Figure S1. Alignment of *SRY* sequences from representative patients with the reference (normal) sequence from the GenBank.

AS-9 GAATCTGGTAG--AAGTG-AGTTTTGGATAGTAAAA-TAAGTTTCGAA-CTCTGGCACCT 55

AS-11 GAATCTGGTAG--AAGTG-AGTTTTGGATAGTAAAA-TAAGTTTCGAA-CTCTGGCACCT 55

NORMAL GAATCTGGTAG--AAGTG-AGTTTTGGATAGTAAAA-TAAGTTTCGAA-CTCTGGCACCT 55

AS-15 GAATCTGGTAG--AAGTG-AGTTTTGGATAGTAAAA-TAAGTTTCGAA-CTCTGGCACCT 55

AS-29 GAATCTGGTAG--AAGTG-AGTTTTGGATAGTAAAA-TAAGTTTCGAA-CTCTGGCACCT 55

AS-28 GAATCTGGTAG--AAGTG-AGTTTTGGATAGTAAAAATAAGTTTCGA--CTCTGGCAC-T 54

AS-17 GAATCTGGTAG--AAGTG-AGTTTTGGATAGTAAAA-TAAGTTTCGAA-CTCTGGCACCT 55

AS-18 GAATCTGGTAAGAAAGTGGAGTTTTGGATAGTAAAATAAAGTTTCGAAACTCTGGCACCT 60

\*\*\*\*\*\*\*\*\*\* \*\*\*\*\* \*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\* \*\*\*\*\*\*\*\*\* \*\*\*\*\*\*\*\*\* \*

AS-9 TTC--AATTTTGTCGCACTCTCCTTGTTTTTGACAATGCAATCATATGCTTCTGCTATGT 113

AS-11 TTC--AATTTTGTCGCACTCTCCTTGTTTTTGACAATGCAATCATATGCTTCTGCTATGT 113

NORMAL TTC--AATTTTGTCGCACTCTCCTTGTTTTTGACAATGCAATCATATGCTTCTGCTATGT 113

AS-15 TTC--AATTTTGT**T**GCACTCTCCTTGTTTTTGACAATGCAATCATATGCTTCTGCTATGT 113

AS-29 TTC--AATTTTGTCGCACTCTCCTTGTTTTTGACAATGCAATCATATGCTTCTGCTATGT 113

AS-28 TTC--AATTTTGTCGCACTCTCCTTGTTTTTGACAATGCAATCATATGCTTCTGCTATGT 112

AS-17 TTC--AATTTTGTCGCACTCTCCTTGTTTTTGACAATGCAATCATATGCTTCTGCTATGT 113

AS-18 TTTCAAATTTTGTCGCACTCTCCTTGTTTTTGACAATGCAATCATATGCTTCTGCTATGT 120

\*\* \*\*\*\*\*\*\*\* \*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*

AS-9 TAA-GCGTATTCAA-CAGCGATGATTACAGTCCAGCTGTGCAAGAGAATATTCCCGCTCT 171

AS-11 TAA-GCGTATTCAA-CAGCGATGATTACAGTCCAGCTGTGCAAGAGAATATTCCCGCTCT 171

NORMAL TAA-GCGTATTCAA-CAGCGATGATTACAGTCCAGCTGTGCAAGAGAATATTCCCGCTCT 171

AS-15 TAA-GCGTATTCAA-CAGCGATGATTACAGTCCAGCTGTGCAAGAGAATATTCCCGCTCT 171

AS-29 TAA-GCGTATTCAA-CAGCGATGATTACAGTCCAGCTGTGCAAGAGAATATTCCCGCTCT 171

AS-28 TAA-GCGTATTCAAACAGCGATGATTACAGTCCAGCTGTGCAAGAGAATATTCCCGCTCT 171

AS-17 TAA-GCGTATTCAA-CAGCGATGATTACAGTCCAGCTGTGCAAGAGAATATTCCCGCTCT 171

AS-18 TAAAGCGTATTCAA-CAGCGATGATTACAGTCCAGCTGTGCAAGAGAATATTCCCGCTCT 179

\*\*\* \*\*\*\*\*\*\*\*\*\* \*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*

