Childhood myelodysplastic syndromes

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Clinics and pathology

Disease

Very heterogeneous:

I. idiopathic MDS
II. secondary MDS: to previous chemo- and/or radio-therapy.
III. ‘genetic MDS’: cases associated with a congenital genetic disease, such as:
- Neurofibromatosis type 1 (Von Recklinhausen) (MIM 16220): an hamartoneoplastic syndrome,
- Kostmann syndrome (MIM 20270): also called congenital neutropenia,
- Bloom syndrome (MIM 21090): a chromosome instability syndrome,
- Dubowitz syndrome (MIM 22337): mimicks Bloom’s, but without chromosome instability,
- Fanconi anaemia (MIM 22765): a chromosome instability syndrome,
- Schwachman syndrome (MIM 26040): with pancreatic insufficiency, and risk of leukaemia,
- Pearson disease (MIM 26056) and other mitochondrial diseases: they often share pancreatic insufficiency, bone marrow pancytopenia with myelodysplastic features but maintained polyclonality, muscular and other ubiquitous manifestations,
- Familial monosomy 7,
- Familial platelet storage pool deficiency,
- Unbalanced constitutional karyotypes, including +21, +8, del(11q), del(21q) miscellaneous conditions.

Phenotype / cell stem origin

RA, RARS (very rare), RAEB, RAEBT, CMMML, ‘Juvenile CML’, ‘Infantile Monosomy 7’; ‘non classifiable cases according to the FAB’; with variable proportions according to the studies.

Epidemiology

10% of haematological malignancies in children; median age: 2 to 5 yrs; sex ratio: balanced for some, male predominance (in RAEB±T or CMMML) for others.

Prognosis

CR is obtained; however, median survival is about 3 yrs, while 1/3 of the cases may be considered as cured; good prognostic features are: young age, female sex, normal karyotype, and some of the genetic predisposing factors; worse prognosis is found in secondary MDS, RAEB and RAEBT, cases with +8, +19, t(1;7).

Cytogenetics

Cytogenetics, morphological

A normal karyotype or a monosomy 7 (intermediate prognosis) are found in 30% -or more- of cases each; others are: +8, +21, t(1,7), del(6q).

References


[No authors listed]. Forty-four cases of childhood myelodysplasia with cytogenetics, documented by the Groupe Français de Cytogénétique Hématologique. Leukemia 1997 Sep; 11(9):1478-85.

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