

The diagram illustrates the workflow for genetic analysis, starting from an undiagnosed patient and branching into traditional and modern methods.

Top Workflow:

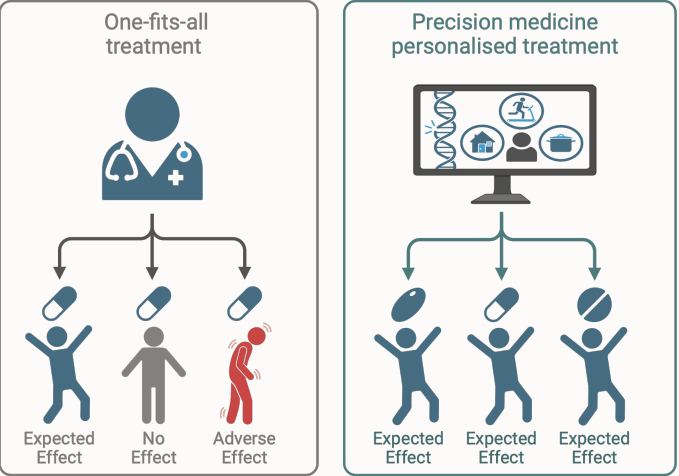
- Undiagnosed patient:** Represented by an illustration of a patient in a hospital bed.
- Minimal blood sample:** Represented by a test tube containing a red liquid.
- Genetic material:** Represented by a pipette tip and a circular DNA molecule.

Bottom Workflow:

- Genetic material:** Represented by a blue double helix structure.
- Next Generation Sequencing:** Represented by an illustration of a sequencing machine.

Analysis Methods:

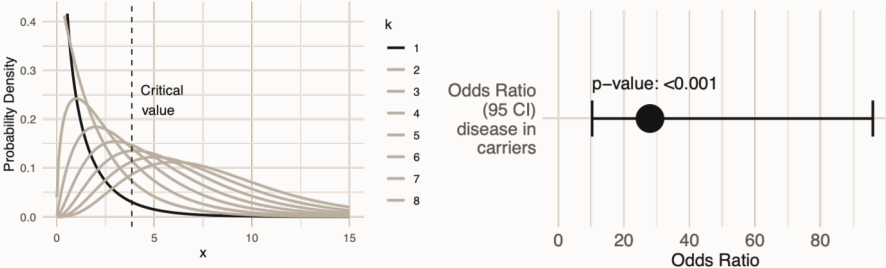
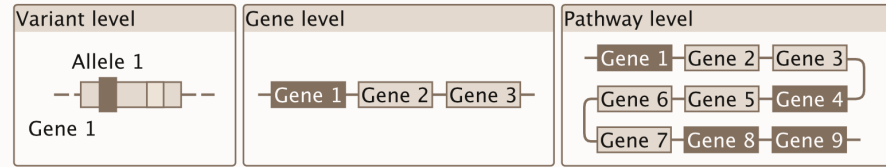
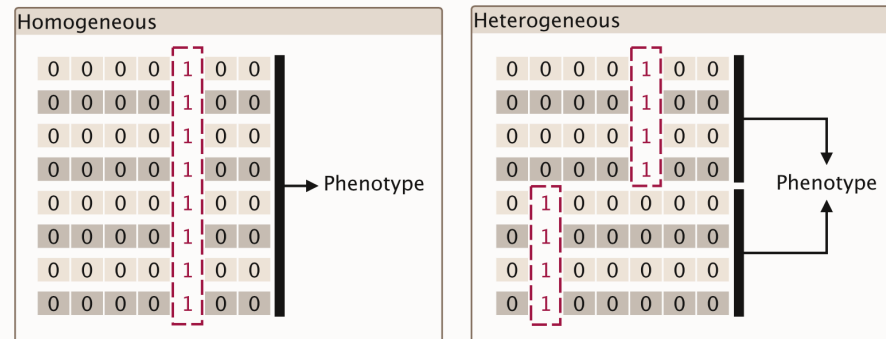
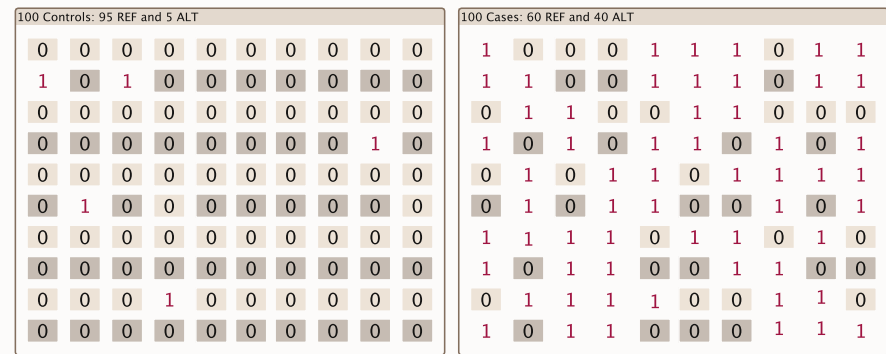
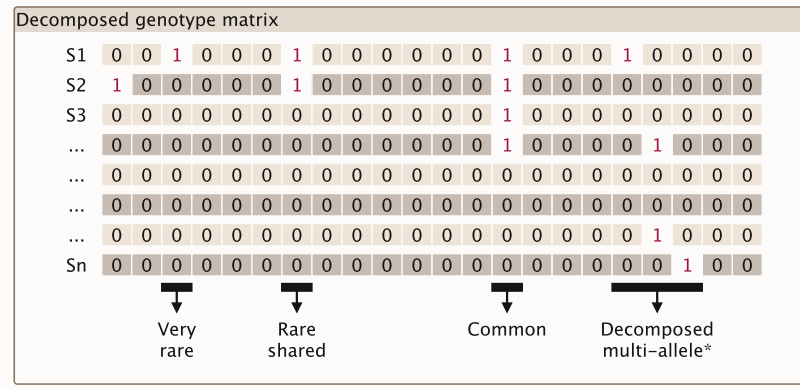
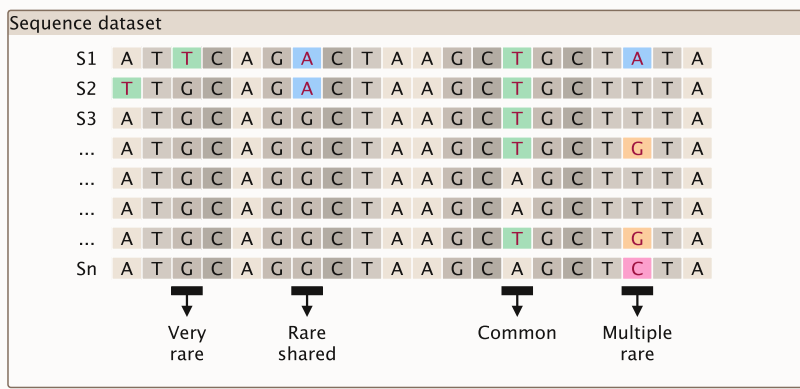
- Traditional clinical genetics:** A box containing an illustration of a person at a computer and the text: "Manual processing and interpreting data is complex and lengthy." Above the person is a sequence: `TTAGGAGCTCAATATCATGCGCAT`.
- GenomeSwift bioinformatics workflow:** A box containing an illustration of a computer monitor displaying a neural network and the text: "Automated analysis is faster and more powerful." To the right of the monitor is a sequence: `TTAATATCATGCGCAT`, `TCAAAATATCATGCG`, `TCAATATCATGCGCAT`, `TCAATATCATGCGCAT`, `TCAATATCATGCGCAT`, `TCAATATCATGCGCAT`.



