Leucemia acuta mieloide
Contributi dell'ematologia bolognese

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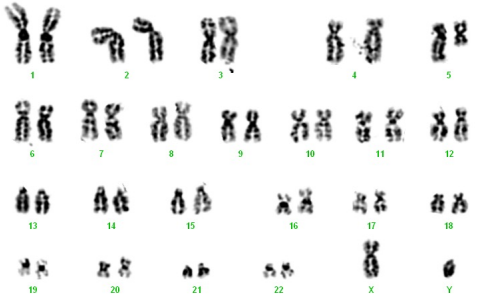
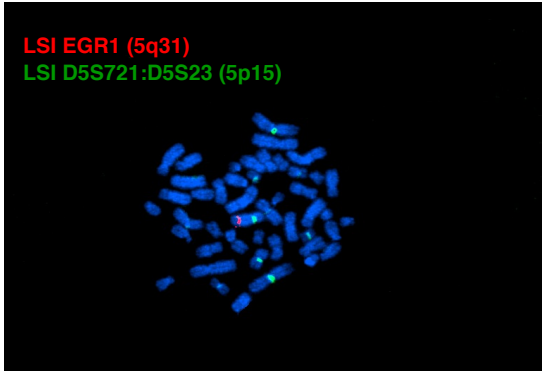
Disclosures of Name Surname

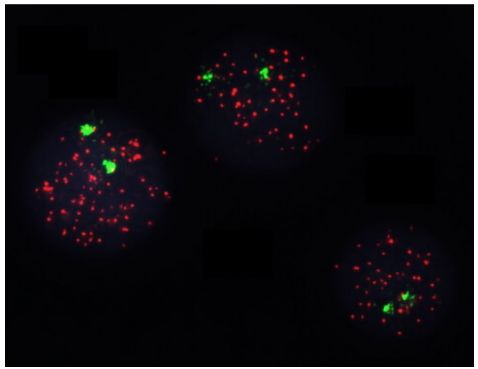
Company name	Research support	Employee	Consultant	Stockholder	Speakers bureau	Advisory board	Other

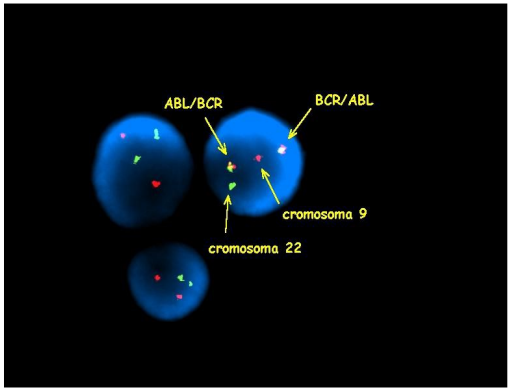
Attività trasversale in :

- Leucemia mieloide cronica
- **Leucemia acuta mieloide** e linfoide
- Neoplasie mieloproliferative Ph-
- Linfomi non-Hodgkin
- Mieloma multiplo – MGUS
- Leucemia linfatica cronica
- Neoplasie mieloidi/linfoidi con eosinofilia









GIMEMA WP CML

Centralizzazione dati citogenetici e citogenetico molecolari

Protocolli clinici (TKI) 021 – 022 – 023

Chronic myeloid leukemia: a prospective comparison of interphase fluorescence in situ hybridization and chromosome banding analysis for the definition of complete cytogenetic response: a study of the GIMEMA CML WP

Deletions of the Derivative Chromosome 9 Do Not Influence the Response and the Outcome of Chronic Myeloid Leukemia in Early Chronic Phase Treated With Imatinib Mesylate: GIMEMA CML Working Party Analysis

Variant Philadelphia translocations: molecular-cytogenetic characterization and prognostic influence on frontline imatinib therapy, a GIMEMA Working Party on CML analysis

2009-2012

Additional chromosomal abnormalities in Philadelphia-positive clone: adverse prognostic influence on frontline imatinib therapy: a GIMEMA Working Party on CML analysis

