

# Genetic instability in HSPC subpopulations of umbilical cord blood from patients with acute lymphoblastic leukemia

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**Supplemental table 1: Percentage of TEL and AML1 rearrangements analyzed using DNA FISH.** Data show percentages of TEL/AML1 translocations, TEL and AML1 duplications, duplications of TEL segment, duplications of AML1 segment, deletions of TEL or AML1, rearrangements of TEL, rearrangements of AML1, and total **rearrangements** in each analyzed UCB HSPC subpopulation from patients and control subjects.

| Sample    | TEL/AML1 | Subpopulation | TEL/AML1 trans. (%) | TEL/AML1 dupl. (%) | Gain of TEL (%) | Gain of AML1 (%) | Deletion TEL or AML1 (%) | Rearrangement of TEL (%) | Rearrangement of AML1 (%) | TOTAL (%) |
|-----------|----------|---------------|---------------------|--------------------|-----------------|------------------|--------------------------|--------------------------|---------------------------|-----------|
| Patient 1 | positive | MNC           | 0                   | 0                  | 0               | 0,3              | 0                        | 0                        | 0                         | 0,3       |
|           |          | Sp1           | 0                   | 0                  | 0               | 0                | 0                        | 0                        | 0                         | 0         |
|           |          | Sp2           | 0                   | 0                  | 0               | 0                | 0                        | 0                        | 0                         | 0         |
|           |          | Sp3           | 0                   | 0                  | 0,3             | 0,3              | 0                        | 0                        | 0                         | 0,6       |
|           |          | Sp4           | 0                   | 0                  | 0,4             | 0,4              | 0                        | 0                        | 0                         | 0,8       |
|           |          | Sp5           | 1,5                 | 0                  | 1,25            | 1,75             | 1,5                      | 0                        | 0                         | 6         |
|           |          | Sp6           | 0                   | 0                  | 0               | 0,25             | 0                        | 0                        | 0                         | 0,25      |
|           |          | Sp7           | 0                   | 0                  | 0               | 0                | 0                        | 0                        | 0                         | 0         |
|           |          | Sp8           | 0                   | 0                  | 0               | 0                | 0                        | 0                        | 0                         | 0         |
| Patient 2 | positive | MNC           | 0                   | 0                  | 0,25            | 0                | 0                        | 0,5                      | 0                         | 0,75      |

|           |          |     |      |      |       |       |   |   |     |       |
|-----------|----------|-----|------|------|-------|-------|---|---|-----|-------|
|           |          | Sp5 | 0    | 0    | 0,2   | 0,1   | 0 | 0 | 0   | 0,3   |
|           |          | Sp6 | 0,1  | 0    | 0,2   | 0,2   | 0 | 0 | 0   | 0,5   |
|           |          | Sp7 | 0    | 0,2  | 0     | 0,2   | 0 | 0 | 0   | 0,4   |
|           |          | Sp8 | 0    | 0    | 0,4   | 0,4   | 0 | 0 | 0   | 0,8   |
| Patient 3 | negative | MNC | 0,25 | 0    | 0     | 0     | 0 | 0 | 0   | 0,25  |
|           |          | Sp1 | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |
|           |          | Sp2 | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |
|           |          | Sp5 | 0    | 0,25 | 0,5   | 0     | 0 | 0 | 0   | 0,75  |
|           |          | Sp6 | 0    | 0,3  | 0,3   | 0,3   | 0 | 0 | 0,3 | 1,2   |
|           |          |     |      |      |       |       |   |   |     |       |
| Patient 4 | negative | MNC | 0    | 0    | 0,2   | 0,4   | 0 | 0 | 0   | 0,6   |
|           |          | Sp1 | 0    | 0    | 0,167 | 0     | 0 | 0 | 0   | 0,167 |
|           |          | Sp2 | 0    | 0    | 0,25  | 0     | 0 | 0 | 0   | 0,25  |
|           |          | Sp3 | 0,2  | 0    | 0     | 0,6   | 0 | 0 | 0   | 0,8   |
|           |          | Sp4 | 0,2  | 0    | 1     | 1     | 0 | 0 | 0   | 2,2   |
|           |          | Sp5 | 0    | 0    | 0,6   | 0     | 0 | 0 | 0   | 0,6   |
|           |          | Sp6 | 0    | 0    | 0     | 0,667 | 0 | 0 | 0   | 0,667 |
|           |          | Sp7 | 0,4  | 0    | 0,6   | 1,2   | 0 | 0 | 0   | 2,2   |
|           |          | Sp8 | 0,2  | 0    | 0,2   | 0     | 0 | 0 | 0   | 0,4   |
| Control 1 | negative | Sp1 | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |
|           |          | Sp2 | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |
|           |          | Sp3 | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |
|           |          | Sp5 | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |
|           |          | Sp6 | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |
|           |          | Sp7 | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |
|           |          | Sp8 | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |
| Control 2 | negative | Sp1 | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |
|           |          | Sp2 | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |
|           |          | Sp5 | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |
|           |          | Sp6 | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |
|           |          | Sp7 | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |
|           |          | Sp8 | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |
| Control 3 | negative | MNC | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |
|           |          | Sp1 | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |
|           |          | Sp5 | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |
|           |          | Sp6 | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |
| Control 4 | negative | MNC | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |
|           |          | Sp1 | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |
|           |          | Sp2 | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |
|           |          | Sp5 | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |
|           |          | Sp6 | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |
|           |          | Sp7 | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |
|           |          | Sp8 | 0    | 0    | 0     | 0     | 0 | 0 | 0   | 0     |

