

# Typing of SNPs around the *SHOX* gene of the Xp/Yp pseudoautosomal region

The location of each SNP used in the linkage disequilibrium and sperm recombination studies is given in basepairs relative to Genbank entry U82668. [Markers shown in green were independently identified and submitted to dbSNP subsequent to this work]. Allele frequencies were determined from genotypes of 99 unrelated UK Caucasian semen donors (UK), 40 Saami samples (S) and 35 Vlax Roma (VR), established by hybridisation of appropriate 1 to 3kb genomic PCR products with each of the indicated allele-specific oligonucleotides (ASOs). The SNP or indel site in each ASO is indicated in red. Alu repeat sequences are shown in blue. "-" = not determined.

| <u>SNP</u> | <u>location</u> | <u>allele</u> | <u>freq</u> |          |           | <u>allele-specific oligo.</u> |
|------------|-----------------|---------------|-------------|----------|-----------|-------------------------------|
|            |                 |               | <u>UK</u>   | <u>S</u> | <u>VR</u> |                               |
| +6/597     | 4557            | C             | 0.87        | -        | 0.74      | 5' GAAACATCGCTTGGGTAT 3'      |
|            |                 | T             | 0.13        | -        | 0.26      | 5' GAAACATCGCTTGGGTAT 3'      |
| +6/1323    | 5880            | C             | 0.03        | -        | 0.06      | 5' GGGAAAGCTTTCCTTCGT 3'      |
|            |                 | G             | 0.97        | -        | 0.94      | 5' GGGAAAGGTTTCCTTCGT 3'      |
| +4/165     | 36312           | A             | 0.64        | -        | 0.74      | 5' CAGGACGCATGGCTGCC 3'       |
|            |                 | G             | 0.36        | -        | 0.26      | 5' CAGGACGC GTGGCTGCC 3'      |
| +4/223     | 36370           | C             | 0.44        | -        | 0.60      | 5' CCAAGCGCAAGCCTCCAG 3'      |
|            |                 | G             | 0.56        | -        | 0.40      | 5' CCAAGCGGAAGCCTCCAG 3'      |
| +4/375     | 36522           | C             | 0.85        | -        | 0.80      | 5' GGCCAGCCCCGGCCTCC 3'       |
|            |                 | T             | 0.15        | -        | 0.20      | 5' GGCCAGCCCCAGCCTCC 3'       |
| +4/500L    | 36649           | 3             | 0.71        | -        | 0.44      | 5' CTGCAGAGGGTCCC GGAG 3'     |
|            |                 | 4             | 0.29        | -        | 0.56      | 5' TGCAGAGGGTCCCC GGAG 3'     |
| +4/1063    | 37210           | C             | 0.28        | -        | 0.14      | 5' TGCGGGACGTGGTGACCG 3'      |
|            |                 | G             | 0.72        | -        | 0.86      | 5' TGCGGGAGGTGGTGACCG 3'      |
| +701       | 42851           | C             | 0.83        | 0.83     | 0.76      | 5' CGGCCTCCGCCGCCGCCG 3'      |
|            |                 | T             | 0.17        | 0.17     | 0.24      | 5' CGGCCTCTGCCGCCGCCG 3'      |
| +876       | 43026           | A             | 0.25        | 0.28     | 0.38      | 5' GCGGCCCGGATCCCCCCC 3'      |
|            |                 | G             | 0.75        | 0.72     | 0.62      | 5' GCGGCCCGGGTCCCCCCC 3'      |
| +1096      | 43246           | A             | 0.17        | 0.23     | 0.24      | 5' GGGGGACTCGGGCGCCTG 3'      |
|            |                 | G             | 0.83        | 0.77     | 0.76      | 5' GGGGGGCTCGGGCGCCTG 3'      |
| +2190      | 44340           | C             | 0.77        | 0.78     | 0.66      | 5' CAACCTCCGCCTCCTGGG 3'      |
|            |                 | T             | 0.23        | 0.22     | 0.34      | 5' CAACCTCTGCCTCCTGGG 3'      |
| +2378      | 44528           | C             | 0.28        | 0.37     | 0.16      | 5' GTGCTGGGACGACAGGCG 3'      |
|            |                 | T             | 0.72        | 0.63     | 0.84      | 5' GTGCTGGGATGACAGGCG 3'      |
| +2784      | 44933           | A             | 0.08        | 0.19     | 0.19      | 5' CAAGGTTATCAGAGCGCA 3'      |
|            |                 | G             | 0.92        | 0.81     | 0.81      | 5' CAAGGTTGTCAGAGCGCA 3'      |
| +2900      | 45050           | A             | 0.20        | 0.18     | 0.34      | 5' AGAGAGAGGGGCTGGCC 3'       |
|            |                 | G             | 0.80        | 0.82     | 0.66      | 5' AGAGAGAAGGGCTGGCC 3'       |
| +3571      | 45721           | C             | 0.64        | 0.66     | 0.71      | 5' GACCTTTTGAAAGTGAGA 3'      |
|            |                 | T             | 0.36        | 0.34     | 0.29      | 5' GACCTTTTGAAGTGAGA 3'       |
| +3837      | 45987           | C             | 0.95        | 0.96     | 1.00      | 5' AAGCCTGCGGGTGTTTGA 3'      |
|            |                 | T             | 0.05        | 0.04     | 0.00      | 5' AAGCCTGTGGGTGTTTGA 3'      |
| +3955      | 46105           | A             | 0.58        | 0.55     | 0.73      | 5' CTTGGGGAGAAGGAATGG 3'      |
|            |                 | G             | 0.42        | 0.45     | 0.27      | 5' CTTGGGGGAAGGAATGG 3'       |
| +3983      | 46133           | A             | 0.36        | 0.45     | 0.30      | 5' CTTGCAGGCACATGTCTG 3'      |
|            |                 | G             | 0.64        | 0.55     | 0.70      | 5' CTTGCAGGCACGTGTCTG 3'      |
| +3990      | 46140           | A             | 0.63        | 0.55     | 0.74      | 5' TGTCTCTATAGCAGGTCGA 3'     |

|          |       |   |      |      |      |    |                    |    |
|----------|-------|---|------|------|------|----|--------------------|----|
| +5053    | 47203 | C | 0.76 | 0.81 | 0.71 | 5' | GCTTTTGCCTTATGGACC | 3' |
|          |       | G | 0.24 | 0.19 | 0.29 | 5' | GCTTTTGCCTTATGGACC | 3' |
| +5683    | 47833 | C | 0.43 | 0.55 | 0.56 | 5' | ATGGGTATATCGTATATA | 3' |
|          |       | T | 0.57 | 0.45 | 0.44 | 5' | ATGGGTATATCGTATATA | 3' |
| +6085    | 48235 | C | 0.75 | 0.61 | 0.39 | 5' | ACCACCCCTCCTCCCTC  | 3' |
|          |       | T | 0.25 | 0.39 | 0.61 | 5' | ACCACCCCTCCTCCCTC  | 3' |
| +6139    | 48289 | A | 0.41 | 0.42 | 0.61 | 5' | GATGAGCAGAGACTATTA | 3' |
|          |       | G | 0.59 | 0.58 | 0.39 | 5' | GATGAGCAGAGACTATTA | 3' |
| +6195    | 48345 | C | 0.84 | 0.93 | 0.99 | 5' | CGCTGCCCATGAGACCA  | 3' |
|          |       | T | 0.16 | 0.07 | 0.01 | 5' | CGCTGCCCATGAGACCA  | 3' |
| +6211    | 48361 | C | 0.93 | 0.99 | 0.94 | 5' | CAGGCACCGGGGGGCGGA | 3' |
|          |       | T | 0.07 | 0.01 | 0.06 | 5' | CAGGCACCGGGGGGCGGA | 3' |
| +6236    | 48386 | C | 0.76 | 0.95 | 0.84 | 5' | TGGGTGTCCGCAGAGGGA | 3' |
|          |       | G | 0.24 | 0.05 | 0.16 | 5' | TGGGTGTCCGCAGAGGGA | 3' |
| +6260    | 48410 | A | 0.13 | -    | 0.11 | 5' | GGCAGAGCCTTCCTCCGC | 3' |
|          |       | C | 0.87 | -    | 0.89 | 5' | GGCAGAGCCTTCCTCCGC | 3' |
| +6485    | 48635 | A | 0.23 | 0.68 | 0.14 | 5' | CTGGGGAACCCGTCAAAA | 3' |
|          |       | C | 0.77 | 0.32 | 0.86 | 5' | CTGGGGAACCCGTCAAAA | 3' |
| +6630    | 48770 | G | 0.11 | -    | -    | 5' | CTTGCTCGGGATTTTGCT | 3' |
|          |       | T | 0.89 | -    | -    | 5' | CTTGCTCGGGATTTTGCT | 3' |
| +7445    | 49585 | C | 0.90 | 0.98 | 0.84 | 5' | ATCGTTACGATTAATCA  | 3' |
|          |       | G | 0.10 | 0.02 | 0.16 | 5' | ATCGTTACGATTAATCA  | 3' |
| +7982    | 50132 | A | 0.86 | 0.95 | 0.91 | 5' | AATAAAACAAACATGGG  | 3' |
|          |       | C | 0.14 | 0.05 | 0.09 | 5' | AATAAAACAAACATGGG  | 3' |
| +8965    | 51115 | C | 0.26 | 0.29 | 0.39 | 5' | CTTTGCTCGGAAACAGGA | 3' |
|          |       | G | 0.74 | 0.71 | 0.61 | 5' | CTTTGCTCGGAAACAGGA | 3' |
| +9177    | 51327 | C | 0.93 | 0.98 | 0.93 | 5' | CACAGTGCCTAAGGGTGG | 3' |
|          |       | T | 0.07 | 0.02 | 0.07 | 5' | CACAGTGCCTAAGGGTGG | 3' |
| +9595    | 51745 | C | 0.67 | 0.85 | 0.66 | 5' | TCCCCTGCGCTATTGCAC | 3' |
|          |       | T | 0.33 | 0.15 | 0.34 | 5' | TCCCCTGCGCTATTGCAC | 3' |
| +9697    | 51847 | G | 0.63 | 0.71 | 0.56 | 5' | GTCCTCAGGACCCTTGGT | 3' |
|          |       | T | 0.37 | 0.29 | 0.44 | 5' | GTCCTCAGGACCCTTGGT | 3' |
| +3/341   | 61494 | A | 0.31 | -    | 0.47 | 5' | TGTTCCGTTGTAACGCCG | 3' |
|          |       | T | 0.69 | -    | 0.53 | 5' | TGTTCCGTTGTAACGCCG | 3' |
| +3/663   | 61817 | C | 0.65 | -    | 0.56 | 5' | GATGTTGCTATTAGATTT | 3' |
|          |       | G | 0.35 | -    | 0.44 | 5' | GATGTTGCTATTAGATTT | 3' |
| +3/849   | 62003 | C | 0.06 | -    | 0.04 | 5' | AAAGAGACGGGACAGATT | 3' |
|          |       | T | 0.94 | -    | 0.96 | 5' | AAAGAGACGGGACAGATT | 3' |
| +3/1035  | 62189 | A | 0.23 | -    | 0.20 | 5' | CAGCCAGACCCCGAAAT  | 3' |
|          |       | G | 0.77 | -    | 0.80 | 5' | CAGCCAGACCCCGAAAT  | 3' |
| +3/1239  | 62393 | C | 0.37 | -    | 0.37 | 5' | GAGGACACAGGACTCTGG | 3' |
|          |       | G | 0.63 | -    | 0.63 | 5' | GAGGACACAGGACTCTGG | 3' |
| +3/1248  | 62401 | A | 0.49 | -    | 0.56 | 5' | TGTCCTCACCTCTGGGTT | 3' |
|          |       | G | 0.51 | -    | 0.44 | 5' | TGTCCTCACCTCTGGGTT | 3' |
| +35/1652 | 62931 | C | 0.53 | -    | -    | 5' | GTATGTGCATTTGTTATT | 3' |
|          |       | G | 0.47 | -    | -    | 5' | GTATGTGCATTTGTTATT | 3' |
| +35/2287 | 63568 | A | 0.24 | -    | -    | 5' | GGTACCCAGTGTGCCCC  | 3' |
