The American Journal of Human Genetics Volume 85

Supplemental Data

Gene Conversion between the Male-Specific Regions of the Y and the X Chromosomes at a Translocation Hotspot

Zoë H. Rosser, Patricia Balaresque, and Mark A. Jobling

Contains:

Figure S1: DNA sequence alignment of X reference sequence, X with annotated SNPs, and Y reference sequence around *HAS*

Figure S2: DNA sequence alignment of Chimpanzee X sequence (ChX_HSA), Gorilla X sequence (GoX_HSA), Human X reference sequence (HuX_HSA), Human Y reference sequence (HuY_HSA), and Chimpanzee Y sequence (ChY_HSA) around *HSA*

Table S1: Primer sequences

Table S2: Additional DNA samples sequenced around GSVs 40, 41, using Y-specific primers A4F and A2R

Figure S1: DNA sequence alignment of X reference sequence, X with annotated SNPs, and Y reference sequence around *HSA*

Corresponds to chrX:3,681,612-3,683,501 and chrY:7,155,737-7,157,796 in Build 36.1.

- GSVs are numbered (1-66) below the alignment
- X-specific primers in magenta, Y-specific primers in blue
- 246-bp identity block in lilac
- GSVs showing evidence of conversion highlighted in blue
- dbSNP X-SNPs highlighted in green; those corresponding to GSVs highlighted in yellow
- Perfect Myers recombination motif[7] (CCNCCNTNNCCNC) boxed in black
- CpG dinucleotides at GSV sites boxed in red

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XrefSeq         CAGAGTCCTATAATACATAGGGGGGTTTGTCTTTCAAAAAGAAAGAAGGAAGGGAGGGAA 60
XrefSNPs        CAGAGTCCTATAATRCATAGGGGGGTTTGTCTTTCAAAAAGAAAGAAGGAAGGGAGGGAA 60
YrefSeq         CAGAGTCCTGTAATACATAAGTGAGTTTGTCTTTCAAAAAGAAAGAAGGAAGGGAGGGAA 60
                ********* **** **** * * ************************************

XrefSeq         GGGGGGGACCTACCATTTATTAAGAAGCTTGTCTACATTAGGCAATAAATGAGCTTTTCC 120
XrefSNPs        GGGGGGGACCTACCATTTATTAAGAAGCTTGTCTACRTTAGGCAATAAATGAGCTTTTCC 120
YrefSeq         GGGGGTGAACTACGATTTATTAAGTAGCTTGTCTACATTAGGCAATAAGT-AGCTTTTCC 119
                ***** ** **** ********** *********** *********** * *********
                     1  2    3          4                       5 6

XrefSeq         CAAATTCCCAAATGAAATCCATGTTAGTTTTATAATGCTGCTGTAAAAAA--TCACCACA 178
XrefSNPs        MAAATTCCCAAATGAAATCYATGTTASTTTTATAATGCTGCTGTAAAAAA--TCACCACA 178
YrefSeq         CAAATTCCAAAATGAAATCCATGTTAGTTTTGTAACGCTGCTGTAAAAAAAATCACCACA 179
                 ******* ********** ****** **** *** **************  ********
                        7                      8   9             10,11

XrefSeq         AACGTATGTGCCTAACACAAACAAATGTATTACAGTTTTAGAGGTCAGAAGCCCCACACG 238
XrefSNPs        AACGTATGTGCCTAACACAAACAAATGTATTACAGTTTTAGAGGTCAGAAGCCCCACACG 238
YrefSeq         AACGTATGTGCCTAACACTAACAAATGTATTATAGTTTTGGAGGTCAGAAGCCCTACACA 239
                ****************** ************* ****** ************** ****
                                  12           13      14             15   16

XrefSeq         AGGCTCATGGAGATAAAATTAAGGTGTCGACTGGGCACAGTGGTTCACACTGGTAATCCC 298
XrefSNPs        AGGCTCATGGAGATAAAATTAAGGTGTCGACTGGGCACAGTGGTTCACACTGGTAATCCC 298
YrefSeq         AGGTTCATGGAGTTAAAATTAAGGTGTTGACTGGGTACAGTGGCTGACACCTGTAATCTC 299
                *** ******** ************** ******* ******* * ****  ****** *
                   17      18             19     20      21 22  23,24    25

XrefSeq         AGCACTTTGGGAGGCTGAGGCAGGAGGATTGCTTGAGGCTAGGAGTTCACGACCAGCCTA 358
XrefSNPs        AGCACTTTGGGAGGCTGAGGCAGGAGGATTGCTTGAGGCTAGGAGTTCACGACCAGCCTA 358
YrefSeq         AGCACTTTGGGAGGCTGAGGCAGGAGGATTGCTTGAGGCCAGGAGTTCAACACCAGCCTG 359
                *************************************** *********  ********
                                                      26        27,28     29

XrefSeq         GGCAATAT-GCAAGACTCCCTGTCTACAAAAAAATACTAAAAATTAGCTGAGTGTTGTGG 417
XrefSNPs        GGCAATAT-GCAAGACTCCCTGTCTACAAAAAAATACTAAAAATTAGCTGAGTGTTGTGG 417
YrefSeq         GACACTATAGCAAGACTCCCTGTCTACAAAAAG-TACCAAAAATTAGCTGGGTATGTTGG 418
                * ** *** ***********************  *** ************ ** *  ***
                30 31  32                     33,34 35           36 37 38,39

XrefSeq         TGCCTGCCTGTAGTCCCAGCTACTTGGGAGGGTGAGGTGGAAGGATCGCTTGAGCCCAGG 477
XrefSNPs        TGCCTGCCTGYAGTCCCAGCTACTTGGGAGGGTGAGGTGGAAGGATCGCTTGAGCCCAGG 477
YrefSeq         TGCCTGCCTGTAGTCCCAGCTACTTGGGAGGGTGAGGTGGAAGGATCGCTTGAGCCCAGG 478
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Figure S1 continued

