Nom du filtre global

Groupes d’utilisateurs y ayant accès

Requis

Appliqué aux séquençages

Analyse en trio dont le nom ne commence pas par "brca"

Nom d’une étape du filtre

db externes

Populations utilisées

db interne

Filtre par allele count

Moins de 5000 occurrences pour chaque population

Filtre par fréquence

Thresholds dans les gènes AD

< 0,5 % dans les db externes et < 5 % dans la db inhouse

Thresholds pour les autres

Exceptions pour "sauver" des variations

Si classification interne ≥ 3

ou si sur Clinvar, davantage de soumissions pathogènes que bénins avec une créance d'au moins deux étoiles,

la variation ne sera pas filtrée.

Filtre les variations hors des régions d'épissage ou UTR définies

Ici, celles situées avant les 20 bases précédant un début d'exon et après les 6 bases suivant une fin d'exon

… sauf exceptions


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        "FIN": 5000,
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        "OTH": 5000,
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    },
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          "combinations": [["pathogenic", ">", "benign"]]
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          "transcript_ablation",
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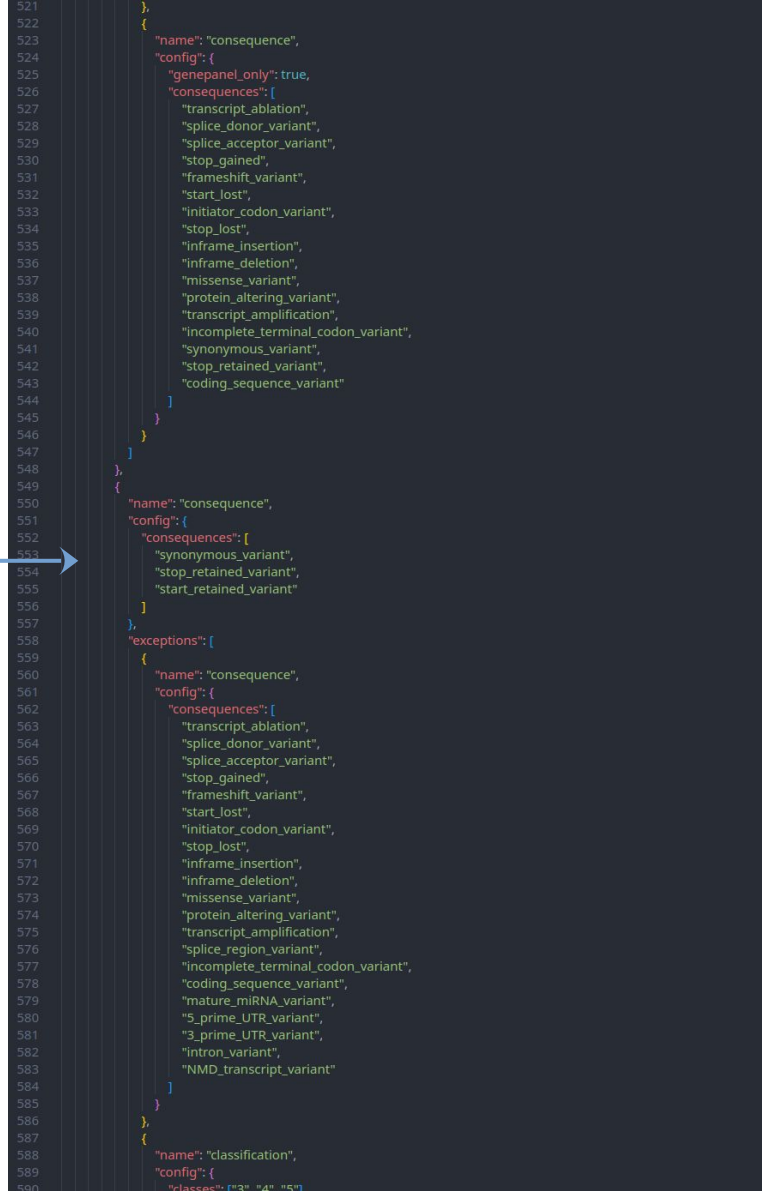
Filtre selon les conséquences estimées par VEP

