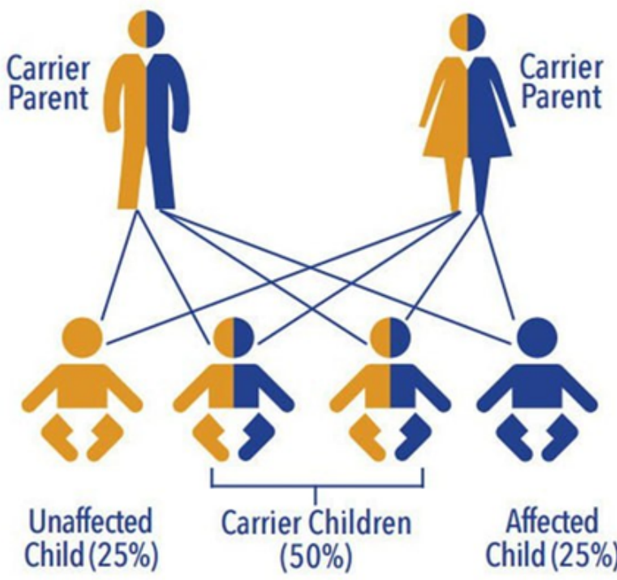
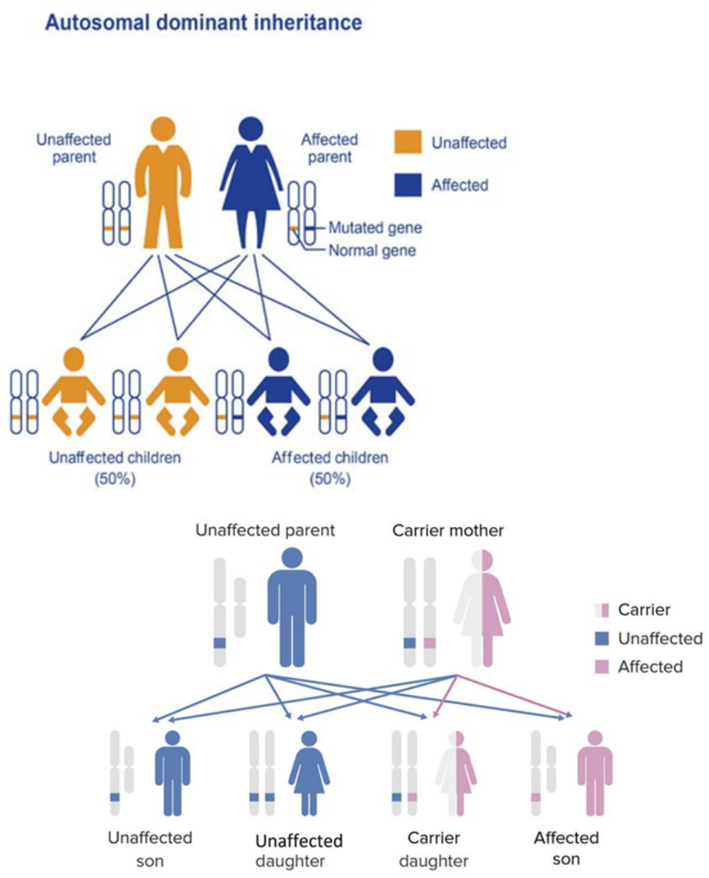
# COUPLE CARRIER SCREENING

Carrier screening aims to determine the carrier status of healthy parents for recessive disorders, including X-linked recessive diseases. Even the most complex scenarios are well covered in a single carrier screening filter in ADVAT Pipeline so that cases can be effortlessly solved in a matter of time.

## 1.AUTOMATED SCREENING FOR RECESSIVE DISORDERS

A single-click workflow considers factors such as the affection status and genotype so that only variants that fit the segregation pattern for carrier screening are displayed. Therefore, no manual work in ruling out variants based on segregation.



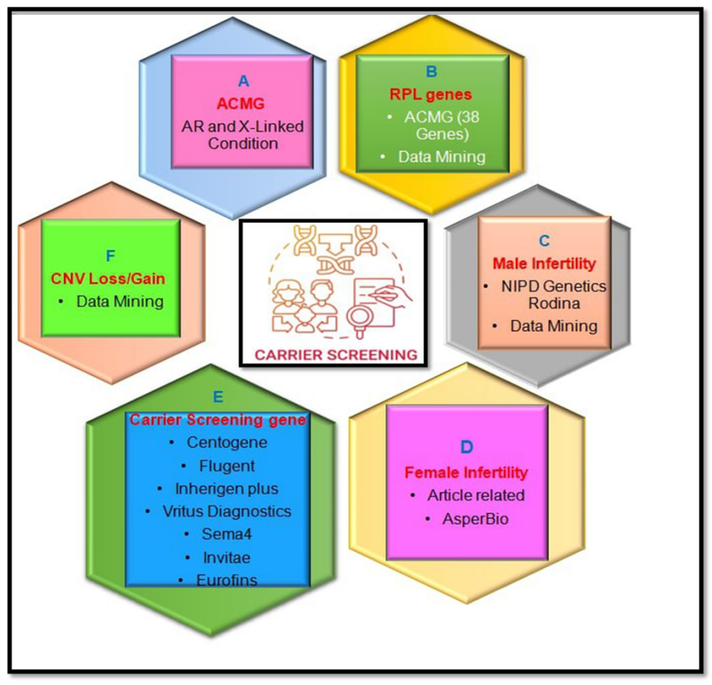

## 2.SCREEN CASES WITH A FAMILY HISTORY EFFORTLESSLY

Screen cases with a family history swiftly by recording the clinical information. In cases where family history and phenotypes are known, our proprietary HPO similarity score helps a great deal in identification of causative variants. Prioritize the list of variants with respect to the score and focus on the most relevant variants first.