**Supplemental table 2: RT qPCR analysis.** Table shows concentration of isolated total RNA, amount of RNA per cell, results of RT-qPCR and standard PCR for patients (P1-4) and healthy controls (K4-7) and finally the validation of PFG positivity performed by sequencing of qPCR product subcloned into sequencing vector.

|                 | RNA              | RNA       | TEL-AML1 |                 |            |
|-----------------|------------------|-----------|----------|-----------------|------------|
|                 | [ng/μl]          | [pg/cell] | RT-qPCR  | standard<br>PCR | sequencing |
| <b>Patient1</b> |                  |           |          |                 |            |
| MNC             | 82.7             | 0.92      | -        | -               | -          |
| #1              | 9.4 <sup>A</sup> | 0.75      | 0/3      | -               | -          |
| #2              | 3.4 <sup>A</sup> | 0.09      | 3/3      | 3/3             | 2/3        |
| #3              | 5.2 <sup>A</sup> | 0.78      | 0/3      | -               | -          |
| #4 <sub>F</sub> | 151.3            | 4.5       | 0/3      | -               | -          |

|                 |       |      |     |   |   |
|-----------------|-------|------|-----|---|---|
| #4              | 100.0 | 3.46 | 0/3 | - | - |
| #5 <sub>F</sub> | 170.7 | 6.16 | 0/3 | - | - |
| #5              | 121.6 | 5.86 | 0/3 | - | - |
| #6              | 136.8 | 3.83 | 0/3 | - | - |
| #7              | 122.1 | 3.7  | 0/3 | - | - |
| #8              | 163.9 | 4.41 | 0/3 | - | - |

## P2

|     |                     |      |     |     |   |
|-----|---------------------|------|-----|-----|---|
| MNC | 85.7                | 1.29 | -   | -   | - |
| #2  | 1.6 <sup>A</sup>    | 0.05 | 0/3 | -   | - |
| #5  | 351.6               | 5.5  | 0/3 | -   | - |
| #6  | 240.9               | 3.93 | 0/3 | -   | - |
| #7  | 8.6 <sup>lost</sup> | 0.99 | -   | -   | - |
| #8  | 2.7 <sup>A</sup>    | 0.57 | 1/3 | 0/1 | - |

## P3

|     |                  |      |     |     |     |
|-----|------------------|------|-----|-----|-----|
| MNC | 12.5             | 0.47 | 0/3 | -   | -   |
| #5  | 97.5             | 3.7  | 2/3 | 2/2 | 2/2 |
| #6  | 9.5 <sup>A</sup> | 1.35 | 0/3 | -   | -   |

## P4

|     |                   |      |     |     |     |
|-----|-------------------|------|-----|-----|-----|
| MNC | 274.1             | 3.04 | 0/3 | -   | -   |
| #2  | 12.2 <sup>A</sup> | 0.45 | 1/3 | 1/1 | 1/1 |
| #3  | 109.8             | 2.4  | 1/3 | 1/1 | 1/1 |
| #4  | 45.5              | 3.5  | 1/3 | 1/1 | 0/1 |
| #5  | 159.7             | 4.2  | 0/3 | -   | -   |
| #6  | 208.4             | 8.6  | 0/3 | -   | -   |
| #7  | 149.9             | 9.7  | 0/3 | 2/3 | 2/2 |
| #8  | 138.6             | 7.2  | 0/3 | 1/3 | 1/1 |

