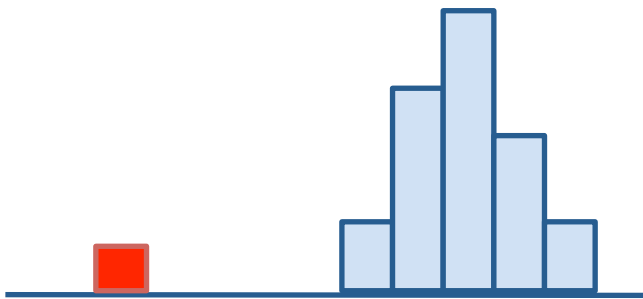


### 1. Patient fibroblasts (n=105)

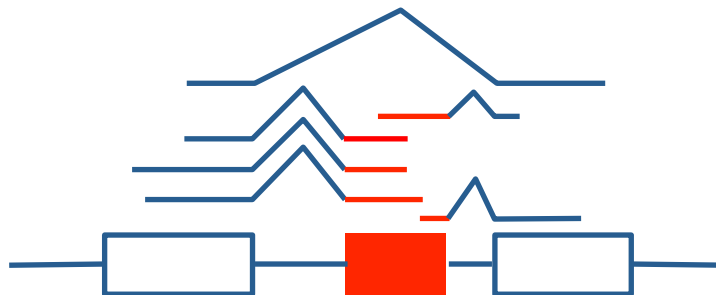


### 2. RNA sequencing

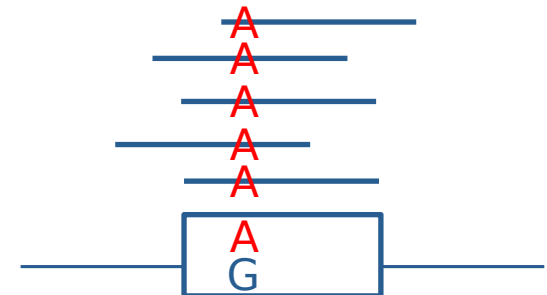
Aberrant expression



Aberrant splicing

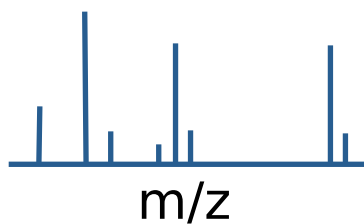


Mono-allelic expression

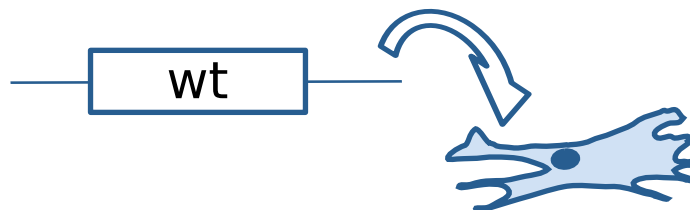


### 3. Functional and biochemical validation

Proteomics



Complementation



Supplementation

