

Associations between dysplastic findings and somatic mutations in de novo acute myeloid leukemia (AML)



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Patients with de novo AML without WHO-defined cytogenetic abnormalities





- 81% (n=137) normal karyotype
- 18% (n=31) abnormal karyotype



Targeted sequencing on bone marrow aspirates for recurrent mutations associated with myeloid malignancies:

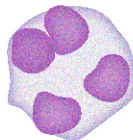
- DNA methylation
- Epigenetic regulators
- Transcription factors
- Cohesin complex
- RAS pathway
- Spliceosome pathway

Degree of dysplasia according to mutational pathways

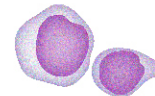
- **Cohesin pathway mutations**  a higher degree of megakaryocytic dysplasia (q=0.046)
 - STAG2 mutations  marginally with greater overall megakaryocytic dysplasia (q=0.064) and marginally with greater overall myeloid lineage dysplasia (q=0.052)
- **RAS pathway mutations**  marginally with greater degree of megakaryocytic dysplasia
 - RIT1 mutations  marginally with greater overall myeloid lineage dysplasia (q=0.056)

Dysplastic features in de novo AML

Megakaryocytes with separated nuclear lobes



Small size megakaryocytes



Megakaryocytes with hypogranular cytoplasm and abnormal nuclear lobulation



Dysplastic erythroid cells with irregular nuclear contours

