

# Constitutional SAMD9L mutations cause familial myelodysplastic syndrome and transient monosomy 7

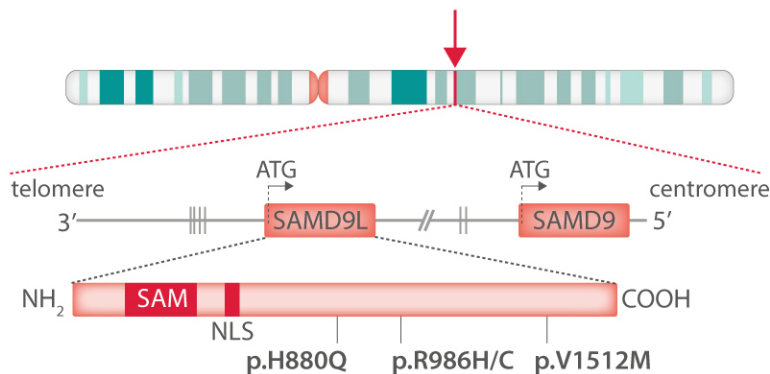


From 4 unrelated pedigrees with myelodysplastic syndrome and loss of 7/7q

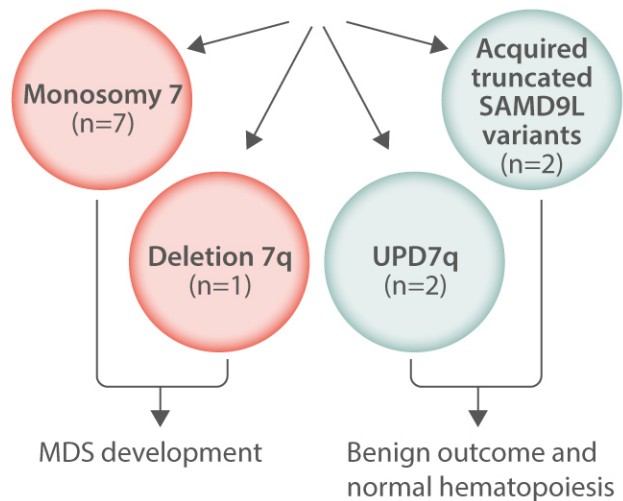
Median age 2.1 (1-42) years

Thrombocytopenia w/ or w/o cytopenia and hypocellular marrow

## Heterozygous missense mutations in SAMD9L gene (7q21)



## Mechanisms of clonal escape from SAMD9L mutations



- ➔ Description of the somatic landscape likely contributing to MDS progression
- ➔ Observation of transient monosomy 7
- ➔ Occurrence of non random revertant mosaicism leading to complete hematological recovery