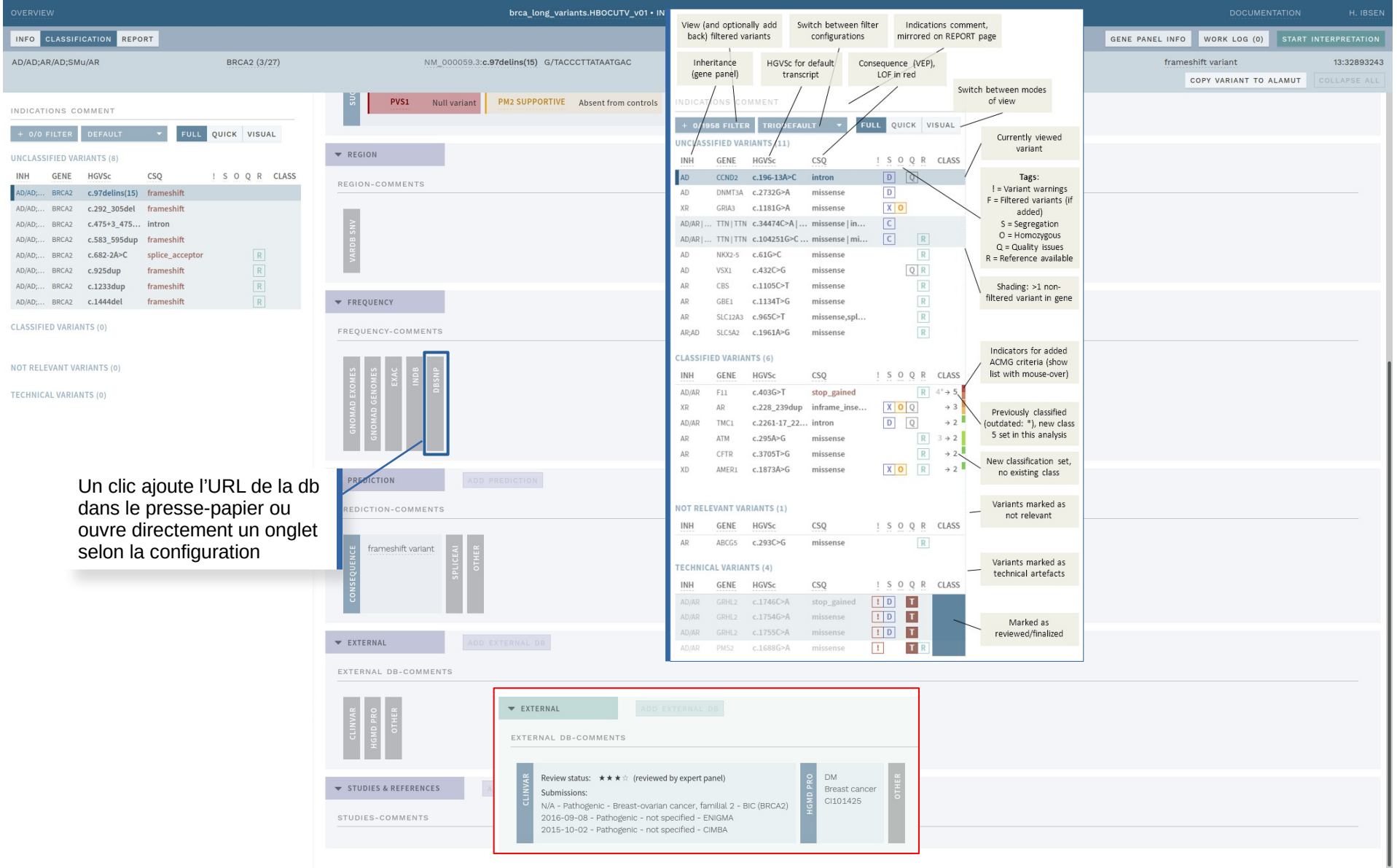
Équivaut à un mode read-only

Liste de variations retenues

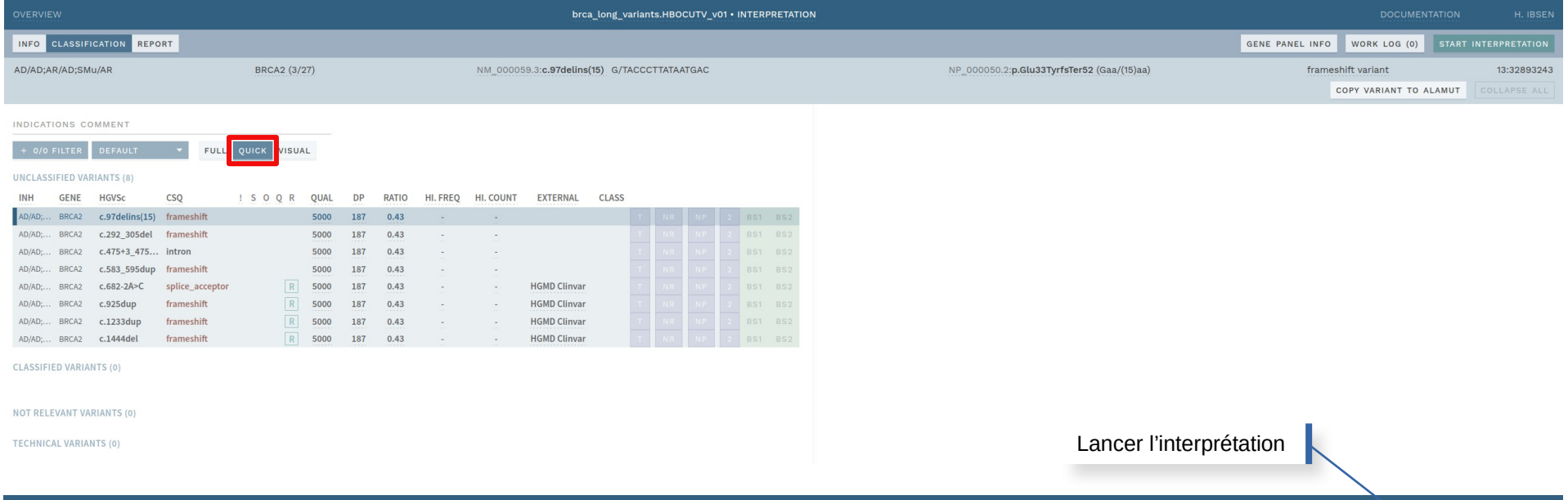
Détails de la variation *c.97delins(15)*



