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1 VATK tool descriptions

- Online Variant Annotation (OVA) Annotates given vcf file using Ensembl VEP API.
- Multi Genome Analysis (MGA) Filters index individuals variants with parents and siblings based on given inheritance pattern.
- Variant Filter (VF) Filters annotation file by given parameters.

1.1 Online Variant Annotation (OVA)

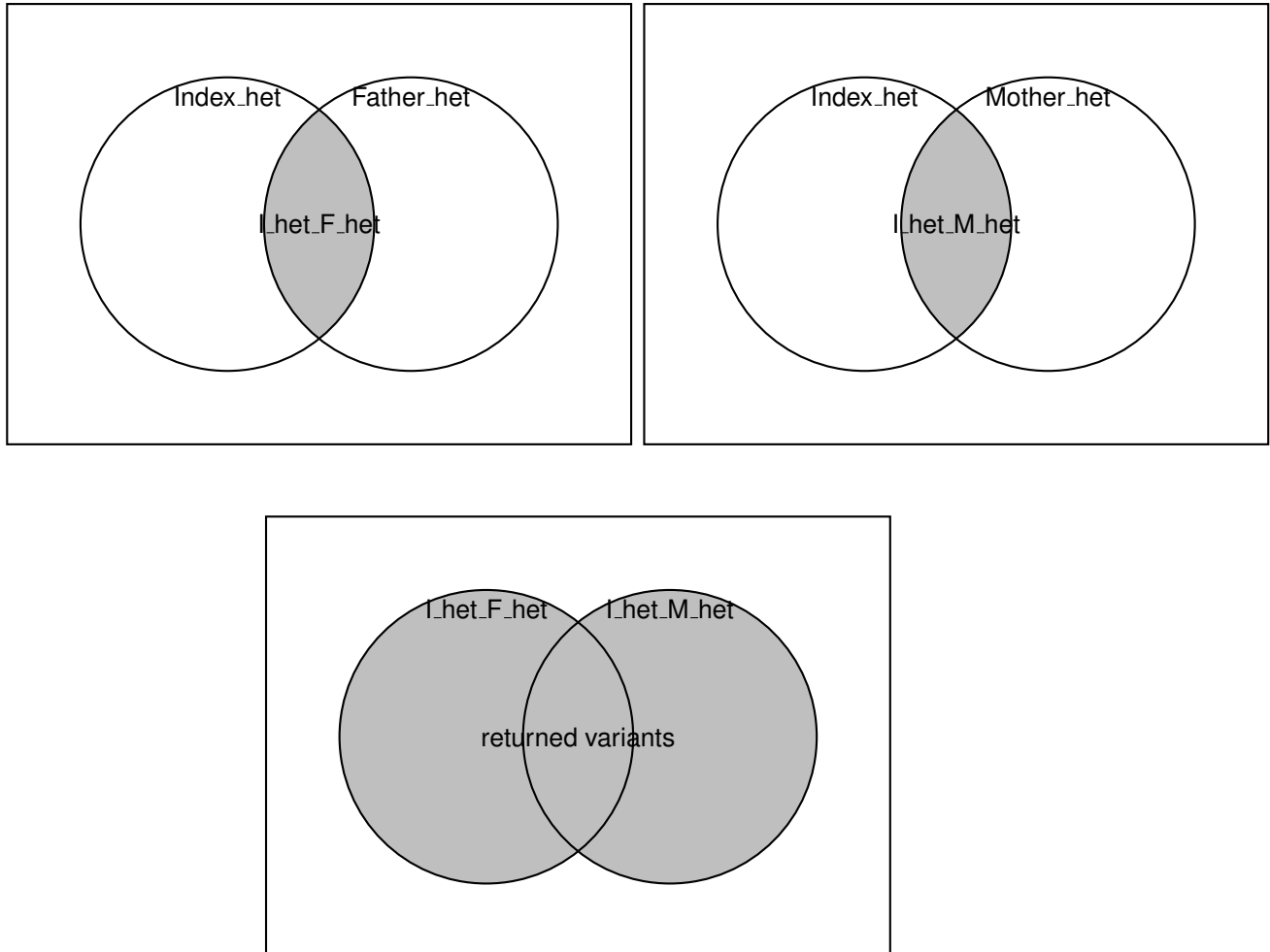
Output columns as follows:

