Classification of germline RUNX1 variants predisposing to myeloid neoplasms
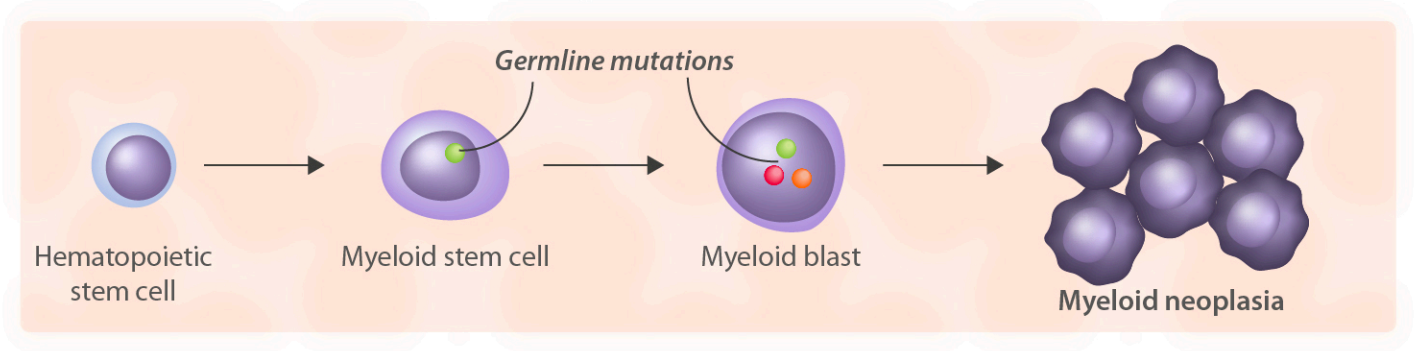
Myeloid differentiation

Germline mutations

Hematopoietic stem cell

Myeloid stem cell

Myeloid blast

Myeloid neoplasia

Detection of germline variants
- Next generation sequencing (NGS)
- Microarrays

The 2015 American College of Medical Genetics and Genomics (ACMG) and Association for Molecular Pathology (AMP) guidelines for sequence variant interpretation

Benign (BEN)

Likely Benign (LBEN)

Variant of Uncertain Significance (VUS)

Likely Pathogenic (LPATH)

Pathogenic (PATH)