**Appendix 1: Primer pairs, primer sequences and their genomic localization.**

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **STS Mapping – Centromeric breakpoint** | | | | | |
| **Forward Primer** | Sequence (5’-3’) | Coordinate‡ | **Reverse Primer** | Sequence (5’-3’) | Coordinate |
| SPATA7-up-28kb-F | gtccatgcaattgccaaac | 88,823,667 | SPATA7-up-28kb-R | gcctacttggatgggactga | 88,823,879 |
| SPATA7-up-33kb-F | gggtctgacaagatcccaca | 88,818,613 | SPATA7-up-33kb-R | ccctctgcatgagcagtttc | 88,818,859 |
| SPATA7-up-38kb-F | aatcccagccgaaaacttct | 88,813,547 | SPATA7-up-38kb-R | agggtcccgattgattctct | 88,813,761 |
| **STS Mapping – Telomeric breakpoint** | | | | | |
| **Forward Primer** | Sequence (5’-3’) | Coordinate | **Reverse Primer** | Sequence (5’-3’) | Coordinate |
| SPATA7-i5-6\_1-F | tgttcaggcctctgtcacaa | 88,883,327 | SPATA7\_Ex6-1\_R\* | taactgaaaccggtgggaat | 88,892,848 |
| SPATA7-i5-6\_2-F | aatttttggcagagaattgttca | 88,884,387 |
| SPATA7-i5-6\_3-F | tgtcccagtaggtggacaat | 88,885,296 |
| SPATA7-i5-6\_4-F | attttcccctgcagattaagg | 88,886,389 |
| SPATA7-i5-6\_5-F | ggaggtgggaggtctcaat | 88,887,383 |
| SPATA7-i5-6\_6-F | ttgggcaactgggagaatag | 88,888,387 |
| SPATA7-i5-6\_7-F | tgctgtcaccctcagaacc | 88,889,416 |
| SPATA7-i5-6\_8-F | ccgctacccatccaattcta | 88,890,173 |
| SPATA7-i5-6\_9-F | ggctctgcctctgttcaaat | 88,891,409 |
| SPATA7-i5-6\_10-F | tcgtttcagaaaggaaattgatt | 88,892,346 |
| **Deletion Spanning Assays** | | | | | |
| **Forward Primer** | Sequence (5’-3’) | Coordinate | **Reverse Primer** | Sequence (5’-3’) | Coordinate |
| SPATA7-up-1kb-F | ccccagggaatatttcacaat | 88,851,145 | SPATA7\_Ex6-1\_R\* | taactgaaaccggtgggaat | 88,892,848 |
| SPATA7-up-5kb-F | ggtcagcacagtgcacattt | 88,847,153 |
| SPATA7-up-10kb-F | cttcagccatttgtgtgtgg | 88,841,770 |
| SPATA7-up-22kb-F | aacacattttcacactgttgga | 88,829,398 |
| SPATA7\_Del-F§ | atagacccccagcttccagt | 88,828,321 | SPATA7\_Del-R§ | attggggtgggggtaatact | 88,892,011 |

‡ Physical map coordinate of the 5’ end of the primer on the human genome assembly (GRCh37/hg19) of chromosome 14.   
\* Reverse primer used in combination with the listed forward primers.  
§ Primers for a diagnostic breakpoint junction PCR (fragment size 283 bp).