Isolated del 20q defines a subgroup of MDS patients with lower blast counts and more frequent thrombocytopenia

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Abstract

Background: isolated del 20q is common in MDS, and considered of good prognosis, but large series of MDS with isolated del 20q have been reported. In addition, it has been suggested that MDS with isolated del 20q may be associated with only mild dysplastic features, sometimes causing difficulties to distinguish them from other causes of thrombocytopenia. The aim of the study is to describe the clinical features and outcome of MDS patients with isolated del 20q.

Methods: a restricted analysis to patients with isolated del 20q diagnosed between 1990 and 2008 in 5 GFM centers were studied. Eighty-six of these patients were enrolled in the GFM registry. Results: Median age of the 64 MDS patients with isolated del20q was 72 (48-92), with 41 males and 23 females. They included 34 RA, 8 RAEB, 10 RAEB-T, 10 CMML and 15 RARS. Fifty-two percent had < 5% BM blasts in the two cohorts. Characteristics were compared between isolated del 20q and non del 20q. Platelet count (109/l) 3.1 3.2 0.9

Results

Comparison of isolated del 20q patients with non del 20q patients of the GFM registry

Clinical features of MDS patients with isolated del20q (n=64) were compared to the GFM registry containing 1388 karyotyped non del20q patients (Table 1). Analysis revealed that 33 (52%) of isolated del20q patients had less than 5% of marrow blasts, compared to 38% in the non del 20q patients included in the GFM registry (p=0.08). The median platelet count was significantly higher in patients with isolated del 20q patients (mean 212 G/l, p=0.014). Reticulocyte count (mean 84 vs 52 G/l, p=0.012) was significantly higher than in non del 20q patients. Significant findings were also observed for the white blood cell (WBC) count (mean 6.7 vs 5.6, p=0.004) and significantly higher reticulocyte count (mean 84 vs 52 G/l, p=0.014) for del20q and no significant differences for other clinical parameters confirming the unselected analysis above (Table 2).

Table 1. Comparison of isolated del20q patients with the non del20q cohort of the GFM registry

<table>
<thead>
<tr>
<th>Del 20q</th>
<th>Non del 20q</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>n</td>
<td>64</td>
<td>1388</td>
</tr>
<tr>
<td>WBC (109/l)</td>
<td>5.4</td>
<td>5.8</td>
</tr>
<tr>
<td>ANC (109/l)</td>
<td>3.1</td>
<td>3.2</td>
</tr>
<tr>
<td>Hb (g/l)</td>
<td>10.6</td>
<td>10.1</td>
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<tr>
<td>Platelet (109/l)</td>
<td>156</td>
<td>212</td>
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<tr>
<td>Reticulocyte count</td>
<td>84</td>
<td>52</td>
</tr>
</tbody>
</table>

Morphologic analysis of bone marrow from patients with isolated del20q display dysplasia similar to other MDS subtypes

Cytochemical analysis of bone marrow from MDS patients with isolated del20q and significant thrombocytopenia was compared to patients with other MDS subtypes excluding patients with 5q syndrome. Bone marrow from these purely thrombocytopenic MDS patients displayed dysplastic features to different degrees and in some patients in all lineages. Similar morphologic of MDS was found predominately in the erythroid lineage in both del20q and non del20q patients.

Summary and Discussion

Our study demonstrates that the MDS with isolated del 20q displays some specific clinical features. Del20q is characterized by the absence of excess of blasts in >5% of the cases and by lower platelet counts than other MDS. About 25% of them have isolated thrombocytopenia confirming in a large cohort earlier studies of MDS patients with isolated thrombocytopenia mimicking immune Thrombocytopenic Purpura (ITP) at clinical presentation. Morphologic analysis of bone marrow aspirates in the cohort showed the significant dysplasia in bone marrow is characteristic for ITP. Our study may help to distinguish MDS from ITP patients. Our study suggests that del20q constitutes a specific category of MDS similar to isolated del20q.

The authors have no relevant conflicts of interest

References