

The American Journal of Human Genetics, Volume 88

Supplemental Data

A Recurrent Deletion of *DPY19L2* Causes Infertility in Man

by Blocking Sperm Head Elongation and Acrosome Formation

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Figure S1. Position of the 54 Described CNVs in Regard with *DPY19L2* LCR1 and 2

This illustration shows the physical localization of the 54 CNV centred on *DPY19L2* described in the CNV CHOP database. Only 7 (indicated by an asterisk) of the 108 breakpoints fall outside the range of *DPY19L2* LCR 1 and 2 described in the manuscript.

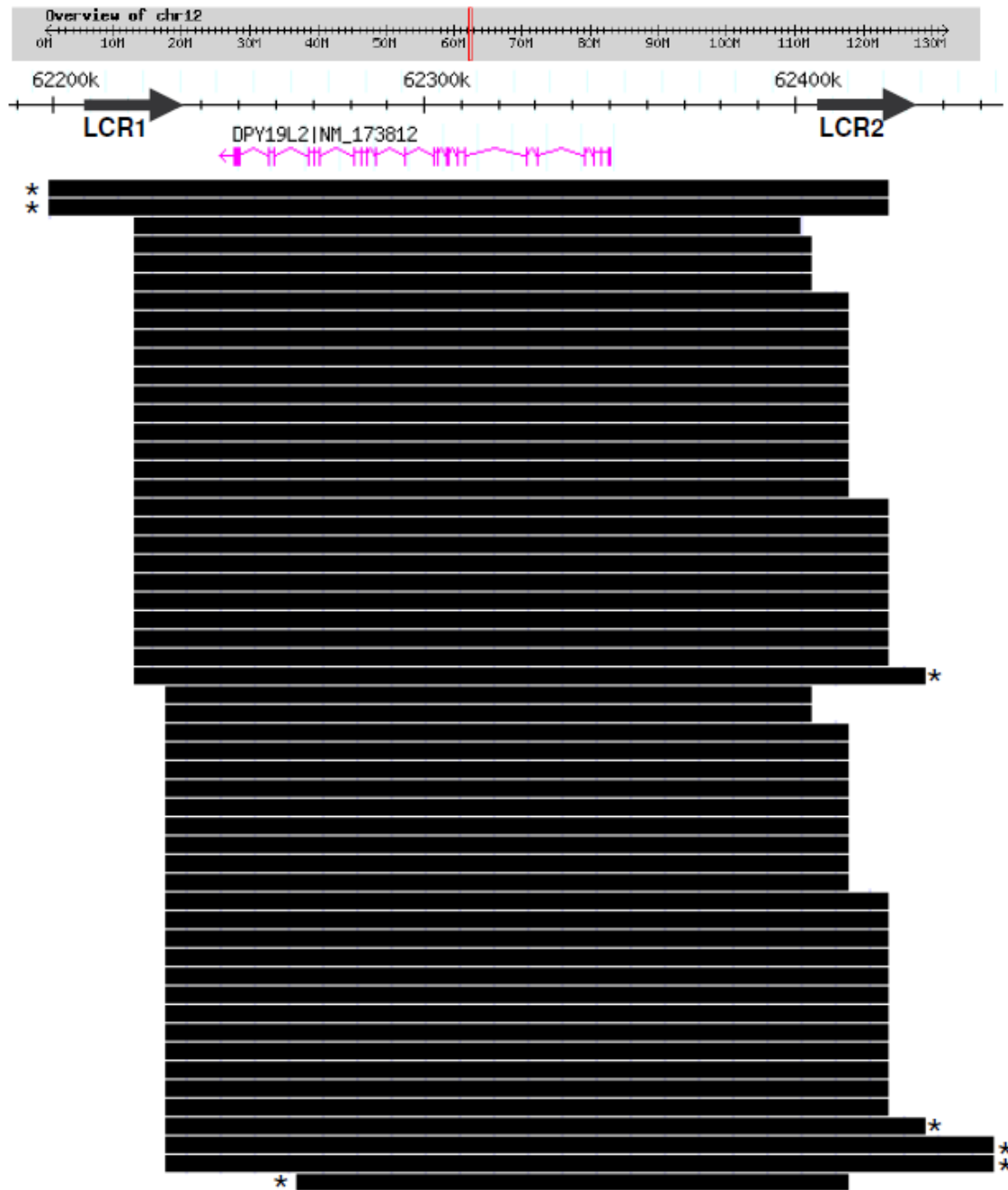


Table S1. Sequence of the PCR Primers, Size of the Amplified Products, Hybridization Temperature (Hyb.) and Genomic Localization on Chromosome 12 from NCBI36/hg18

Locus amplified	5' to 3' DNA sequence of the forward (F) and reverse primers (R)	Size (nt)	Hyb. (°C)	Localization
a) centromeric to <i>DPY19L2</i>	F: CCCTCAAGCAATTCTCTTTG R: GATTTGATTTGGGGCCTAA	288	60	62,216,631- 62,216,919
b) centromeric to <i>DPY19L2</i>	F: GATTAAAGTTCTGGGGTGAG R: GCAGCCTATTACTTCCGATA	444	60	62,233,265- 62,233,709
c) exon 22	F: GTGTCTGTTATTAAAGCTTGTG R: ATTGTCTCTAGACAGCAATACAT	313	57	62,240,454- 62,240,767
d) exon 11	F: AACCTCCTCAAGTGACTTAG R : TTGGCCAAGAGTCATT	516	57	62,297,046- 62,297,562
e) exon 1	F: GGCCAAGTTCTTTCTACTCGGAC R: GACCCAGCTCCACCATACTCCTT	504	65	62,347,964- 62,348,468