AS-9 CCGGAGAAGCTCTTCCTTCCTTTGCACTGAAAGCTGTAACTCTAAGTATCAGTGTGAAAC 231

AS-11 CCGGAGAAGCTCTTCCTTCCTTTGCACTGAAAGCTGTAACTCTAAGTATCAGTGTGAAAC 231

NORMAL CCGGAGAAGCTCTTCCTTCCTTTGCACTGAAAGCTGTAACTCTAAGTATCAGTGTGAAAC 231

AS-15 CCGGAGAAGCTCTTCCTTCCTTTGCACTGAAAGCTGTAACTCTAAGTATCAGTGTGAAAC 231

AS-29 CCGGAGAAGCTCTTCCTTCCTTTGCACTGAAAGCTGTAACTCTAAGTATCAGTGTGAAAC 231

AS-28 CCGGAGAAGCTCTTCCTTCCTT-GCACTGAAAGCTGTAACTCTAAGTATCAGTGTGAAAC 230

AS-17 CCGGAGAAGCTCTTCCTTCCTTTGCACTGAAAGCTGTAACTCTAAGTATCAGTGTGAAAC 231

AS-18 CCGGAGAAGCTCTTCCTTCCTTTGCACTGAAAGCTGTAACTCTAAGTATCAGTGTGAAAC 239

\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\* \*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*

AS-9 GGGAGAAAACAGTAAAGGCAACGTCCAGGATAGAGTGAAGCGACCCATGAACGCATTCAT 291

AS-11 GGGAGAAA**G**CAGTAAAGGCAACGTCCAGGATAGAGTGAAGCGACCCATGAACGCATTCAT 291

NORMAL GGGAGAAAACAGTAAAGGCAACGTCCAGGATAGAGTGAAGCGACCCATGAACGCATTCAT 291

AS-15 GGGAGAAAACAGTAAAGGCAACGTCCAGGATAGAGTGAAGCGACCCATGAACGCATTCAT 291

AS-29 GGGAGAAAACAGTAAAGGCAACGTCCAGGATAGAGTGAAGCGACCCATGAACGCATTCAT 291

AS-28 GGGAGAAAACAGTAAAGGCAACGTCCAGGATAGAGTGAAGCGACCCATGAACGCATTCAT 290

AS-17 GGGAGAAAACAGTAAAGGCAACGTCCAGGATAGAGTGAAGCGACCCATGAACGCATTCAT 291

AS-18 GGGAGAAAACAGTAAAGGCAAC**A**TCCAGGATAGAGTGAAGCGACCCATGAACGCATTCAT 299

\*\*\*\*\*\*\*\* \*\*\*\*\*\*\*\*\*\*\*\*\* \*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*

AS-9 CGTGTGGTCTCGCGATCAGAGGCGCAAGATGGCTCTAGAGAATCCC-AGAATGCGAAACT 350

AS-11 CGTGTGGTCTCGCGATCAGAGGCGCAAGATGGCTCTAGAGAATCCCCAGAATGCGAAACT 351

NORMAL CGTGTGGTCTCGCGATCAGAGGCGCAAGATGGCTCTAGAGAATCCC-AGAATGCGAAACT 350

AS-15 CGTGTGGTCTCGCGATCAGAGGCGCAAGATGGCTCTAGAGAATCCC-AGAATGCGAAACT 350

AS-29 CGTGTGGT**T**TCGCGATCAGAGGCGCAAGATGGCTCTAGAGAATCCC-AGAATGCGAAACT 350

AS-28 CGTGTGGTCTCGCGATCAGAGGCGCAAGATGGCTCTAGAGAATCCC-AGAATGCGAAACT 349

AS-17 CGTGTGGTCTCGCGATCAGAGGCGCAAGATGGCTCTAGAGAATCCC-AGAATGCGAAACT 350

AS-18 CGTGTGGTCTCGCGATCAGAGGCGCAAGATGGCTCTAGAGAATCCC-AGAATGCGAAACT 358

\*\*\*\*\*\*\*\* \*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\* \*\*\*\*\*\*\*\*\*\*\*\*\*

AS-9 CAGAGATCAGCAAGCAGCTGGG-ATACCAGTGGAAAATGCTTACTG-AAGCCGAAAAATG 408

AS-11 CAGAGATCAGCAAGCAGCTGGGGATACCAGTGGAAAATGCTTACTGGAAGCCGAAAAATG 411

NORMAL CAGAGATCAGCAAGCAGCTGGG-ATACCAGTGGAAAATGCTTACTG-AAGCCGAAAAATG 408

AS-15 CAGAGATCAGCAAGCAGCTGGG-ATACCAGTGGAAAATGCTTACTG-AAGCCGAAAAATG 408

AS-29 CAGAGATCAGCAAGCAGCTGGG-ATACCAGTGGAAAATGCTTACTG-AAGCCGAAAAATG 408

AS-28 CAGAGATCAGCAAGCAGCTGGG-ATACCAGTGGAAAATGCTTACTG-AAGCCGAAAAATG 407

AS-17 CAGAGATCAGCAAGCAGCTGGG-ATACCAGTGGAAAATGCTTACTG-AAGCCGAAAAATG 408

AS-18 CAGAGATCAGCAAGCAGCTGGG-ATACCAGTGGAAAATGCTTACTG-AAGCCGAAAAATG 416

\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\* \*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\* \*\*\*\*\*\*\*\*\*\*\*\*\*

