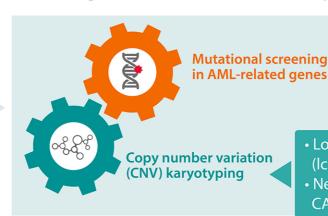
## Development of a NSG platform to enable fast and reliable genetic aberrations that are of critical importance for diagnosis, prognosis and therapy in adult acute myeloid leukemia



Diagnostic approach in acute myeloid leukemia (AML)

## Whole genome Next Generation Sequencing (NGS) technology





- Aberrations
- Translocations
- Short sequencing variants
- FLT3-ITDs
- KMT2A-PTDs
- Low coverage whole genome sequencing (Ic-WGS) analysis
- New algorithm for the detection of CNVs:
  CAI[N] (chromosomal aberration identifier [numerical])

## Advantages of NGS technology

- Identification of chromosomal gains and losses at high resolution (1Mb)
- Need of only 5-10% genome coverage for detection
- >90% sensitivity and specificity
- No need of short term culture of the sample material
- Implementation even at small diagnosis centers



22 Primary AML patient samples



- Correct classification of 9/9 numerically normal karyotypes
- Identification of 30/31 (97%) copy number variations