

anemia with excess blasts and CML in blast crisis.

Cytogenetically, 54% of AML patients with t(8;21) had additional chromosome abnormalities, and most of them were similar to those described in the literature: sex chromosome loss (38%), del 9q (12,9%), del 7q(6%) and trisomy 8 (6%).

In three cases, we found different additional cytogenetic abnormalities. Using fluorescence in situ hybridization (FISH), two of these showed in the analysis an atypical fusion between AML1 and ETO genes.

The first case was a 12-year-old girl. She showed an additional translocation, t(1;1)(q25;p36). The patient was transplanted and is in complete remission after 9 years from diagnosis.

The second case was a 77-year-old man recently diagnosed. He has a variant of the specific translocation, involving chromosomes 7, 8 and 21, with loss of critical region 7q31 and loss of the derivative 8. At this moment, he is in complete remission.

The third patient, a 17-year-old man, has at diagnosis the typical translocation t(8;21)(q22;q22). He was transplanted and relapsed after 4 months with additional anomalies, del(2)(p23),add(3)(p13)+ mar. FISH analysis at that time showed atypical AML1/ETO rearrangement. The patient died after 2 years from diagnosis.

The prognostic value of the additional uncommon chromosomes abnormalities associated with this translocation t(8;21) is under discussion. Further studies with more patients are needed to determine the clinical relevance of these anomalies.

## 6.P61

### Epigenetic silencing of the tumor suppressor HIC1 and SFRP2 genes in prostate carcinomas

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although the prevalence of this modification remains poorly understood. Hypermethylated in cancer 1 (HIC1) gene is a putative tumor suppressor gene on chromosome 17p13.3 and epigenetically inactivated in different cancers.

**Methods:** In this study, using bisulphite-modifying-based reverse hybridisation technique, a total of 30 prostate tumour specimens were investigated for promoter methylation status of tumour suppressor HIC1, DAPK1, SFRP2, p16 and MGMT genes.

**Results:** High frequency of promoter hypermethylation was found in HIC1 (70.9%) and SFRP2 (58.3%) genes in the tumour samples examined. Tumour specimens also showed hypermethylated promoter domain for the DAPK1 and MGMT tumour suppressor genes.

**Conclusions:** These findings showed that tissue-specific epigenetic silencing of tumour suppressor HIC1 and SFRP2 genes may play a crucial role in tumour progression and recurrence in prostate carcinomas.

**Keywords:** HIC1, SFRP2, MGMT, Prostate carcinoma, Suppressor genes, Epigenetic silencing

## 6.P62

### Cytogenetic analyses in a group of patients with myelodysplastic syndromes

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Myelodysplastic syndromes (MDS) constitute a heterogeneous group of clonal disorders of haematopoietic stem cell diseases characterised by dysplasia and ineffective haematopoiesis in one or more of the major myeloid cell lines. This disease occurs predominantly in older adults where the median age at diagnosis is approximately 70 years. The aim of this study was to evaluate the data from cytogenetic analyses in 425 patients with myelodysplastic syndromes. This population was constituted by 212 females and 213 males; the median age at diagnosis is 66 years. Numerical and structural chromosomal abnormalities were documented for each patient and subdivided according to the number

of additional abnormalities. From the 425 cytogenetic analyses, 97 (22.8%) were abnormal. The results for the abnormal population were as follows: 73.2% had only one anomaly, 9.3% had two anomalies, and 17.5% had a complex karyotype. In the abnormal population, the most frequent isolated anomaly observed was the trisomy 8 (17.5%), followed by the deletion of chromosome 5 (13.4%), the loss of chromosome Y (11.3%) and the deletion of chromosome 20 (8.2%). When associating these anomalies with complex karyotypes, the most frequent anomaly observed was the deletion of chromosome 5 (24.7%). Overall, these results are different from those in the literature; however, the deletion of chromosome 5 is still the most recurrent anomaly in this syndrome. We can also conclude that the loss of chromosome Y is not always associated with age, but is one of the anomalies that characterize this group of pathologies. All these anomalies were found by cytogenetic analysis, a low-cost technique that allows clinicians to use this important prognostic tool to evaluate and make a more accurate clinical decision for patients with MDS.

**Keywords:** Myelodysplastic syndromes, Cytogenetic data review

## 6.P63

### Loss of the Y chromosome in male patients with haematological disorders

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The clinical association between loss of the Y (LOY) chromosome and haematological disorders has been continuously debated because both phenomena can be age-related. In order to understand the relationship between the LOY chromosome and the different haematological diseases, we retrospectively analysed cytogenetic results of 1,241 male patients from 1995 to 2010. Seventy-eight patients (6.3%) showed LOY. Of the 78 patients without Y chromosome, 15 (19.2%) had B cell lymphomas (B lymphomas), 12 (15.4%) had myelodysplastic syndromes (MDS), 10

(12.8%) had chronic myelogenous leukaemia (CML), 10 (12.8%) had acute myeloid leukaemia (AML), 8 (10.3%) had myeloproliferative neoplasms (MN), 6 (7.7%) had chronic lymphocytic leukaemia (CLL), 5 (6.4%) had multiple myeloma (MM), 5 (6.4%) had other mature B cell neoplasms (BN), 3 (3.8%) had MDS/MN, 3 (3.8%) had other mature T cell neoplasms (TN), and 1 (1.3%) had acute lymphoblastic leukaemia (ALL). We did not observe the LOY chromosome in the 15 patients (1.2% of all patients studied) with Hodgkin's disease. These percentages can be different if we consider only the pathology in which the LOY was found: 4.1% of all patients with MN, 5.7% of all patients with MDS, 9.3% in the patients with AML, 12.5% in patients with ALL, 5.8% in patients with CLL, 5.8% in B lymphoma patients, 8.2% in the CML patients, 4.5% in MM patients, 9.1% in BN patients, 9.1% in MDS/MN patients and 15.8% in TN patients. Twenty-five patients (32.1%) had the LOY associated with other cytogenetic anomalies. There are few reports of LOY associated with haematological disorders since this has been considered mainly an age-related event. Therefore, the tendency of LOY diseases to be associated that seems apparent in our data indicates that careful consideration should be taken when evaluating male patients with LOY.

**Keywords:** Haematological diseases, Loss of the Y chromosome

## 6.P64

### A new case of UBE2A gene deletion revealed by aCGH in a patient with MCA/MR and untreatable myelodysplastic syndrome

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