

Pipeline: DNA SNV INDEL v1

The pipeline starts with **Cohort DNA WGS**, followed by **GATK** and **Annotate VEP**. The output of **Annotate VEP** is categorized into **QV 1** (dashed box) and **QV 2** (dashed box).

QV 1 leads to a central box with four categories: **Pathogenic**, **Likely pathogenic**, **Uncertain**, **Likely benign**, and **Benign**. This box also receives input from **QV 2**.

QV 2 leads to a box with four categories: **Pathogenic**, **Likely pathogenic**, **Uncertain**, **Likely benign**, and **Benign**. This box also receives input from the central box.

The central box branches into four main analysis paths:

- Pathway VSAT SKAT-O**: Leads to **Pathway-level result**, which is then annotated to produce **Result + annotation**.
- Gene VSAT SKAT-O**: Leads to **Gene-level result**, which is then annotated to produce **Result + annotation**.
- Single variant test Plink**: Leads to **SVAT result**, which is then annotated to produce **Result + annotation**.
- Other (ML/DL etc.)**: Leads to **Result**.

The final results are categorized into three groups:

- Traditional clinical genetics**: Includes **Annotation-based result** and **Result + annotation**.
- Statistical genomics**: Includes **SVAT result**, **Gene-level result**, and **Pathway-level result**.
- New analytical analysis**: Includes **Result**.

QV set in pipeline code (i.e. QV 1)

The diagram illustrates the components of the QV set in pipeline code and user documentation.

Pipeline code

- General project variables
- QV variables
 - QV step 1
 - QV step ...
 - QV step ...
 - QV step n

User docs

- QV description file
- Brief explanation and definitions for user

A plus sign (+) is shown between the General project variables and the QV variables. A double-headed arrow connects the QV settings (under QV variables) and the Brief explanation and definitions for user.

The diagram is divided into two main sections: 'Pipeline code' on the left and 'User docs' on the right, separated by a vertical dashed line.

Pipeline code:

- Contains a large '+' sign on the left.
- A document icon labeled 'QV 2 variables' contains a list of steps: 'QV step 1', 'QV step ...', 'QV step ...', and 'QV step n'.
- Below this is an orange box labeled 'QV settings'.

User docs:

- Contains a document icon labeled 'QV 2 description file'.
- Below this is an orange box labeled 'Brief explanation and definitions for user'.

A double-headed arrow connects the 'QV settings' box in the Pipeline code section to the 'Brief explanation and definitions for user' box in the User docs section.