| Control 2        | RNA               | RNA       | TEL-AML1 |       |            | MLL <sub>2</sub> -AF4 |       |            | Bcr-Abl p190 |       |            |
|------------------|-------------------|-----------|----------|-------|------------|-----------------------|-------|------------|--------------|-------|------------|
|                  | [ng/μl]           | [pg/cell] | qPCR     | stPCR | sequencing | qPCR                  | stPCR | sequencing | qPCR         | stPCR | sequencing |
| MNC              | 142.7             | 4.2       | 0/3      | -     | -          | 1/3                   | 0/1   | -          | 0/3          | -     | -          |
| #5               | 443.3             | 5.5       | 0/3      | -     | -          | -                     | -     | -          | -            | -     | -          |
| #6               | 259.7             | 5.9       | 0/3      | -     | -          | -                     | -     | -          | -            | -     | -          |
| #7               | 80.4              | 5.2       | 0/3      | -     | -          | -                     | -     | -          | -            | -     | -          |
| #8               | 85.0              | 4.6       | 0/3      | -     | -          | -                     | -     | -          | -            | -     | -          |
| <b>Control 3</b> |                   |           |          |       |            |                       |       |            |              |       |            |
| MNC              | 144.6             | 1.44      | 0/3      | -     | -          | 0/3                   | -     | -          | 1/3          | 0/1   | -          |
| #5               | 237.1             | 5.0       | 0/3      | -     | -          | -                     | -     | -          | -            | -     | -          |
| #6               | 229.6             | 5.9       | 0/3      | -     | -          | -                     | -     | -          | -            | -     | -          |
| <b>Control 4</b> |                   |           |          |       |            |                       |       |            |              |       |            |
| MNC              | 277.7             | 2.77      | 0/3      | -     | -          | 0/3                   | -     | -          | 0/3          | -     | -          |
| #5               | 13.6 <sup>A</sup> | 1.10      | 0/3      | -     | -          | -                     | -     | -          | -            | -     | -          |

|                   |                   |      |     |   |   |   |   |   |   |   |   |
|-------------------|-------------------|------|-----|---|---|---|---|---|---|---|---|
| #5 <sup>lys</sup> | 8.7 <sup>A</sup>  | 1.00 | 0/3 | - | - | - | - | - | - | - | - |
| #6                | 14.4 <sup>A</sup> | 0.99 | 0/3 | - | - | - | - | - | - | - | - |
| #6 <sup>lys</sup> | 27.1 <sup>A</sup> | 0.57 | 0/3 | - | - | - | - | - | - | - | - |

#### Control 5

|     |                    |      |     |     |     |     |   |   |     |   |   |
|-----|--------------------|------|-----|-----|-----|-----|---|---|-----|---|---|
| MNC | 85.8               | 0.86 | 0/3 | -   | -   | 0/3 | - | - | 0/3 | - | - |
| #5  | 34.4 <sup>A</sup>  | 1.3  | 3/3 | 0/3 | -   | -   | - | - | -   | - | - |
| #6  | 18.3 <sup>A</sup>  | 1.1  | 1/3 | 1/1 | 0/1 | -   | - | - | -   | - | - |
| #7  | 141.3              | 3.4  | 0/3 | -   | -   | -   | - | - | -   | - | - |
| #7  | 141.3 <sup>A</sup> |      | 3/3 | 1/3 | 0/1 | -   | - | - | -   | - | - |
| #7  | 247.6              | 3.2  | 0/3 | -   | -   | 0/3 | - | - | 0/3 | - | - |
| #7  | 247.6 <sup>A</sup> |      | 0/3 | -   | -   | -   | - | - | -   | - | - |

<sup>A</sup> cDNA was amplified (usually due to low RNA concentration); <sup>lys</sup> lysed erythrocytes in the sample; stPCR, standard PCR - used for amplification of qPCR product (for subcloning into a sequencing vector).

**Supplemental table 3: Percentage of BCR and ABL rearrangements analyzed using DNA FISH.** Table shows the percentages of BCR/ABL translocations, BCR and ABL duplications, duplications of BCR segment, duplications of ABL segment, deletions of BCR or ABL, rearrangements of BCR, rearrangements of ABL and total rearrangements in each analyzed UCB MNC subpopulation for patients and control subjects.