… sauf exceptions



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          "inframe_deletion",
          "missense_variant",
          "protein_altering_variant",
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          "incomplete_terminal_codon_variant",
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          "NMD_transcript_variant"
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    {
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      "config": {
```

Filtre les variations C>T, T>C, delCC, delTT, delCT et delTC dans le tractus polypyrimidine, la région précisée ici entre 20 et 3 bases précédant un début d'exon

Filtre les variations avec une balance allélique < 25 %

Filtre par ségrégation

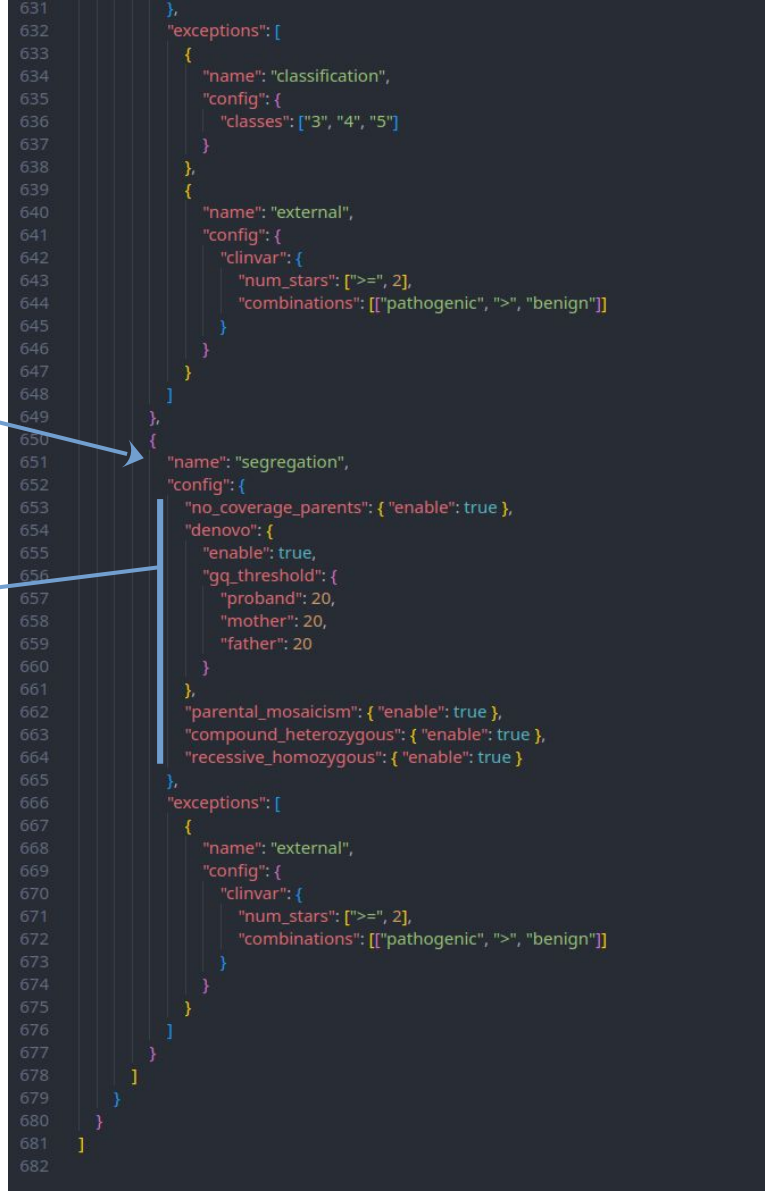
Garde les variations dont le génotype d'un parent est manquant

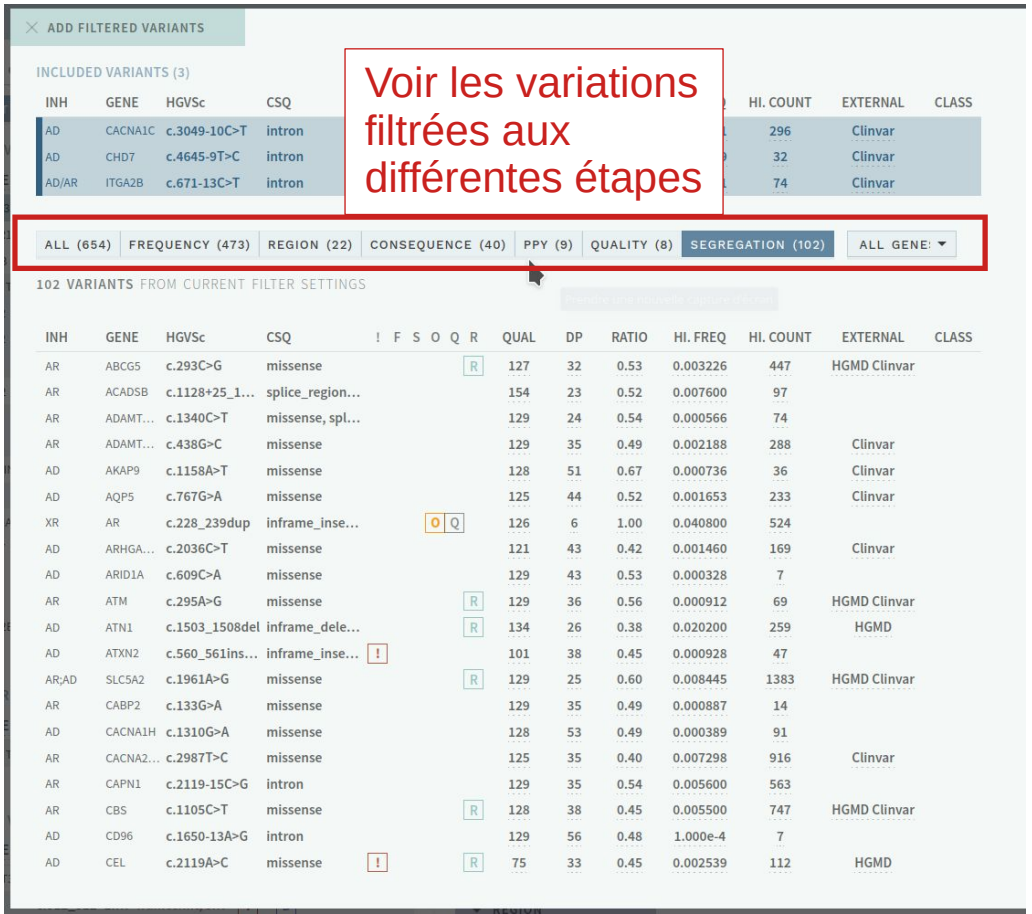
Garde les variations estimées de novo avec des GQ > 20

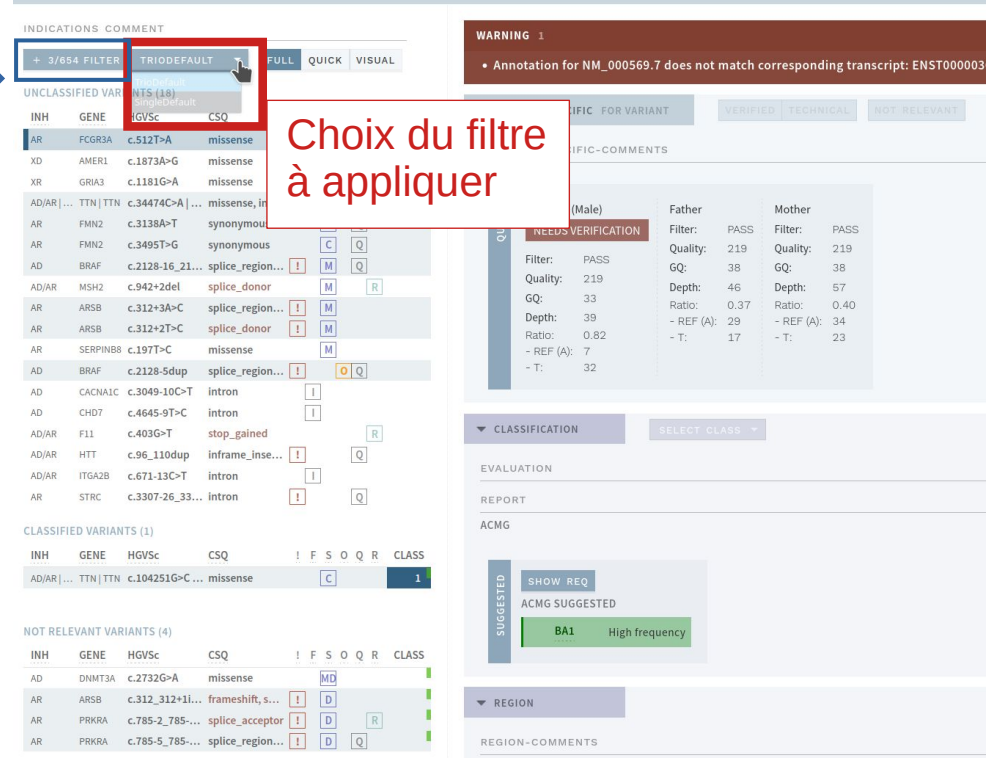
Garde les variations pouvant être issues d'une mosaïque parentale

Garde les variations pouvant causer une hétérozygotie composite

Garde les variations pouvant causer une maladie récessive




ADD FILTERED VARIANTS
INCLUDED VARIANTS (3)
INH GENE HGVSc CSQ HI. COUNT EXTERNAL CLASS
AD CACNA1C c.3049-10C>T intron 296 Clinvar
AD CHD7 c.4645-9T>C intron 32 Clinvar
AD/AR ITGA2B c.671-13C>T intron 74 Clinvar
Voir les variations filtrées aux différentes étapes