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XrefSeq         AGCTCAAGGCTGCAGTGAGCCGTGATTGTAGCACTGTACTCCAGCCTGGGTGACAGAGAA 537
XrefSNPs        AGCTCAAGGCTGCAGTGAGCCGTGATTGTAGCACTGTACTCCAGCCTGGGTGACAGAGAA 537
YrefSeq         AGCTCAAGGCTGCAGTGAGCCGTGATTGTAGCACTGTACTCCAGCCTGGGTGACAGAGAG 538
                *********************************************************** 
                                                                          40
XrefSeq         AGTTTCTGTCTCTTATGAAACCCAGGAGTGCTGGTTCTTTCTGGAGGCTGTGGGGAGGAT 597
XrefSNPs        AGTTTCTGTCTCTTATGAAACCCAGGAGTGCTGGTTCTTTCTGGAGGCTGTGGGGAGGAT 597
YrefSeq         AGATTCTGTCTCTTACAAAACCCAGGAGTGCTGGTTCTTTCTGGAGGCTGTGGGGAGGAT 598
                ** ************  *******************************************
                 41           42,43

XrefSeq         CCGATTCCTTGCCTTCTTTAGCTTCCAGCGTTTCCAAGCTCATGGCTCTCCCTCACTCGC 657
XrefSNPs        CCGATTCCTTGCCTTCTTTAGCTTCCAGCGTTTCCAAGCTCATGGCTCTCCCTCACTCGC 657
YrefSeq         CCG-TTTCCTGCCTTTTTTAGTTTCCAGCATTTCCAAGCTCATGGCTCTCCCTCACTCGC 657
                *** ** * ****** ***** ******* ******************************
                  44 45 46     47    48     49

XrefSeq         ATCTCAGCTTCCATTTTCATGTCTCCTTCTCCAACTTGGACCCTCCTGCCTCCCTCTTAG 717
XrefSNPs        ATCTCAGCTTCCATTTTCATGTCTCCTTCTCCAACTTGGACCCTCCTGCCTCCCTCTTAG 717
YrefSeq         ATCTCAGCTTCCATTTTCATGTCTCCTTCTCCAACTTGGACCCTCCTGCCTCCCTCTTAG 717
                ************************************************************

XrefSeq         AAGGAGCCTGTGACGGCATTTAGGGCCAACCTGGGTCATCCAGGGGCATCTCCCATCTCA 777
XrefSNPs        AAGGAGCCTGTGACGGCATTTAGGGCCAACCTGGGTCATCCAGGGGCATCTCCCATCTCA 777
YrefSeq         AAGGAGCCTGTGACGGCATTTAGGGCCAACCTGGGTCATCCAGGGGCATCTCCCATCTCA 777
                ************************************************************

XrefSeq         AGATCCTTAACCTCATCTCATCTGCAAAGTCCTCCTGACCGTGCAAGATCAGCTGTTCCC 837
XrefSNPs        AGATCCTTAACCTCATCTCATCTGCAAAGTCCTCCTGACCGTGCAAGATCAGCTGTTCCC 837
YrefSeq         AGATCCTTAACCTCATCTCATCTGCAAAGTCCTCCTGACCGTGCAAGATCAGCTGTTCCC 837
                ************************************************************

XrefSeq         AGGTTCCAGGGATTCAGATGTGGACATCTTTGGGGAATCATTATTCTACCTATCACAGTA 897
XrefSNPs        AGGTTCCAGGGATTCAGATGTGGACATCTTTGGGGAATCATTATTCTACCTATCACAGTA 897
YrefSeq         AGGTTCCAGGGATTCAGATGTGGACATCTTTGGGGAGCCATTATTCTACCTATCACAGTA 897
                ************************************  **********************
                                                  50,51

XrefSeq         GCCTATATGCTTTTTCCCCCAAAACTCTCTGAAGTAACCACTGTTATCTCATCAGGCAAA 957
XrefSNPs        GCCTATATGCTTTTTYCCCCAAAACTCTCTGAAGTAACCACTGTTATCTCATCAGGCAAA 957
YrefSeq         GCCTATATGCTTTTTCCCCCAAAACTCTCTGAAGTAACCACTGTTACCTCATCAGGCAAA 957
                *************** ****************************** *************
                                                             52

XrefSeq         AATTCTGAGACCCAGCCAAGTGAATTACTGTGGATAATATCACGTCAACTAGAAAGTGGC 1017
XrefSNPs        AATTCTGAGACCCAGCCAAGTGAATTACTGTGGATAATATCACGTCAACTAGAAAGTGGC 1017
YrefSeq         AATTCTGAGACCCAGCCAAGTGAATTACTGTGGATAATATCACGTCAACTAGAAAGTGGC 1017
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XrefSeq         CCCAGGTAGTCAGACCATATGCTTTTTCTTTTTCCTTCAACTTTTATTTTAAGTCCGGGG 1077
XrefSNPs        CCCAGGTAGTCARACCATATGCTTTTTCTTTTTCCTTCAACTTTTATTTTAAGTCCGGGG 1077
YrefSeq         CCCAGGTAGTCAAACCATATGCTTTTTCTTTTTCCTTCAACTTTTATTTTAAGTCCGGGG 1077
                ************ ***********************************************
                            53

XrefSeq         GTATCAGTGCAGGCTGTGCAGGTTGTTTCAGAGGCAAACGTGGGCCATGCTGCTTTGTTG 1137
XrefSNPs        GTATCAGTGCAGGCTGTGCAGGTTGTTTCAKAGGCAAACGTGGGCCATGCTGCTTTGTTG 1137
YrefSeq         GTATCAGTGCAGGCTGTGCAGGTTGTTTCATAGGCAAACGTGGGCCATGCTGCTTTGTTG 1137
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                                             54
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Figure S1 continued

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XrefSeq       TTTTTCACTTTTTAATAATCGCCATTCCGACTGGCATGAGATAGCAACTCATTATGGTTT 1197
XrefSNPs      TTTTTCACTTTTTAATAATCGCCATTCCGACTGGCATGAGATARCAACTCATTATGGTTT 1197
YrefSeq       TTTTTCACTTTTTAATAATCGCCATTCCGACTGGCATGAGATAACAACTCATTATGGTTT 1197
              ******************************************* ****************
                                                         55

XrefSeq       TCATGTACATTTCATTACCCAGGTATTGAGCCTGGCATCCATTAGCTATTCTTCCTGATG 1257
XrefSNPs      TCATGTACATTTCATTACCCAGGTATTGAGCCTGGCATCCATTAGCTATTCTTCCTGATG 1257
YrefSeq       TCATGTACATTTCATTACCCAGGTATTGAGCCTGGCATCCATTAGCTATTCTTCCTGATG 1257
              ************************************************************