AS-9 GCCATTCTTCCAGGAGGCACAGAAATTAC**C**GGCCCATGCACAGAGAGAAATACCCGAATT 468

AS-11 GCCAT-CTTCCAGGAGGCACAGAAATTACAGGCC-ATGCACAGAGAGAAATACCCGAATT 469

NORMAL GCCATTCTTCCAGGAGGCACAGAAATTACAGGCC-ATGCACAGAGAGAAATACCCGAATT 467

AS-15 GCCATTCTTCCAGGAGGCACAGAAATTACAGGCC-ATGCACAGAGAGAAATACCCGAATT 467

AS-29 GCCATTCTTCCAGGAGGCACAGAAATTACAGGCC-AT**C**CACAGAGAGAAATACCCGAATT 467

AS-28 GCCATTCTTCCAGGAGGCACAGAAATTACAGGCC-ATGCACAGAGAGAAATACCCGAATT 466

AS-17 GCCATTCTTCCAGGAGGCACAGAAATTACAGGCC-ATG**T**ACAGAGAGAAATACCCGAATT 467

AS-18 GCCATTCTTCCAGGAGGCACAGAAATTACAGGCC-ATGCACAGAGAGAAATACCCGAATT 475

\*\*\*\*\* \*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\* \*\*\*\* \*\* \*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*

AS-9 ATAAGTATCGACCTCGTCGGAAGGCGAAGATGCTGCCGAAGAATTGCAGTTTGCTTCCCG 528

AS-11 ATAAGTATCGACCTCGTCGGAAGGCGAAGATGCTGCCGAAGAATTGCAGTTTGCTTCCCG 529

NORMAL ATAAGTATCGACCTCGTCGGAAGGCGAAGATGCTGCCGAAGAATTGCAGTTTGCTTCCCG 527

AS-15 ATAAGTATCGACCTCGTCGGAAGGCGAAGATGCTGCCGAAGAATTGCAGTTTGCTTCCCG 527

AS-29 ATAAGTATCGACCTCGTCGGAAGGCGAAGATGCTGCCGAAGAATTGCAGTTTGCTTCCCG 527

AS-28 ATAAGTATCGACCTCGTCGGAAGGCGAAGATGCTGCCGAAGAATTGCAGTTTGCTTCCCG 526

AS-17 ATAAGTATCGACCTCGTCGGAAGGCGAAGATGCTGCCGAAGAATTGCAGTTTGCTTCCCG 527

AS-18 ATAAGTATCGACCTCGTCGGAAGGCGAAGATGCTGCCGAAGAATTGCAGTTTGCTTCCCG 535

\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*

AS-9 CAGATCCCGCTTCGGTACTCTGCAGCGAAG-GCAACTG-ACAACAGGTTGTACAGGGATG 586

AS-11 CAGATCCCGCTTCGGTACTCTGCAGCGAAGTGCAACTGGACAACAGGTTGTACAGGGATG 589

NORMAL CAGATCCCGCTTCGGTACTCTGCAGCGAAGTGCAACTGGACAACAGGTTGTACAGGGATG 587

AS-15 CAGATCCCGCTTCGGTACTCTGCAGCGAAGTGCAACTGGACAACAGGTTGTACAGGGATG 587

AS-29 CAGATCCCGCTTCGGTACTCTGCAGCGAAGTGCAACTGGACAACAGGTTGTACAGGGATG 587

AS-28 CAGATCCCGCTTCGGTACTCTGCAGCGAAGTGCAACTGGACAACAGGTTGTACAGGGATG 586

AS-17 CAGATCCCGCTTCGGTACTCTGCAGCGAAGTGCAACTGGACAACAGGTTGTACAGGGATG 587

AS-18 CAGATCCCGCTTCGGTACTCTGCAGCGAAGTGCAACTGGACAACAGGTTGTACAGGGATG 595

\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\* \*\*\*\*\*\*\* \*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*

