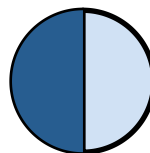


Exome sequenced patients

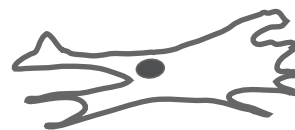
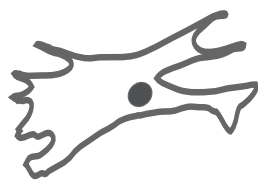
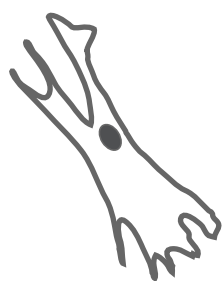


Genetic diagnosis



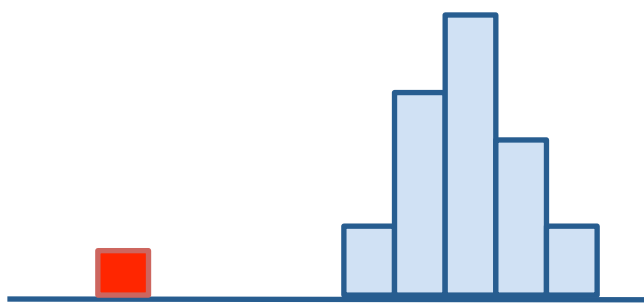
No genetic diagnosis

### 1. Patient fibroblasts

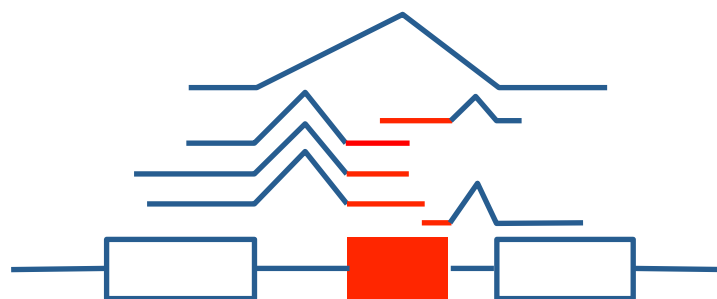


### 2. RNA sequencing

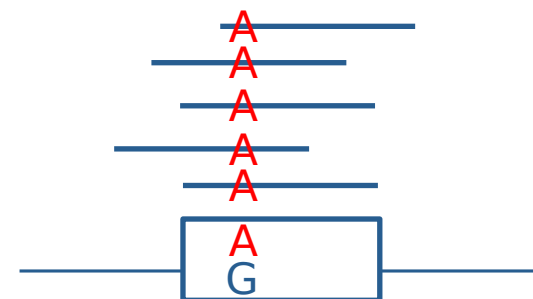
Aberrant expression



Aberrant splicing

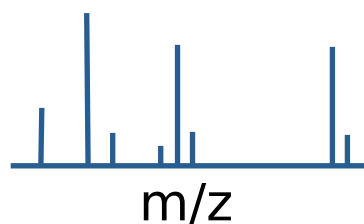


Mono-allelic expression

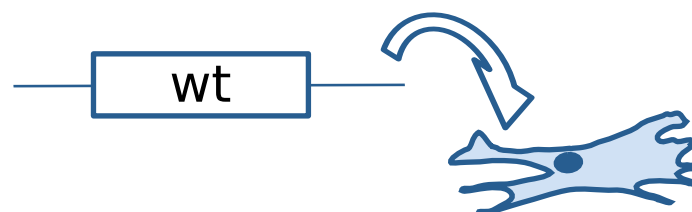


### 3. Functional and biochemical validation

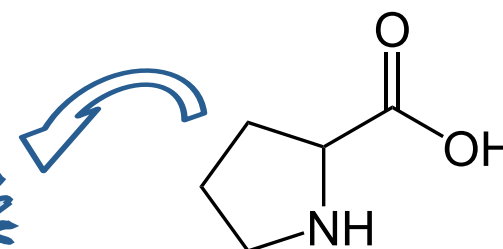
Proteomics



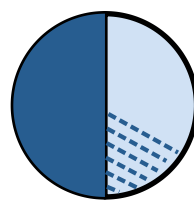
Complementation



Supplementation



Genetic diagnosis



No genetic diagnosis

New genetic diagnosis