

American Journal of Human Genetics, Volume 86

Supplemental Data

Mechanisms for Nonrecurrent Genomic Rearrangements

Associated with CMT1A or HNPP:

Rare CNVs as a Cause for Missing Heritability

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Figure S1. The breakpoint sequences of nonrecurrent CMT1A or HNPP rearrangements (NCBI build 36). Microhomologies are shown in red. The insertions are framed. The DNA templates for the SRS event in subject SP951 are highlighted.

A) Subject A23
chr17:15101705-15101764 **TTCATTTACTCAGTTGCCAACTCACCATGCA**AAACCACACGTAATTTCTGGGATGGCTT
A23 **TTCATTTACTCAGTTGCCAACTCACCATGCA**AGACCAGCCAAATGTAGCTCAGCGGATGC
chr17:15106879-15106938 **GAGAAACTGGCTCCTCAGCCACTCTGGGA**AGACCAGCCAAATGTAGCTCAGCGGATGC

B) Subject A26
chr17:14553322-14553381 **TAATTATGTCAATGCATCTTGTAGAAAAA**AGTGTTCTGTTACTTCATAATTTGACCTTC
A26 **TAATTATGTCAATGCATCTTGTAGAAAAA**GATCAAAACAAAAGAGTTGTTATTGAAGCGC
chr17:15089562-15089621 **CACTAATATATTTTATTGGGTTGTCAAT**GATCAAAACAAAAGAGTTGTTATTGAAGCGC

C) Subject A29
chr17:15096958-15097017 **TCCAGTATTCTGACCCATTCTGCTACACGTC**AGGTCCACACAAATTAACAAAAAGCAGG
A29 **TCCAGTATTCTGACCCATTCTGCTACACGTC**CCCGAGTCTGGAGAGACCTGCAGAATTTGG
chr17:15110257-15110316 **GGCATAAAGACTTCCTCTTGTGTTAGGGA**TCCCGAGTCTGGAGAGACCTGCAGAATTTGG

D) Subjects C1292 and C2405
chr17:14215217-14215276 **GTGATCTGCCTGCCTT**AGCCTCCCAAAGTGCTGGGATTACAGGGGTGAGCCACCGCACCTT
C1292/C2405 **GTGATCTGCCCGCCTT**AGCCTCCCAAAGTGCTGGGATTACAGGCATGAGCCACTGCGCCC
chr17:15509333-15509274 **GTGATCTGCCCGCCTC**AGCCTCCCAAAGTGCTGGGATTACAGGCATGAGCCACTGCGCCC
AluY:AluY ***** ** * *****
chr17:14534640-14534581 **CTCATTCTGAGGGCAGCATTAAC**TAAATACCAAAGCCAAAGATACAAGAAGATAAAAT
C1292/C2405 **CTCATTCTGAGGGCAGC**TTTGATCCATTTTGAGTTACAGCTCACACCACTGTGTACAG
chr17:14886192-14886251 **TCCCGAGTCCACCGGCTAGGGAGCTGGAAT**TCTGTTCTCACACCACTGTGTACAG

E) Subject C3011
chr17:17856598-17856657 **ATAGTCCAGCTACTCAGGA**TGCTGAGGTGGGAGGATGGCTTGAGCCAGGAGGTGGAGG
C3011 **ATAGTCCAGCTACTCAGGA**GACCGAGGCAGGAGAATGGCCACAAACATGGCAGTTCTT
chr17:17730614-17730673 **CTAAAAACAGTTT**CAACGCCCACTGTCGGAGGCTGGGAAAAAACACATGGCAGTTCTT

F) Subjects C3159 and C4316
chr17:16072633-16072692 **TAGCCAGGATGGTCTCGA**TCTCCTGACCTCGTGATCCGCCCGCCTTGGCCTCCCAAAGTG
C3159/C4316 **TAGCCAGGATGGTCTCGA**TCTCCTGACCTCGTGATCCGCCCGCCTTGGCCTCCCAAAGTG
chr17:15025026-15025085 **TAGCCAGGATGGTCTCA**ATCTCCTGACCTCGTGATCCGCCCGCCTTGGCCTCCCAAAGTG
AluY:AluY ***** ***** ** *

G) Subject SP54C
chr17:15468600-15468659 **TTTTTCTATTTTCAGTAGAGATGGA**GTTTACCATGTTGGTCAGGCTGCTCTCGAACTCC
SP54C **TTTTTCTATTTTCAGTAGAGATGGA**GTTTACCATATTTGGCCAGGCTGATCTCGAACTCC
chr17:15057050-15057109 **TTTTTGTATTTT**TAGTAGACGAGGTTTACCATATTGGCCAGGCTGATCTCGAACTCC
AluSg:AluSg ***** * *****

H) Subject SP951
chr17:15328198-15328257 **ATGAGTGACTATTATGTGCCAGGCACCATACTAGTAGCTAGAAATAAAACATATAATGAT**
SP951 **ATGAGTGACTATTATGTGCC**TAAAATTATCTTTAGTCATTAAATTTAGTCATTATTTAT
chr17:14890718-14890777 **TAATAGTGAATAGAATTTCTT**TAAAATTATCTTTAGTCATTAAATTTAATCTTACTTCA
chr17:14890706-14890765 **GTATTTTAACTTAATAGTGAATAGAATTTCTT**TAAAATTATCTTTAGTCATTAAATTTAT