XrefSeq       CTCTCCCTCCCCCTGCGCCCCCTTACAACAGGCCCCAGAGTGTGCTCTTCCCCACAATGT 1317
XrefSNPs      CTCTCCYTCCCCCTGCGCCCCCTTACAACAGGCCCCAGAGTGTGCTCTTCCCCACAATGT 1317
YrefSeq       CTCTCCTTCCCCCTGCGCCCCCTTACAACAGGCCCCAGAGTGTGCTCTTCCCCACAATGT 1317
              ****** *****************************************************
                    56

XrefSeq       GTCCATGTGTTCTCATCGTTCATCTCCCAATTATAAGTGAGAACATGCAGTGTTTGGTTT 1377
XrefSNPs      GTCCATGTGTTCTCATCGTTCAKCTCCCAATTATAAGTGAGAACATGCAGTGTTTGGTTT 1377
YrefSeq       GTCCATGTGTTCTCATCGTTCAGCTCCCAATTATAAGTGAGAACATGCAGTGTTTGGTTT 1377
              ********************** *************************************
                                    57

XrefSeq       TCTCTTCCTGTGTTAGTTTCCTGAGGATAACAGCTTCCAGCTTCATCCACATTCCTGCAA 1437
XrefSNPs      TCTCTTCCTGTGTTAGTTTCCTGAGGATAACAGCTTCCAGCTTCATCCACRTTCCTGCAA 1437
YrefSeq       TCTCTTCCTGTGTTAGTTTCCTGAGGATAACAGCTTCCAGCTTCATCCACGTTCCTGCAA 1437
              ************************************************** *********
                                                                58

XrefSeq       AGGACATAATCTCACTCCTTTTTATGGCTGCATAGTATTCCATGGTGTCTATGGACCACA 1497
XrefSNPs      AGGACATAATCTCACTCCTTTTTATGGCTGCATAGTATTCCATGGTGTCTATGGACCACA 1497
YrefSeq       AGGACATAATCTCACTCCTTTTTATGGCTGCATAGTATTCCATGGTGTCTATGGACCACA 1497
              ************************************************************

XrefSeq       TTTTCTCTGTCCAGTCTACCACTGATGGTCATTTGGATTGATCCCGTGTTGTTGCTATCA 1557
XrefSNPs      TTTTCTCTRTCCAGTCTACCACTGATGGTCATTTGGRTTGATCCCGTGTTGTTGCTATCA 1557
YrefSeq       TTTTCTCTATCCAGTCTACCACTGATGGTCATTTGGGTTGATCCCGTGTTGTTGCTATCA 1557
              ******** *************************** ***********************
                      59                          60

XrefSeq       TGAATAGTGCTGCAGTGAACATACACGTCTATGTATCTTTATAATAGAGTAATTTACAGT 1617
XrefSNPs      TGAATARTGCTGCAGTGAACATACACGTCTATGTATCTTTATAATRGAGTAATTTAYAGT 1617
YrefSeq       TGAATAATGCTGCAGTGAACATACACGTCTATGTATCTTTATAATAGAGTAATTTATAGT 1617
              ****** ************************************** ********** ***
                    61                                                62

XrefSeq       CCTTTGAGTATATACCCAGCAATAGGATTGGTGGGTTAAATGATAGTTTTGCCTCTAGAT 1677
XrefSNPs      CCTTTGAGTATATACCCAGCAATAGGATTGGTGGGTTAAATGATAGTTTTGCCTCTAGAT 1677
YrefSeq       CCTTTGAGTATATACCCAGCAATAGGATTGGTGGGTTAAATGATAGTTTTGCCTCTAGAT 1677
              ************************************************************

XrefSeq       CTTTGAGAAATCACCACACTCTCCCACAATGGTTGAACTAATTTACACTCCCACCAATAG 1737
XrefSNPs      CTTTGAGAAATCACCACACTCTCCCACAATGGTTGAACTAATTTACACTCCCACCAATAG 1737
YrefSeq       CTTTGAGAAATCACCACACTCTCCCACAATGGTTGAACTAATTTACACTCCCACCAATAG 1737
              ************************************************************

XrefSeq       TGTAAAAGCCTTCCTTTTCCTCTGCAACCTCACCAGCTTCTGTTGGTTTTTGACTTTTTA 1797
XrefSNPs      TGTAAAAGCCTTCCTTTTCCTCTGCAACCTCACCAGCTTCTGTTGGTTTTTGACTTTTTA 1797
YrefSeq       TGTAAAAGCCTTCCTTTTTCTCTGCAACCTCACCAGCTTCTGTTTGTTTTTCACTTTTTA 1797
              ****************** ************************* ****** ********
                                63                        64     65
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Figure S1 continued

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XrefSeq         ATAATTGCCATTCTGACTGGTGTGAGATGGTCTCTCATTGTGGTTTTGATTTGCATTCCT 1857
XrefSNPs        ATAATTGCCATTCTGACTGGTGTGAGATGGTCTCTCATTGTGGTTTTGATTTGCATTCCT 1857
YrefSeq         ATAATTGCCATTCTGACTGGCGTGAGATGGTCTCTCATTGTGGTTTTGATTTGCATTCCT 1857
                ******************** ***************************************
                                    66

XrefSeq         CTAATCAGACCATATCTTATCTTATTGTAAGTC 1890
XrefSNPs        CTAATCAGACCATATCTTATCTTATTGTAAGT- 1889
YrefSeq         CTAATCGGACCATATCCTATCTTCTTGCAAGTA 1890
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Figure S2: DNA sequence alignment of Chimpanzee X sequence (ChX_HSA), Gorilla X sequence (GoX_HSA), Human X reference sequence (HuX_HSA), Human Y reference sequence (HuY_HSA), and Chimpanzee Y sequence (ChY_HSA) around *HSA*

Corresponds to chrX:3,681,612-3,683,501 and chrY:7,155,737-7,157,796 in human Build 36.1

- Human 246-bp identity block in lilac
- Human GSVs showing evidence of conversion in **red**
- Sites showing simple X-Y divergence highlighted in green; those showing clear evidence of gene conversion from inter-specific comparisons highlighted in yellow; those interpretable as X-GSVs arising prior to human-chimp divergence in blue

Conversion types are indicated, based on the most parsimonious deductions, assuming no recurrent mutation. Ambiguity is introduced by alternative assumptions that can be made about ancestral states prior to the human-chimp divergence. Note that identifying X-to-Y conversions unambiguously is not possible in the absence of a gorilla Y sequence.

