# Alyssa

# Bonnes idées

- Graphe de filtres =  stratégie : suite d'opération (briques en drag and drop)
- Flagger les opérations manuelles dans le graphe (expertise utilisateur ajoutée ou en attente)
- Valider une stratégie (fige la config)
- Partager une stratégie (publique)
- Etiqueter une stratégie (clinical domains : à quoi sert cette stratégie)
- Représentation de l'ACMG
- Représentations des overlaps en % d'une boite
- Gestion des d'identifiants entre les manip (CGH034 <-> VCF18)
- Waiting analyses

# Accueil de l'analyse

Les informations sont très similaires à celles qu'on avait décrites :
Statut de l'analyse, date, génome, strétgie de filtres utilisées, input
types (VCF, CNV, CGH...)

# Accueil de l'analyse

Les informations sont très similaires à celles qu'on avait décrites :
Fichiers et contenus.
Phénotype.
Structure familiale.

## Lab Results

| Status | | Source | Regions of low coverage | File Type | # Var. pos. in Lab Result | # Var. pos. used in Analysis | # Variants in Analysis |
|---|---|---|---|---|---|---|---|
| Ready | ⓘ | DI_WESA_20-36228.gatk.haplotype.snp_indel.filtred2.vcf (20-36228-A-02-00) | | VCF File | 57 721 | 57 721 | 64 137 |
| Ready | | IntervalBasedReport_20488.xls (20-20488-252206093341_1_2) | | Cytogenomics Interval File | 19 | 19 | 19 |

## Phenotype

| Code | Phenotype Trait | Modifiers | Actions |
|---|---|---|---|
| No phenotype traits found. | | | |

## Family Members

| Patient | Affected status | Lab Results | # Var. pos. in Lab Result | # Var. pos. used in Analysis |
|---|---|---|---|---|
| 37647 (Mother) | unaffected | DI_WESA_20-36228.gatk.haplotype.snp_indel.filtred2.vcf (20-37647-A-02-00) | 57 701 | 62 077 |
| 37917 (Father) | unaffected | DI_WESA_20-36228.gatk.haplotype.snp_indel.filtred2.vcf (20-37917-A-02-00) | 57 661 | 62 303 |

# Accueil de l'analyse

Les informations sont très similaires à celles qu'on avait décrites :
Sources de BDD utilisées (nécessaire uniquement dans le rapport)

| Sources | | |
|---|---|---|
| **Platform Dataset** | 33, RefSeq Transcripts v96 + RefSeqGene released 2019-10-22 | |
| | OMIM® Copyright © 1966-2019 Johns Hopkins University - All rights reserved. | |
| **Genome build** | GRCh37.p13 | |
| **Annotation Sources** | **Name** | **Description** |
| | 1000Genomes | 1000 Genomes Phase1 release v3.20101123 |
| | 1000GenomesPhase3 | 1000 Genomes Phase 3 release v5 (10 September 2014), including GRCh38 data |
| | CGA | ClinGen CNV Atlas 2019-10-07 |
| | CGDS | ClinGen Dosage Sensitivity Map 2019-08-12 |
| | CGDS_GENE | ClinGen Gene Curation Dosage Sensitivity Map - 2019-07-15 |
| | CIViC | CIViC - Clinical Interpretations of Variants in Cancer - release 01-Oct-2019 |
| | COSMIC | COSMIC release v90 |
| | ClinVar | NCBI ClinVar 2019-10 |
| | DGV | Database of Genomic Variants 2016-05-15 |
| | DPOP | DECIPHER population CNVs v9.23 |
| | DSYN | DECIPHER syndromes 2019-10-05 |
| | ESP6500 | Variants in the ESP6500SI-V2 dataset of the exome sequencing project (ESP), annotated with SeattleSeqAnnotation137. |
| | ExAC | ExAC release 1.0 - including GRCh38 from liftover data |
| | JAX | Somatic gene variant annotations and related content powered by The Jackson Laboratory Clinical Knowledgebase (JAX-CKB™) - version 20200124 |
| | OMIM | OMIM 2019-10-25 |
| | dbNSFP | dbNSFP v3.0b2: Database of functional predictions for non-synonymous SNPs |
| | dbSNP | dbSNP build 151 |
| | gnomAD | gnomAD release 2.0.2 - with additional multi-allelic insertions and GRCh38 statistics from the lift-over vcf files |

# Accueil de l'analyse

Les informations sont très similaires à celles qu'on avait décrites :
Audit trail

