

# INPUTS

Trio's BAM/ CRAM (father, mother, child),  
Reference FASTA

## VARIANT CALLING

Samtools mpileup

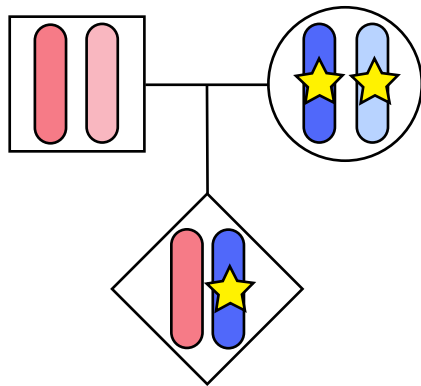
## VARIANT GROUPING

Group A variants : One parent is 0/0, the other is 1/1

Group B variants: One parent is 0/0, the other is 0/1

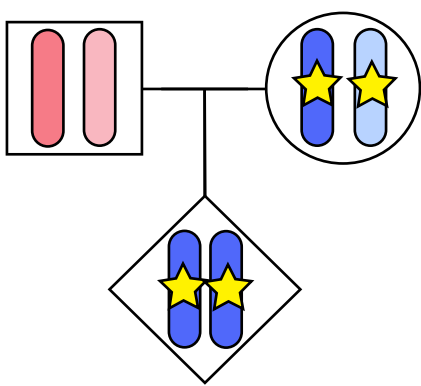
## UPD PATTERN DETECTION

### Normal



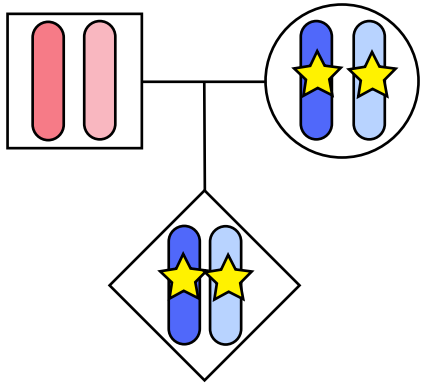
Child's Expected  
VAF=0.5

### UPD isodisomy



Child's Expected  
VAF=1

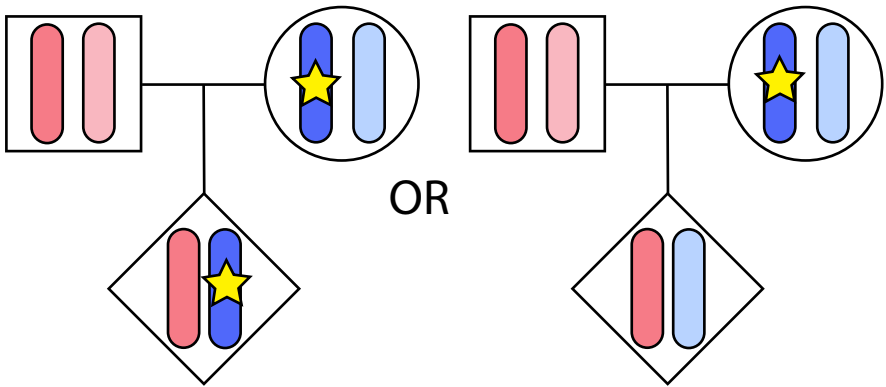
### UPD heterodisomy



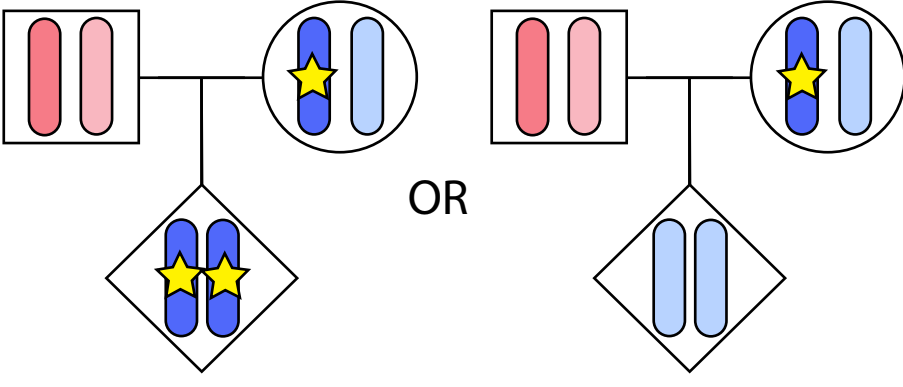
Child's Expected  
VAF=1

GroupA variant

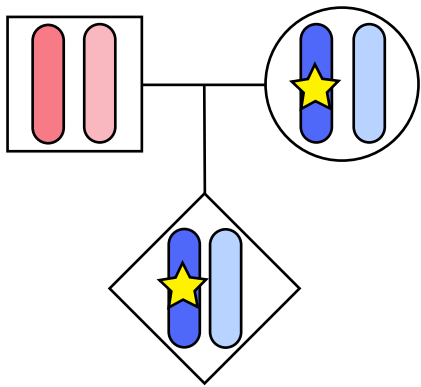
GroupB variant



Child's Expected  
VAF=0.5 or 0



Child's Expected  
VAF=1 or 0



Childs' Expected  
VAF= 0.5

## OUTPUTS

UPD Classification (BED),  
VAF & Depth Plot,  
Segmentation result (TSV),  
SNP list