***a*:** Human Y-to-X conversion
***b*:** Human GSV then conversion, direction unknown; or, opposite human and chimp conversions
***c*:** Chimp GSV then conversion, direction unknown; or, opposite human and chimp conversions
***d*:** X-GSV arising in chimp-human common ancestor followed by human X-to-Y conversion; or, opposite human and gorilla conversions

Sources of great ape *HSA* sequences are as follows:

- **ChY_HSA:** the chimpanzee ortholog of the Y-specific human *HSA* sequence was retrieved as a single hit by BLAST search with the human Y copy as query. ChY_HSA is part of contig NW_001252928.1.
- **ChX_HSA**: the chimpanzee ortholog of the X-specific human *HSA* sequence was retrieved as a gapped hit by BLAST search with the human X copy as query, part of contig NW_001251693.1. The 463-bp gap in this sequence was filled by searching the trace archive with the available chimp X sequence, and retrieving a reverse-complement bridging sequence, ti|1255561336.
- **GoX_HSA**: the gorilla ortholog of the X-specific human *HSA* sequence was retrieved by searching the trace archive with the human X sequence, and retrieving four sequences allowing an assembly, as follows: ti|2019343614 reverse-complement; ti|167684544; ti|2037316608; ti|2033233512 reverse-complement. The gorilla sequenced was a female, so all retrieved traces must map to the X.

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ChX_HSA         CAGAGTCCTATAATACATAGGGGGGTTTGTCTTTCAAAAAGAAAGAAGGAAGGGAGGGAA 60
GoX_HSA         CAGAGTCCTATAATACATAGGTGGGTTTGTCTTTCAAAAAGAAAGAAGGAAGGGAGGGAA 60
HuX_HSA         CAGAGTCCTATAATACATAGGGGGGTTTGTCTTTCAAAAAGAAAGAAGGAAGGGAGGGAA 60
HuY_HSA         CAGAGTCCTGTAATACATAAGTGAGTTTGTCTTTCAAAAAGAAAGAAGGAAGGGAGGGAA 60
ChY_HSA         CAGAGTCCTATAATACATAAGTGAGTTTGTCTTTCAAAAAGAAAGAAGGAAGGGAAGGAA 60
                ********* ********* * * ******************************* ****

ChX_HSA         GGGGGGGACCTACCATTTATTAAAAAGCTTGTCTACATTAGGCAATAAATGAGCTTTTCC 120
GoX_HSA         GGGGGGGACCTACCATTTATTAAGAAGCTTGTCTACATTAGGCAATAAATGAGCTTTTCC 120
HuX_HSA         GGGGGGGACCTACCATTTATTAAGAAGCTTGTCTACATTAGGCAATAAATGAGCTTTTCC 120
HuY_HSA         GGGGGTGAACTACGATTTATTAAGTAGCTTGTCTACATTAGGCAATAAGT-AGCTTTTCC 119
ChY_HSA         GGGGGTGAACTACGATTTATTAAGTAGCTTGTCTACATTAGGCAATAAGT-AGCTTTTCC 119
                ***** ** **** *********  *********************** * *********

ChX_HSA         CAAATTCCCAAATGAAATCCATGTTAGTTTTATAATGCTGCTGTAAAAAA--TCACCACA 178
GoX_HSA         CAAATTCCCAAATGAAATCCATGTTAGTTTTATAATGCTGCTGTAAAAAA--TCACCACA 178
HuX_HSA         CAAATTCCCAAATGAAATCCATGTTAGTTTTATAATGCTGCTGTAAAAAA--TCACCACA 178
HuY_HSA         CAAATTCCAAAATGAAATCCATGTTAGTTTTGTAACGCTGCTGTAAAAAAAATCACCACA 179
ChY_HSA         CAAATTCCAAAATGAAATCCATGTTAGTTTTGTAACGCTGCTGTAAAAAAA-TCACCACA 178
                ******** ********************** *** **************  ********

ChX_HSA         AACGTATGTGCCTAACACAAACAAATGTATTACAGTTTTAGAGGTCAGAAGCCCCACACG 238
GoX_HSA         AACGTATGTGCCTAACACAAACAAAAGTATTACAGTTTTAGAGGTCAGAAGCCCCACACG 238
HuX_HSA         AACGTATGTGCCTAACACAAACAAATGTATTACAGTTTTAGAGGTCAGAAGCCCCACACG 238
HuY_HSA         AACGTATGTGCCTAACACTAACAAATGTATTATAGTTTTGGAGGTCAGAAGCCCTACACA 239
ChY_HSA         AACGTATGTGCCTAACACTAACAAATGTATTATAGTTTTGGAGGTCAGAAGCCCTACACA 238
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Figure S2 continued

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ChX_HSA         AGGCTCATGGAGATAAAATTAAGGTGTCGACTGGGCACAGTGGTTCACACTGGTAATCCC 298
GoX_HSA         AGGCTCATGGAGATAAAATTAAGGTGTCGACTGGGCACAGTGGTTCACACTGGTAATCCC 298
HuX_HSA         AGGCTCATGGAGATAAAATTAAGGTGTCGACTGGGCACAGTGGTTCACACTGGTAATCCC 298
HuY_HSA         AGGTTCATGGAGTTAAAATTAAGGTGTTGACTGGGTACAGTGGCTGACACCTGTAATCTC 299
ChY_HSA         AGGTTCATGGAGTTAAAATTAAGGTGTTGACTGGGTACAGTGGCTGACACCTGTAATCTC 298
                *** ******** ************** ******* ******* * ****  ****** *

ChX_HSA         AGCACTTTGGGAGGCTGAGGCAGGAGGACTGCTTGAGGCTAGGAGTTCAAGACCAGCCTA 358
GoX_HSA         AGCACTTTGGGAGGCTGAGGCAGGAGGATTGCTTGAGGCTAGGAGTTCAAGACCAGCCTA 358
HuX_HSA         AGCACTTTGGGAGGCTGAGGCAGGAGGATTGCTTGAGGCTAGGAGTTCACGACCAGCCTA 358
HuY_HSA         AGCACTTTGGGAGGCTGAGGCAGGAGGATTGCTTGAGGCCAGGAGTTCAACACCAGCCTG 359
ChY_HSA         AGCACTTTGGGAGGCTGAGGCAGGAGGATTGCTTGAGGCCAGGAGTTCAACACCAGCCTG 358
                **************************** ********** *********  ********