ALL (654) FREQUENCY (473) REGION (22) CONSEQUENCE (40) PPY (9) QUALITY (8) SEGREGATION (102) ALL GENE
102 VARIANTS FROM CURRENT FILTER SETTINGS
INH GENE HGVSc CSQ ! F S O Q R QUAL DP RATIO HI. FREQ HI. COUNT EXTERNAL CLASS
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
AR ADAMT... c.1340C>T missense, spl... 129 24 0.54 0.000566 74
AR ADAMT... c.438G>C missense 129 35 0.49 0.002188 288 Clinvar
AD AKAP9 c.1158A>T missense 128 51 0.67 0.000736 36 Clinvar
AD AQP5 c.767G>A missense 125 44 0.52 0.001653 233 Clinvar
XR AR c.228_239dup inframe_inse... O Q 126 6 1.00 0.040800 524
AD ARHGA... c.2036C>T missense 121 43 0.42 0.001460 169 Clinvar
AD ARID1A c.609C>A missense 129 43 0.53 0.000328 7
AR ATM c.295A>G missense R 129 36 0.56 0.000912 69 HGMD Clinvar
AD ATN1 c.1503_1508del inframe_dele... R 134 26 0.38 0.020200 259 HGMD
AD ATXN2 c.560_561ins... inframe_inse... ! 101 38 0.45 0.000928 47
AR;AD SLC5A2 c.1961A>G missense R 129 25 0.60 0.008445 1383 HGMD Clinvar
AR CABP2 c.133G>A missense 129 35 0.49 0.000887 14
AD CACNA1H c.1310G>A missense 128 53 0.49 0.000389 91
AR CACNA2... c.2987T>C missense 125 35 0.40 0.007298 916 Clinvar
AR CAPN1 c.2119-15C>G intron 129 35 0.54 0.005600 563
AR CBS c.1105C>T missense R 128 38 0.45 0.005500 747 HGMD Clinvar
AD CD96 c.1650-13A>G intron 129 56 0.48 1.000e-4 7
AD CEL c.2119A>C missense ! R 75 33 0.45 0.002539 112 HGMD


INDICATIONS COMMENT