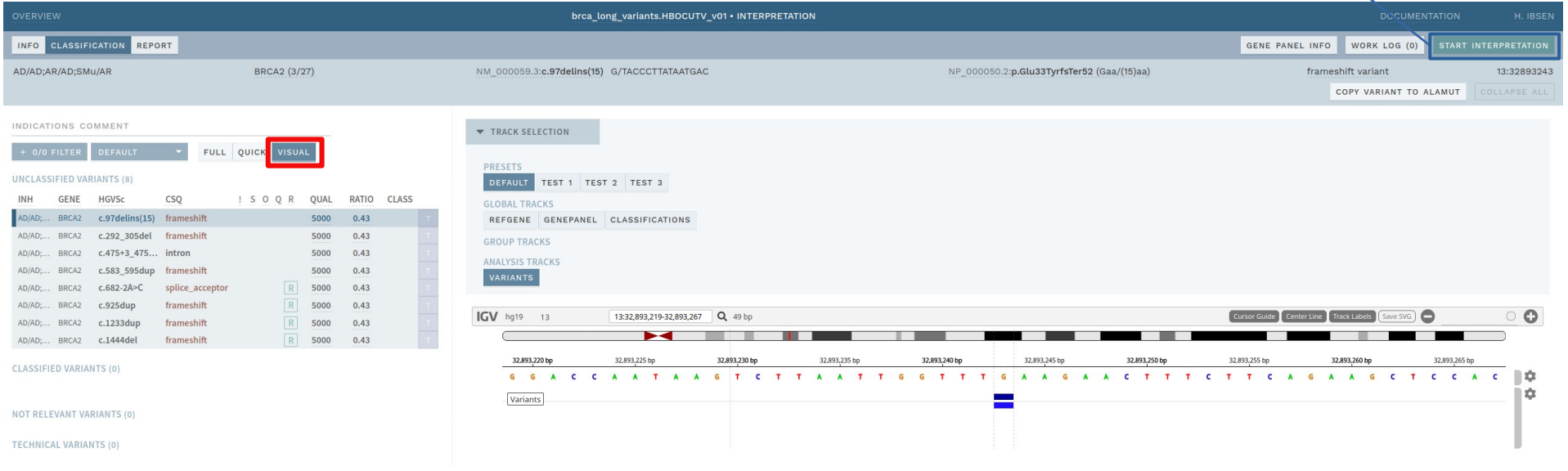

Gène (exon/total)
Protéine et codon touché
Transcrits alternatifs
Historique de l'anayse
Les analyses sur ELLA reposent sur des panels de gènes (quitte à inclure l'ensemble de l'exome)
OVERVIEW
brca_long_variants.HBOCUTV_v01 • INTERPRETATION
DOCUMENTATION
H. IBSEN
INFO
CLASSIFICATION
REPORT
GENE PANEL INFO
WORK LOG (0)
START INTERPRETATION
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
COLLAPSE ALL
NM_000059.3:c.51_52del AC/-
HGVSc
NM_000059.3(BRCA2):c.51_52del
NM_001136571.1(ZAR1L):
NM_001136571.2(ZAR1L):
PVS1
NP_000050.2:p.Arg18LeufsTer12 (ACa/a)
PROTEIN
NP_000050.2(BRCA2):p.Arg18LeufsTer12
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} ()
{Prostate cancer} (AD)
BS1/BA1 CUTOFF
External: 0.0005/0.008 (overridden)
Internal: 0.0005/0.008 (overridden)
GENE COMMENTS
EDIT
NOT RELEVANT VARIANTS (0)
TECHNICAL VARIANTS (0)
EVALUATION
REPORT
ACMG
SELECT CLASS
GENE PANEL INFO
CLOSE
INFO:
Panel: HBOCUTV_v01
Genes: 7
Transcripts: 7
GENE LIST:
COPY ALL
COPY ALL WITH TRANSCRIPTS
FILTER
BRCA1
BRCA2
CDH1
PALB2
PTEN
STK11
TP53
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 EXOMES
GNOMAD GENOMES
EXAC
INDB
DBSNP
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 13:50
PRIORITY SET TO HIGH
NEW MESSAGE
ADD MESSAGE

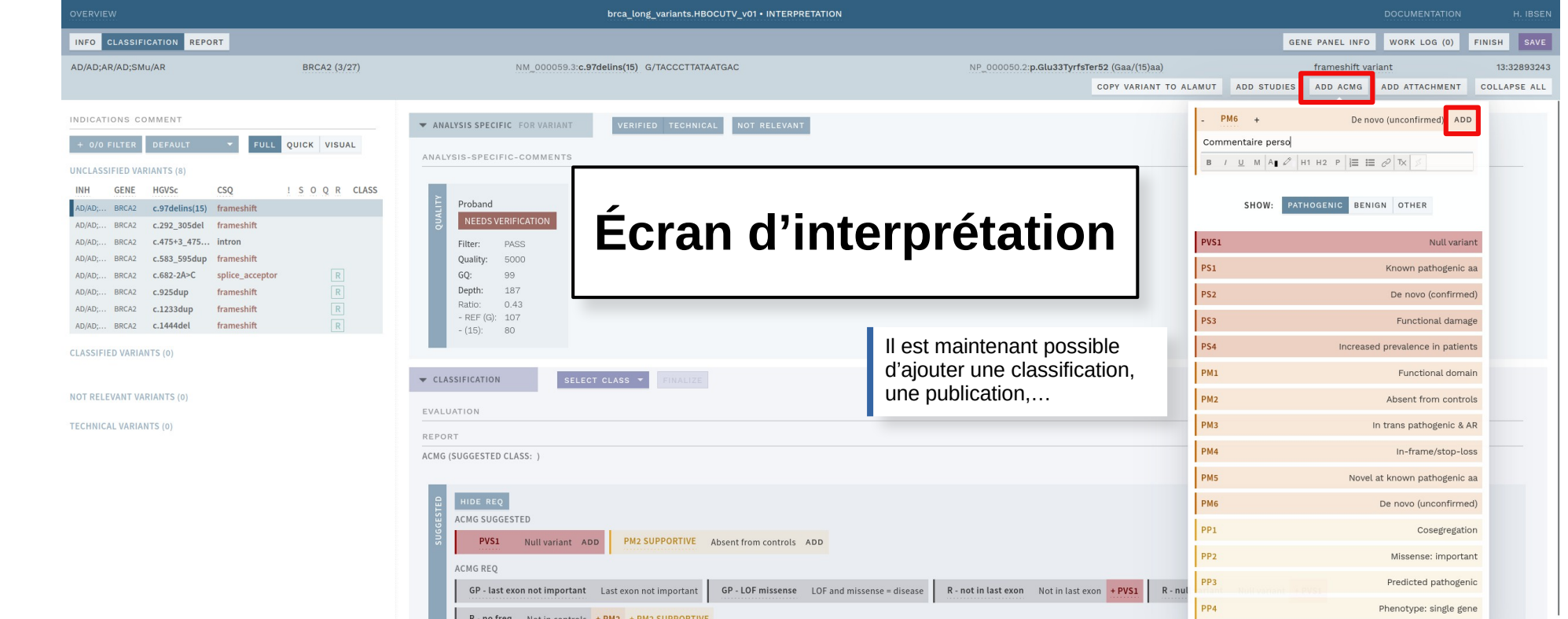


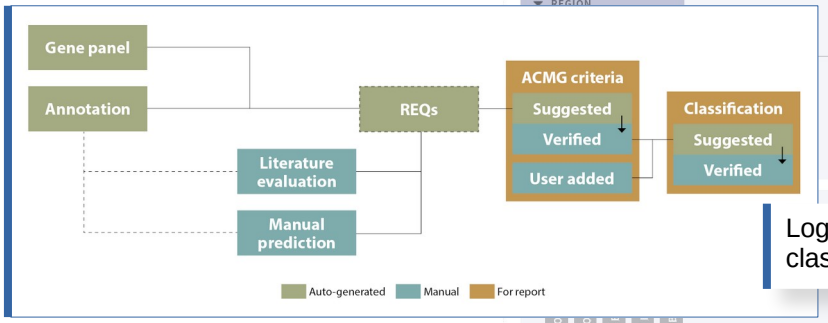
Un clic ajoute l'URL de la db dans le presse-papier ou ouvre directement un onglet selon la configuration