ChX_HSA         GGCAATAT-GCAAGACTCCCTGTCTACAAAAAA-TACTAAAAATTAGCTGAGTGTTGTGG 416
GoX_HSA         GGCAATAT-GCAAGACTCCCTGTCTACAAAAAA-TACTAAAAATTAGCTGAGTGTTGTGG 416
HuX_HSA         GGCAATAT-GCAAGACTCCCTGTCTACAAAAAAATACTAAAAATTAGCTGAGTGTTGTGG 417
HuY_HSA         GACACTATAGCAAGACTCCCTGTCTACAAAAAG-TACCAAAAATTAGCTGGGTATGTTGG 418
ChY_HSA         GACACTATAGCAAGATTCCCTGTCTACAAAAAG-TACCAAAAATTAGCTGAGTATGTTGG 417
                * ** *** ****** ****************  *** ************ ** *  ***

ChX_HSA         TGCCTGCCTGTAGTCCCAGCTACTTGGGAGGGTGAGGTGGAAGGATCGCTTGAGCCCAGG 476
GoX_HSA         TGCCTGCCTGTAGTCCCAGCTACTTGGGAGGGTGAGGTGGAAGGATCGCTTGAGCCCAGG 476
HuX_HSA         TGCCTGCCTGTAGTCCCAGCTACTTGGGAGGGTGAGGTGGAAGGATCGCTTGAGCCCAGG 477
HuY_HSA         TGCCTGCCTGTAGTCCCAGCTACTTGGGAGGGTGAGGTGGAAGGATCGCTTGAGCCCAGG 478
ChY_HSA         TGCCTGCCTGTAGTCCCAGCTACTTGGGAGGGTGAGGTGGAAGGATCAGTTGAGCCCAGG 477
                ***********************************************  ***********

ChX_HSA         AGCTCAAGGCTGCATTGAGCCGTGATTGTAGCACTGTACTCCAGCCTGGGTGACAGAGAA 536
GoX_HSA         AGCTCAAGGCTGCAGTGAGCCGTGATTGTAGCACTGTACTCCAGCCTGGGTGACAGAGAA 536
HuX_HSA         AGCTCAAGGCTGCAGTGAGCCGTGATTGTAGCACTGTACTCCAGCCTGGGTGACAGAGAA 537
HuY_HSA         AGCTCAAGGCTGCAGTGAGCCGTGATTGTAGCACTGTACTCCAGCCTGGGTGACAGAGAG 538
ChY_HSA         AGATCAAGGCTGCAGTAAGCCATAATTGTAGTACTGTACTCCAGCCTGGGTGACAGAGAG 537
                ** *********** * **** * ******* ***************************

ChX_HSA         AGTTTCTGTCTCTTATGAAACCCATCAGTGCTGGTTCTTTCTGGAGGCTGTGGGGAGGAT 596
GoX_HSA         AGTTTCTGTCTCTTATGAAACCCATCAGTGCTGGTTCTTTCTGGAGGCTGTGGGGAGGAT 596
HuX_HSA         AGTTTCTGTCTCTTATGAAACCCAGGAGTGCTGGTTCTTTCTGGAGGCTGTGGGGAGGAT 597
HuY_HSA         AGATTCTGTCTCTTACAAAACCCAGGAGTGCTGGTTCTTTCTGGAGGCTGTGGGGAGGAT 598
ChY_HSA         AGATTCTGTCTCTTACAAAACCCAGGAGTGCTGGTTCTTTCTGGAGGCTGTGGGGAGGAT 597
                ** ************  *******  **********************************

                                        aa

ChX_HSA         CTATTTCCTTGCCTTTTTTAGCTTCCAGCATTTCCAAGCTCATGGCTCTCCCTCACTCGC 656
GoX_HSA         CCGTTTCCTTGCCTTTTTTAGTTTCCAGCATTTCCAGGCTCAAGGCTCTCCCTCACTCGC 656
HuX_HSA         CCGATTCCTTGCCTTCTTTAGCTTCCAGCGTTTCCAAGCTCATGGCTCTCCCTCACTCGC 657
HuY_HSA         CCGTTTCCT-GCCTTTTTTAGTTTCCAGCATTTCCAAGCTCATGGCTCTCCCTCACTCGC 657
ChY_HSA         CTGTTTCCTTGCGTTTTTTAGTTTCCAGCATTTCCAGGCTCAAGGCTCTCCCTCACTCGC 657
                '   ***** ** ** ***** ******* ****** ***** *****************

                 c                                   d     d

ChX_HSA         ATCTCAGCTTTC-----CATGTCTCCCTTCTCCAATTTAGGACCCGTCCTGCCTCCCTCT 711
GoX_HSA         ATCTCAGCTTCCATTTTCATGTCTCC-TTCTCCAATTT-GGACCC-TCCTGCCTCCCTCT 713
HuX_HSA         ATCTCAGCTTCCATTTTCATGTCTCC-TTCTCCAACTT-GGACCC-TCCTGCCTCCCTCT 714
HuY_HSA         ATCTCAGCTTCCATTTTCATGTCTCC-TTCTCCAACTT-GGACCC-TCCTGCCTCCCTCT 714
ChY_HSA         ATCTCAGCTTCCATTTTGATGTCTCC-TTCTCCAATTT-GGACCC-TCCTGCCTCCCTCT 714
                ********** *      ******** ******** ** ****** **************
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Figure S2 continued

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ChX_HSA         -AGAAGGAGCCTGTGACGGCATTTAGGGCCCAACCTGGGTCATCCAGGGGCATCTCCCAT 770
GoX_HSA         TAGAAGGAGCCTGTGACGGCATTTAGGGCC-AGCCTGGGTCATCCAGGGGCATCTCCCAT 772
HuX_HSA         TAGAAGGAGCCTGTGACGGCATTTAGGGCC-AACCTGGGTCATCCAGGGGCATCTCCCAT 773
HuY_HSA         TAGAAGGAGCCTGTGACGGCATTTAGGGCC-AACCTGGGTCATCCAGGGGCATCTCCCAT 773
ChY_HSA         TAGAAGGAGCTTGTGACGGCATTTAGGGCC-AACCTGGGTCATCCAGGGGCATCTCCCAT 773
                 ********* ******************* * ***************************

