**eTable 1.** **Sequences of all primers used in this study.**

|  |  |  |  |
| --- | --- | --- | --- |
| **primers** | **Sequence 5’-3’** | **Genome sequence（hg19）** | **function** |
| seq-F1 | CGACCTCTGCTTACTACGACC | chr11:101817177-101817548(-)::chr11:99900659-99900828(+) | breakpoint PCR primers |
| seq-R1 | CTACAGCCATGTTCTTTACTATTTG |
| seq-F2 | TAGCAGTGTTCACCGCATCT | chr11:100571297-100571704(+)::chr11:99900287-99900660(-) |
| seq-R2 | CCTGGGCAACAGCGAGA |
| seq-F3 | TGGGAATGTTGTTTGCTTCT | chr11:98496146-98496655(-)::chr2:26068500-26069072(+) |
| seq-R3 | CTTTAGAAATGCCCTCTTTACTG |
| seq-F4 | CCTTTCGGATTCCATTTATGA | chr11:98495268-98496150(+)::chr11:101816900-101817176(-) |
| seq-R4 | CTCTTCTTCGCTACTGTGCC |
| seq-F5 | GCCAATGCTCCTTTACCAT | chr11:100571719-100572047(-)::chr2:26067963-26068500(-) |
| seq-R5 | CATTATTTCTTCTGGGACTATCTGT |
| ASXL2\_F | GTTCACTAGAGGCCAAACAT |  | qPCR primers |
| ASXL2\_R | GCTACGGATTGCCTTACC |  |

**eTable 2.** **Affected genes identified via ONT from III-1.**

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
|  | **Gene** | **Transcript** | **Localization (hg19)** | **Chr** | **SV** |
| A | ASXL2 | NM\_018263 | Intron: 1 / 12 | chr2:26068499-26068500 | 2p23.3 | TRA |
| B | CNTN5 | NM\_014361 | Intergenic region | chr11:98496146-98496150 | 11q22.1 |
| D | ARHGAP42 | NM\_152432 | Intron: 1 / 23 | chr11:100571709-100571717 |
| C | CNTN5 | NM\_014361 | Exon: 10-25 / 25 | chr11:99900655-100571709 | 11q22.1 | INV |
| ARHGAP42 | NM\_152432 | Exon: 1 / 24 |
| E | ARHGAP42 | NM\_152432 | Exon: 2-24 / 24 | chr11:100571717-101817177 | 11q22.1 |
| TMEM133 | NM\_032021 | Exon: 1 / 1 |
| PGR | NM\_000926 | Exon: 1-8 / 8 |
| TRPC6 | NM\_004621 | Exon: 1-12 / 12 |
| ANGPTL5 | NM\_178127 | Exon: 1-8 / 8 |
| CEP126 | NM\_020802 | Exon: 1-3 / 11 |