

Custom workflow (HPC)



1. Germline VCF
2. Annotations
 - VEP
 - ANNOVAR with InterVar
 - AutoPVS1
 - ClinVar (optional)

CAVATICA Workflow (AWS)



1. Kids First Germline Workflow
 - VEP- and ClinVar-annotated VCF
2. Pathogenicity preprocessing
 - ANNOVAR with InterVar
 - AutoPVS1

AutoGVP: Automated Germline Variant Pathogenicity (R, Docker)

ClinVar annotation

Yes

No

2-stars or more

Yes

No

P, LP, B, LB,
or VUS

Yes

No

1-star

Yes

No

Conflicting
Interpretation

Yes

No

ClinVar
Classification

Modified
InterVar
Classification

ClinVar
Classification
(Majority call; if tie,
most recent evaluated)

ClinVar
Classification

Modified
InterVar
Classification

Modified
InterVar
Classification