

# **VARIANT CALLING**

## **Whole Genome Sequencing**



# Whole

## Genome Sequencing

Virilizer (Vir), first isolated from *Drosophila melanogaster*, plays a role in sex determination. In humans, its homologue and METTL3 are involved in N6-adenosine methylation (m<sup>6</sup>A) writing. As m<sup>6</sup>A of mRNA is an important process in most eukaryotes, it is very important to understand its role and the status of factors accompanying this modification. Whole genome sequencing enables the genome-wide mutation profiling in the *vir-1* mutant of *Arbidopsis*.



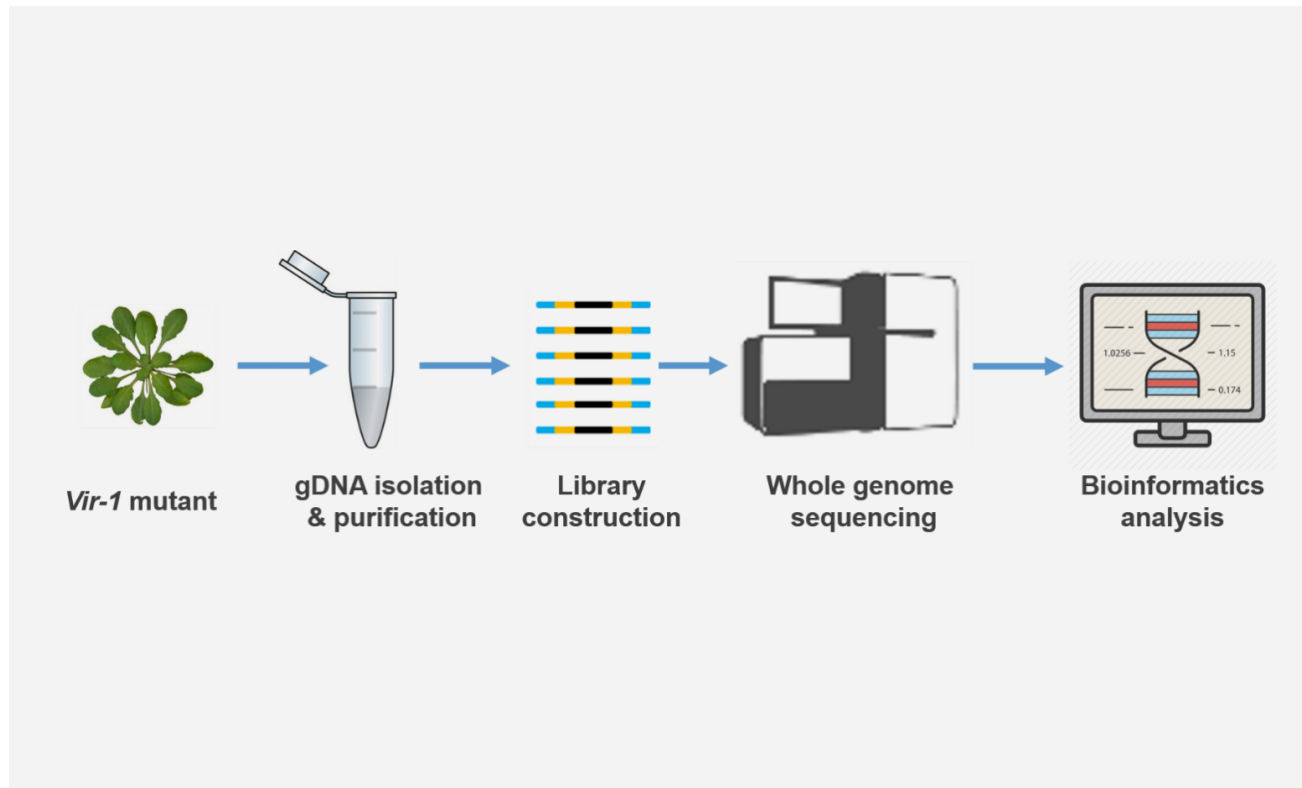
### Features

- ◆ Single base-pair resolution
- ◆ Genome-wide variation profiling (SNPs, InDels, CNVs, SVs)
- ◆ *De novo* assembly or reference-based assembly



