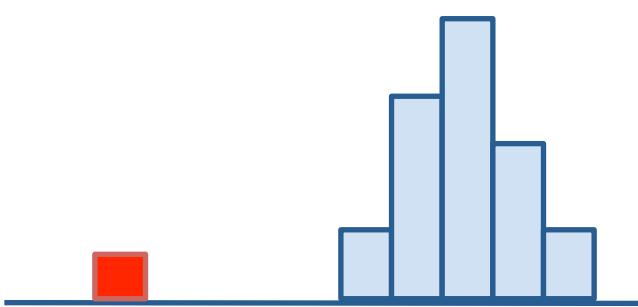


1. Patient fibroblasts (n=105)

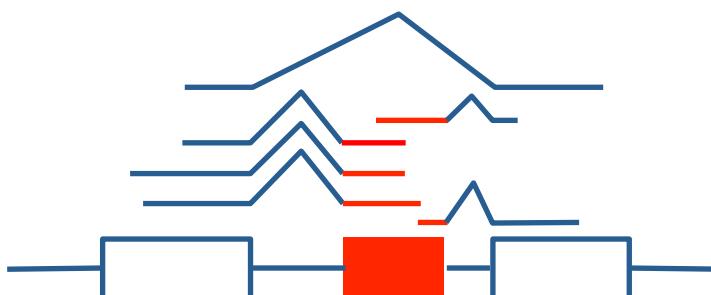


2. RNA sequencing

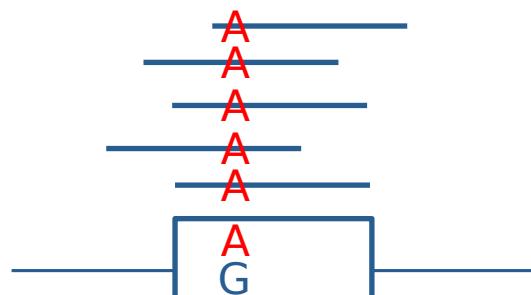
Aberrant expression



Aberrant splicing

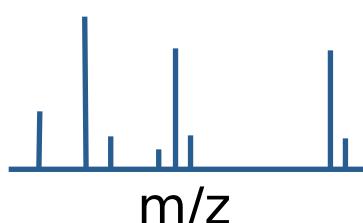


Mono-allelic expression

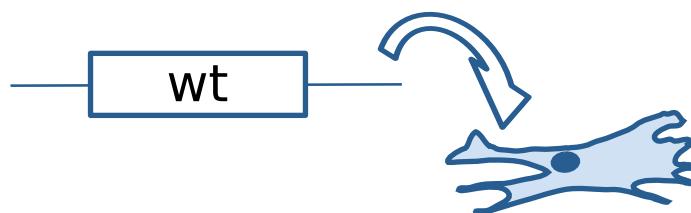


3. Functional and biochemical validation

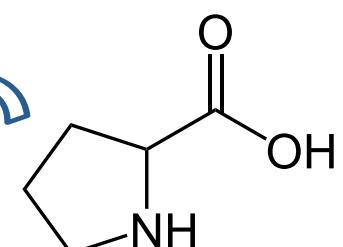
Proteomics



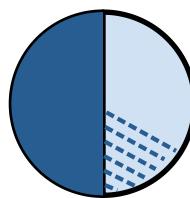
Complementation



Supplementation



Genetic diagnosis



No genetic diagnosis

New genetic diagnosis