| Sample    | BCR/ABL  | Subpopulation | BCR/ABL trans. (%) | BCR/ABL dupl. (%) | Gain of BCR (%) | Gain of ABL (%) | Deletion BCR or ABL (%) | Rearrangement of BCR (%) | Rearrangement of ABL (%) | TOTAL (%) |
|-----------|----------|---------------|--------------------|-------------------|-----------------|-----------------|-------------------------|--------------------------|--------------------------|-----------|
| Patient 1 | negative | MNC           | 0                  | 0                 | 0               | 0               | 0                       | 0                        | 0                        | 0         |
|           |          | Sp2           | 1                  | 0                 | 1,5             | 0               | 0                       | 0                        | 0                        | 2,5       |
|           |          | Sp3           | 0                  | 0                 | 0               | 0               | 0                       | 0                        | 0                        | 0         |
|           |          | Sp4           | 0                  | 0                 | 0               | 0               | 0                       | 0                        | 0                        | 0         |
|           |          | Sp5           | 0                  | 0                 | 0               | 0,4             | 0                       | 0                        | 0                        | 0,4       |
|           |          | Sp6           | 0                  | 0                 | 0               | 0               | 0                       | 0                        | 0                        | 0         |
|           |          | Sp7           | 0                  | 0                 | 0               | 0               | 0                       | 0                        | 0                        | 0         |
|           |          | Sp8           | 0,4                | 0                 | 0,4             | 0,8             | 0                       | 0                        | 0                        | 1,6       |
| Patient 2 | negative | Sp5           | 0                  | 0                 | 0,3             | 0,3             | 0                       | 0                        | 0                        | 0,6       |
|           |          | Sp6           | 0                  | 0                 | 0               | 0               | 0                       | 0                        | 0                        | 0         |
|           |          | Sp7           | 0                  | 0                 | 0               | 0               | 0                       | 0                        | 0                        | 0         |
| Patient 3 | negative | MNC           | 0                  | 0                 | 0               | 0               | 0                       | 0                        | 0                        | 0         |
|           |          | Sp5           | 0                  | 0                 | 1               | 0               | 0                       | 0                        | 0                        | 1         |
|           |          | Sp6           | 0                  | 0                 | 0,667           | 0,667           | 0                       | 0                        | 0                        | 1,334     |
| Patient 4 | negative | Sp1           | 0,5                | 0                 | 1,25            | 1               | 0                       | 0                        | 0                        | 2,75      |
|           |          | Sp3           | 0,333              | 0                 | 0               | 0,667           | 0                       | 0                        | 0                        | 1         |
|           |          | Sp4           | 1,2                | 0                 | 1,8             | 1               | 0                       | 0                        | 0                        | 4         |
|           |          | Sp5           | 0                  | 0                 | 0,4             | 0,2             | 0                       | 0                        | 0                        | 0,6       |
|           |          | Sp6           | 0                  | 0                 | 6               | 0,6             | 0                       | 0                        | 0                        | 6,6       |
|           |          | Sp7           | 0                  | 0                 | 1,2             | 0,4             | 0                       | 0                        | 0                        | 1,6       |
|           |          | Sp8           | 0,4                | 0                 | 0,4             | 0,8             | 0                       | 0                        | 0                        | 1,6       |
|           |          | MNC           | 0                  | 0                 | 0               | 0               | 0                       | 0                        | 0                        | 0         |
| Control 3 | negative | Sp1           | 0                  | 0                 | 0               | 0               | 0                       | 0                        | 0                        | 0         |
|           |          | Sp2           | 0                  | 0                 | 0               | 0               | 0                       | 0                        | 0                        | 0         |
|           |          | Sp3           | 0                  | 0                 | 0               | 0               | 0                       | 0                        | 0                        | 0         |
| Control 5 | negative | Sp5           | 1,5                | 0                 | 0               | 0               | 0                       | 0                        | 0                        | 1,5       |
|           |          | Sp6           | 0                  | 0                 | 0               | 0               | 0                       | 0                        | 0                        | 0         |

|                  |                 |     |   |   |   |   |   |   |   |   |
|------------------|-----------------|-----|---|---|---|---|---|---|---|---|
|                  |                 | Sp7 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
|                  |                 | Sp8 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| <b>Control 6</b> | <b>negative</b> | Sp1 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
|                  |                 | Sp2 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
|                  |                 | Sp3 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
|                  |                 | Sp4 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
|                  |                 | Sp5 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
|                  |                 | Sp6 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
|                  |                 | Sp7 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
|                  |                 | Sp8 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| <b>Control 7</b> | <b>negative</b> | Sp3 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
|                  |                 | Sp5 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
|                  |                 | Sp6 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
|                  |                 | Sp7 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
|                  |                 | Sp8 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |

**Supplemental table 4: Percentage of MLL rearrangements analyzed using DNA FISH.** Table shows the percentages of MLL rearrangements, MLL duplications, gains of red signal, gains of green signal, deletions of MLL, and total rearrangements in each analyzed UCB HSPC subpopulation of patients and control subjects.