- CHR
- LOC
- REF
- ALT
- ZYGO
- AD
- DP
- gene_symbol
- transcript_id
- id
- minor_allele_freq
- gnomad_maf
- consequence_terms
- biotype
- polyphen_prediction
- polyphen_score
- sift_prediction
- sift_score
- hgvs_c
- hgvs_p

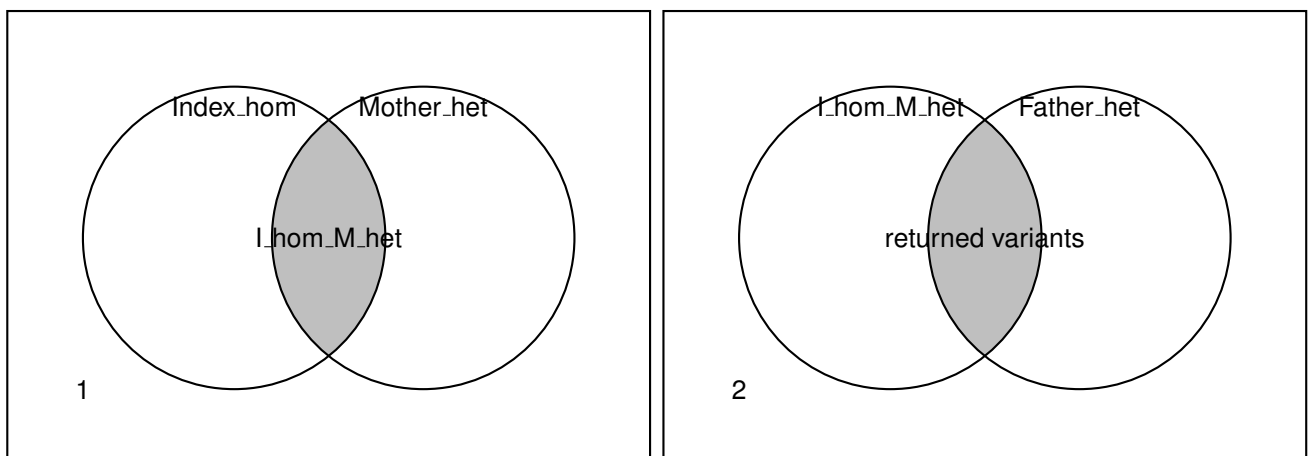
- phenotypes
- clin_sig
- pubmed

1.2 Multi Genome Analysis (MGA)

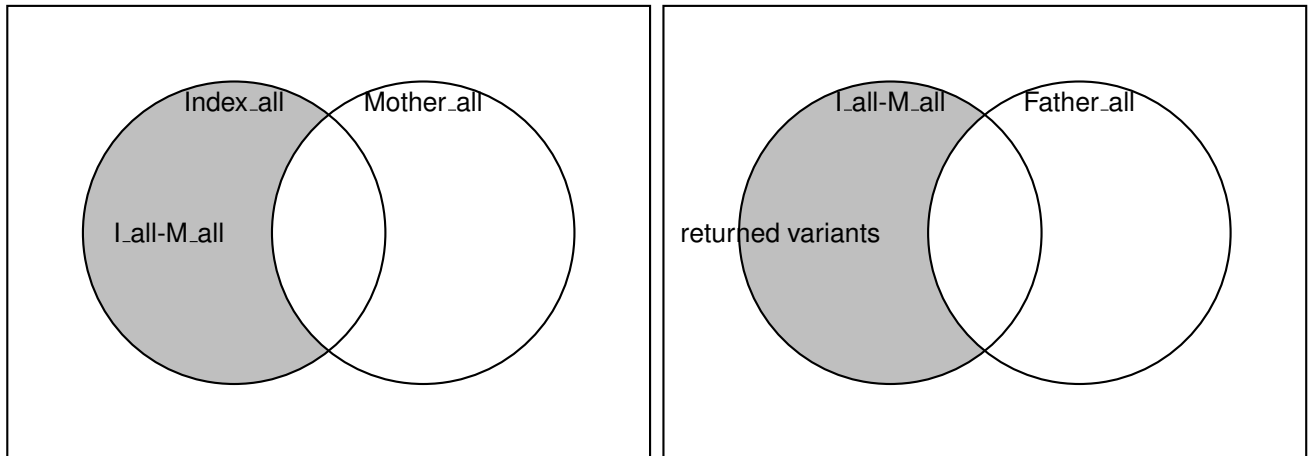