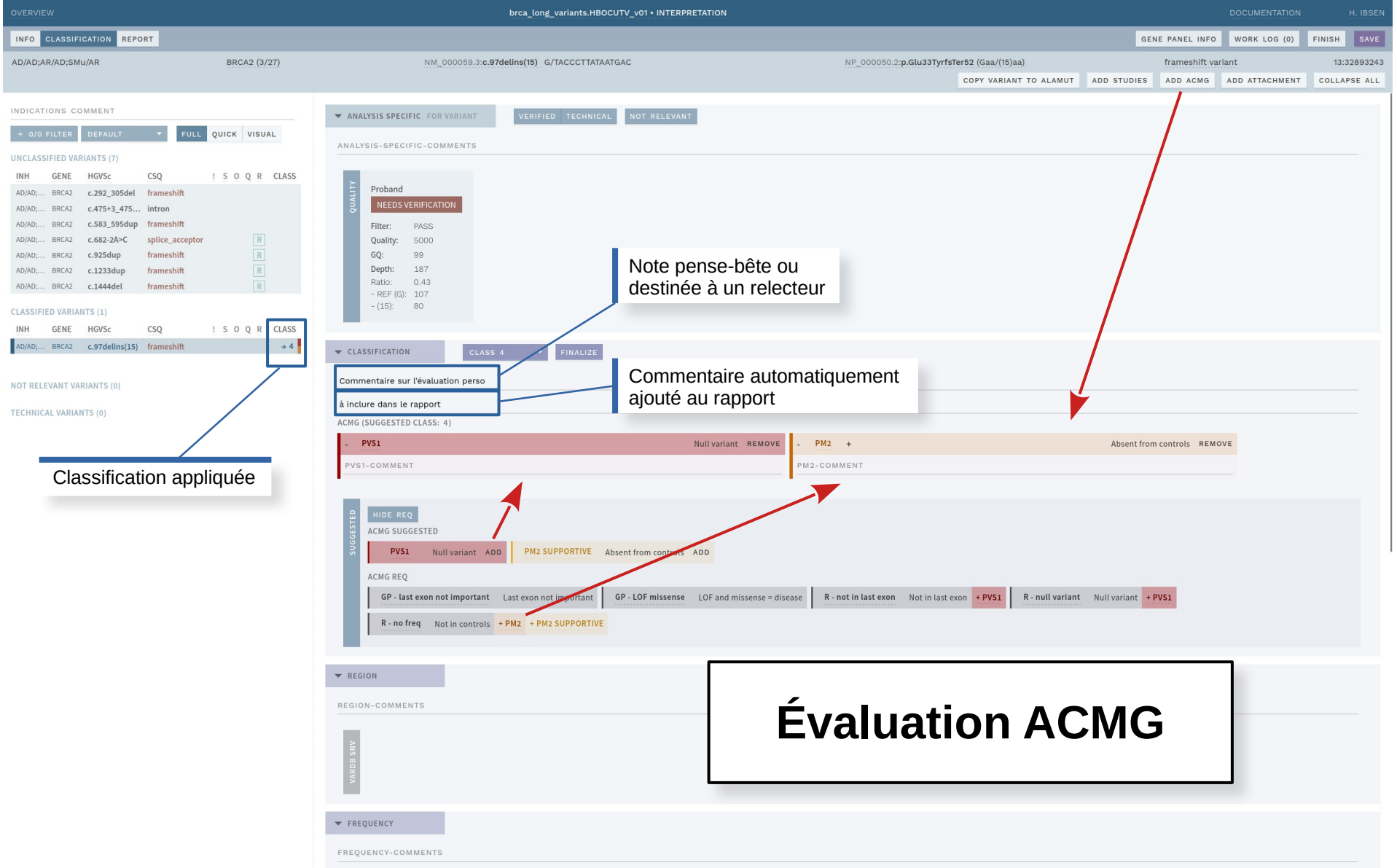
Lancer l'interprétation

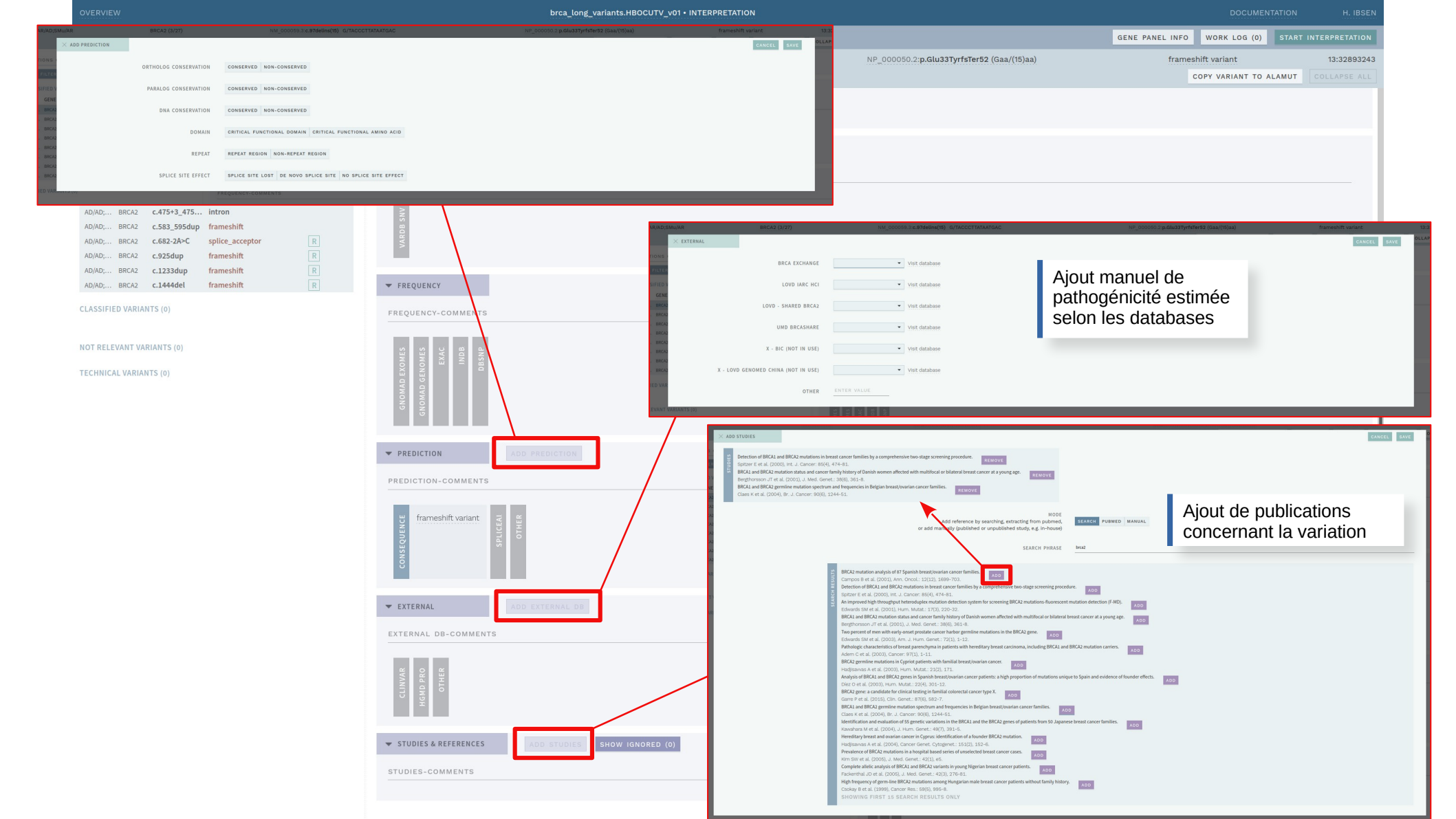


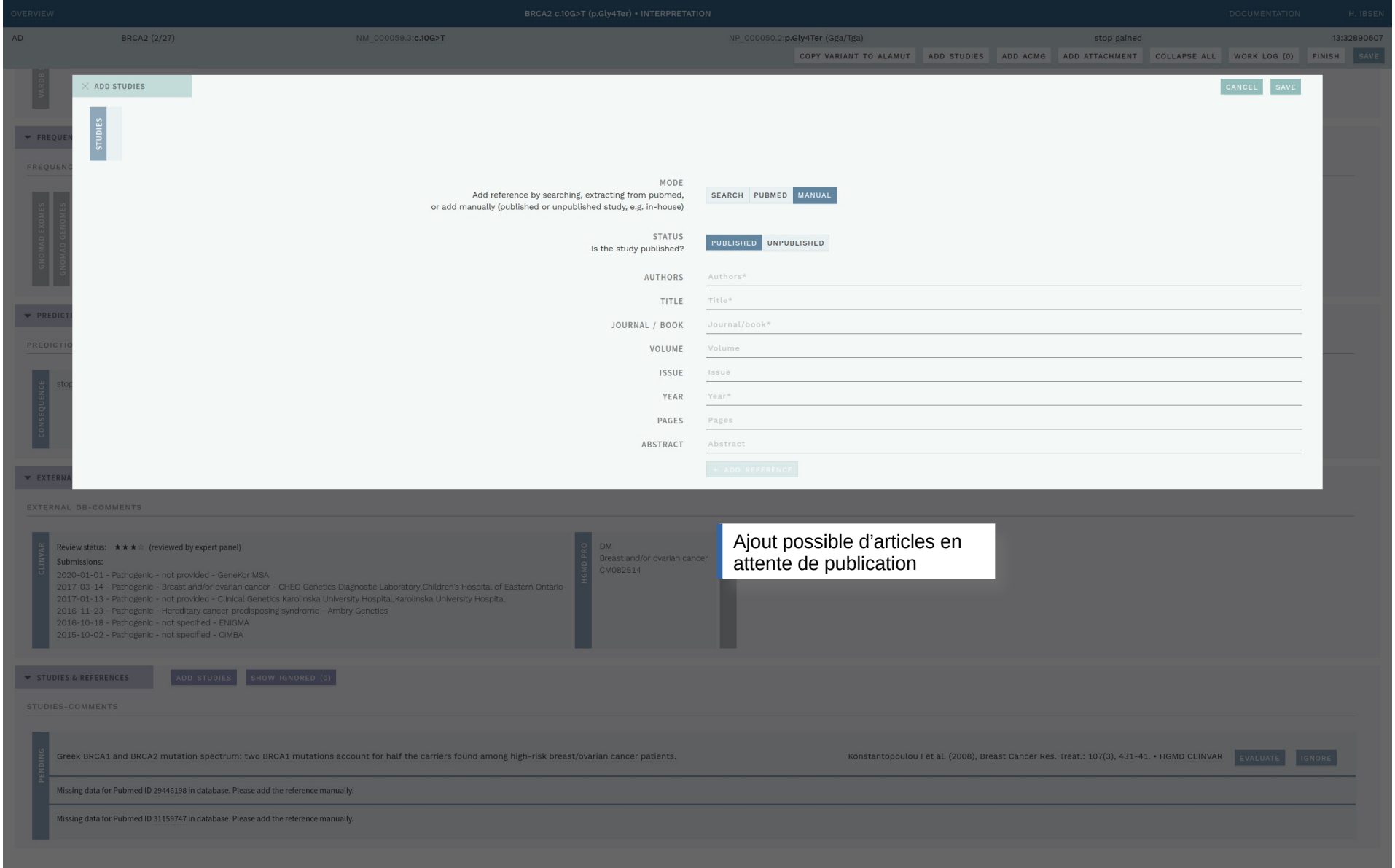


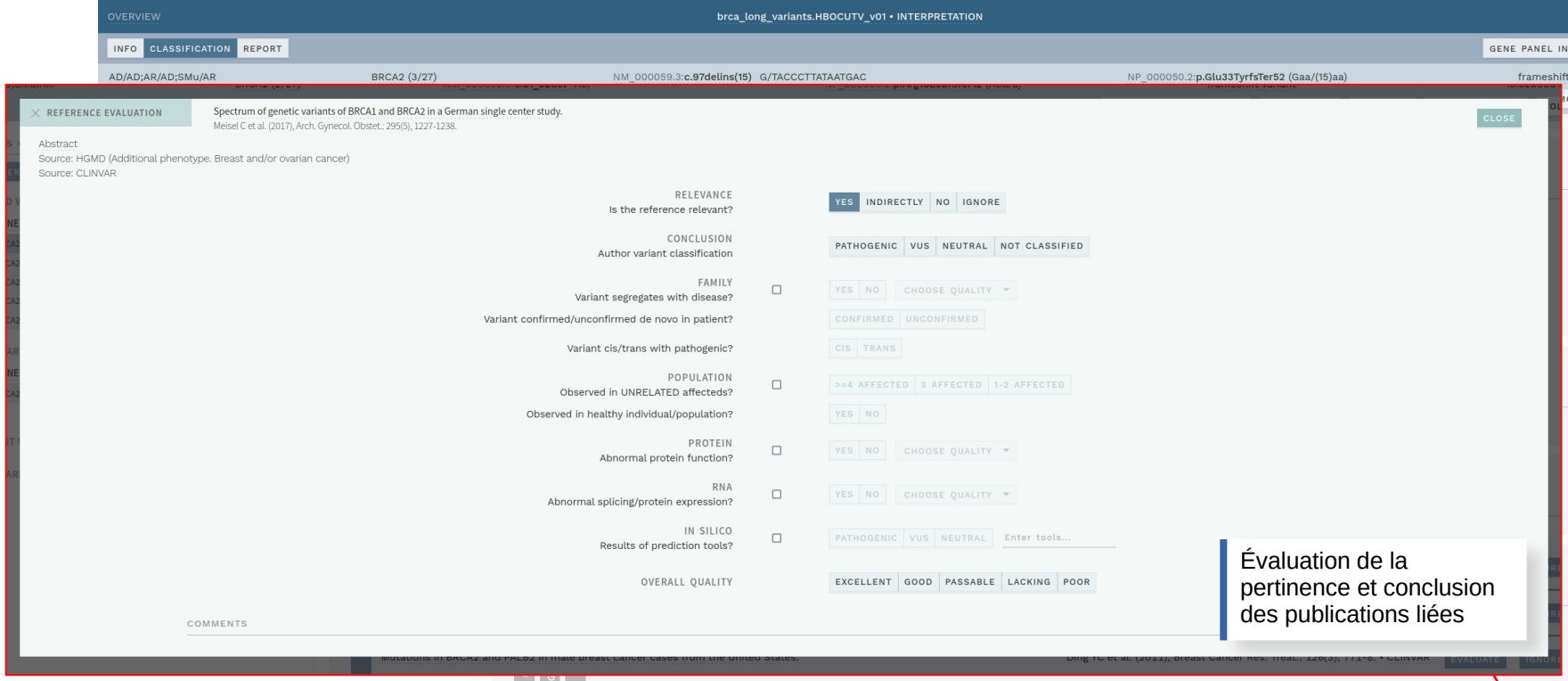


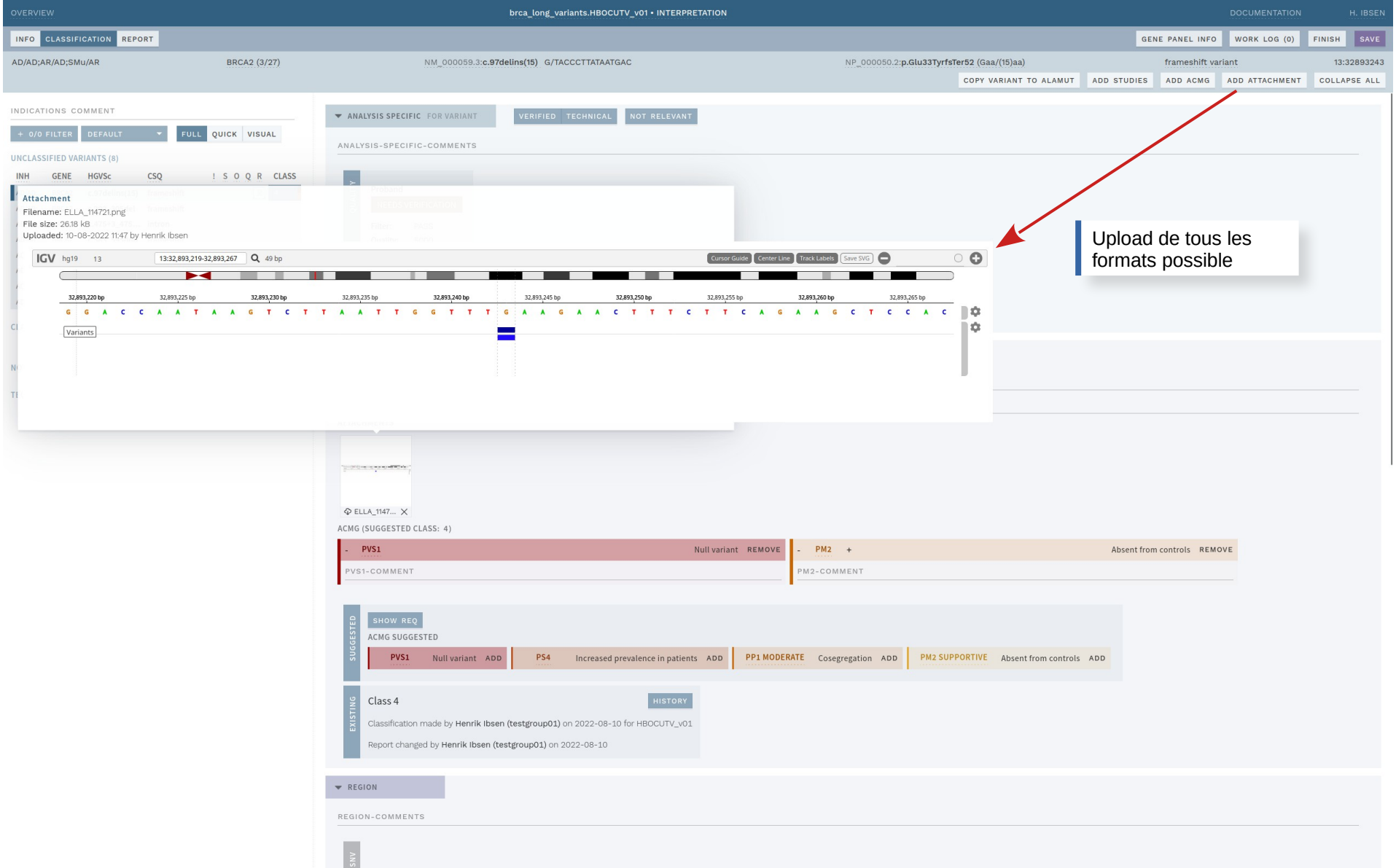
Logiciel focalisé sur la classification ACMG


OVERVIEW
brca_long_variants.HBOCUTV_v01 • INTERPRETATION
DOCUMENTATION
H. IBSEN
INFO
CLASSIFICATION
REPORT
GENE PANEL INFO
WORK LOG (0)
FINISH
SAVE
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 (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
PVS1-COMMENT
- PM2 + Absent from controls REMOVE
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
REGION
REGION-COMMENTS
VARDB SNV
Évaluation ACMG
FREQUENCY
FREQUENCY-COMMENTS


OVERVIEW
brca_long_variants.HBOCUTV_v01 • INTERPRETATION
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
ADD PREDICTION
CANCEL
SAVE
ORTHOLOG CONSERVATION
CONSERVED
NON-CONSERVED
PARALOG CONSERVATION
DNA CONSERVATION
DOMAIN
CRITICAL FUNCTIONAL DOMAIN
CRITICAL FUNCTIONAL AMINO ACID
REPEAT
REPEAT REGION
NON-REPEAT REGION
SPLICE SITE EFFECT
SPLICE SITE LOST
DE NOVO SPLICE SITE
NO SPLICE SITE EFFECT
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)
VARDB SNV
FREQUENCY
FREQUENCY-COMMENTS
GNOMAD EXOMES
GNOMAD GENOMES
EXAC