| Sample            | MLL             | Subpopulation | MLL rearrangement (%) | MLL dupl. (%) | Gain of red (%) | Gain of green (%) | Deletion (%) | TOTAL (%) |
|-------------------|-----------------|---------------|-----------------------|---------------|-----------------|-------------------|--------------|-----------|
| <b>Patient 1</b>  | <b>negative</b> | Sp1           | 0                     | 0             | 0               | 0                 | 0            | 0         |
|                   |                 | Sp3           | 0                     | 0             | 0               | 0                 | 0            | 0         |
|                   |                 | Sp5           | 0                     | 0             | 0,1             | 0,1               | 0            | 0,2       |
|                   |                 | Sp6           | 0                     | 0             | 0               | 0                 | 0            | 0         |
|                   |                 | Sp7           | 0                     | 0             | 0               | 0,2               | 0            | 0,2       |
|                   |                 | Sp8           | 0                     | 0,1           | 0               | 0                 | 0            | 0,1       |
| <b>Patient 2</b>  | <b>negative</b> | MNC           | 0                     | 0             | 0               | 0                 | 0            | 0         |
|                   |                 | Sp4           | 0                     | 0,333         | 0,333           | 0                 | 0            | 0,666     |
|                   |                 | Sp5           | 0                     | 0             | 0               | 0,4               | 0            | 0,4       |
|                   |                 | Sp6           | 0                     | 0             | 0               | 0                 | 0            | 0         |
|                   |                 | Sp7           | 0                     | 0             | 0               | 0                 | 0            | 0         |
| <b>Patient 3</b>  | <b>negative</b> | MNC           | 6,667                 | 0             | 0               | 3,333             | 3,333        | 13,333    |
|                   |                 | Sp5           | 1,5                   | 0             | 2,5             | 0                 | 1            | 5         |
|                   |                 | Sp6           | 4,167                 | 0             | 3,333           | 0                 | 0            | 4,167     |
| <b>Patient 4</b>  | <b>negative</b> | Sp3           | 0,333                 | 5,2           | 3,6             | 5,2               | 0            | 14,333    |
|                   |                 | Sp4           | 0                     | 0             | 0               | 1                 | 0            | 1         |
|                   |                 | Sp5           | 0,667                 | 0             | 3               | 3                 | 0            | 6,667     |
|                   |                 | Sp6           | 6,667                 | 0             | 10              | 14,667            | 0            | 31,334    |
|                   |                 | Sp7           | 0,6                   | 0             | 1,6             | 0,4               | 0            | 2,6       |
|                   |                 | Sp8           | 0,2                   | 0             | 2               | 3                 | 0            | 5,2       |
| <b>Control 8</b>  | <b>negative</b> | MNC           | 0                     | 0             | 0               | 0                 | 0            | 0         |
|                   |                 | Sp1           | 0                     | 0             | 0               | 0                 | 0            | 0         |
|                   |                 | Sp2           | 0                     | 0             | 0               | 0                 | 0            | 0         |
|                   |                 | Sp5           | 0                     | 0             | 0               | 0                 | 0            | 0         |
|                   |                 | Sp6           | 0,125                 | 0             | 0               | 0                 | 0            | 0,125     |
|                   |                 | Sp7           | 0                     | 0             | 0               | 0                 | 0            | 0         |
| <b>Control 9</b>  | <b>negative</b> | Sp8           | 0                     | 0             | 0               | 0                 | 0            | 0         |
|                   |                 | MNC           | 0                     | 0             | 0               | 0                 | 0            | 0         |
|                   |                 | Sp1           | 0                     | 0             | 0               | 0                 | 0            | 0         |
|                   |                 | Sp2           | 0                     | 0             | 0               | 0                 | 0            | 0         |
|                   |                 | Sp3           | 0                     | 0             | 0               | 0                 | 0            | 0         |
|                   |                 | Sp5           | 0                     | 0             | 0               | 0                 | 0            | 0         |
|                   |                 | Sp6           | 0                     | 0             | 0               | 0                 | 0            | 0         |
| <b>Control 10</b> | <b>negative</b> | Sp7           | 0                     | 0             | 0               | 0                 | 0            | 0         |
|                   |                 | MNC           | 0                     | 0             | 0               | 0                 | 0            | 0         |
|                   |                 | Sp1           | 0                     | 0             | 0               | 0                 | 0            | 0         |
|                   |                 | Sp2           | 0                     | 0             | 0               | 0                 | 0            | 0         |
|                   |                 | Sp3           | 0                     | 0             | 0               | 0                 | 0            | 0         |
|                   |                 | Sp5           | 0                     | 0             | 0               | 0                 | 0            | 0         |
|                   |                 | Sp6           | 0                     | 0             | 0               | 0                 | 0            | 0         |
|                   |                 | Sp7           | 0                     | 0             | 0               | 0                 | 0            | 0         |

|  |  |     |   |   |   |   |   |   |
|--|--|-----|---|---|---|---|---|---|
|  |  | Sp8 | 0 | 0 | 0 | 0 | 0 | 0 |
|--|--|-----|---|---|---|---|---|---|

### Supplemental table 5: Change in the number of TEL and AML1 aberrant cells before and after expansion.

Data show the results for all aberrant subpopulations for each patient. Table shows the time of expansion, derived doubling time, number of cells before and after the expansion, number of positive cells, derived total positive cells in expanded samples and in the samples before expansion.