f) telomeric to <i>DPY19L2</i>	F: CTACTATTGTCTCAACCAAATATGT R : GGAATTCAAGACTAGCCTAGAA	610	58	62,410,682- 62,411,292
g) telomeric to <i>DPY19L2</i>	F : GGCATGGAAGTTACCACAAGAA R : GTTACACCACTGGCCGTCTC	750	65	62,424,210- 62,424,960

Table S2. Results of a Blastn Search for the SNP Sequences Localized in the *DPY19L2* Deleted Region

The position of the SNP is indicated in bold and the localization on chromosome 12 or 7 of the first nucleotide of each sequence is the indicated in bp (NCBI36/hg18).

Reference	Blast result	Localization
<u>rs7301605</u>	ATATAAATACACACTTCGAATAGCTAC G ATTGCCTCAGAATTTGTAAAATGT	12:62,304,630
	 ATATAAATACACACTTCGAATAGCTAC G ATTGCCTCAGAATTTGTAAAATGT	7:102,668,788
<u>rs4763094</u>	CCAAACAGGATCATATTAGAGCAATT A CTCTGCATCTACTCTGGTATTGGAT	12:62,261,438
	 CCAAACAGAACCATATTAGAGCAATT A CTCTGCATCTACTCTGGTATTGGAT	7:102,623,852
<u>rs11175119</u>	GGGCATGTTGGTAGGCACCTATAAA C CAGCTACTCAGGAAGCTGAGGCAGG	12:62,370,370
	 AGGCATGGTGGTGGGCACCTATAAA C CAGCTACTCAGGAAGCTAAGGCAGG	7:35,212,543
<u>rs11175120</u>	TCTTAAAAAAT- - - -A-A-AAATAAAGTGT A TAGTTTGATCAGTTTGGGCATATGTA	12:62,370,508
	 TCTTAAAAAATAAAAATAGAAATAAAGTGT A TAGTTTGATCAGTTTGGGCATATGTA	7:35,212,680
<u>rs11175121</u>	ATTGCCACAAGCACAATAACAAAA A TACCTAACACCTACAAAAGCTAGCTTC	12:62,370,575
	 ACTGCCACAAGCACAATAACAAAA A TCTAACACCTACAAAAGCTAGCTTC	7:35,212,753