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
ADD STUDIES
SHOW IGNORED (0)
STUDIES-COMMENTS
EXTERNAL
BRCA EXCHANGE
LOVD IARC HCI
LOVD - SHARED BRCA2
UMD BRCASHARE
X - BIC (NOT IN USE)
X - LOVD GENOMED CHINA (NOT IN USE)
OTHER
ENTER VALUE
Visit database
Ajout manuel de pathogénicité estimée selon les databases
ADD STUDIES
STUDIES
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: 89(4), 474-81.
BRCA1 and BRCA2 mutation status and cancer family history of Danish women affected with multifocal or bilateral breast cancer at a young age.
Bergthorsson JT et al. (2001), J. Med. Genet.: 38(6), 361-8.
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.
REMOVE
MODE
Add reference by searching, extracting from pubmed, or add manually (published or unpublished study, e.g. in-house)
SEARCH
PUBMED
MANUAL
SEARCH PHRASE
brca2
SEARCH RESULTS
BRCA2 mutation analysis of 87 Spanish breast/ovarian cancer families.
Campos B et al. (2001), Ann. Oncol.: 12(12), 1699-703.
ADD
Ajout de publications concernant la variation
SHOWING FIRST 15 SEARCH RESULTS ONLY


OVERVIEW
BRCA2 c.10G>T (p.Gly4Ter) • INTERPRETATION
DOCUMENTATION
H. IBSEN
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
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.