| PATIENT SAMPLE INFO |              |                                                           | SORTING AND EXPANSION            |                                  |                       |                   |                       | PFG ANALYSIS FISH                       |                                    |                                              |                                            |                                            | ABERRATIONS ANALYSIS FISH                   |       |       |  |  |
|---------------------|--------------|-----------------------------------------------------------|----------------------------------|----------------------------------|-----------------------|-------------------|-----------------------|-----------------------------------------|------------------------------------|----------------------------------------------|--------------------------------------------|--------------------------------------------|---------------------------------------------|-------|-------|--|--|
| Patient             | Sorted subp. | Number of cells after overnight incubation and filtration | Number of cells before expansion | Number of cells after expansion) | Time of expansion (h) | Doubling time (h) | Number of cell cycles | Relative positive cells after expansion | PFG positive cells after expansion | Absolute PFG positive cells before expansion | Relative aberrant positive after expansion | Absolute aberrant positive after expansion | Absolute aberrant positive before expansion | cells | cells |  |  |
| Patient 1           | 3            | 57,5 x 10 <sup>6</sup>                                    | 1218                             | 400000                           | 312                   | 37,32             | 8,36                  | 0                                       | 0                                  | 0                                            | 0,006                                      | 2400                                       | 7,31                                        |       |       |  |  |
|                     | 4            | 57,5 x 10 <sup>6</sup>                                    | 8300                             | 1500000                          | 312                   | 41,61             | 7,50                  | 0                                       | 0                                  | 0                                            | 0,008                                      | 12000                                      | 66,4                                        |       |       |  |  |
|                     | 5            | 57,5 x 10 <sup>6</sup>                                    | 11697                            | 1330000                          | 312                   | 45,69             | 6,83                  | 0,015                                   | 19950                              | 175,46                                       | 0,06                                       | 79800                                      | 701,82                                      |       |       |  |  |
|                     | 6            | 57,5 x 10 <sup>6</sup>                                    | 2225                             | 1870000                          | 312                   | 32,12             | 9,72                  | 0                                       | 0                                  | 0                                            | 0,0025                                     | 4675                                       | 5,56                                        |       |       |  |  |
| Patient 2           | 5            | 20 x 10 <sup>6</sup>                                      | 5600                             | 240000                           | 288                   | 53,12             | 5,42                  | 0                                       | 0                                  | 0                                            | 0,003                                      | 720                                        | 16,8                                        |       |       |  |  |
|                     | 6            | 20 x 10 <sup>6</sup>                                      | 1500                             | 2320000                          | 288                   | 27,18             | 10,59                 | 0,001                                   | 2320                               | 1,5                                          | 0,005                                      | 11600                                      | 7,5                                         |       |       |  |  |
|                     | 7            | 20 x 10 <sup>6</sup>                                      | 1794                             | 460000                           | 288                   | 35,99             | 8,00                  | 0                                       | 0                                  | 0                                            | 0,004                                      | 1840                                       | 7,18                                        |       |       |  |  |
|                     | 8            | 20 x 10 <sup>6</sup>                                      | 791                              | 240000                           | 288                   | 34,93             | 8,25                  | 0                                       | 0                                  | 0                                            | 0,008                                      | 1920                                       | 6,33                                        |       |       |  |  |
| Patient 3           | 5            | 5 x 10 <sup>6</sup>                                       | 1647                             | 990000                           | 312                   | 33,80             | 9,23                  | 0                                       | 0                                  | 0                                            | 0,0075                                     | 7425                                       | 12,35                                       |       |       |  |  |
|                     | 6            | 5 x 10 <sup>6</sup>                                       | 476                              | 360000                           | 312                   | 32,63             | 9,56                  | 0                                       | 0                                  | 0                                            | 0,012                                      | 4320                                       | 5,71                                        |       |       |  |  |
| Patient 4           | 3            | 45 x 10 <sup>6</sup>                                      | 10310                            | 1580000                          | 312                   | 42,98             | 7,26                  | 0,002                                   | 3160                               | 20,62                                        | 0,008                                      | 12640                                      | 82,48                                       |       |       |  |  |
|                     | 4            | 45 x 10 <sup>6</sup>                                      | 1159                             | 580000                           | 312                   | 34,79             | 8,97                  | 0,002                                   | 1160                               | 2,32                                         | 0,022                                      | 12760                                      | 25,50                                       |       |       |  |  |
|                     | 5            | 45 x 10 <sup>6</sup>                                      | 10000                            | 1430000                          | 312                   | 43,576202         | 7,1598713             | 0                                       | 0                                  | 0                                            | 0,006                                      | 8580                                       | 60                                          |       |       |  |  |
|                     | 6            | 45 x 10 <sup>6</sup>                                      | 4528                             | 920000                           | 312                   | 40,695921         | 7,6666161             | 0                                       | 0                                  | 0                                            | 0,00667                                    | 6136,4                                     | 30,20176                                    |       |       |  |  |
|                     | 7            | 45 x 10 <sup>6</sup>                                      | 2092                             | 660000                           | 312                   | 37,583844         | 8,3014394             | 0,004                                   | 2640                               | 8,368                                        | 0,022                                      | 14520                                      | 46,024                                      |       |       |  |  |
|                     | 8            | 45 x 10 <sup>6</sup>                                      | 4035                             | 770000                           | 312                   | 41,181889         | 7,576146              | 0,002                                   | 1540                               | 8,07                                         | 0,004                                      | 3080                                       | 16,14                                       |       |       |  |  |

