Monosomal karyotype (MK) in myeloid malignancies
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Published in Atlas Database: April 2011
Online updated version : http://AtlasGeneticsOncology.org/Anomalies/MonoKaryoID1574.html
DOI: 10.4267/2042/46038
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Identity

Note
Monosomal karyotype is defined as the presence of at least 2 autosomal monosomies or a single autosomal monosomy associated with at least one structural abnormality.

Clinics and pathology

Disease
Acute myeloid leukemia (AML), myelodysplastic syndrome (MDS), primary myelofibrosis (PMF)

Epidemiology
In AML, the frequency of monosomal karyotype increases with age: 4% in patients ≤ 30 years; 6-10% in patients ≤ 60 years and 13-20% above 60 years.

The frequency of MK cases is significantly higher in t-AML compared with de novo AML (24% vs 10%).

In MDS, MK frequency among patients with a complex karyotype is 83%. In children with advanced MDS MK frequency is 15%.

In PMF patients, MK is present in 42% of complex karyotype cases.

Clinics
Monosomal karyotype is associated with prior chemotherapy or history of abnormal blood counts (Estey, 2010).

Prognosis
Very poor prognosis, worse with advanced age.

In comparison with non-MK patients, AML, MDS and PMF patients with a monosomal karyotype have lower overall survival rates. In MDS and PMF, the risk of leukemic transformation is higher in MK patients than in patients with non-MK complex karyotypes.

In MK-AML, overall survival at 4 years after diagnosis is 3-4% (vs 26-27% for non-CBF, non-MK AML patients) and event-free survival 2%. Overall survival decreases with advancing age: 17-40% for patients ≤ 30 years, 3-4% ≤ 60 years and 1% in patients older than 60 years. Complete remission rate is generally low, and also worse in older groups (24-52% under 60 years and 13-34% in patients older than 60 years).

In adult MDS patients with MK, 2-year survival is 6% (vs 23% for complex karyotype without monosomies) and 1-year leukemia risk is 32% (vs 14% for complex karyotype without monosomies). In advanced childhood MDS, the presence of a MK does not seem to be an independent adverse prognostic factor.

In PMF, the median survival for MK patients is 6 months, 2-year survival rate is 17% and 2-year leukemic transformation rate is 29% (vs 24 months, 51% and 8,3% in complex karyotype without monosomies).

As with most prognostic factors, the significance of monosomal karyotype seems to depend on the treatment strategy (Itzykson et al., 2011; Löwenberg et al., 2011).
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Cytogenetics

Note
Monosomal karyotype group does not include AML with t(15;17)(q22;q21) or CBF abnormalities (t(8;21)(q22;q22); inv(16)(p13q22)/t(16;16)(p13;q22)); monosomies of sex chromosomes (-X,-Y) are excluded due to apparent lack of negative prognostic effect.

Cytogenetics morphological
Two or more autosomal monosomies or one autosomal monosomy associated with at least one structural abnormality. The most frequent autosomal monosomies in MK involve the chromosomes 7, 5, 17 and 18.

Genes involved and proteins

Note
Genes involved are unknown.

References


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This article should be referenced as such: