

## **BCOR and BCORL1 mutations in pediatric acute myeloid leukemia**

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## Supplemental data

**Supplementary table 1a. Overview of *BCOR* primers**

PCR cycle: 10'95°C, 40x(15"95°C, 1'60°C, 30"72°C), 10'72°C

| Exon                 | Direction | Primer                        |
|----------------------|-----------|-------------------------------|
| 2                    | Forward   | 5'-GGCTCCCAGACCACTGT-3'       |
|                      | Reverse   | 5'-GCGGGAAAGCTCTTCTCT-3'      |
| 3                    | Forward   | 5'-GCGGAGGGTTAAGGACA-3'       |
|                      | Reverse   | 5'-GGGGCCTTGTCCCTCTC-3'       |
| 4.1                  | Forward   | 5'-GCCGAAGCCTGTCTT-3'         |
|                      | Reverse   | 5'-GGCAGCCGCAGATAAC-3'        |
| 4.2                  | Forward   | 5'-GCCAGCGACAAACAGAG-3'       |
|                      | Reverse   | 5'-GGGGGCAACAGGAGA-3'         |
| 4.3                  | Forward   | 5'-CCACGCCTATCCTCAC-3'        |
|                      | Reverse   | 5'-AGCTTGAAAGCATCTACATC-3'    |
| 4.4                  | Forward   | 5'-AGCGGTTCAAGACAGAAAA-3'     |
|                      | Reverse   | 5'-GCCCACGTGCTGAATAA-3'       |
| 4.5                  | Forward   | 5'-CACCGATGCTGTCATCAC-3'      |
|                      | Reverse   | 5'-GTGCCAGGAAACAGACT-3'       |
| 4.6                  | Forward   | 5'-GAGGGCATTGCTGTAAGTC-3'     |
|                      | Reverse   | 5'-TCGCTTTGACAACAGTCTT-3'     |
| 4.7                  | Forward   | 5'-TCCCCACCGACAAGAA-3'        |
|                      | Reverse   | 5'-GGGGGTACATCCACA-3'         |
| 4.8                  | Forward   | 5'-TGGCCCTGCTGTAACTT-3'       |
|                      | Reverse   | 5'-CCCCAATCCTGTTACACA-3'      |
| 5                    | Forward   | 5'-AAAGGTTGCTTAAAGGGATAGA-3'  |
|                      | Reverse   | 5'-ACCATGGCCCACAAACT-3'       |
| 6                    | Forward   | 5'-TTTGGGCACTTTCTTGA-3'       |
|                      | Reverse   | 5'-GGTGCCACCATTATAAG-3'       |
| 7                    | Forward   | 5'-CCCTCCCTCTGAAAGTT-3'       |
|                      | Reverse   | 5'-CCGCACATCCACATCTC-3'       |
| 8                    | Forward   | 5'-CCCCCACCCCCATTAGT-3'       |
|                      | Reverse   | 5'-CCCGCATACCTTGTCA-3'        |
| 9                    | Forward   | 5'-CGCCCGCTTCTTC-3'           |
|                      | Reverse   | 5'-AAGCCGGGGTCAAGAG-3'        |
| 10                   | Forward   | 5'-CTCTCCACCGCAGGTTG-3'       |
|                      | Reverse   | 5'-CTCGCCCACCACAGTC-3'        |
| 11                   | Forward   | 5'-GCCCGAGAGGTTCTCAG-3'       |
|                      | Reverse   | 5'-ATTGGGAGCTTACATCTACATT-3'  |
| 12                   | Forward   | 5'-TGGCGGTGACTGTGC-3'         |
|                      | Reverse   | 5'-TCGGCTGCTCTCCTAAAA-3'      |
| 13                   | Forward   | 5'-CTTCCAGCCTGTATGAAT-3'      |
|                      | Reverse   | 5'-GCCACCACCACTTTC-3'         |
| 14                   | Forward   | 5'-CTGGGAAAGAACATTGTTATTAA-3' |
|                      | Reverse   | 5'-CCCCCCACCAACTG-3'          |
| 15                   | Forward   | 5'-GGCGCACTTTCATTTAC-3'       |
|                      | Reverse   | 5'-CCAGCTTGCTCACCACTG-3'      |
| RT-qPCR <sup>1</sup> | Forward   | 5'-ACCGATTCAAATGTGTCACT-3'    |
|                      | Reverse   | 5'-GCCACCTCTCTTTCTT-3'        |

<sup>1</sup> PCR cycle; 10'95°C, 40x(15"95°C, 1'60°C)

**Supplementary table 1b. Overview of *BCORL1* primers.**

| PCR cycle; 10'95°C, 40x(15''95°C, 1'60°C, 30''72°C), 10'72°C |           |                                |
|--|-----------|--------------------------------|
| Exon   | Direction | Primer                         |
| 2  | Forward   | 5'-CTTCCCAGGTTAGACCTTAAT-3'    |
|  | Reverse   | 5'-CCCAGGCCCTATTGTATG-3'       |
| 3  | Forward   | 5'-CCCCCTCACAAAGTTACTACAG-3'   |
|  | Reverse   | 5'-AGCCAGCCAATTGTGTC-3'        |
| 4.1  | Forward   | 5'-CGGGCCTCAGGACAC-3'          |
|  | Reverse   | 5'-GGGCTGGCAGAGGACT-3'         |
| 4.2  | Forward   | 5'-AGGCCAGCAACAGCAG-3'         |
|  | Reverse   | 5'-AGGGGGTTCGAGTCAGA-3'        |
| 4.3  | Forward   | 5'-TGTTCCAGTCCAAGTTGCCACTTC-3' |
|  | Reverse   | 5'-ATGGGTGTAGGGCTGGAGTAAA-3'   |
| 4.4  | Forward   | 5'-TGCCCACGCTCATCTC-3'         |
|  | Reverse   | 5'-TGGAGGCAGGATATATACC-3'      |
| 4.5  | Forward   | 5'-CCGGCCTCCTTCAGTT-3'         |
|  | Reverse   | 5'-CAGGGGAGCCTGTTCA-3'         |
| 4.6  | Forward   | 5'-CCCGAGCTCCGTTCTT-3'         |
|  | Reverse   | 5'-CAGGCAGGAGGTGACATT-3'       |
| 4.7  | Forward   | 5'-CCCCCATGCCTGTGT-3'          |
|  | Reverse   | 5'-GTGGGTTCCACAAAGAGA-3'       |
| 4.8  | Forward   | 5'-AGCCCCATCTCCATCATTG-3'      |
|  | Reverse   | 5'-AGGCCCCTGATTCCATTC-3'       |
| 4.9  | Forward   | 5'-CTGCCAACGCTTATGAAG-3'       |
|  | Reverse   | 5'-GCCAACACAGGACTGTGT-3'       |
| 5  | Forward   | 5'-ACCCCTGGAGAGCTTCT-3'        |
|  | Reverse   | 5'-CTGCCCTCACCACTGTG-3'        |
| 6  | Forward   | 5'-TGGGCTTCTGGCTTTAAG-3'       |
|  | Reverse   | 5'-GTGGCAATATTGAGGACT-3'       |
| 7  | Forward   | 5'-GTCCAGGGCATTCACTTC-3'       |
|  | Reverse   | 5'-AGCACCGGGTTCTGTG-3'         |
| 8  | Forward   | 5'-GGGCTGGGAGCTGTCT-3'         |
|  | Reverse   | 5'-CGCCACACACACCTCTA-3'        |
| 9  | Forward   | 5'-GTGGCTCAGGACTGATGTT-3'      |
|  | Reverse   | 5'-GGCAAGGTCTTTGAGTGA-3'       |
| 10   | Forward   | 5'-TTCCCTGATGCAGTAGC-3'        |
|  | Reverse   | 5'-GCCCAAGGACAACAGT-3'         |
| 11   | Forward   | 5'-TGGCAGTCTCTGACTGAGAA-3'     |
|  | Reverse   | 5'-TGAGGGATGGCTGTGTC-3'        |
| 12   | Forward   | 5'-GTGCCCTCTGAAGGGATGT-3'      |
|  | Reverse   | 5'-AGCCTTGTATGTGTGATG-3'       |
| 13   | Forward   | 5'-TGGGCTTGCAGAGTGT-3'         |
|  | Reverse   | 5'-GGTCGGGGAAAGACAAG-3'        |
| RT-qPCR <sup>1</sup>   | Forward   | 5'-TGCCTCCAGAAAGACAGT-3'       |
|  | Reverse   | 5'-CCACCGCATCATGAAC-3'         |

<sup>1</sup> PCR cycle; 10'95°C, 40x(15''95°C, 1'60°C)

**Supplementary table 2. Characteristics of pediatric CN-AML cases (n=48) screened for *BCOR* and *BCORL1* mutations.**

Each column represent a single case. A colored field means this case is positive for a mutation or translocation in the corresponding gene(s). A white field indicates the case was tested negative for a mutation or translocation in the corresponding gene(s).

**Supplementary figure 1. *BCOR* expression levels determined by RT-qPCR per cytogenetic subgroup.** Graphs showing expression levels of *BCOR* for a selected cohort of pediatric AML patients (n=65, initial cohort). Each dot represents a patient and horizontal bars represent median expression for every cytogenetic subgroup. Black indicates wild-type for *BCOR*, red indicates mutated for *BCOR*. 11q23 indicates *MLL*-rearranged or *MLL*-ptd; t(8;21), the *RUNXI/RUNXIT1* translocation; t(15;17), *PML/RARA*; inv(16), *CBFB/MYH11*. Other or unknown indicates patient group without the other mentioned aberrations. Expression levels of two *BCOR*-mutated cases seemed lower than the other pediatric AML cases, but the 4 mutated cases did not significantly deviate from the non-mutated pediatric AML patients.

### Supplementary figure 1