1.2.1 trio AD



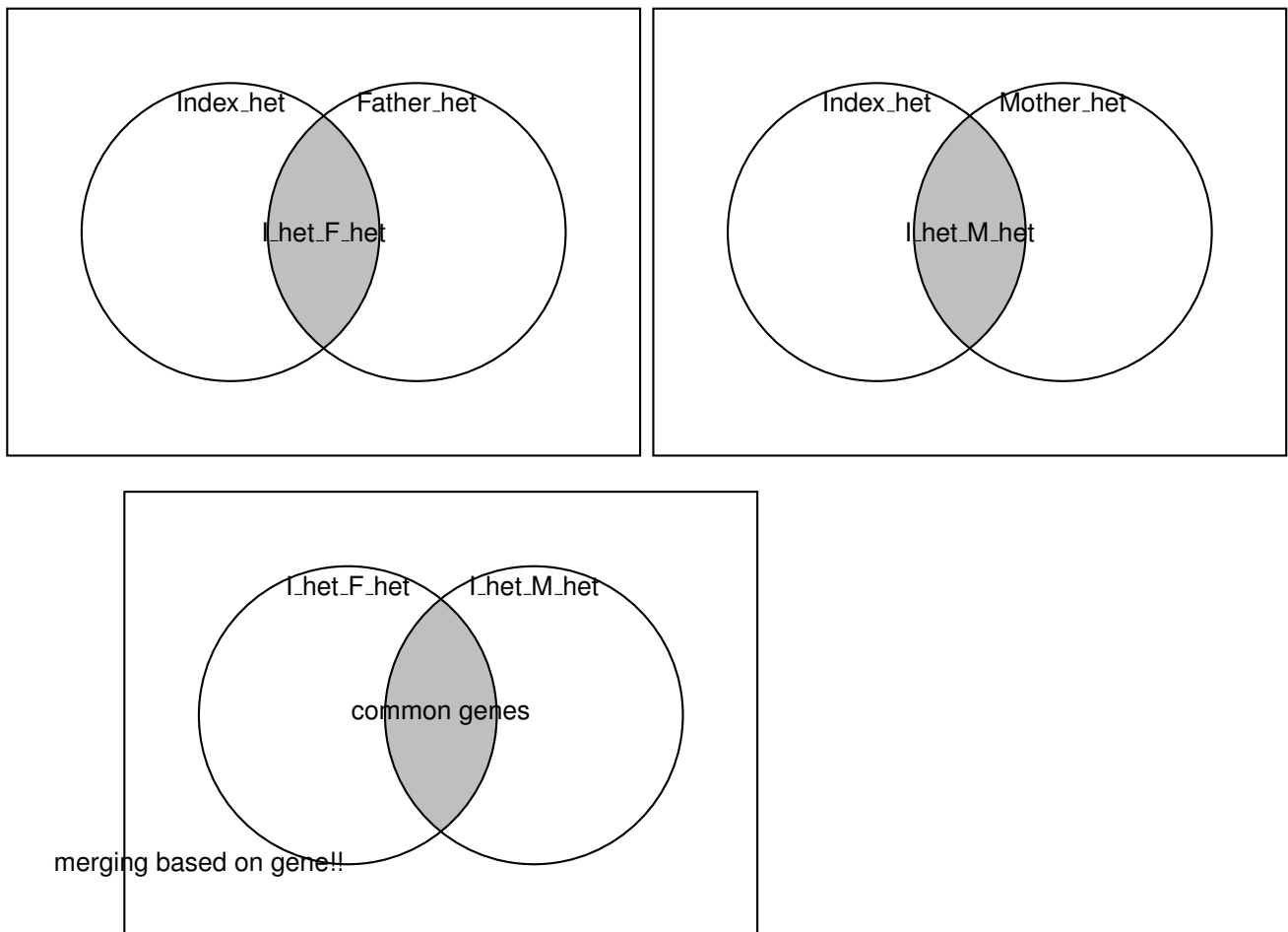
1.2.2 trio AR

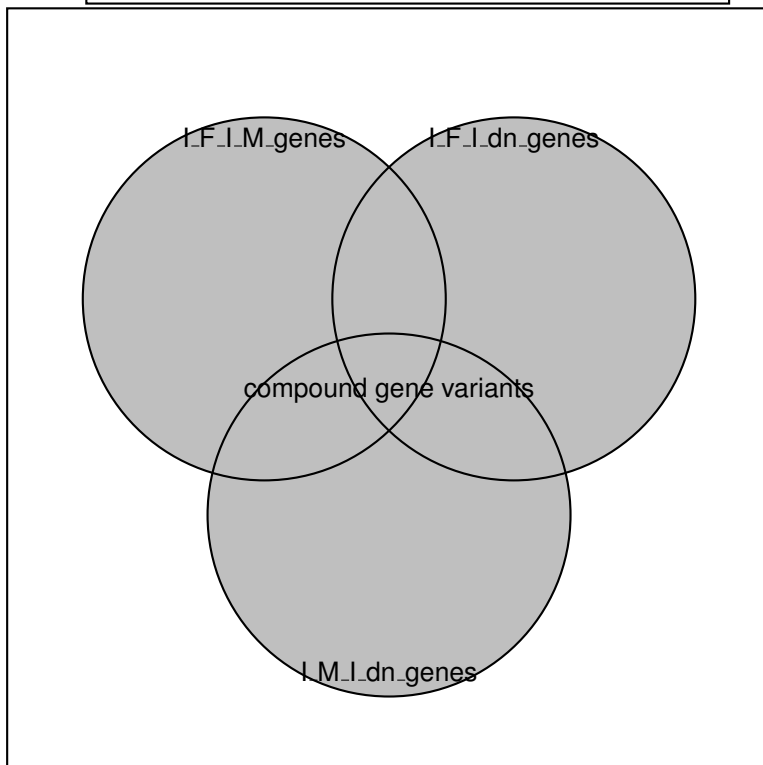
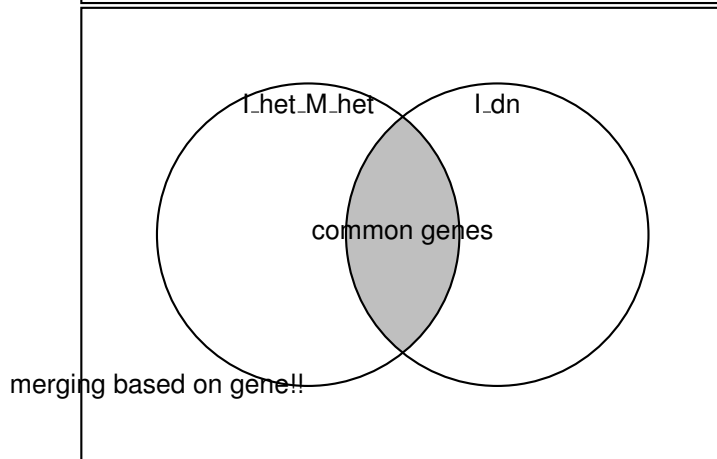
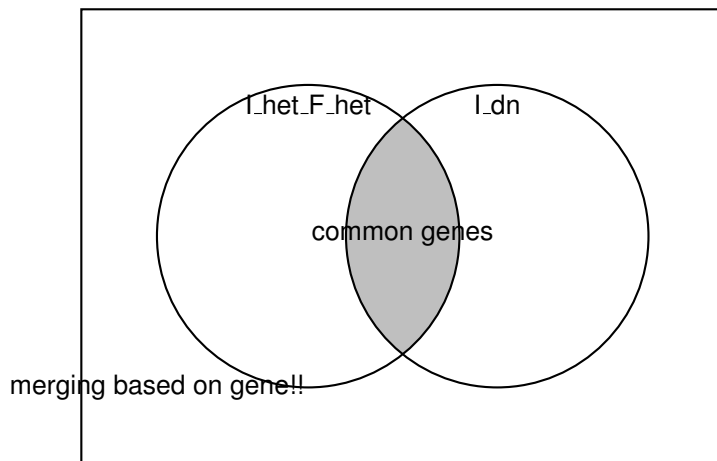