### Supplemental table 6: Change in the number of BCR and ABL aberrantcells before and after expansion.

Data show the results for all aberrant subpopulations for each patient. Table shows the time of expansion, derived doubling time, number of cells before and after the expansion, relative positive cells, derived total positive cells in expanded samples and in the samples before expansion.

| PATIENT SAMPLE INFO |              |                                                           | SORTING AND EXPANSION            |                                 |                       |                   |                  |                                         | PFG ANALYSIS FISH                  |                                          |                                    |                                                  | ABERRATIONS ANALYSIS FISH                        |                                                   |  |  |
|---------------------|--------------|-----------------------------------------------------------|----------------------------------|---------------------------------|-----------------------|-------------------|------------------|-----------------------------------------|------------------------------------|------------------------------------------|------------------------------------|--------------------------------------------------|--------------------------------------------------|---------------------------------------------------|--|--|
| Patient             | Sorted subp. | Number of cells after overnight incubation and filtration | Number of cells before expansion | Number of cells after expansion | Time of expansion (h) | Doubling time (h) | Number of cycles | Relative positive cells after expansion | PFG positive cells after expansion | Absolute positive cells before expansion | PFG positive cells after expansion | Relative aberrant positive cells after expansion | Absolute aberrant positive cells after expansion | Absolute aberrant positive cells before expansion |  |  |
| Patient 1           | 5            | 57,5 x 10 <sup>6</sup>                                    | 11697                            | 1330000                         | 312                   | 45,69             | 6,83             | 0                                       | 0                                  | 0                                        | 0,004                              | 5320                                             | 46,79                                            |                                                   |  |  |
|                     | 8            | 57,5 x 10 <sup>6</sup>                                    | 3266                             | 1980000                         | 312                   | 33,75             | 9,24             | 0,004                                   | 7920                               | 13,06                                    | 0,016                              | 31680                                            | 52,26                                            |                                                   |  |  |

|           |   |                      |        |         |     |       |      |         |        |       |         |        |        |
|-----------|---|----------------------|--------|---------|-----|-------|------|---------|--------|-------|---------|--------|--------|
| Patient 2 | 5 | 20 x 10 <sup>6</sup> | 5600   | 240000  | 288 | 53,12 | 5,42 | 0       | 0      | 0     | 0,006   | 1440   | 33,6   |
|           | 5 | 5 x 10 <sup>6</sup>  | 1647   | 990 000 | 312 | 33,80 | 9,23 | 0       | 0      | 0     | 0,01    | 9900   | 16,47  |
| Patient 3 | 6 | 5 x 10 <sup>6</sup>  | 476    | 360 000 | 312 | 32,63 | 9,56 | 0       | 0      | 0     | 0,01334 | 4802,4 | 6,35   |
|           | 3 | 45 x 10 <sup>6</sup> | 10310  | 1580000 | 312 | 42,98 | 7,26 | 0,00333 | 5261,4 | 34,33 | 0,01    | 15800  | 103,1  |
| Patient 4 | 4 | 45 x 10 <sup>6</sup> | 1159   | 580000  | 312 | 34,79 | 8,97 | 0,012   | 6960   | 13,91 | 0,04    | 23200  | 46,36  |
|           | 5 | 45 x 10 <sup>6</sup> | 10 000 | 1430000 | 312 | 43,58 | 7,16 | 0       | 0      | 0     | 0,006   | 8580   | 60     |
|           | 6 | 45 x 10 <sup>6</sup> | 4528   | 920 000 | 312 | 40,70 | 7,67 | 0       | 0      | 0     | 0,066   | 60720  | 298,85 |
|           | 7 | 45 x 10 <sup>6</sup> | 2092   | 660 000 | 312 | 37,58 | 8,30 | 0       | 0      | 0     | 0,016   | 10560  | 33,47  |
|           | 8 | 45 x 10 <sup>6</sup> | 4035   | 770 000 | 312 | 41,18 | 7,58 | 0,004   | 3080   | 16,14 | 0,016   | 12320  | 64,56  |