|          |       | G | 0.76 | -    | -    | 5' | GGTACCCAGTGTGCCCC  | 3' |
| +35/2637 | 63918 | A | 0.43 | -    | -    | 5' | CACCACCACGCCCGGCTC | 3' |
|          |       | G | 0.57 | -    | -    | 5' | CACCACCACGCCCGGCTC | 3' |
| +35/3390 | 64671 | C | 0.16 | -    | -    | 5' | TTTTTGCCTGTTAGCAGA | 3' |
|          |       | G | 0.85 | -    | -    | 5' | TTTTTGCCTGTTAGCAGA | 3' |
| +35/4673 | 65954 | A | 0.56 | -    | -    | 5' | AACAATAATGGCCAAAAC | 3' |
|          |       | G | 0.44 | -    | -    | 5' | AACAATAATGGCCAAAAC | 3' |
| +35/6219 | 67500 | C | 0.38 | -    | -    | 5' | ACCAGCTCATCTACTGGT | 3' |
|          |       | T | 0.62 | -    | -    | 5' | ACCAGCTCATCTACTGGT | 3' |
| +35/6445 | 67725 | C | 0.17 | -    | -    | 5' | TAGCTCACGCTTGTTGTA | 3' |
|          |       | T | 0.83 | -    | -    | 5' | TAGCTCACGCTTGTTGTA | 3' |
| +35/6978 | 68259 | A | 0.82 | -    | -    | 5' | CTGGCTGATGCTGTAAT  | 3' |

|         |        |   |      |   |      |    |                     |    |
|---------|--------|---|------|---|------|----|---------------------|----|
| +5/507  | 69507  | A | 0.20 | - | 0.35 | 5' | AGCACAGATTTGTTTCCT  | 3' |
|         |        | G | 0.80 | - | 0.65 | 5' | AGCACAGGTTTGTTCCT   | 3' |
| +5/772  | 69772  | G | 0.80 | - | 0.62 | 5' | GTGGAAGGGCAGATGGA   | 3' |
|         |        | T | 0.20 | - | 0.38 | 5' | GTGGAAGTGGCAGATGGA  | 3' |
| +5/934  | 69935  | A | 0.59 | - | 0.68 | 5' | TGGCGTATCTCGGCTCA   | 3' |
|         |        | C | 0.41 | - | 0.32 | 5' | TGGCGTCTCTCGGCTCA   | 3' |
| +35/375 | 71375  | A | 0.17 | - | -    | 5' | CGTGTGCGCACGTGTGTG  | 3' |
|         |        | G | 0.83 | - | -    | 5' | CGTGTGCGCGCGTGTGTG  | 3' |
| +35/520 | 71520* | C | 0.17 | - | -    | 5' | TGTATTTCTGAGACAGAG  | 3' |
|         |        | T | 0.83 | - | -    | 5' | TGTATTTTGTGAGCAGAG  | 3' |
| +35/801 | 71801  | C | 0.90 | - | -    | 5' | GCCCACCGCCACACCTGG  | 3' |
|         |        | T | 0.10 | - | -    | 5' | GCCCACGTGCCACACCTGG | 3' |
| +35/939 | 71939  | A | 0.37 | - | -    | 5' | GGCCACCACGTCCAGCCT  | 3' |
|         |        | G | 0.63 | - | -    | 5' | GGCCACCGCGTCCAGCCT  | 3' |
| +35/151 | 72151  | A | 0.10 | - | -    | 5' | TAGAGACAGGGCTTCACC  | 3' |
|         |        | G | 0.90 | - | -    | 5' | TAGAGACGGGGCTTCACC  | 3' |
| +35/329 | 72329  | A | 0.20 | - | -    | 5' | GACGTCCAGCGAAGAGAA  | 3' |
|         |        | G | 0.80 | - | -    | 5' | GACGTCCGGCGAAGAGAA  | 3' |
| +7/1400 | 78440  | C | 0.45 | - | -    | 5' | ATTCTGCGGCATGCATC   | 3' |
|         |        | T | 0.55 | - | -    | 5' | ATTCTGTGGCATGCATC   | 3' |
| +7/1593 | 78633  | C | 0.99 | - | -    | 5' | CTGCAGCCGGAAGACGCA  | 3' |
|         |        | T | 0.01 | - | -    | 5' | CTGCAGCTGGAAGACGCA  | 3' |
| +7/1985 | 79025  | A | 0.13 | - | -    | 5' | TTCGAAGATCTCCAGTGC  | 3' |
|         |        | G | 0.87 | - | -    | 5' | TTCGAAGTCTCCAGTGC   | 3' |

\* actually detecting 2 SNPS which are in complete disequilibrium at positions 71517 & 71522 (CG & TA primers did not hybridise to the semen donor panel).