Isolated tetrasomy 8 in AML, MDS and MPD

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Identity

Note
Isolated tetrasomy 8 is relatively rare compared to trisomy 8. A review of literature revealed only 29 cases of solely tetrasomy 8 in hematologic malignancies. Except for the implication of MLL in AML cases, no additional molecular abnormalities have been explored.

Clinics and pathology

Disease
Acute myeloid leukemia (AML)

Phenotype/cell stem origin
FAB subtypes: M0 AML (2 cases) M1 AML (1 case) M2 AML (3 cases) M4 AML (3 cases) M5 AML (11 cases out 20).

Epidemiology
Median age 56 years (range: 17-82 years); sex ratio: 13M/7F.

Clinics
Tetrasomy 8 has been observed in de novo malignant hemopathies as well as in leukaemia with prior history of haematological disorder (4 cases of myelodisplastic syndrome: 2 RA and 2 RAEB), exposure to radiotherapy or treatment with cytotoxic chemotherapy (1 case of each).

Prognosis
Median survival: 15 months.

Disease
Myelodysplastic syndrome (MDS)

Phenotype/cell stem origin
FAB subtypes: refractory anemia (RA) (1 case), refractory anemia with excess of blasts (RAEB) (1 case), refractory anemia with ringed sideroblasts (RARS) (1 case), chronic myelomonocytic leukemia (CMML) (2 cases) and MF (1 case).

Epidemiology
Median age: 68 years (range: 46-91 years); sex ratio: 4M/2F.

Prognosis
Median survival: 5.5 months.

Disease
Myeloproliferative disorder (MPD)

Phenotype/cell stem origin
Subtypes: polycytemia vera (PV) (1 case) and myelofibrosis (MMM) (2 cases).

Epidemiology
Median age: 36 years (range 30-51 years); sex ratio: 0M/3F.

Prognosis
Median survival: 36 months.
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Cytogenetics

Specific sequences for the centromere of chromosome 8. No cryptic MLL rearrangements were detected in all cases.

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