**Table 1:** Main clinical and hematological features of JMML patients with RNAseq analysis in total and by gene expression group

|  |  |  |  |
| --- | --- | --- | --- |
|  | **JMML\_F** | **JMML\_O** | **Total** |
| **n** | 6 | 8 | 16 |
| **Sex Ratio (M/F)** | 5 | 4 | 12/4 |
| **Age at onset (years), median (min-max)** | 1.2 (0.2-3.3) | 3.7 (0,4-8) | 1.2 (0.1-8) |
| **Peripheral blood, median (min-max)** |  |  |  |
| **WBC count, x109/L** | 18.8 (7.1-102.0) | 35.9 (13.0-59.4) | 35.9 (7.1-102.0) |
| **Monocyte count, x109/L** | 3.3 (1.7-35.7) | 6.2 (1.2-9.6) | 5.9 (1.2-35.7) |
| **Hb, g/dl** | 8.3 (7.2-11.3) | 10.1 (8.2-12.2) | 9.7 (7.2-12.2) |
| **Platelets count, x109/L** | 83.5 (15-396) | 54 (24-122) | 63 (15-396) |
| **Patients with myeloid precursors in PB** | 5/6 | 7/8 | 14/16 |
| **Patients with blasts in PB** | 4/6 | 7/7 | 11/15 |
| **Circulating blasts in PB (%)** | 1 (0-7.5) | 2 (1-10) | 1 (0-7.5) |
| **Bone marrow** |  |  |  |
| **Blasts (%)** | 6.5 (2-22) | 8 (3-38) | 5.2 (2-38) |
| **Dysplastic features** | 3/5 | 7/7 | 11/13 |
| **Karyotype** |  |  |  |
| **Normal** | 3/6 | 7/7 | 12/15 |
| **Monosomy 7** | 2/6 | 0/7 | 2/15 |
| **Other clonal alteration** | 1/6 | 0/7 | 1/15 |
| **RAS pathway mutation** |   |  |  |
| ***NRAS*** | 3/6 | 2/8 | 6/16 |
| ***KRAS*** | 2/6 | 0/8 | 3/16 |
| ***PTPN11*** | 0/6 | 5/8 | 5/16 |
| **Other**  | 1/7 | 1/8 | 2/16 |
| **Additional alterations** |   |  |  |
| ***ASXL1*** | 1/6 | 1/8 | 2/16 |
| ***SETBP1*** | 0/6 | 0/8 | 0/16 |
| ***JAK3*** | 0/6 | 1/8 | 1/16 |
| **RAS double mutant** | 0/6 | 2/8 | 2/16 |
| **≥ 2 alterations (SNV, CNV)** | 3/6 | 4/8 | 7/16 |
| **Outcome, n (%)** |   |  |  |
| **HSCT** | 3/6 | 7/8 | 11/16 |
| **Relapse after HSCT** | 1/3 | 0/7 | 1/16 |
| **Alive at last follow up** | 5/6 | 8/8 | 15/16 |

M: male ; F: female ; WBC : white blood cell ; Hb: hemoglobin ; HSCT: hematopoietic stem cell transplant ; LFU: last follow-up ; PB: peripheral blood ; SNV: single nucleotide variation ; CNV: copy number variation