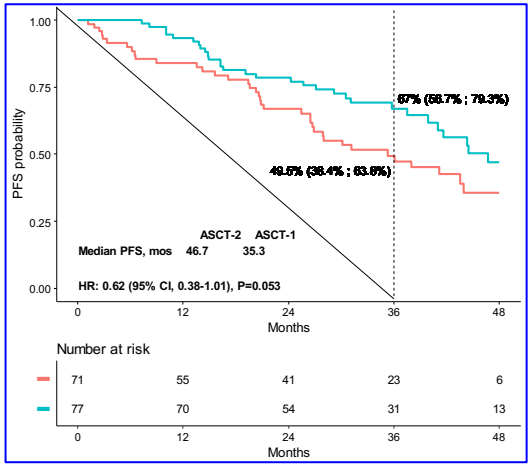
EMN02 - Centralizzazione nazionale studi citogenetico-molecolari

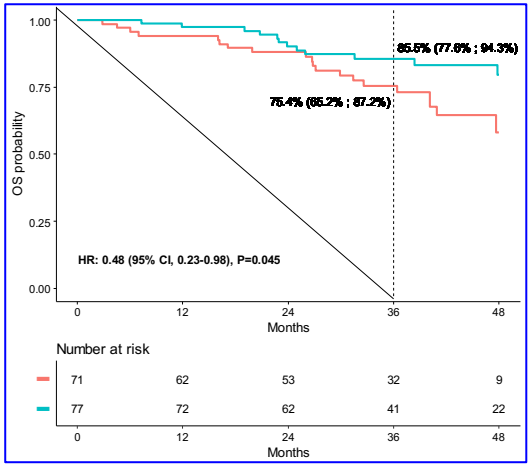
High-risk Cytogenetics in Newly Diagnosed Multiple Myeloma: Prognostic Relevance of Co-segregations and Analysis of the Role of Double versus Single Autotransplantation

461 evaluable patients for FISH analysis of all HRAs

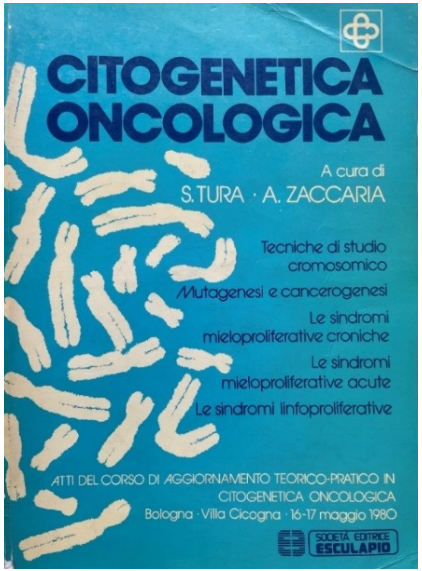
Among the 148 patients with at least 1 HRA, randomization to ASCT-2 significantly prolonged PFS and OS in comparison with ASCT-1

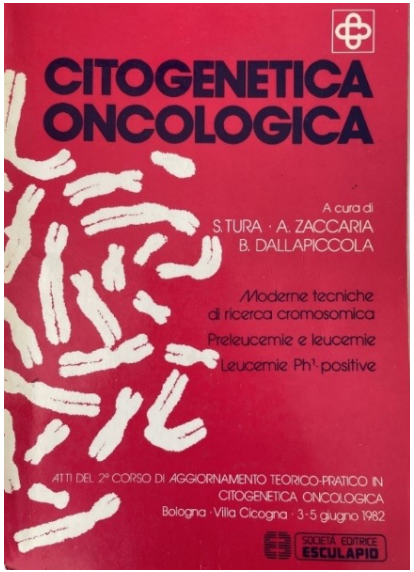
2017





Acute Nonlymphocytic Leukemias and Dysmyelopoietic Syndromes in Patients Treated for Hodgkin's Lymphoma





Chromosome Studies in Patients with Acute Nonlymphocytic or Acute Lymphocytic Leukemia Submitted to Bone Marrow Transplantation—Results of a European Cooperative Study

A.Zaccaria et al

Cytogenetic Analyses in 89 Patients with Secondary Hematologic Disorders—Results of a Cooperative Study

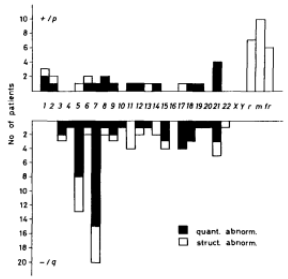
Chromosome studies in acute promyelocytic leukemia. An analysis of 11 cases