OVERVIEW
brca_long_variants.HBOCUTV_v01 • INTERPRETATION
DOCUMENTATION
H. IBSEN
INFO
CLASSIFICATION
REPORT
GENE PANEL INFO
WORK LOG (0)
FINISH
SAVE
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
ADD ATTACHMENT
COLLAPSE ALL
REFERENCE EVALUATION
Spectrum of genetic variants of BRCA1 and BRCA2 in a German single center study.
Meisel C et al. (2017), Arch. Gynecol. Obstet.: 295(5), 1227-1238.
CLOSE
Abstract
Source: HGMD (Additional phenotype. Breast and/or ovarian cancer)
Source: CLINVAR
RELEVANCE
Is the reference relevant?
YES
INDIRECTLY
NO
IGNORE
CONCLUSION
Author variant classification
PATHOGENIC
VUS
NEUTRAL
NOT CLASSIFIED
FAMILY
Variant segregates with disease?
YES
NO
CHOOSE QUALITY
Variant confirmed/unconfirmed de novo in patient?
CONFIRMED
UNCONFIRMED
Variant cis/trans with pathogenic?
CIS
TRANS
POPULATION
Observed in UNRELATED affecteds?
>=4 AFFECTED
3 AFFECTED
1-2 AFFECTED
Observed in healthy individual/population?
YES
NO
PROTEIN
Abnormal protein function?
YES
NO
CHOOSE QUALITY
RNA
Abnormal splicing/protein expression?
YES
NO
CHOOSE QUALITY
IN SILICO
Results of prediction tools?
PATHOGENIC
VUS
NEUTRAL
Enter tools...
OVERALL QUALITY
EXCELLENT
GOOD
PASSABLE
LACKING
POOR
COMMENTS
Évaluation de la pertinence et conclusion des publications liées
STUDIES & REFERENCES
ADD STUDIES
SHOW IGNORED (0)
STUDIES-COMMENTS
EVALUATED
Published studies
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
RE-EVALUATE
IGNORE
EVALUATION
PENDING
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
EVALUATE
IGNORE
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
EVALUATE
IGNORE


OVERVIEW
brca_long_variants.HBOCUTV_v01 • INTERPRETATION
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H. IBSEN
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CLASSIFICATION
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GENE PANEL INFO
WORK LOG (0)
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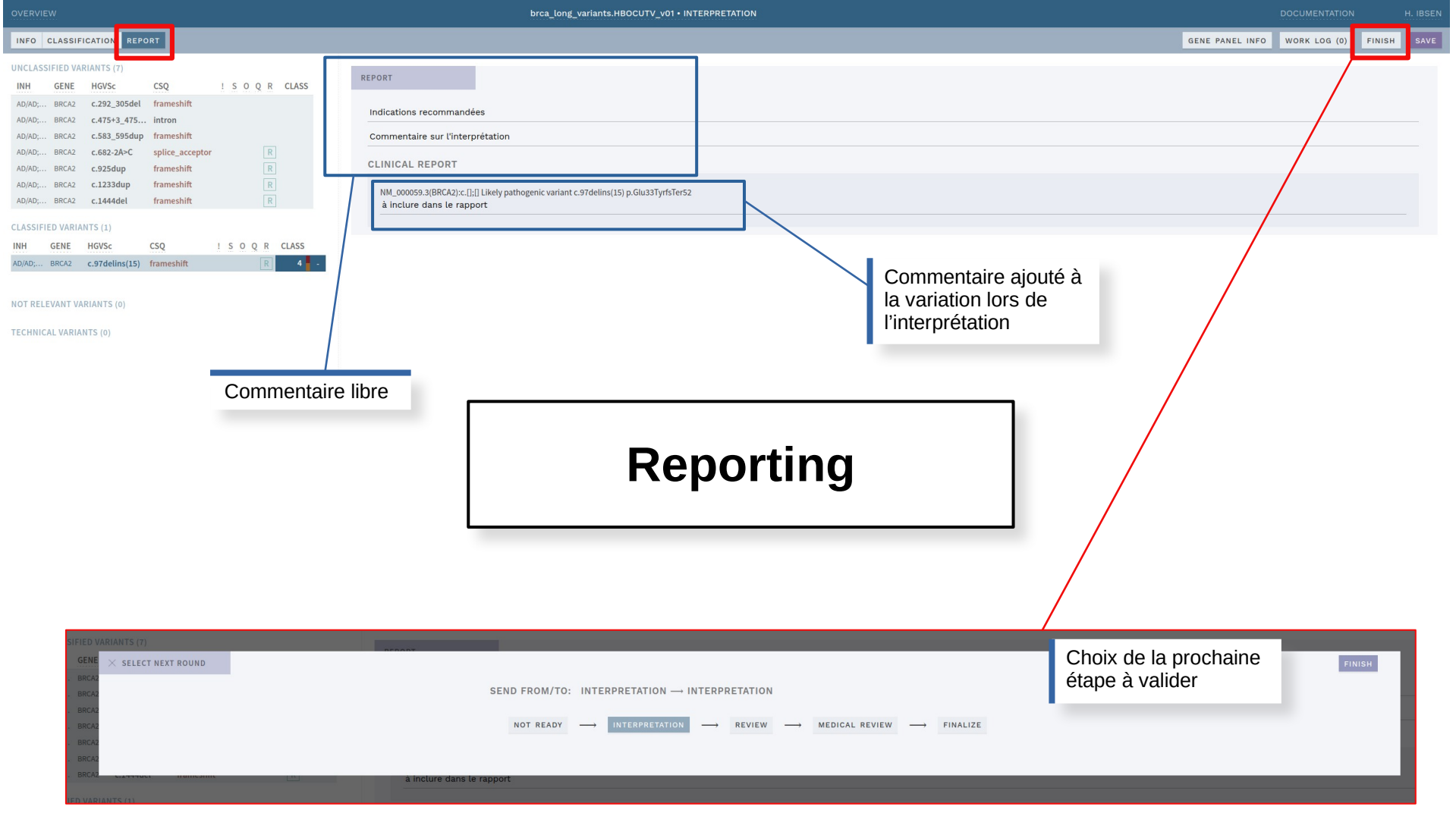
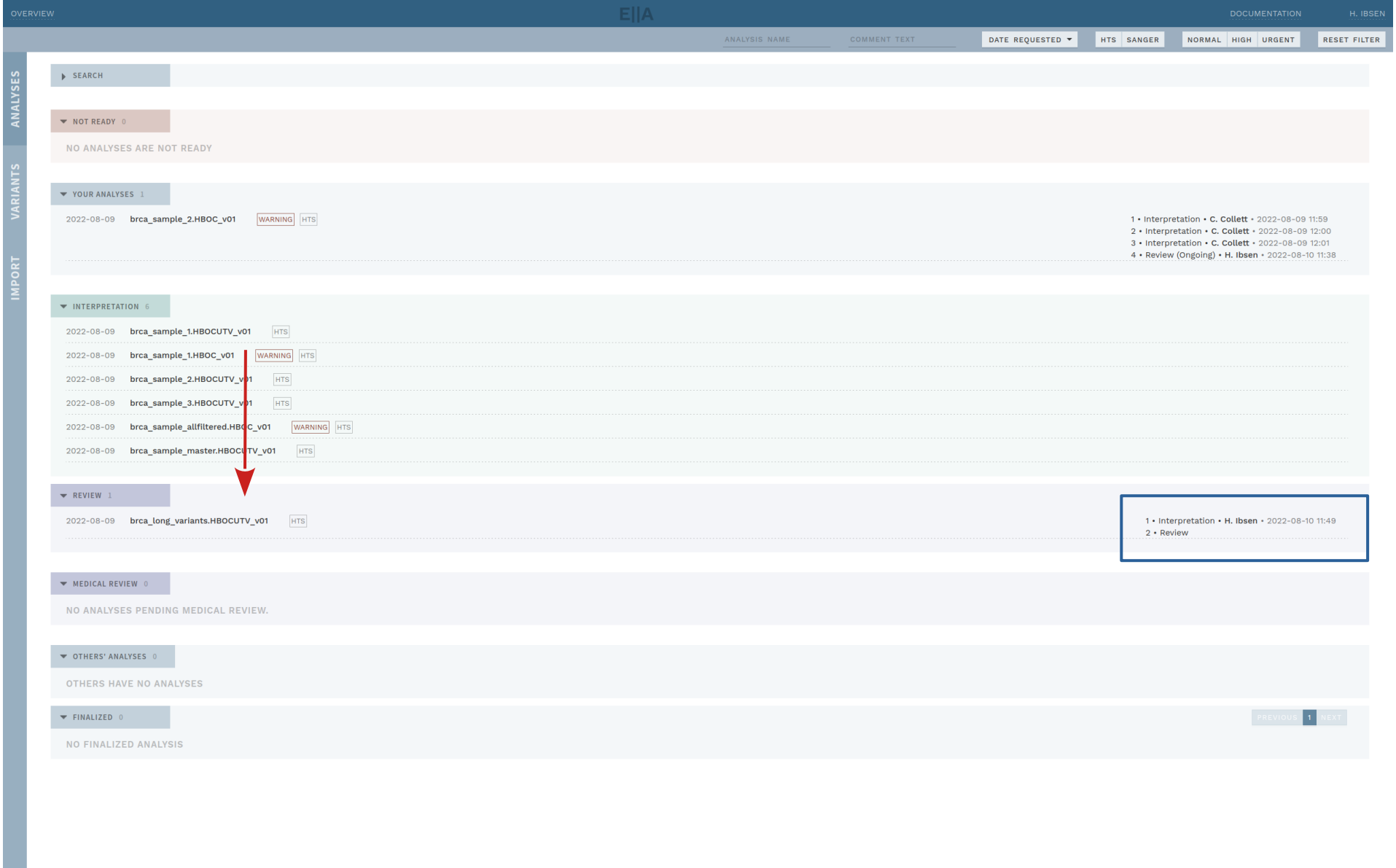
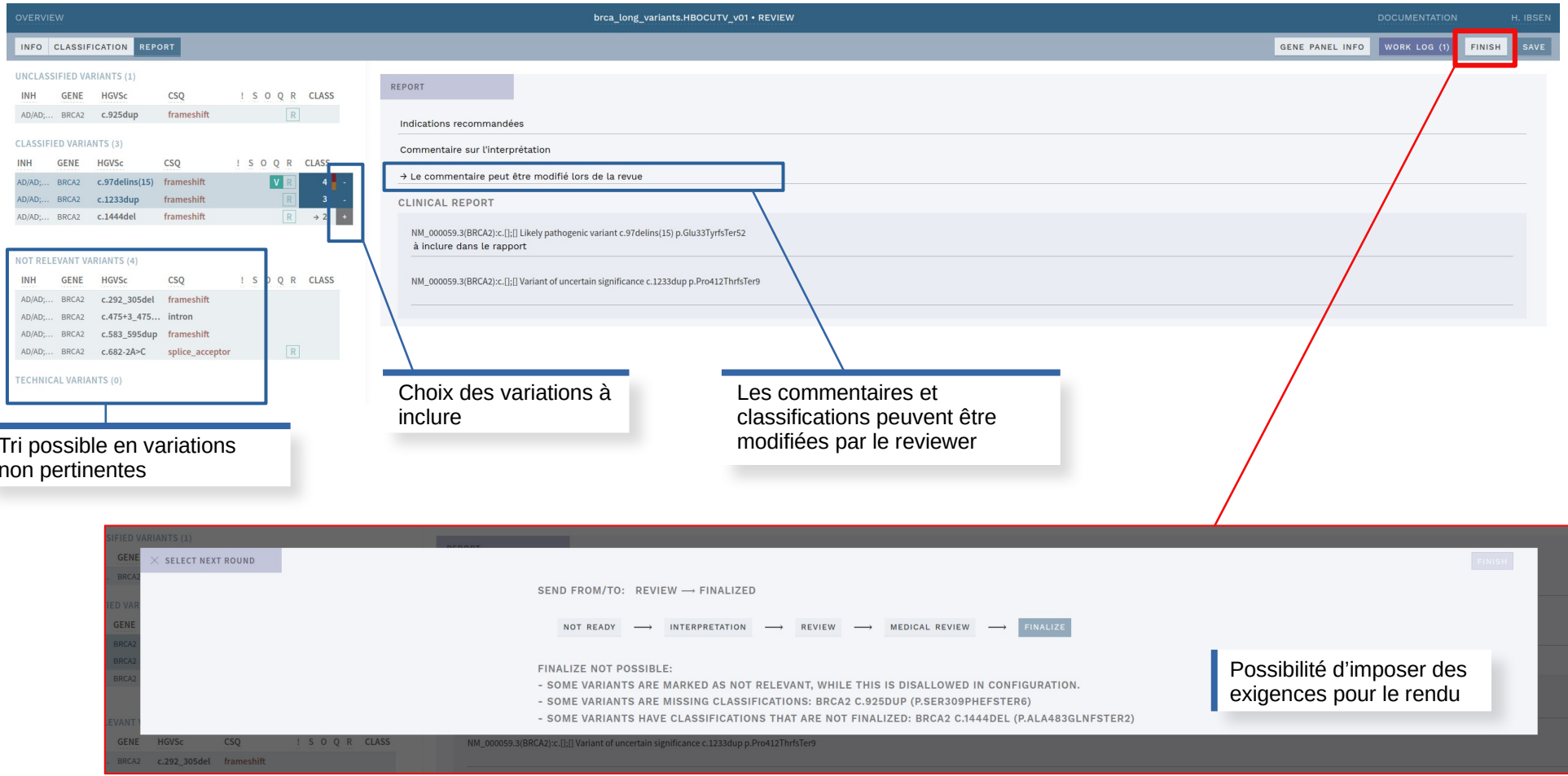
SAVE
AD/AD;AR/AD;SMu/AR
BRCA2 (3/27)
NM_000059.3:c.97delins(15) G/TACCCTTATAATGAC
NP_000050.2:p.Glu33TyrfsTer52 (Gaa/(15)aa)
frameshift variant
13:32893243
COPY VARIANT TO ALAMUT
ADD STUDIES
ADD ACMG
ADD ATTACHMENT
COLLAPSE ALL
INDICATIONS COMMENT
0/0 FILTER
DEFAULT
FULL
QUICK
VISUAL
UNCLASSIFIED VARIANTS (8)
INH
GENE
HGVSc
CSQ
CLASS
ANALYSIS SPECIFIC FOR VARIANT
VERIFIED
TECHNICAL
NOT RELEVANT
ANALYSIS-SPECIFIC-COMMENTS
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
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 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
Upload de tous les formats possible
ELLA_1147...
ACMG (SUGGESTED CLASS: 4)
- PVS1 Null variant REMOVE
PVS1-COMMENT
- PM2 + Absent from controls REMOVE
PM2-COMMENT
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