+ 3/654 FILTER
TRIODEFAULT
FULL QUICK VISUAL
UNCLASSIFIED VARIANTS (18)
INH GENE HGVSc CSQ
AR FCGR3A c.512T>A missense
XD AMER1 c.1873A>G missense
XR GRIA3 c.1181G>A missense
AR FMN2 c.3138A>T synonymous
AR FMN2 c.3495T>G synonymous
AD BRAF c.2128-16_21... splice_region...
AD/AR MSH2 c.942+2del splice_donor
AR ARSB c.312+3A>C splice_region...
AR ARSB c.312+2T>C splice_donor
AR SERPINB8 c.197T>C missense
AD BRAF c.2128-5dup splice_region...
AD CACNA1C c.3049-10C>T intron
AD CHD7 c.4645-9T>C intron
AD/AR F11 c.403G>T stop_gained
AD/AR HTT c.96_110dup inframe_inse...
AD/AR ITGA2B c.671-13C>T intron
AR STRC c.3307-26_33... intron
CLASSIFIED VARIANTS (1)
INH GENE HGVSc CSQ ! F S O Q R CLASS
AD/AR TTN c.104251G>C ... missense 1
NOT RELEVANT VARIANTS (4)
INH GENE HGVSc CSQ ! F 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
AR PRKRA c.785-5_785-... splice_region... D
Choix du filtre à appliquer
WARNING 1
• Annotation for NM_000569.7 does not match corresponding transcript: ENST000003
VERIFIED TECHNICAL NOT RELEVANT
(Male)
NEEDS VERIFICATION
Filter: PASS
Quality: 219
GQ: 33
Depth: 39
Ratio: 0.82
- REF (A): 7
- T: 32
Father
Filter: PASS
Quality: 219
GQ: 38
Depth: 46
Ratio: 0.37
- REF (A): 29
- T: 17
Mother
Filter: PASS
Quality: 219
GQ: 38
Depth: 57
Ratio: 0.40
- REF (A): 34
- T: 23
CLASSIFICATION
SELECT CLASS
EVALUATION
REPORT
ACMG
SUGGESTED
SHOW REQ
ACMG SUGGESTED
BA1 High frequency
REGION
REGION-COMMENTS