## Change History

| Updated on | Updated by | Scope of change ▼ | Action |
|---|---|---|---|
| mars 16, 2021 4:47 PM CET | mlebeurrier | Analysis access | Analysis has been accessed |
| mars 16, 2021 2:37 PM CET | jtaminau | Filtering | Filtered Molecular variants using classification tree bin: ClinVar () Match |
| mars 16, 2021 2:19 PM CET | jtaminau | Analysis access | Analysis has been accessed |
| mars 12, 2021 10:36 AM CET | jtaminau | Analysis access | Analysis has been accessed |
| mars 8, 2021 9:46 AM CET | jmages | Analysis access | Analysis has been accessed |
| mars 5, 2021 3:53 PM CET | jmages | Analysis status | In Progress |
| mars 5, 2021 3:53 PM CET | jmages | Analysis access | Analysis has been accessed |
| mars 5, 2021 3:53 PM CET | system | Analysis status | Available For Analysis |
| mars 5, 2021 3:53 PM CET | jmages | Analysis details | Created analysis with reference: A_36228, summary: '' and description: '' and target panels: None |

# Variant triage = tableau de variants

Graphe de filtre interactif avec les étapes filtrages et les comptages associés.
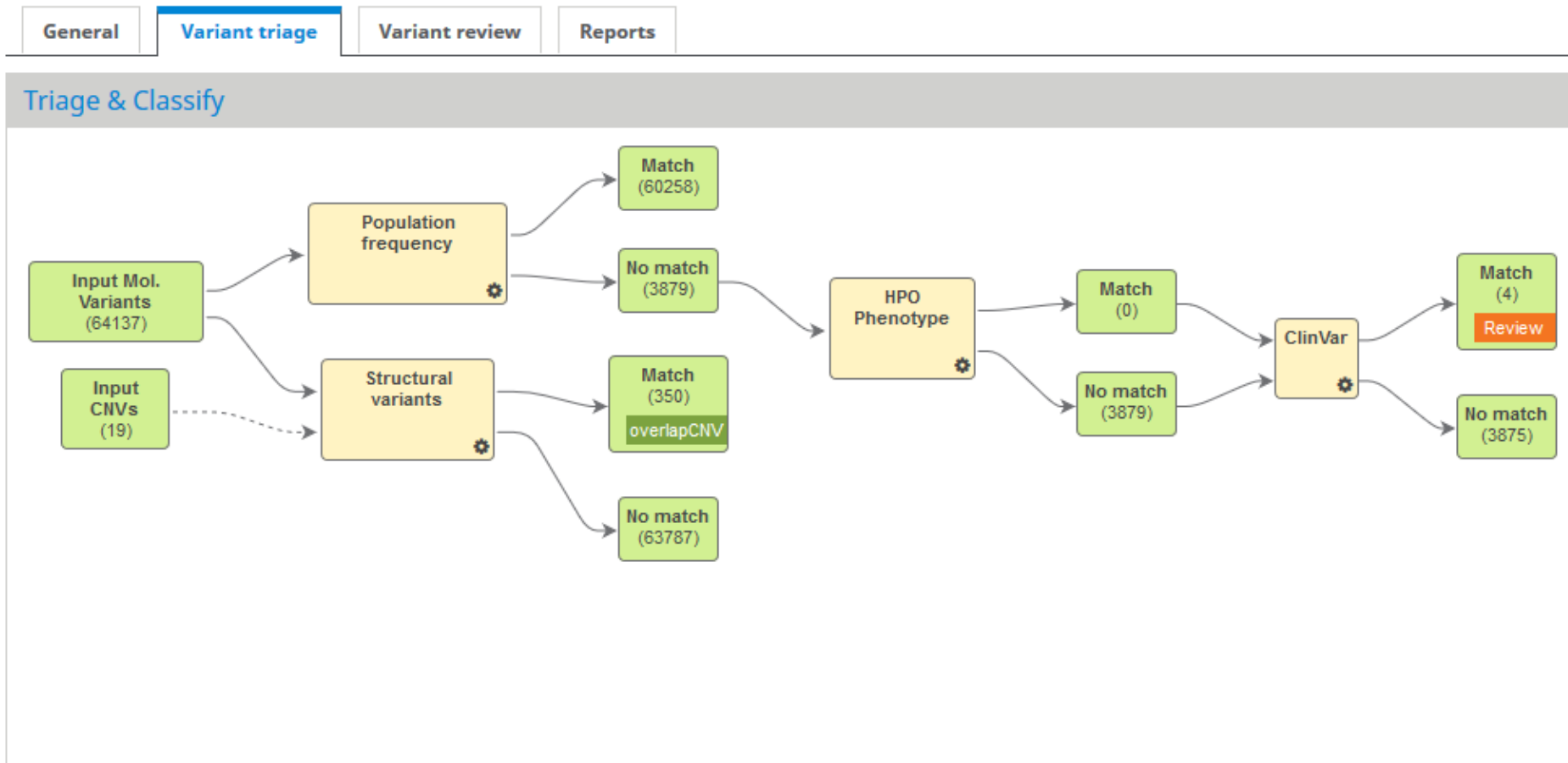
# Editer le graphe