ChX_HSA         CTCAAGATCCTTAACCTCATCACATCTGCAAAGTCCCCCTGATGGTGCAAGATCAGCTGT 830
GoX_HSA         CTCAAGATCCTTAACCTCATCACATCTGCAAAGTCCCCCTGACCGTGCAAGACCAGCTGT 832
HuX_HSA         CTCAAGATCCTTAACCTCATCTCATCTGCAAAGTCCTCCTGACCGTGCAAGATCAGCTGT 833
HuY_HSA         CTCAAGATCCTTAACCTCATCTCATCTGCAAAGTCCTCCTGACCGTGCAAGATCAGCTGT 833
ChY_HSA         CTCAAGATCCTTAACCTCATCACATCTGCAAAGTCCCCCTGATGGTGCAAGATCAGCTGT 833
                ********************* ************** *****  ******** *******
                                     b              b     cc
ChX_HSA         TCCCAGGTTCCAGGGATTCAGATGTGGACATCTTTGGGGAACCATTCTTCTACCTATCAC 890
GoX_HSA         TCCCAGGTTCCAGGGATTCAGATGTGGACATCTTTGGGGAACCATTCTTCTACCTATCAC 892
HuX_HSA         TCCCAGGTTCCAGGGATTCAGATGTGGACATCTTTGGGGAATCATTATTCTACCTATCAC 893
HuY_HSA         TCCCAGGTTCCAGGGATTCAGATGTGGACATCTTTGGGGAGCCATTATTCTACCTATCAC 893
ChY_HSA         TCCCAGGTTCCAGGGATTCAGATGTGGACATCTTTGGGGAACCATTATTCTACCTATCAC 893
                ****************************************  **** *************
                                                              a

ChX_HSA         AGTAGCCTATATGCTTTCCCCCCCAAAACTCTCTGAAGTAACCCCTGTTATCTCATCAGG 950
GoX_HSA         AGTAGCCTATATGCTTTTTTCCCCAAAACTCTCTGAAGTAACCACTGTTATCTCATCAGG 952
HuX_HSA         AGTAGCCTATATGCTTTTTCCCCCAAAACTCTCTGAAGTAACCACTGTTATCTCATCAGG 953
HuY_HSA         AGTAGCCTATATGCTTTTTCCCCCAAAACTCTCTGAAGTAACCACTGTTACCTCATCAGG 953
ChY_HSA         AGTAGCCTATATGCTTTCCCCCCCAAAACTCTCTGAAGTAACCACTGTTATCTCATCAGG 953
                *****************   *********************** ****** *********
                                 cc

ChX_HSA         CAAAAATTCTGAGACCCAGCCAAGTGAATTACTGTGGATAATATCACGTCAACTAGAAAG 1010
GoX_HSA         CAAAAATTCTGAGACCCAGCCAAGTGAATTACTGTGGATAATATCACGTCAACTAGAAAG 1012
HuX_HSA         CAAAAATTCTGAGACCCAGCCAAGTGAATTACTGTGGATAATATCACGTCAACTAGAAAG 1013
HuY_HSA         CAAAAATTCTGAGACCCAGCCAAGTGAATTACTGTGGATAATATCACGTCAACTAGAAAG 1013
ChY_HSA         GAAAAATTCTGAGTCCCAGCCAAGTGAATTACTGTGGATAATATCACGTCAACTAGAAAG 1013
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ChX_HSA         TGGCCCCAGGTAGCCAGACCATATCCTTTTTCTTTTTCCTTCAACTTTTATTTTAAGTCC 1070
GoX_HSA         TGGCCCCAGGTAGTCAGACCATATCCTTTTTCTTTTTCCTTCAACTTTTATTTTAAGTCC 1072
HuX_HSA         TGGCCCCAGGTAGTCAGACCATATGCTTTTTCTTTTTCCTTCAACTTTTATTTTAAGTCC 1073
HuY_HSA         TGGCCCCAGGTAGTCAAACCATATGCTTTTTCTTTTTCCTTCAACTTTTATTTTAAGTCC 1073
ChY_HSA         TGGCCCCAGGTAGTCAGACCATATCTTTTTTCTTTTTCCTTTGACTTTTATTTTAAGTCC 1073
                ************* ** *******  ***************  *****************
                                        b

ChX_HSA         AGGGGTATCAGTGCAGGCTGTGCAGGTTGTTTCATAGGCAAACGTGGGCCATGCTGCTTT 1130
GoX_HSA         AGGGGTATCAGTGCAGGCTGTACAGGTTGTTTCATAGGCAAACGTGGGCCATGCTGCTTT 1132
HuX_HSA         GGGGGTATCAGTGCAGGCTGTGCAGGTTGTTTCAGAGGCAAACGTGGGCCATGCTGCTTT 1133
HuY_HSA         GGGGGTATCAGTGCAGGCTGTGCAGGTTGTTTCATAGGCAAACGTGGGCCATGCTGCTTT 1133
ChY_HSA         AGGGGTATCAGTGCAGGCTGTGCAGGTTGTTTCACAGGCAAACGTGGGCCATGCTGCTTT 1133
                 ******************** ************ *************************
                b

ChX_HSA         GTTGTTTTTCACTTTTTAATAATCGCCATTCCGACTGGCATGAGATAGCAACTCATTATG 1190
GoX_HSA         GTTGTTTTTCACTTTTTAATAATCGCCATTCCGACTGGCATGAGATAGCAACTCATTATG 1192
HuX_HSA         GTTGTTTTTCACTTTTTAATAATCGCCATTCCGACTGGCATGAGATAGCAACTCATTATG 1193
HuY_HSA         GTTGTTTTTCACTTTTTAATAATCGCCATTCCGACTGGCATGAGATAACAACTCATTATG 1193
ChY_HSA         GTTGTTTTTCACTTTTTAATAATCGCCATTCCGACTGGCATGAGATAGCAACTCATTATG 1193
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Figure S2 continued