AS-9 ACTGTACGAAAGCCACACACTCAAGAATGGAGCACCAGCTAGGCCACTTACCGCCCATCA 646

AS-11 ACTGTACGAAAGCCACACACTCAAGAATGGAGCACCAGCTAGGCCACTTACCGCCCATCA 649

NORMAL ACTGTACGAAAGCCACACACTCAAGAATGGAGCACCAGCTAGGCCACTTACCGCCCATCA 647

AS-15 ACTGTACGAAAGCCACACACTCAAGAATGGAGCACCAGCTAGGCCACTTACCGCCCATCA 647

AS-29 ACTGTACGAAAGCCACACACTCAAGAATGGAGCACCAGCTAGGCCACTTACCGCCCATCA 647

AS-28 ACTGTACGAAAGCCACACACTCAAGAATGGAGCACCAGCTAGGCCACTTACCGCCCATCA 646

AS-17 ACTGTACGAAAGCCACACACTCAAGAATGGAGCACCAGCTAGGCCACTTACCGCCCATCA 647

AS-18 ACTGTACGAAAGCCACACACTCAAGAATGGAGCACCAGCTAGGCCACTTACCGCCCATCA 655

\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*

AS-9 ACGCAGCCAGCTCACCGCAGCAACGGGACCGCTACAGCCACTGGACAAAGCTGTAGGACA 706

AS-11 ACGCAGCCAGCTCACCGCAGCAACGGGACCGCTACAGCCACTGGACAAAGCTGTAGGACA 709

NORMAL ACGCAGCCAGCTCACCGCAGCAACGGGACCGCTACAGCCACTGGACAAAGCTGTAGGACA 707

AS-15 ACGCAGCCAGCTCACCGCAGC**G**ACGGGACCGCTACAGCCACTGGACAAAGCTGTAGGACA 707

AS-29 ACGCAGCCAGCTCACCGCAGCAACGGGACCGCTACAGCCACTGGACAAAGCTGTAGGACA 707

AS-28 ACGCAGCCAGCTCACCGCAGCAACGGGACCGCTACAGCCACTGGACAAAGCTGTAGGACA 706

AS-17 ACGCAGCCAGCTCACCGCAGCAACGGGACCGCTACAGCCACTGGACAAAGCTGTAGGACA 707

AS-18 ACGCAGCCAGCTCACCGCAGCAACGGGACCGCTACAGCCACTGGACAAAGCTGTAGGACA 715

\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\* \*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*

AS-9 ATCGGGTAACATTGGCTACAAAGACCTACCTAGATGCTCCTTTTTACGATAACTTACAGC 766

AS-11 ATCGGGTAACATTGGCTACAAAGACCTACCTAGATGCTCCTTTTTACGATAACTTACAGC 769

NORMAL ATCGGGTAACATTGGCTACAAAGACCTACCTAGATGCTCCTTTTTACGATAACTTACAGC 767

AS-15 ATCGGGTAACATTGGCTACAAAGACCTACCTAGATGCTCCTTTTTACGATAACTTACAGC 767

AS-29 ATCGGGTAACATTGGCTACAAAGACCTACCTAGATGCTCCTTTTTACGATAACTTACAGC 767

AS-28 ATCGGGTAACATTGGCTACAAAGACCTACCTAGATGCTCCTTTTTACGATAACTTACAGC 766

AS-17 ATCGGGTAACATTGGCTACAAAGACCTACCTAGATGCTCCTTTTTACGATAACTTACAGC 767

AS-18 ATCGGGTAACATTGGCTACAAAGACCTACCTAGATGCTCCTTTTTACGATAACTTACAGC 775

\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*

AS-9 CCTCACTTTCTTATGTTTAGTTTCAATATTGTTTTCTTTTCTCTGGCTAATAAAGGC 823

AS-11 CCTCACTTTCTTATGTTTAGTTTCAATATTGTTTTCTTTTCTCTGGCTAATAAAGGC 826

NORMAL CCTCACTTTCTTATGTTTAGTTTCAATATTGTTTTCTTTTCTCTGGCTAATAAAGGC 824

AS-15 CCTCACTTTCTTATGTTTAGTTTCAATATTGTTTTCTTTTCTCTGGCTAATAAAGGC 824

AS-29 CCTCACTTTCTTATGTTTAGTTTCAATATTGTTTTCTTTTCTCTGGCTAATAAAGGC 824

AS-28 CCTCACTTTCTTATGTTTAGTTTCAATATTGTTTTCTTTTCTCTGGCTAATAAAGGC 823

AS-17 CCTCACTTTCTTATGTTTAGTTTCAATATTGTTTTCTTTTCTCTGGCTAATAAAGGC 824

AS-18 CCTCACTTTCTTATGTTTAGTTTCAATATTGTTTTCTTTTCTCTGGCTAATAAAGGC 833

\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*