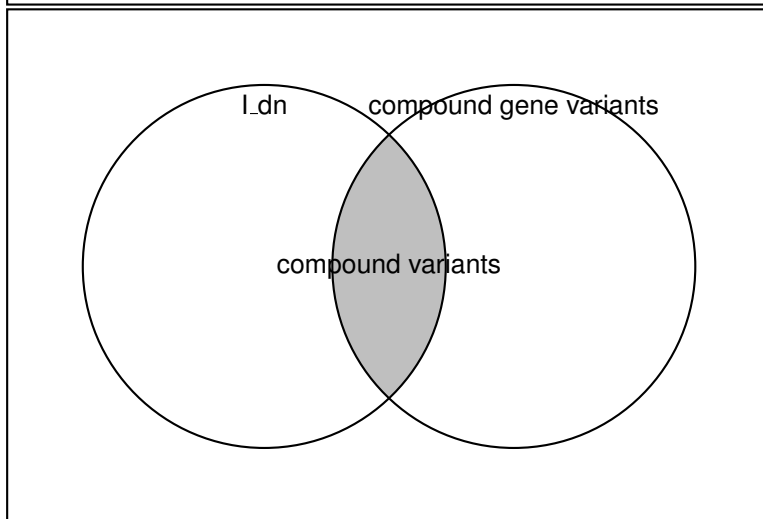
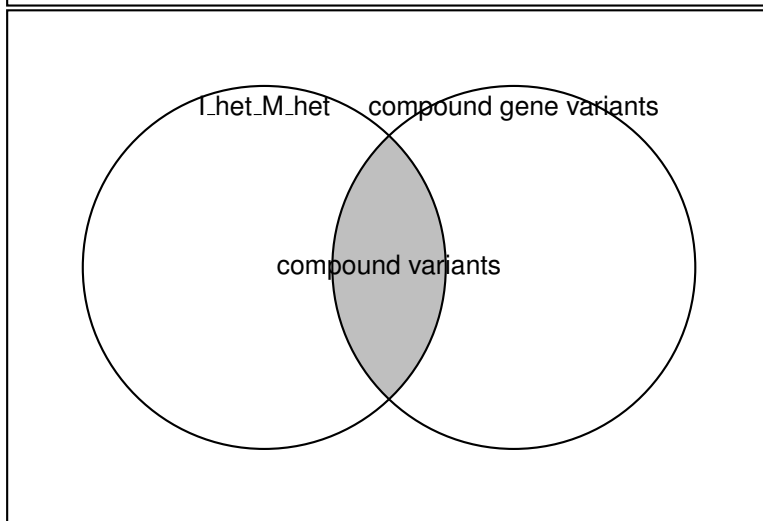
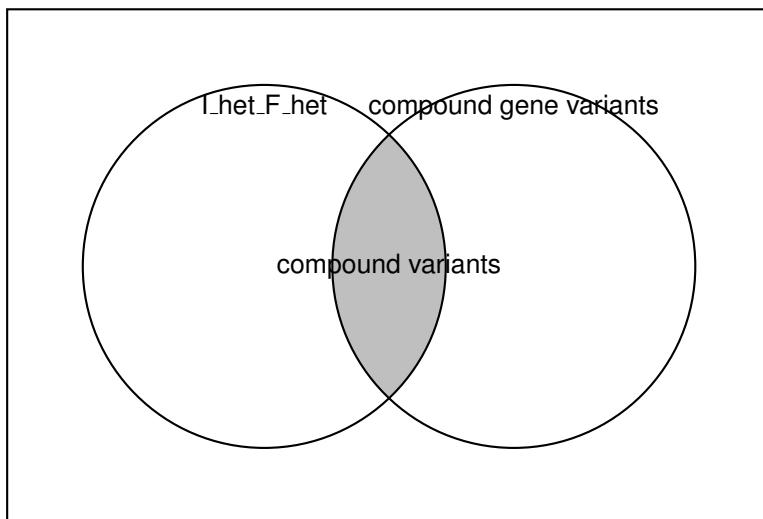
1.2.3 trio DN