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ChX_HSA         GTTTTCATGTACATTTCATTACCCAGGTATTGAGCCTGGCATCCATTAGCTATTCTTCCT 1250
GoX_HSA         GTTTTCATGTACATTTCATTACCCAGGTATTGAGCCTGGCATCCATTAGCTATTCTTCCT 1252
HuX_HSA         GTTTTCATGTACATTTCATTACCCAGGTATTGAGCCTGGCATCCATTAGCTATTCTTCCT 1253
HuY_HSA         GTTTTCATGTACATTTCATTACCCAGGTATTGAGCCTGGCATCCATTAGCTATTCTTCCT 1253
ChY_HSA         GTTTTCATGTACATTTCATTACCCAGGTATTGAGCCTGGCATCCATTAGCTATTCTTCCT 1253
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ChX_HSA         GATGCTCTCCCTCCCCCTGCGCCCCTCACAACAGGCCCCAGAGTGTGCTCTTCCCCACA 1310
GoX_HSA         GATGCTCTCCCTCCCCCTGCGCCCCTTACAACAGGCCCCAGAGTGTGCTCTTCCCCACA 1312
HuX_HSA         GATGCTCTCCCTCCCCCTGCGCCCCTTACAACAGGCCCCAGAGTGTGCTCTTCCCCACA 1313
HuY_HSA         GATGCTCTCCTTCCCCCTGCGCCCCTTACAACAGGCCCCAGAGTGTGCTCTTCCCCACA 1313
ChY_HSA         GATGCTCTCCCTCCCCCTGCGCCCCTCACAACAGGCCCCAGAGTGTGCTCTTCCCCACA 1313
                ********** *************** *********************************
                                          c

ChX_HSA         ATGTGTCCATGTGTTCTCATCGTTCAGCTCCCAATTATAAGTGAGAACATGCAGTGTTTG 1370
GoX_HSA         ATGTGTCCATGTGTTCTCATCGTTCAGCTCCCAATTATAAGTGAGAACACGCAGTGTTTG 1372
HuX_HSA         ATGTGTCCATGTGTTCTCATCGTTCATCTCCCAATTATAAGTGAGAACATGCAGTGTTTG 1373
HuY_HSA         ATGTGTCCATGTGTTCTCATCGTTCAGCTCCCAATTATAAGTGAGAACATGCAGTGTTTG 1373
ChY_HSA         ATGTGTCCATGTGTTCTCATCGTTCAGCTCCCAATTATAAGTGAGAACATGCAGTGTTTG 1373
                ************************** ********************** **********

ChX_HSA         GTTTTCTCTTCCTGTGTTAGTTTCCTGAGGATAACAGCTTCCAGCTTCATCCACGTTCCT 1430
GoX_HSA         GTTTTCTCTTCCTGTGTTAGTTTCCTGAGGATAACAGCTTCCAGCTTCATCCATGTTCCT 1432
HuX_HSA         GTTTTCTCTTCCTGTGTTAGTTTCCTGAGGATAACAGCTTCCAGCTTCATCCACATTCCT 1433
HuY_HSA         GTTTTCTCTTCCTGTGTTAGTTTCCTGAGGATAACAGCTTCCAGCTTCATCCACGTTCCT 1433
ChY_HSA         GTTTTCTCTTCCTGTGTTAGTTTCCTGAGGATAACAGCTTCCAGCTTCATCCACGTTCCT 1433
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ChX_HSA         GCAAAGGACATAATCTCACTCCTTTTTATGGCTGCATAGTGTTCCATGGTGTCTATGGAC 1490
GoX_HSA         GCAAGGGACATAATCTCACTCCTTTTTATGACTGCATAGTATTCTATGGTGTCTATGGAC 1492
HuX_HSA         GCAAAGGACATAATCTCACTCCTTTTTATGGCTGCATAGTATTCCATGGTGTCTATGGAC 1493
HuY_HSA         GCAAAGGACATAATCTCACTCCTTTTTATGGCTGCATAGTATTCCATGGTGTCTATGGAC 1493
ChY_HSA         GCAAAGGACATAATCTCACTCCTTTTTATGGCTGCATAGTGTTCCATGGTGTCTATGGAC 1493
                **** ************************* ********* *** ***************
                                                        c

ChX_HSA         CACATTTTCTCTATCCAGTCTACCACTGATGGTCATTTGGGTTGATCCCGTGTTGTTGCT 1550
GoX_HSA         CACATTTTCTTTATCCAGTCTACCACTGATGGTCATTTATGTTGATCCCATGTTGTTGCT 1552
HuX_HSA         CACATTTTCTCTGTCCAGTCTACCACTGATGGTCATTTGGATTGATCCCGTGTTGTTGCT 1553
HuY_HSA         CACATTTTCTCTATCCAGTCTACCACTGATGGTCATTTGGGTTGATCCCGTGTTGTTGCT 1553
ChY_HSA         CACATTTTCTCTATCCAGTCTACCACTGATGGTCATTTGGGTTGATCCCGTGTTGTTGCT 1553
                ********** * *************************   ******** **********

ChX_HSA         ATCATGAATAGTGCTGCAGTGAACATACACGTCTATGTATCTTTATAATAGAGTAATTTA 1610
GoX_HSA         ACTGTGAATAGTGCTGCAATGAACATACACGTCTATGTATCTTTATAATAGAGTAATTTA 1612
HuX_HSA         ATCATGAATAGTGCTGCAGTGAACATACACGTCTATGTATCTTTATAATAGAGTAATTTA 1613
HuY_HSA         ATCATGAATAATGCTGCAGTGAACATACACGTCTATGTATCTTTATAATAGAGTAATTTA 1613
ChY_HSA         CTCATGAATAGTGCTGCAGTGAACATACACGTCTATGTATCTTTATAATAGAGTAATTTA 1613
                    ****** ******* *****************************************

ChX_HSA         TAGTCCTTTGAGTATATACCCAGCAATAGGATTGGTGGGTTAAATGATAGTTTTGCCTCT 1670
GoX_HSA         TAGTCCTTTGAGTATATACCCAGCAATAGGATTGGTGGGTTAAATGATAGTTTTGCCTCT 1672
HuX_HSA         CAGTCCTTTGAGTATATACCCAGCAATAGGATTGGTGGGTTAAATGATAGTTTTGCCTCT 1673
HuY_HSA         TAGTCCTTTGAGTATATACCCAGCAATAGGATTGGTGGGTTAAATGATAGTTTTGCCTCT 1673
ChY_HSA         TAGTCCTTTGAGTATATACCCAGCAATAGGATTGGTGGGTTAAATGATAGTTTTGCCTCT 1673
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Figure S2 continued