Table 5 Chromosome patterns of nine ANLL patients at relapse after BMT, compared with the pattern at diagnosis

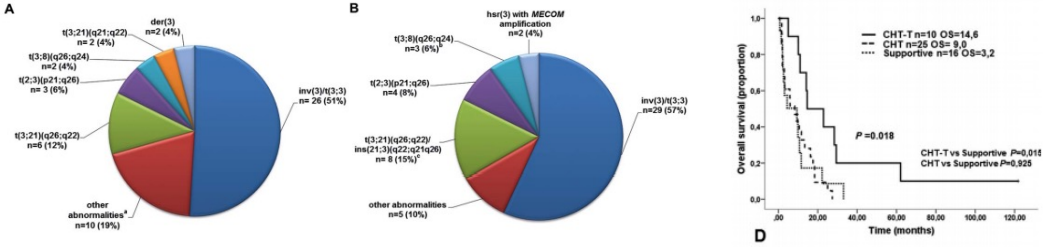
UPN	Sex	Center	Donor Sex	Karyotype at diagnosis	Karyotype at relapse
1	F	Bologna	M	46,XX	46,XX, − 5,t(9;17),14q − , 22q − /46,XX, − 5,6p + ,t(9;17), 12p + ,14q − ,22q −
2	F	Bologna	F	46,XX,11q −	46,XX,11q −
4	M	Bologna	F	46,XY	47,XY, + 8
776 (IN)	F	Ulm	F	N.D.	46,XX, structural abnormalities
5	F	Leuven-Brussels	M	46,XX	50/53,X[X],der(5), + 6, + 8, + 8,9p − ,der(11q), − 12, + 13,16q + , − 17, + 19, + 21, − 21, + mar
19	F	Leuven-Brussels	M	N.D.	46 − 48,XX,del(1),t(15;17), del(17)
7	M	Leuven-Brussels	F	46,XY	46,XY/46,XY,t(1;2)
4	F	Leuven-Brussels	F	46,XX	46,XX/46,XX,10q −
20	F	Leuven-Brussels	F	46,XX, t(11;19)	46,XX,t(11;19)/ 46,XX,t(11;19), 13q + ,4p + ,5q −

UPN, unique patient number; IN, identification number of Ulm patients.

Figure 1 Cumulative chromosome abnormalities of the patients treated with CT + RT for a previous tumor.



Complex Chromosomal Rearrangements Leading to *MECOM* Overexpression are Recurrent in Myeloid Malignancies with Various 3q Abnormalities



Our study highlights the importance of screening for MECOM rearrangements by FISH in patients with any 3q abnormalities as 3q26 rearrangements can be missed by CBA. All MECOM rearrangements were associated with a very short OS irrespective of the type of treatment, and thus underlining the importance of finding innovative therapies targeting MECOM expression

FGFR1 and KAT6A rearrangements in patients with hematological malignancies and chromosome 8p11 abnormalities: biological and clinical features

Since the poor prognosis associated with KAT6A rearrangement, we also suggest to perform FISH analysis to investigate KAT6A gene in hematological malignancies with 8p11 abnormalities without FGFR1 rearrangements, at least in AML without complex karyotype. To date, alloSCT is the only therapeutic choice to obtain long-term survival and should be considered early in patients with FGFR1 and KAT6A rearrangements.

t(5;12)(q31;p13)/*ETV6::ACSL6* and t(6;9)(p23;q34)/*DEK::NUP214* concurrence in acute myeloid leukemia: an unusual association of two rare abnormalities

Moreover, at relapse, both cases presented with eosinophilia, further strengthening the association of t(5;12) with eosinophilia in myeloid malignancies. Given the poor prognosis and the non-responsiveness to tyrosine kinase inhibitors of cases of ETV6::ACSL6 rearrangement, in contrast to cases of ETV6::PDGFRB rearrangement, we recommend the introduction of testing for this abnormality in myeloid malignancies with eosinophilia.