### Applications

- ◆ Create a reference genome
- ◆ Population evolution and phylogenetic studies
- ◆ Disease research, drug discovery and development, and personalized medicine

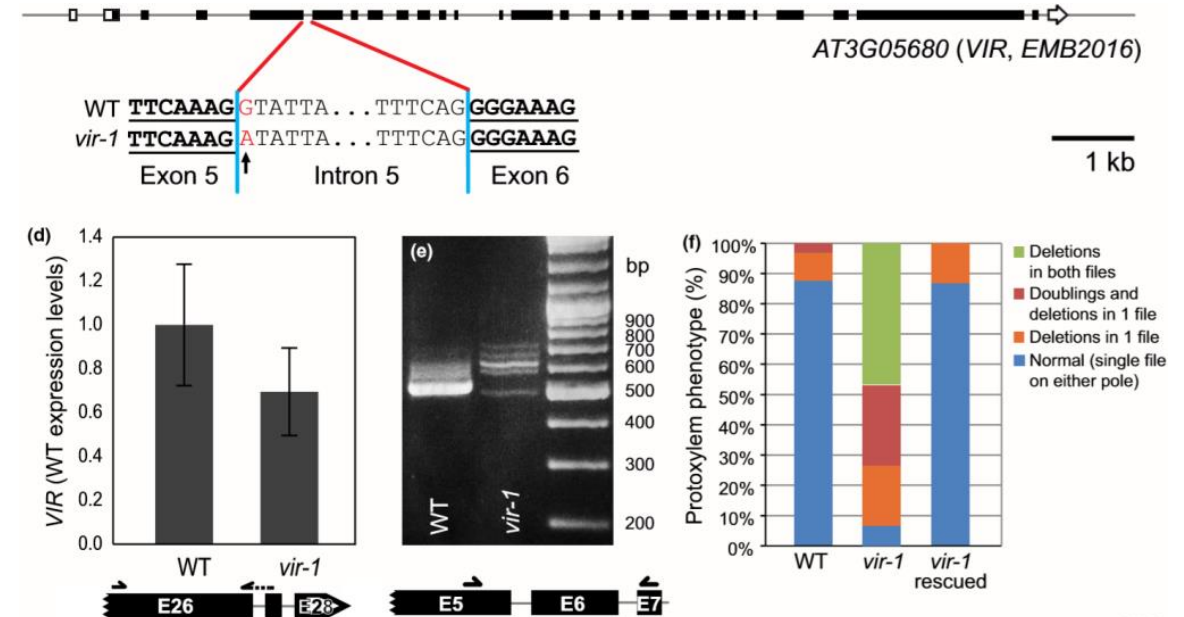


## Assay Overview

The *vir-1* mutant was isolated in an ethyl methanesulfonate (EMS) mutagenesis screening for reduced and irregular *AHP6prom:GFP* expression (*AHP6* is a factor required for protoxylem formation in *Arabidopsis*). Total DNA was extracted and subjected to whole genome sequencing on Illumina instruments, so as to identify the mutations and functions of Virilizer in *Arabidopsis*.

## Data Overview

The *vir-1* mutation (orthologous to the splicing regulator/m6A writer protein Virilizer) is caused by a G-to-A conversion at the first nucleotide of the 5th intron of the VIR gene. Although the *vir-1* mutation does not significantly change VIR expression levels at  $\alpha 0.05$ , it leads to numerous predominantly wrongly spliced VIR transcripts as detected by RT-PCR.



## Conclusions

Researchers use combined methods of genetics, proteomics and RNA biochemistry to investigate a core set of m<sup>6</sup>A write proteins in *A. thaliana*, which includes MTA, MTB, FIP37, Virilizer and the E3 ubiquitin ligase HAKAI.

### Reference:

Růžička K, Zhang M, Campilho A, et al. Identification of factors required for m<sup>6</sup>A mRNA methylation in Arabidopsis reveals a role for the conserved E3 ubiquitin ligase HAKAI. *New Phytologist*, 2017, 215(1): 157-172.