**Supplemental table 7: Change in the number of cells aberrant for MLL rearrangements before and after expansion.** Data show the results for all the aberrant subpopulations for each patient. Table shows the time of expansion, derived doubling time, number of cells before and after the expansion, relative positive cells, derived total positive cells in expanded samples and in the samples before expansion.

| PATIENT SAMPLE INFO |              |                                                           | SORTING AND EXPANSION            |                                 |                       |                   |                  | PFG ANALYSIS FISH                           |                                             |                                              | ABERRATIONS ANALYSIS FISH                        |                                                  |                                                   |
|---------------------|--------------|-----------------------------------------------------------|----------------------------------|---------------------------------|-----------------------|-------------------|------------------|---------------------------------------------|---------------------------------------------|----------------------------------------------|--------------------------------------------------|--------------------------------------------------|---------------------------------------------------|
| Patient             | Sorted subp. | Number of cells after overnight incubation and filtration | Number of cells before expansion | Number of cells after expansion | Time of expansion (h) | Doubling time (h) | Number of cycles | Relative PFG positive cells after expansion | Absolute PFG positive cells after expansion | Absolute PFG positive cells before expansion | Relative aberrant positive cells after expansion | Absolute aberrant positive cells after expansion | Absolute aberrant positive cells before expansion |
| Patient 1           | 5            | 57,5 x 10 <sup>6</sup>                                    | 11697                            | 1330000                         | 312                   | 45,69             | 6,83             | 0                                           | 0                                           | 0                                            | 0,002                                            | 2660                                             | 23,39                                             |
|                     | 7            | 57,5 x 10 <sup>6</sup>                                    | 804                              | 1 720 000                       | 312                   | 28,20             | 11,06            | 0                                           | 0                                           | 0                                            | 0,002                                            | 3440                                             | 1,61                                              |
|                     | 8            | 57,5 x 10 <sup>6</sup>                                    | 3266                             | 1 980 000                       | 312                   | 33,75             | 9,24             | 0                                           | 0                                           | 0                                            | 0,001                                            | 1980                                             | 3,27                                              |
| Patient 2           | 4            | 20 x 10 <sup>6</sup>                                      | 5600                             | 240 000                         | 288                   | 53,12             | 5,42             | 0                                           | 0                                           | 0                                            | 0,007                                            | 1680                                             | 39,20                                             |
|                     | 5            | 20 x 10 <sup>6</sup>                                      | 1500                             | 2 320 000                       | 288                   | 27,18             | 10,59            | 0                                           | 0                                           | 0                                            | 0,004                                            | 9280                                             | 6                                                 |
| Patient 3           | 5            | 5 x 10 <sup>6</sup>                                       | 1647                             | 990 000                         | 312                   | 33,80             | 9,23             | 0,02                                        | 14850                                       | 24,71                                        | 0,05                                             | 49500                                            | 82,35                                             |
|                     | 6            | 5 x 10 <sup>6</sup>                                       | 476                              | 360 000                         | 312                   | 32,63             | 9,56             | 0,04                                        | 15001,2                                     | 19,83                                        | 0,04                                             | 15001,2                                          | 19,83                                             |
| Patient 4           | 3            | 45 x 10 <sup>6</sup>                                      | 10310                            | 1580000                         | 312                   | 42,98             | 7,26             | 0,01                                        | 5261,4                                      | 34,33                                        | 0,14                                             | 226461,4                                         | 1477,73                                           |
|                     | 4            | 45 x 10 <sup>6</sup>                                      | 1159                             | 580000                          | 312                   | 34,79             | 8,97             | 0                                           | 0                                           | 0                                            | 0,01                                             | 5800                                             | 11,59                                             |
|                     | 5            | 45 x 10 <sup>6</sup>                                      | 10 000                           | 1430000                         | 312                   | 43,58             | 7,16             | 0,01                                        | 9538,1                                      | 66,7                                         | 0,07                                             | 95338,1                                          | 666,7                                             |
|                     | 6            | 45 x 10 <sup>6</sup>                                      | 4528                             | 920 000                         | 312                   | 40,70             | 7,67             | 0,07                                        | 61364                                       | 302,02                                       | 0,31                                             | 288272,8                                         | 1418,80                                           |
|                     | 7            | 45 x 10 <sup>6</sup>                                      | 2092                             | 660 000                         | 312                   | 37,58             | 8,30             | 0,01                                        | 3960                                        | 12,55                                        | 0,03                                             | 17160                                            | 54,39                                             |
|                     | 8            | 45 x 10 <sup>6</sup>                                      | 4035                             | 770 000                         | 312                   | 41,18             | 7,58             | 0,01                                        | 1540                                        | 8,07                                         | 0,05                                             | 40040                                            | 209,82                                            |