14q32 rearrangements deregulating *BCL11B* mark a distinct subgroup of T-lymphoid and myeloid immature acute leukemia

DI GIACOMO et al
blood® 2 SEPTEMBER 2021 | VOLUME 138, NUMBER 9

KEY POINTS

- **14q32 rearrangements, activating *BCL11B*, provide a novel biomarker for a new entity among immature acute leukemias.**

Figure 4. Mutational profile of *BCL11B*-a AL. Oncoprint heat map showing all sequence variants detected in *BCL11B*-a AL cases. In addition to somatic mutations (dark green), variants in which the somatic or germline origin could not be definitively assessed (light green) are indicated. Additional gene alterations are represented by different colors. Mutational analysis was not performed in cases 15 and 19 because of lack of material. cnLOH, copy neutral loss of heterozygosity.

4q12 translocations with *GSX2* expression identify a CD7^{+} acute myeloid leukaemia subset

despite heterogeneous molecular breakpoints, GSX2 overexpression is the common denominator of all 4q12 translocations, typically found in CD7+ AML-M0 with bad risk myeloid mutations and poor response to chemotherapy.

DI GIACOMO et al *British Journal of Haematology*, 2015, **171**, 137–151

Novel and Rare Fusion Transcripts Involving Transcription Factors and Tumor Suppressor Genes in Acute Myeloid Leukemia

Overall, by combining different approaches, we described rare fusion events contributing to the complexity of AML and we linked the expression of some chimeras to genomic alterations hitting known genes in AML.

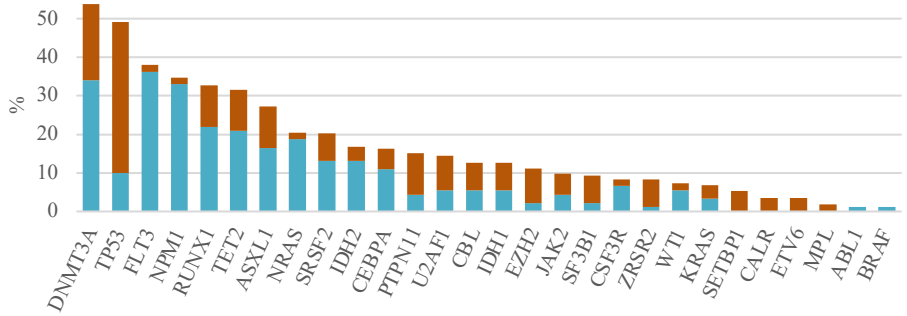
Padella A et al *Cancers* **2019**, *11*, 1951; doi:10.3390/cancers11121951

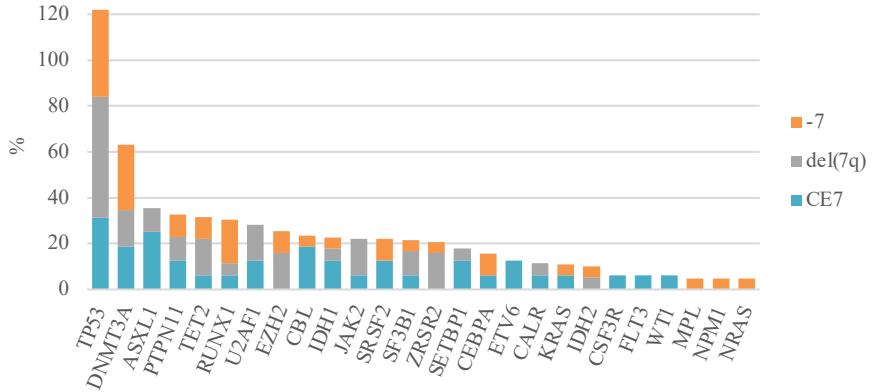
"High Risk Genetic Abnormalities in Acute Myeloid Leukemia and Myelodysplastic Syndrome: Unravelling the Pathogenetic Mechanisms to Improve Diagnostic and Therapeutic Approaches" Progetto di Ricerca Finalizzata

Obiettivi: Il progetto è finalizzato alla miglioramento delle conoscenza dei meccanismi patogenetici in neoplasie mieloidi con alterazioni genetiche ad alto rischio attraverso una caratterizzazione del profilo genetico che potrà portare a trattamenti terapeutici più appropriati:

1) Studio delle alterazioni del cromosoma 7q, del background genetico e della loro evoluzione clonale;

2) Valutare la frequenza e l'impatto prognostico dell'overespressione dei geni ***PRDM16*** **e** ***MECOM*** e caratterizzarne il background genetico per identificare percorsi patogenetici specifici.





grazie!

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