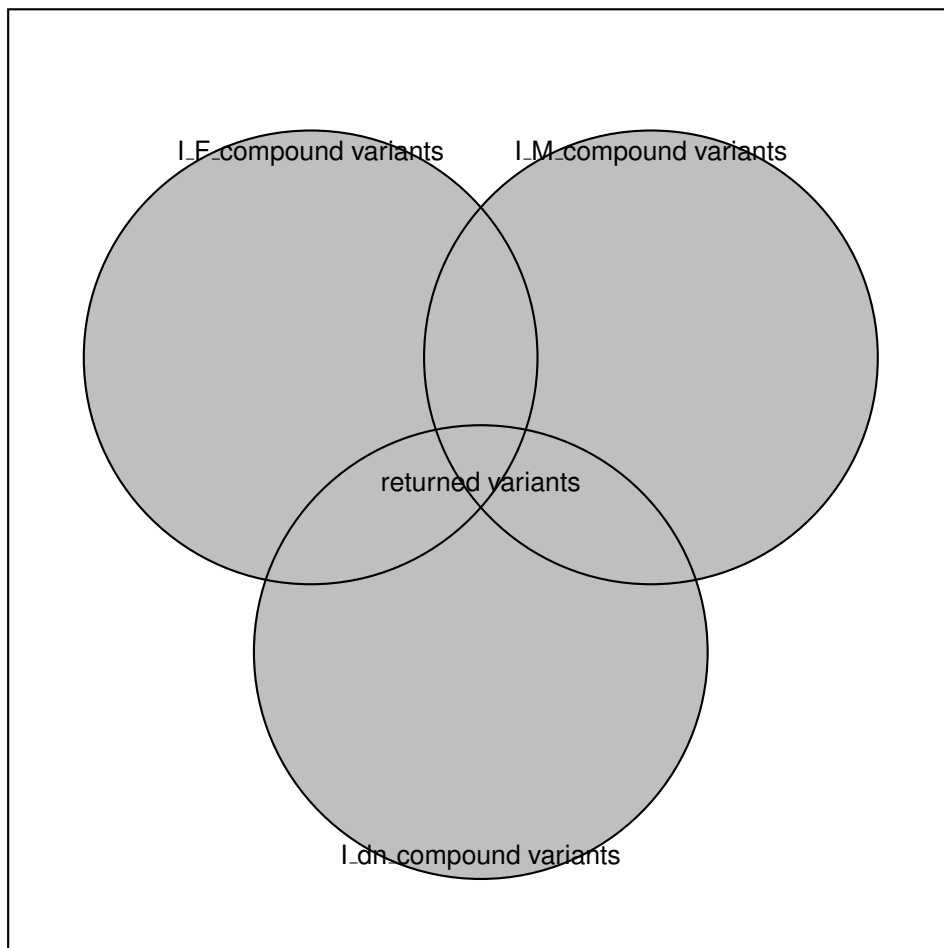


1.2.4 trio CH









1.2.5 siblings unaffected

Returns variants that are not present in unaffected sibling.

1.2.6 siblings affected

Returns variants that are present in affected sibling.

1.3 Variant Filtering (VF)

Can be used to filter based on columns of the annotation file. (see examplefilter file comments for info.)

2 Example Uses

OVA examples

```
VATK OVA -i input.vcf -o output.csv
```

MGA examples

```
VATK MGA -i index.csv -m mother.csv -f father.csv -a trio -p AR -o output.csv  
VATK MGA -d pedigree.csv -a siblings -o output.csv
```

VF generic filter examples

```
VATK VF -i index.csv -z 1/1 -f 0.01 -r 20 -b protein_coding -o output.csv
```

VF Specific filter examples

```
VATK VF -t filterfile.txt
```

VATK examplefile

```
VATK examplefile --help  
VATK examplefile -e both
```

VATK updateomim

```
VATK updateomim --help  
VATK updateomim -k apikey
```