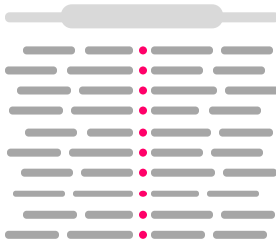


WGS e.g. with protein coding region

Reference genome



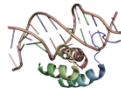
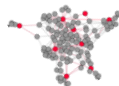
Aligned reads ~ 30X



Pathogenic variant
e.g. G>C

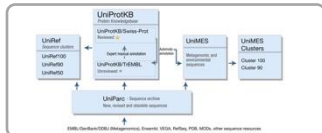
ATGCAGCTACGGAC...

Variant incl.
knowledge
base data



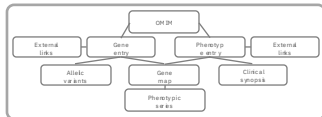
Pathway

Protein
function



OMIM

ClinVar



more ...