

INPUTS

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

VARIANT CALLING

Samtools mpileup

VARIANT GROUPING

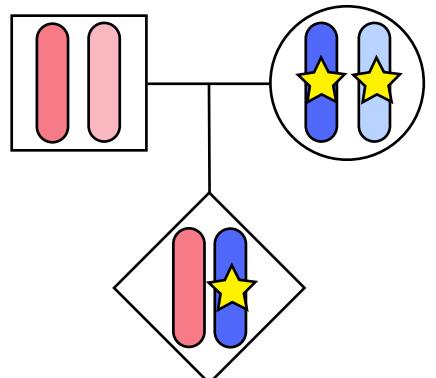
Group A variants : One parent is 0/0, the other is 1/1

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

UPD PATTERN DETECTION

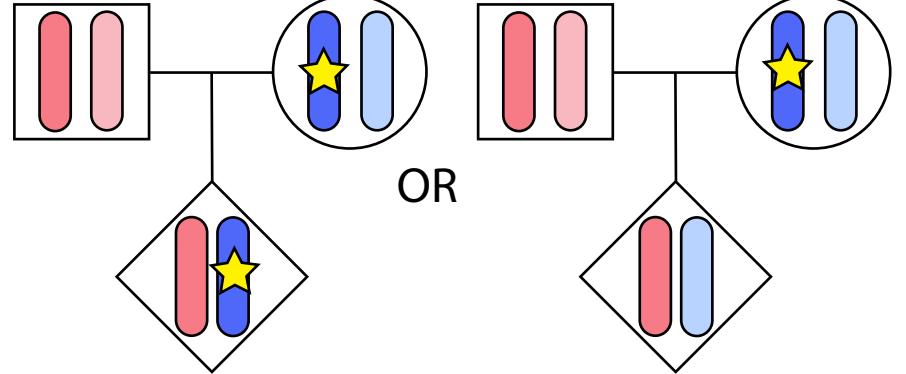
Normal

GroupA variant



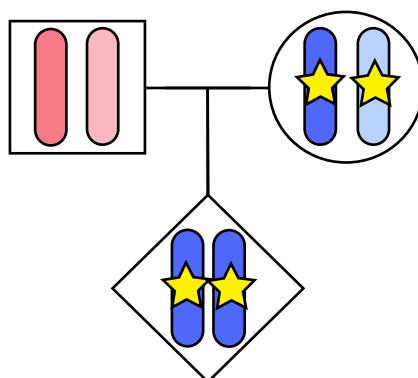
Child's Expected
 $VAF=0.5$

GroupB variant



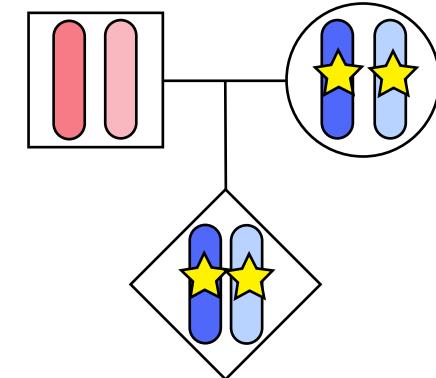
Child's Expected
 $VAF=0.5 \text{ or } 0$

UPD isodisomy

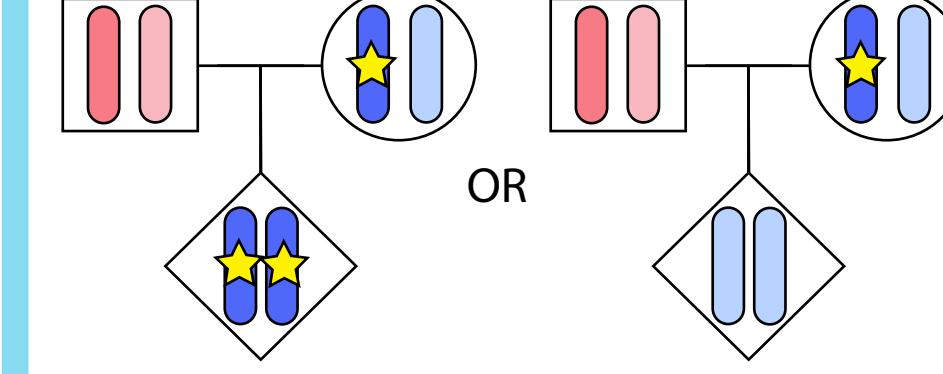


Child's Expected
 $VAF=1$

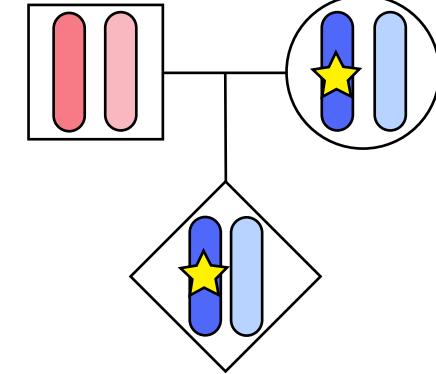
UPD heterodisomy



Child's Expected
 $VAF=1$



Child's Expected
 $VAF=1 \text{ or } 0$



Child's Expected
 $VAF=0.5$

OUTPUTS

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