

Supplementary table 5: Summary of aberrations on *ERG* gDNA and mRNA identified in *ERG*-related patients carrying the *ERG* intragenic deletion.

| SAMPLE | N. BREAK POINTS | EXON 2-PROXIMAL BREAKPOINT | | INSERT | EXON 10-PROXIMAL BREAKPOINT | | | ABERRANT <i>ERG</i> TRANSCRIPTS |
|--------|-----------------|----------------------------|------------------|------------|-----------------------------|----------|--------|---------------------------------|
| | | POSITION | SEQUENCE | SEQUENCE | SEQUENCE | POSITION | INTRON | |
| 1 | 1 | 222327 | AAAGCCCAAGT | GGCCC | CCTAAACGTTGAGGCCAA | 271826 | 9 | EX2-EX10 |
| 2 | 1 | 222327 | AAAGCCCAAGT | AGAGGGCC | AACGTTGAGGCCAA | 271830 | 9 | EX2-EX10 |
| 3 | 1 | 222331 | AAAGCCCAAGTGTC | CCCCCAG | CGTTGAGGCCAA | 271832 | 9 | EX2-EX10 |
| 4 | 1** | ND | ND | ND | ND | ND | ND | EX2-EX10 |
| 5 | 1** | ND | ND | ND | ND | ND | ND | EX2-EX10, EX2-EX9, EX2-EX8 |
| 6 | 1 | 222322 | AAAGCCC | ATC | TGTTTATCTT | 269825 | 7 | EX2-EX10*, EX2-EX9, EX2-EX8 |
| 7 | 1 | 222329 | AAAGCCCAAGTGT | CCCCCCT | TGTGTGTCAGTCTCTGTGCCAAA | 271290 | 9 | EX2-EX10 |
| 8 | 2** | ND | ND | ND | ND | ND | ND | EX2-EX9*, EX2-EX8* |
| | | ND | ND | ND | ND | ND | ND | |
| 9 | 4 | 222332 | AAAGCCCAAGTGTTCA | AACC | CTTCTCAGCCTTC | 272238 | 9 | EX2-EX10*, EX2-EX9, EX2-EX8 |
| | | 222332 | AAAGCCCAAGTGTTCA | no insert | ATTCCTAAACGTTGAGGCCAA | 271822 | 9 | |
| | | 222325 | AAAGCCCAA | no insert | ACGTTGAGGCCAA | 271831 | 9 | |
| | | 222327 | AAAGCCCAAGT | CCCCCCC | TCTTCTCAGCCTTC | 272237 | 9 | |
| 10 | 6 | 222324 | AAAGCCCA | GGTAGG | CTCTGTGCCAAA | 271301 | 9 | EX2-EX10, EX2-EX9, EX2-EX8 |
| | | 222329 | AAAGCCCAAGTGT | CCCC | GTCTTCTCAGCCTTC | 272234 | 9 | |
| | | 222321 | AAAGCC | TCGTTGCGCG | TCTTCTCAGCCTTC | 272237 | 9 | |
| | | 222330 | AAAGCCCAAGTGTT | TAGGGAG | ACGTTGAGGCCAA | 271831 | 9 | |
| | | 222326 | AAAGCCCAAG | CGTTA | TATTCCTAAACGTTGAGGCCAA | 271821 | 9 | |
| | | 222331 | AAAGCCCAAGTGTC | no insert | CCTAAACGTTGAGGCCAA | 271826 | 9 | |
| 11 | 14 | 222323 | AAAGCCCC | CGGCG | CTTCTCAGCCTTC | 272238 | 9 | EX2-EX10, EX2-EX9, EX2-EX8 |
| | | 222327 | AAAGCCCAAGT | CGAA | CTTCTCAGCCTTC | 272238 | 9 | |
| | | 222327 | AAAGCCCAAGT | C | CTTCTCAGCCTTC | 272238 | 9 | |
| | | 222329 | AAAGCCCAAGTGT | CCCTCCCC | GTCTTCTCAGCCTTC | 272234 | 9 | |
| | | 222323 | AAAGCCCC | C | GTCTTCTCAGCCTTC | 272234 | 9 | |
| | | 222323 | AAAGCCCC | no insert | CGTCTTCTCAGCCTTC | 272233 | 9 | |
| | | 222327 | AAAGCCCAAGT | no insert | GTCTTCTCAGCCTTC | 272234 | 9 | |
| | | 222327 | AAAGCCCAAGT | TCCC | GTCTTCTCAGCCTTC | 272234 | 9 | |
| | | 222328 | AAAGCCCAAGTG | CCTCCTAG | GGCAA | 271838 | 9 | |
| | | 222329 | AAAGCCCAAGTGT | CGAGG | CTTCTCAGCCTTC | 272238 | 9 | |
| | | 222332 | AAAGCCCAAGTGTTCA | GGC | GTCTTCTCAGCCTTC | 272234 | 9 | |
| | | 222332 | AAAGCCCAAGTGTTCA | GGG | CTTCTCAGCCTTC | 272236 | 9 | |
| | | 222331 | AAAGCCCAAGTGTC | CCCGAGG | CCTAAACGTTGAGGCCAA | 271826 | 9 | |
| | | 222327 | AAAGCCCAAGT | TCACACC | CAGTCTGTGCCAAA | 271297 | 9 | |
| 12 | ND | ND | ND | ND | ND | ND | ND | EX2-EX10 |
| 13 | ND | ND | ND | ND | ND | ND | ND | EX2-EX10 |
| 14 | ND | ND | ND | ND | ND | ND | ND | EX2-EX10 |

Different breakpoints, position on the gene *ERG*, sequences and inserts identified by analysis of gDNA in 11 patients are listed. Type of aberrant *ERG* transcripts identified by analysis of mRNA in 14 patients are listed. *ERG* gene ID 2078, NG_029732.1; exons numbering according to *ERG* isoform 1 (NM_182918). **ERG* transcripts inferred by the size of PCR products and not confirmed by sequencing; **Breakpoints inferred by the size of PCR products and not confirmed by sequencing.