OVERVIEW
brca_long_variants.HBOCUTV_v01 • INTERPRETATION
DOCUMENTATION
H. IBSEN
INFO
CLASSIFICATION
REPORT
GENE PANEL INFO
WORK LOG (0)
FINISH
SAVE
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.[];[] Likely pathogenic variant c.97delins(15) p.Glu33TyrfsTer52
à inclure dans le rapport
Commentaire ajouté à la variation lors de l'interprétation
Commentaire libre
Reporting
SELECT NEXT ROUND
SEND FROM/TO: INTERPRETATION ⟶ INTERPRETATION
NOT READY ⟶ INTERPRETATION ⟶ REVIEW ⟶ MEDICAL REVIEW ⟶ FINALIZE
FINISH
Choix de la prochaine étape à valider


OVERVIEW
EIIA
DOCUMENTATION
H. IBSEN
ANALYSIS NAME
COMMENT TEXT
DATE REQUESTED
HTS
SANGER
NORMAL
HIGH
URGENT
RESET FILTER
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
PREVIOUS
1
NEXT

Revue

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

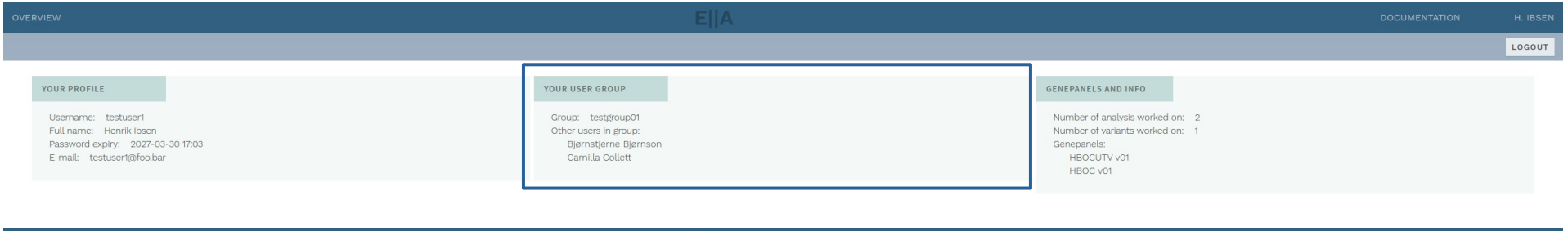


Choix des variations à inclure

Les commentaires et classifications peuvent être modifiées par le reviewer

Tri possible en variations non pertinentes

Possibilité d'imposer des exigences pour le rendu



≠

Gestion des droits

ANALYSIS INFO

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

SAMPLES

PROBAND	Sample name:	HG002	MOTHER	Sample name:	HG004	FATHER	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+1i…	frameshift, s…	!	D				
AR	PRKRA	c.785-2_785-…	splice_acceptor	!	D			R	
AR	PRKRA	c.785-5_785-…	splice_region…	!	D		Q		
AR	FCGR3A	c.512T>A	missense	!	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…	!	M		Q		
AD/AR	MSH2	c.942+2del	splice_donor		M			R	
AR	ARSB	c.312+3A>C	splice_region…	!	M				
AR	ARSB	c.312+2T>C	splice_donor	!	M				
AR	SERPINB8	c.197T>C	missense		M				
AD	BRAF	c.2128-5dup	splice_region…	!		O	Q		
AD/AR	F11	c.403G>T	stop_gained					R	
AD/AR	HTT	c.96_110dup	inframe_inse…	!			Q		
AR	STRC	c.3307-26_33…	intron	!			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