I) Subjects SP3672 and SP3840
chr17:15329758-15329817 **GGCCAGGCTAGTTTCCA**ACTCCTGACCTCAAGTGCTCCGCCACCTAGGCCTCCCAAAGT
SD11/SD14 **GGCCAGGCTAGTTTCCA**ACTCCTGACCTCAGGATGGTAGTTAAGAGCTCACATCCTAGTA
chr17:15143634-15143693 **AAAAAAAAAAAAAAAAAAAAA**ACAGTATAGGATGGTAGTTAAGAGCTCACATCCTAGTA

J) Subject SPR1
chr17:15311588-15311647 **GTGCTCTCAATCCCACTTCTCTTGGC**TCTCTTTTGCTGCAGCTAACCATCCTCTTGCC
SPR1 **GTGCTCTCAATCCCACTTCTCTTGGC**TCTCTAAAGGAGGAGAGTGGTTAAGGCCAGAGA
chr17:15118163-15118222 **TGGACTGTAATAGTTGGCTCAGTTTCA**TCTCTAAAGGAGGAGAGTGGTTAAGGCCAGAGA

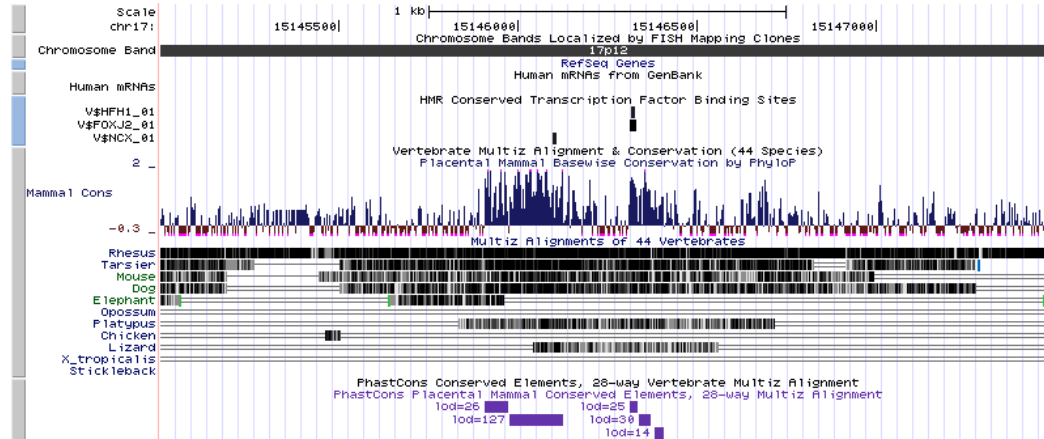
K) Subject SPR2
chr17:15079005-15079064 **TGCTCCAAGGCACCCCACTAATTTTT**AAATCATTFTTTGTAGAGATGGCATCTCGCTTG
SPR2 **TGCTCCAAGGCACCCCACTAATTTTT**CATTTGCTGAGACTGGCAAGATTTAGTTTCTCTAA
chr17:15096316-15096375 **GGAGACCACCAAGGGCTCAGTTGGGGC**TGCTGAGACTGGCAAGATTTAGTTTCTCTAA

Figure S2. The conservation tracks of the duplicated upstream regions of *PMP22* in the human UCSC Genome Browser. *Above*, the user tracks show the duplication regions in subjects SPR1, SD11, and SD14, which involved the upstream of the *PMP22* gene. *Middle*, plots of conservation scores and sequence alignments of available vertebrate genomes. The red arrows show the most conserved noncoding sequences (CNS) with log-odds scores of >100 within the overlapped duplication region. *Bottom*, the 28-way “PhastCons Placental Mammal Conserved Elements” track shows predicted conserved elements and log-odds scores.

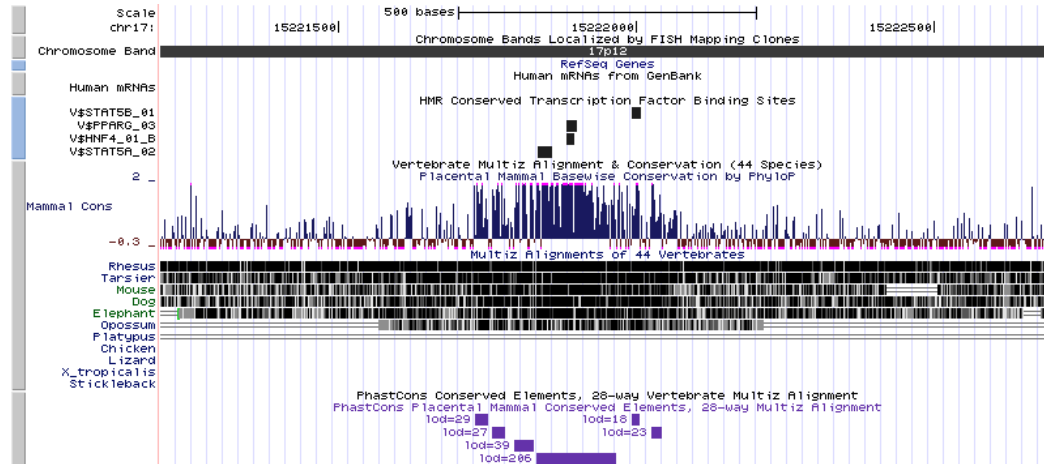


Figure S3. The UCSC Genome Browser tracks of conserved transcription factor binding sites within the highly conserved noncoding sequences (CNS 1-3) upstream of *PMP22*.

CNS1



CNS2



CNS3