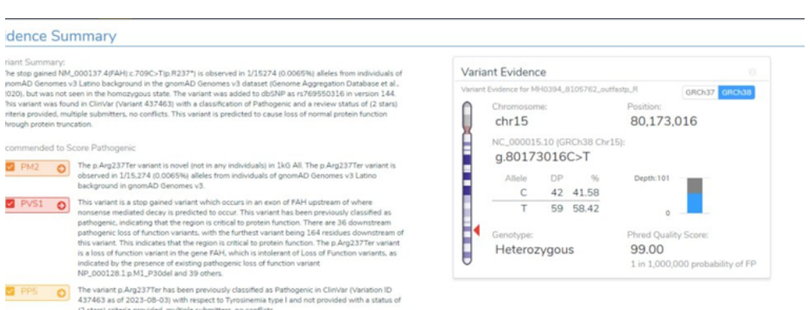
## 3.VIRTUAL PANELS TO APPLY ALL TIERS

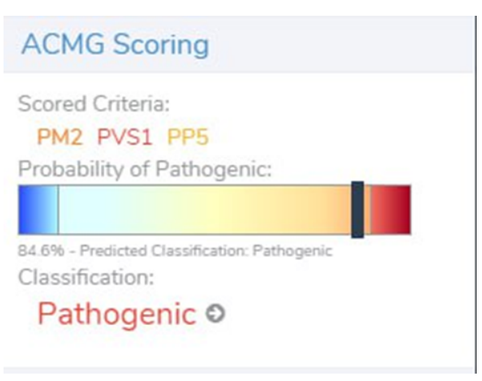
ADVAT have cutomized gene panel that has been designed by the laboratory or one that has been recommended by organizations such as ACMG and ACOG.

2271-genes cover our carrier screening panel targets the full coding region of all genes, enabling the analysis of >36,000 nonbenign ClinVar variants for single-nucleotide variants (SNVs), insertions and deletions (indels).




## 4.ACMG CALCULATOR IN ADVAT

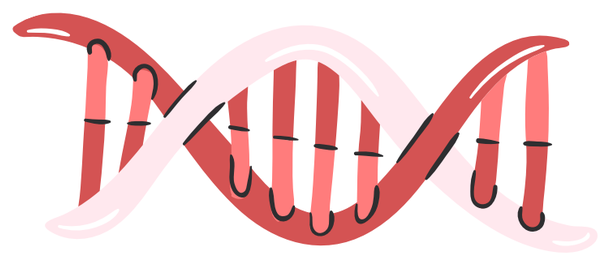


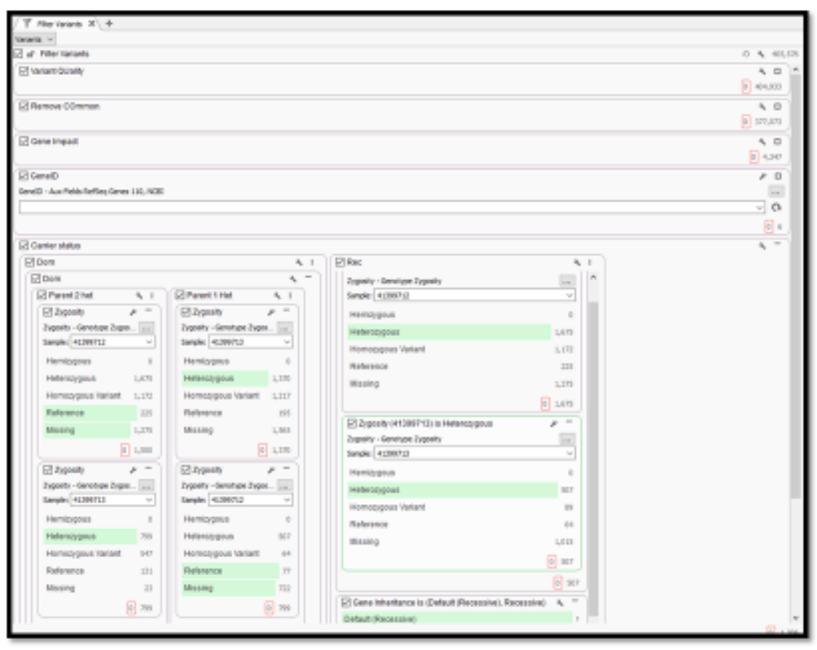

ADVAT Pipeline have facilitates clinical variant interpretation in concordance with the ACMG standards and guidelines. The evidence for classification becomes part of our ACMG knowledge base.

# EXPANDED CARRIER SCREENING GENES

- Carrier panel genes (2271)
- X- linked  genes (16)
- Pan ethnic gene (12)
- Male infertility genes (126)
- Female Infertility genes (144)
- RPL genes (38)
- PCOS genes (15)
- Primary Ovarian insufficiency genes(25)



VarSeq Couple carrier Workflow-Major filter Steps



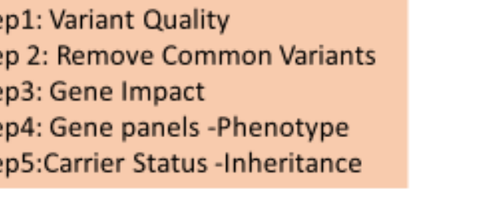
- Step1: Variant Quality
- Step 2: Remove Common Variants
- Step3: Gene Impact
- Step4: Gene panels -Phenotype
- Step5:Carrier Status -Inheritance

Step1

Step2



Step3



Step4



Step5

- Listed Significant Variants-Couple Carrier & Affected(ACMG&ClinVar)

Innovation Award Winner - 2023
Dr. S. Muthukumaran
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