Drag and drop une étape (depuis la liste accessible) de filtrage puis la relier aux data.
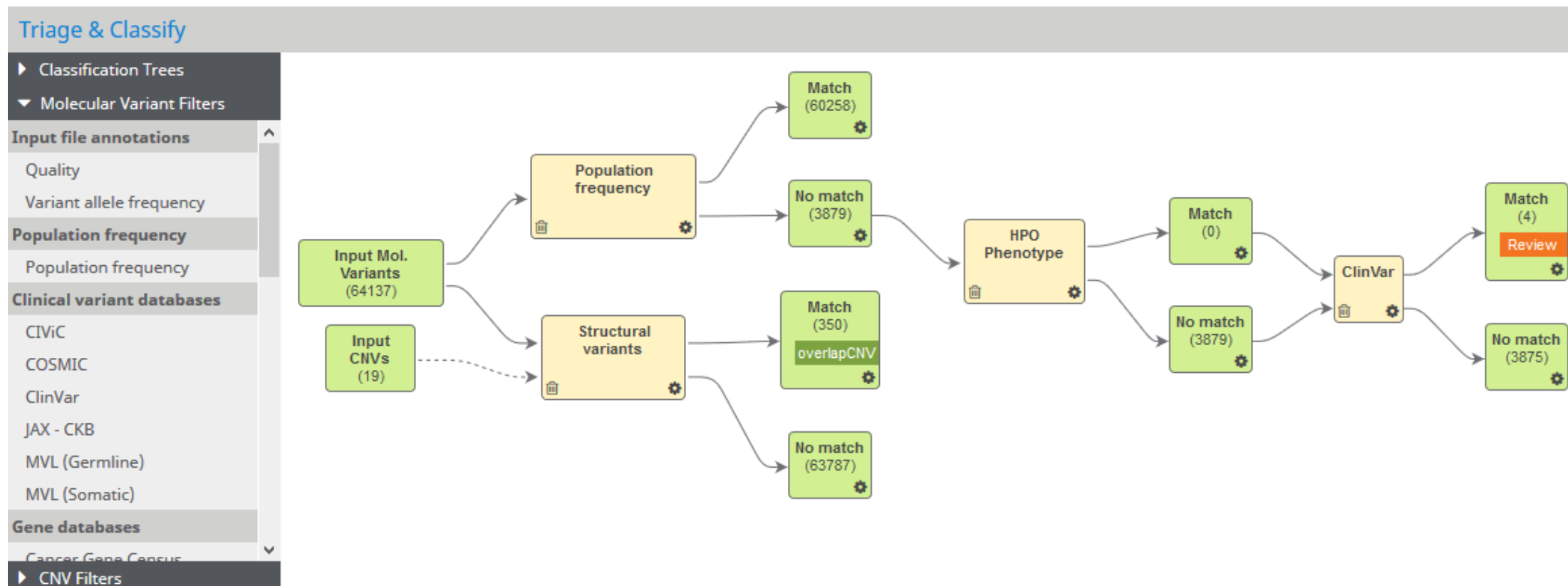
# Voir le tableau de variants d'une étape

# Où retrouver un variant dans la stratégie

# Voir les annotations pour un variant

## External databases

| | |
|---|---|
| **dbSNP** | rs146571352 |
| **dbSNP validated** | yes |
| **dbSNP average heterozygosity** | 0.001 |
| **dbSNP suspect false snp** | no |
| **dbSNP clinical significance** | Likely pathogenic |
| **ClinVar Id** | 209185, 209185, 209185, 209185 |
| **ClinVar clinical significance** | probable-pathogenic, other, other, unknown |
| **ClinVar disease** | Combined oxidative phosphorylation deficiency 13, not specified, not provided, Hypermetropia |
| **COSMIC Id** | COSM7384835 |
| **COSMIC sample count** | 2 |
| **COSMIC tumor type (sample count)** | Pulmonary adenocarcinoma - non-small cell lung cancer (NSCLC) (1) |

## HGVS nomenclature

| | |
|---|---|
| **RefSeqGene** | NG_033012.1 |
| **HGVS cDNA-level nomenclature** | NM_033109.5:c.1525G>A |
| **HGVS genomic-level nomenclature** | NC_000002.11:g.55874559C>T |
| **HGVS protein-level nomenclature** | NP_149100.2:p.Val509Ile |

## Quality

| | |
|---|---|
| **Observed allele 1 depth** | 18 |
| **Observed allele 2 depth** | 30 |
| **Allelic depth** | C = 18 |
| | T = 30 |
| **Call quality** | 1376.16 |
| **Genotype quality** | 99 |
| **Filter status** | PASS |