Variant effect

Variant Impact

- High
- Moderate
- Low
- Modifier
- Proxy



Evident pathogenicity

Predicted pathogenicity

A

Number of genes

No. ACMG criteria (P) variants per gene

B

No. variants

No. ACMG criteria assigned (P)

C

No. samples

No. ACMG criteria assigned (P)

D

No. variants

Pathogenicity thresholds passed

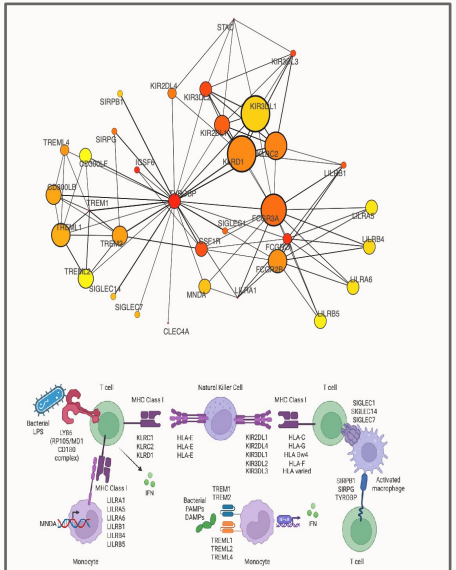
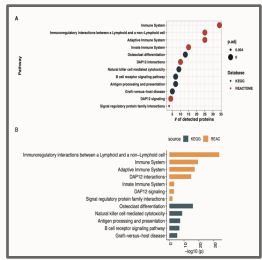
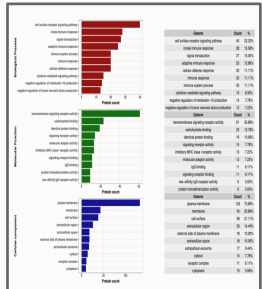
E

Strong pathogenic
Moderate pathogenic
Supporting pathogenic

F

BayesDel_noAF, BayesDel_noAF, CADD, DANN, EGEN, EGEN-PC, FATHMM, FATHMM-MKL, FATHMM-XF, LRT, M-CAP, MetaLR, MetaLRN, MetaSVM, MetaSnooper, MutationTaster, phastCons100way, reVEL, PhyloP-HGV, PolyPhen-HMM, PROVEAN, REVEL, SIFT

In silico prediction score



- DNA variation
- Interpret effect
- Molecular analysis
- Complex biological effects
- Final interpretation
- Standards and guidelines