VARDB SNV

FREQUENCY

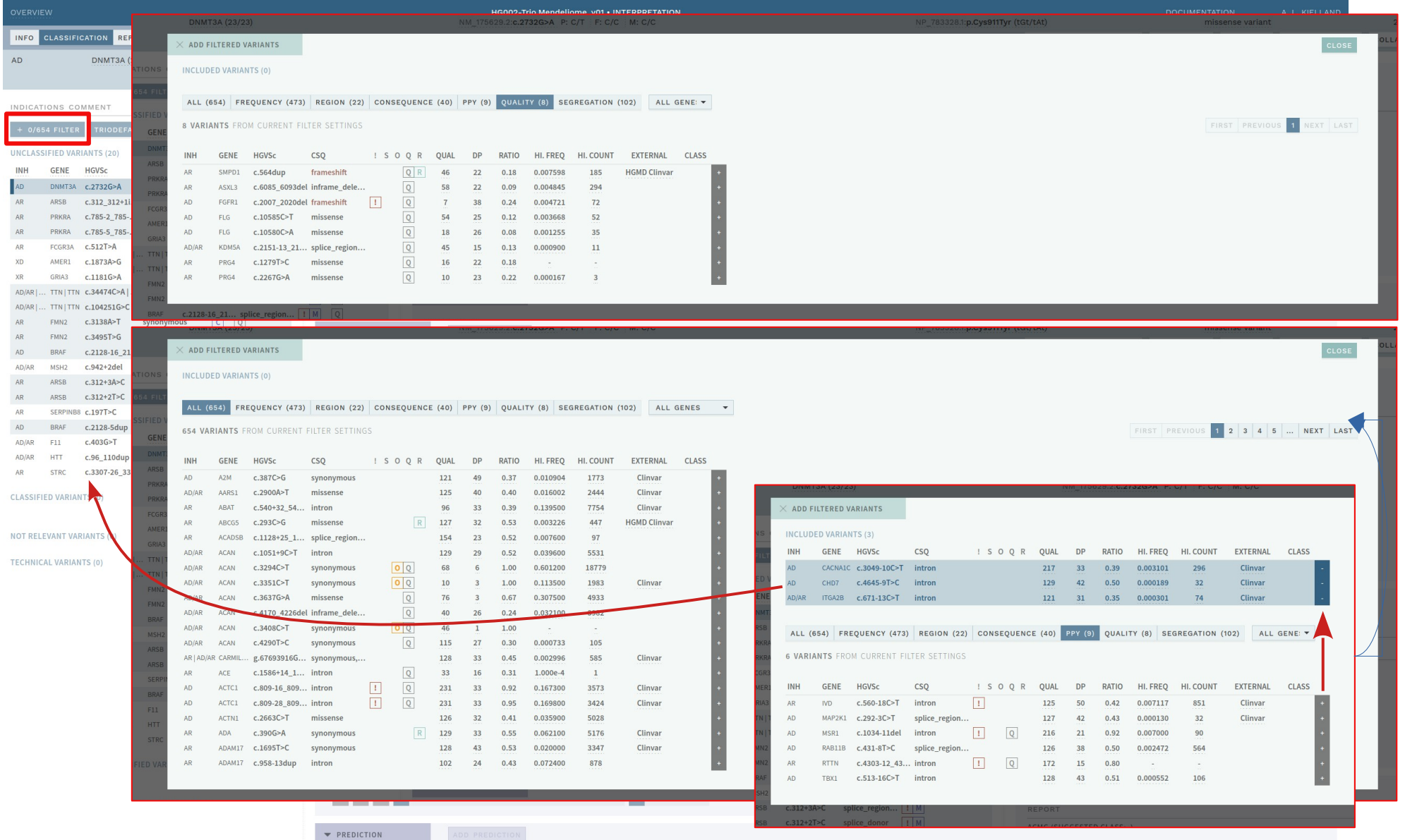
FREQUENCY-COMMENTS

GNOMAD EXOMES GNOMAD GENOMES EXAC INDB

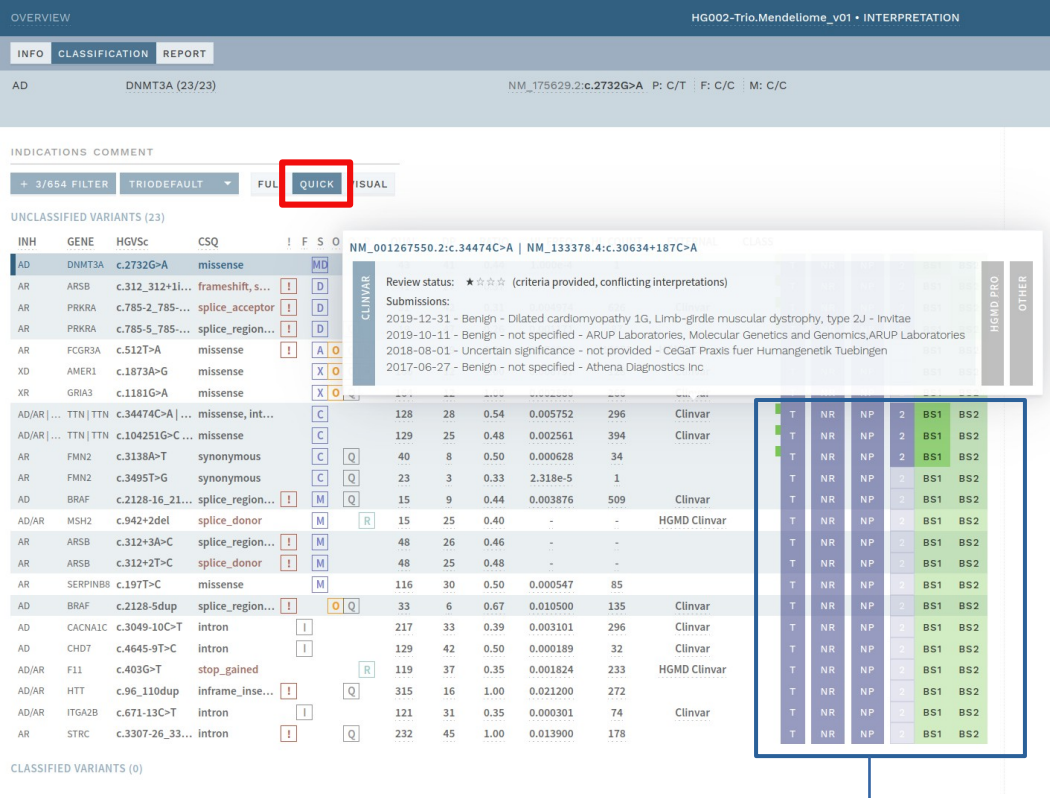
POP	COUNT	NUM	HOM	FREQ
OUSWES:	1	12840	0	1.000e-4
OUSWGS:				
OUSWES indications:				
- PU:		1		