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ChX_HSA         AGATCTTTGAGAAATCACCACACTCTCCCACAATGGTTGAACTAATTTACATTCCCACCA 1730
GoX_HSA         AGATCTTTGAGAAATCACCACACTCTCCCACAATGGTTGAACTAATTTACACTCCCACCA 1732
HuX_HSA         AGATCTTTGAGAAATCACCACACTCTCCCACAATGGTTGAACTAATTTACACTCCCACCA 1733
HuY_HSA         AGATCTTTGAGAAATCACCACACTCTCCCACAATGGTTGAACTAATTTACACTCCCACCA 1733
ChY_HSA         AGATCTTTGAGAAATCACCACACTCTCCCACAATGGTTGAACTAATTTACATTCCCACCA 1733
                *************************************************** ********
                                                                   c

ChX_HSA         ATAGTGTAAAAGCCTTCCTTTTTCTCTGCAACCTCACCAGCTTCTGTTGGTTTTTGACTT 1790
GoX_HSA         ATAGTGTAAAAGCCTTCCTTTTTCTCTGCAACCTCACCAGCTTCTGTTGGTTTTTGACTT 1792
HuX_HSA         ATAGTGTAAAAGCCTTCCTTTTCCTCTGCAACCTCACCAGCTTCTGTTGGTTTTTGACTT 1793
HuY_HSA         ATAGTGTAAAAGCCTTCCTTTTTCTCTGCAACCTCACCAGCTTCTGTTTGTTTTTCACTT 1793
ChY_HSA         ATAGTGTAAAAGCCTTCCTTTTTCTCTGCAACCTCACCAGCTTCTGTTGGTTTTTGACTT 1793
                ********************** ************************* ****** ****

ChX_HSA         TTTAATAATTGCCATTCTGACCGGTGTGAGATGGTCTCTCATTGTGGTTTTGATTTGCAT 1850
GoX_HSA         TTTAATAATTGCCATTCTGACTGGTGTGAGATGGTCTCTCATTGTGGTTTTGATTTGCAT 1852
HuX_HSA         TTTAATAATTGCCATTCTGACTGGTGTGAGATGGTCTCTCATTGTGGTTTTGATTTGCAT 1853
HuY_HSA         TTTAATAATTGCCATTCTGACTGGCGTGAGATGGTCTCTCATTGTGGTTTTGATTTGCAT 1853
ChY_HSA         TTTAATAATTGCCATTCTGACGGGTGTGAGATGGTCTCTCATTGTGGTTTTGATTTGCAT 1853
                ********************* ** ***********************************

ChX_HSA         TCCTCTAATCAGACCATATCTTATCTTATTGTAAGT- 1886
GoX_HSA         TCCTCTAATCAGACCATATCTTATCTTACTGTAAGTC 1889
HuX_HSA         TCCTCTAATCAGACCATATCTTATCTTATTGTAAGTC 1890
HuY_HSA         TCCTCTAATCGGACCATATCCTATCTTCTTGCAAGTA 1890
ChY_HSA         TCCTCTAATCAGACCATATCTTATCTTATTGTAAGT- 1889
                ********** ********* ******  ** ****
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Table S1: Primer sequences

* X- and Y-specificity was verified using male and female samples, and X- and Y-specific somatic hybrid cell-line DNAs.

† PCR conditions were: 94°C 30s, 64°C 30s, 70°C 1min 30s; 32 cycles, with the buffer system of Jeffreys et al. (1990) Cell 60, 473-485.

Sequenced amplicons in all cases encompassed switches from GSVs normally specific for one sex chromosome, to GSVs normally specific for the other, so PCR product contamination could be excluded as an explanation for apparent conversions.

Primer	**Sequence (5´ to 3´)**	**Notes**
Y-HSAf1*†	CAG AGT CCT GTA ATA CAT AAG TGA	Y-specific HSA amplification
Y-HSAr*†	TGC AAG AAG ATA GGA TAT GGT CC	Y-specific HSA amplification
X-HSAf2*†	CAG AGT CCT ATA ATA CAT AGG GGG	X-specific HSA amplification
X-HSAr1*†	GAC TTA CAA TAA GAT AAG ATA TGG TCT	X-specific HSA amplification
HSAseq4	ATG GAA GCT GAG ATG CGA GT	Universal sequencing primer
HSAseq6	CTG GTT CTT TCT GGA GGC TG	Universal sequencing primer
HSAseq15	CTC ATC AGG CAA AAA TTC TGA	Universal sequencing primer
HSAseq16	CAA CCT GCA CAG CCT GCA CTG	Universal sequencing primer
HSAseq17	GTG AGA ACA TGC AGT GTT TGG	Universal sequencing primer
HSAseq18	ACG TGT ATG TTC ACT GCA GCA	Universal sequencing primer
A4F*	GGT ACA GTG GCT GAC ACC T	Y-specific primer for targeted resequencing around GSVs 40, 41
A2R*	TGC TGG AAA CTA AAA AAG GCA G	Y-specific primer for targeted resequencing around GSVs 40, 41

Table S2: Additional DNA samples sequenced around GSVs 40, 41, using Y-specific primers A4F and A2R

Following discovery of an X-to-Y conversion tract covering GSVs 40-43 in YCC4, an additional 32 hgQ chromosomes from diverse populations were analysed (not shown), but no other examples of this conversion tract were observed.

Name	Population	Haplogroup
GB1757	British	A1a
YCC35	Namibian, Tsumkwe	A2b
YCC5	Namibian, Tsumkwe	A2c
YCC38	Namibian, Tsumkwe	A3b1
YCC42	S. African, Zulu	B2a1a
KRG7	Central Asian	C3c
YCC8	Zaire, Mbuti	E2b1a1
YCC55	Russian, Adygean	G2a*
H744	Nepalese	H1a*
H542	Nepalese	H1a*
YCC56	Russian, Adygean	J2a12
YCC10	Melanesian Nasioi	M2a
GB1078	British	T
GB1151	British	T
H1930	Bhutanese	L
H1973	Bhutanese	L
YCC77	Japanese	N1a
YCC47	Siberian Yakut	N1c*
YCC51	Siberian Yakut	N1c1b
YCC67	Chinese, Han	O1a1*
YCC69	Cambodia Khmer	O2a*
YCC4	US, Amerindian	Q*(xQ1a1,Q1a2,Q1a3a)
YCC70	Russian	R1a1