## Population

| | |
|---|---|
| **ESP6500 allele frequency** | 15/13006=0.001 |
| **ESP65000 genotype frequency** | 15/6503=0.002 |
| **ExAC allele frequency** | 91/121358=0.001 |
| **gnomAD allele frequency** | 262/276804=0.001 |
| **gnomAD genotype frequency** | 258/138402=0.002 |

## Conservation

| | |
|---|---|
| **GERP++ neutral rate** | 5.53 |
| **GERP++ RS score** | 5.53 |
| **PhyloP score** | 0.871 |
| **SiPhy score** | 19.811 |

## Substitution

| | |
|---|---|
| **MutationTaster score** | 1 |
| **MutationTaster prediction** | Disease causing |
| **MutationAssessor score** | 0.625 |
| **MutationAssessor prediction** | Neutral |
| **FATHMM Score** | 0.56 |

## Coding

| | |
|---|---|
| **LRT score** | 0 |
| **LRT prediction** | Deleterious |
| **LRT omega** | 0 |

## Missense

| | |
|---|---|
| **BLOSUM45** | 3 |
| **BLOSUM62** | 3 |
| **BLOSUM80** | 3 |
| **SIFT score** | 0.136 |
| **PolyPhen2 score HumDiv** | 0.992 |
| **PolyPhen2 score HumVar** | 0.867 |
| **PolyPhen2 prediction HumDiv** | Probably damaging |
| **PolyPhen2 prediction HumVar** | Possibly damaging |
| **PROVEAN Score** | -0.83 |
| **PROVEAN prediction** | Neutral |

## Splice

| | |
|---|---|
| **Positions from nearest splice site** | 30 |

## Custom fields

No annotations found.

## Repetitive regions

No annotations found.

# Voir les infos pour un gene



| Gene | Position | Ref | Patient | Read Depth | Inheritance mode | Inh. from | Family Mother | Father | Read Depth Mother | Father | CNV | Type | Transcript | cDNA | Location | Exon | Effect | Protein | CT Labels | External Databases | Simil. | Info | Classification | Asses |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| PNPT1 | 2:55,874,559 | C | T \| C | 48 | Comp. Het. cand. | ♀ | C T | C C | 47 | 41 | | snp | NM_033109.5 | c.1525G>A | exonic | 19 | nonsynonymous | p.Val509Ile | | | | | | |

## Gene Info

| | |
|---|---|
| **HGNC Symbol** | PNPT1 |
| **Gene name** | polyribonucleotide nucleotidyltransferase 1 |
| **Synonyms** | PNPase, OLD35, old-35 |
| **Ensembl ID** | ENSG00000138035 |
| **Chromosomal location** | 2p16.1 |
| **Genomic location** | 2:55 861 196-55 920 980 |
| **Strand** | - |
| **Biotype** | gene with protein product |

## Gene-Disease Relationships

| | |
|---|---|
| **OMIM® Morbid gene MIM** | 610316 |
| **OMIM® Morbid phenotype** ❶ | Combined oxidative phosphorylation deficiency 13, 614932 (3), Autosomal recessive |
| | Deafness, autosomal recessive 70, 614934 (3), Autosomal recessive |
| **NCBI gene ID** | 87178 |
| **ClinVar disease** ❶ | Deafness, autosomal recessive 70 C1824925, NCBI curation |
| | Combined oxidative phosphorylation deficiency 13 C3554129, NCBI curation |
| | Combined oxidative phosphorylation deficiency CN228601, OMIM (PS609060) |

### Cancer Gene Census

## HPO phenotype traits

Abnormal corpus callosum morphology
Abnormal corpus striatum morphology
Absent speech
Ankle flexion contracture
Autosomal recessive inheritance
Cataract
Show all

## ClinGen Dosage Sensitivity Map

No annotations found.

# Flag human annotation

Cliquer sur  n'importe quelle étape de la stratégie permet d'ajouter une catégorie, d'ajouter ces variants aux panier. Permet d'identifier où l'utilisateur intervient.

# CNV tableau de variants
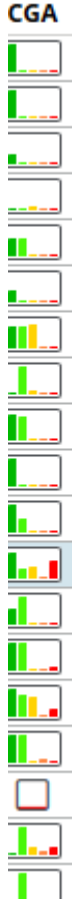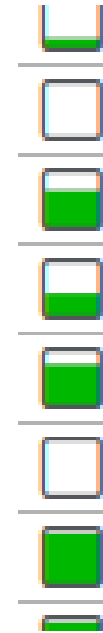
# ACMG representation

Proportion of ACMG classes in th regions :

Benign : 10%

Likely benign : 10 %

…

Pathogenic : 50%

# Coverage of regions

- % of coverage of the element :
- Not only 0, 50, 100%

# Interface de filtre pas top

# Le panier = variant review

Affiner la définition du panier. Ici human manageable variant list. Ajout d'informations tel que la classification les commentaires l'interprétation.
Mais surtout ajout au rapport du variant classification finale.