DBSNP rs906113912

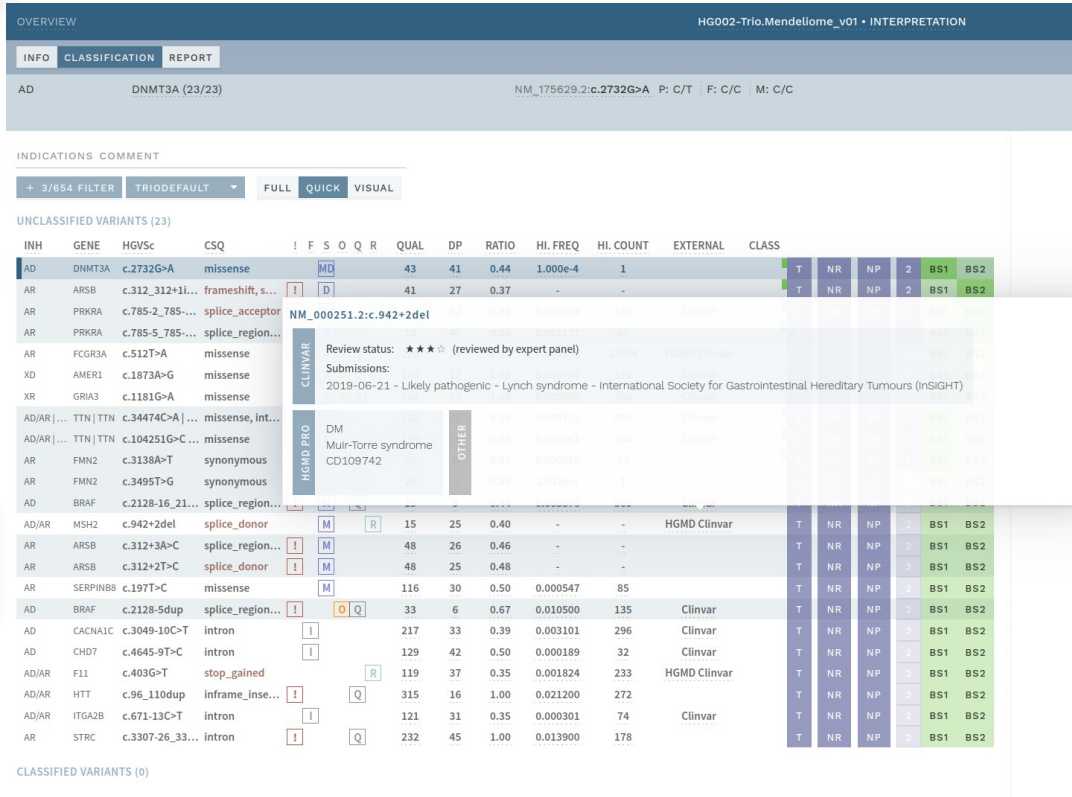
PREDICTION ADD PREDICTION

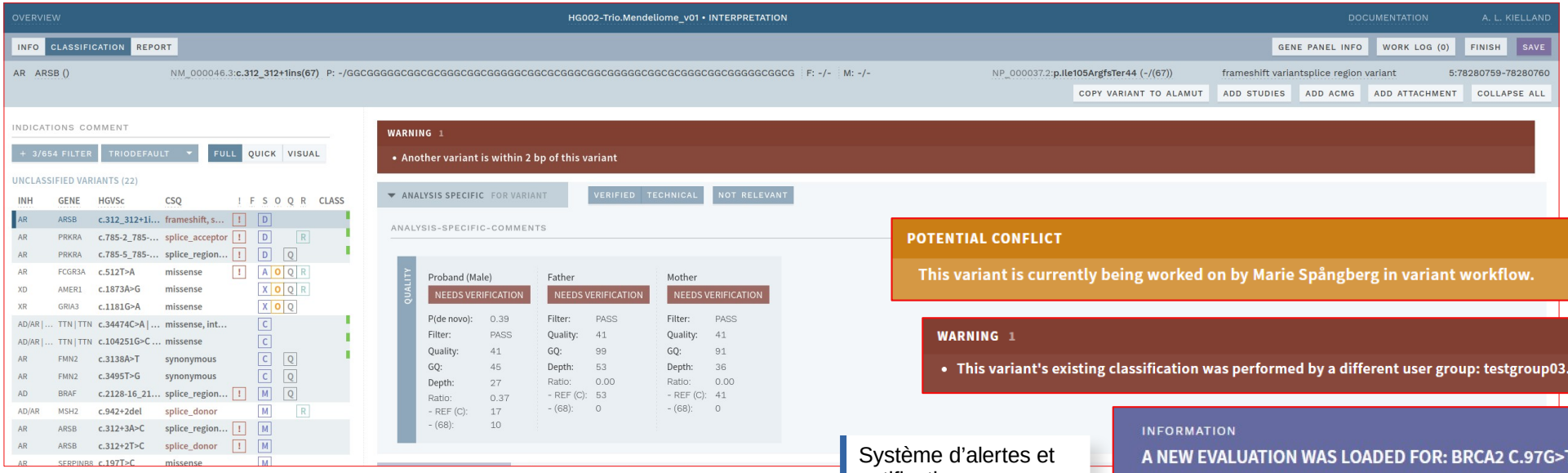

OVERVIEW
HG002-Trio Mendeliome_v01 • INTERPRETATION
DOCUMENTATION
INFO
CLASSIFICATION
AD
DNMT3A (
INDICATIONS COMMENT
+ 0/654 FILTER
TRIODEFA
UNCLASSIFIED VARIANTS (20)
INH GENE HGVSc
AD DNMT3A c.2732G>A
AR ARSB c.312_312+1
AR PRKRA c.785-2_785-
AR PRKRA c.785-5_785-
AR FCGR3A c.512T>A
XD AMER1 c.1873A>G
XR GRIA3 c.1181G>A
AD/AR | ... TTN | TTN c.34474C>A |
AD/AR | ... TTN | 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 SERPINB8 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 VARIANTS (
NOT RELEVANT VARIANTS (
TECHNICAL VARIANTS (0)
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
ADD FILTERED VARIANTS
CLOSE
INCLUDED VARIANTS (0)
ALL (654) FREQUENCY (473) REGION (22) CONSEQUENCE (40) PPY (9) QUALITY (8) SEGREGATION (102) ALL GENE
8 VARIANTS FROM CURRENT FILTER SETTINGS
FIRST PREVIOUS 1 NEXT LAST
INH GENE HGVSc CSQ ! S O Q R QUAL DP RATIO HI. FREQ HI. COUNT EXTERNAL CLASS
AR SMPD1 c.564dup frameshift Q R 46 22 0.18 0.007598 185 HGMD Clinvar
AR ASXL3 c.6085_6093del inframe_dele... Q 58 22 0.09 0.004845 294
AD FGFR1 c.2007_2020del frameshift ! Q 7 38 0.24 0.004721 72
AD FLG c.10585C>T missense Q 54 25 0.12 0.003668 52
AD FLG c.10580C>A missense Q 18 26 0.08 0.001255 35
AD/AR KDM5A c.2151-13_21... splice_region... Q 45 15 0.13 0.000900 11
AR PRG4 c.1279T>C missense Q 16 22 0.18 - -
AR PRG4 c.2267G>A missense Q 10 23 0.22 0.000167 3
c.2128-16_21... splice_region... ! M Q
synonymous C Q
ADD FILTERED VARIANTS
CLOSE
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
FIRST PREVIOUS 1 2 3 4 5 ... NEXT LAST
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 R 127 32 0.53 0.003226 447 HGMD Clinvar
AR ACADSB 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 O Q 68 6 1.00 0.601200 18779
AD/AR ACAN c.3351C>T synonymous O Q 10 3 1.00 0.113500 1983 Clinvar
AD/AR ACAN c.3637G>A missense Q 76 3 0.67 0.307500 4933
AD/AR ACAN c.4170_4226del inframe_dele... Q 40 26 0.24 0.032100 9582
AD/AR ACAN c.3408C>T synonymous O Q 46 1 1.00 - -
AD/AR ACAN c.4290T>C synonymous Q 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 Q 33 16 0.31 1.000e-4 1
AD ACTC1 c.809-16_809... intron ! Q 231 33 0.92 0.167300 3573 Clinvar
AD ACTC1 c.809-28_809... intron ! Q 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 R 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
ADD FILTERED VARIANTS
INCLUDED VARIANTS (3)
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 GENE
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 ! Q 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 ! Q 172 15 0.80 - -
AD TBX1 c.513-16C>T intron 128 43 0.51 0.000552 106
c.312+3A>C splice_region... ! M
c.312+2T>C splice_donor ! M
REPORT
PREDICTION
ADD PREDICTION

Classification rapide

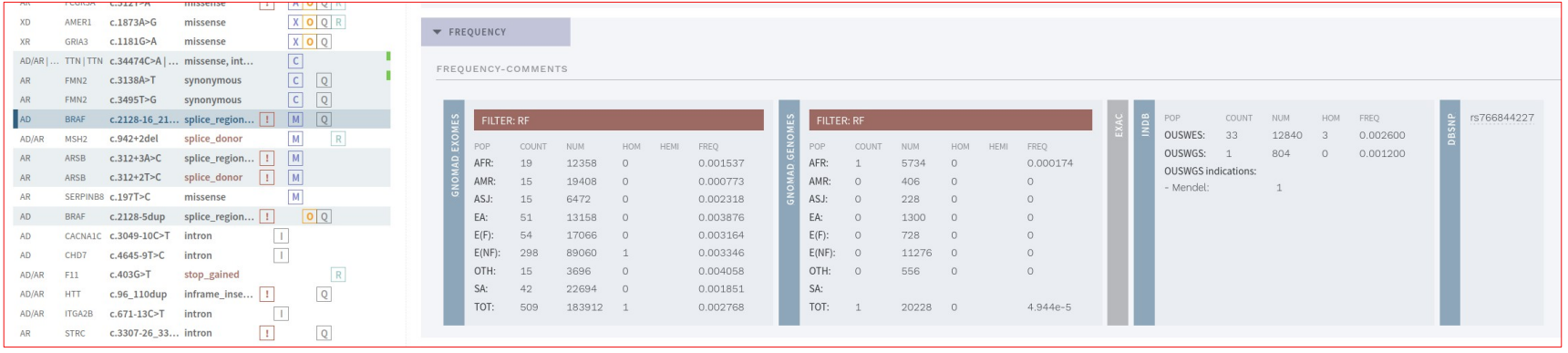


Classification bénigne/non pertinente en un clic





Système d'alertes et notifications



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 appliquer 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 }
    },
}
```