

Supplementary Table S4.1: Determination of the lineage specificity of the pericentric inversions that distinguish the human and chimpanzee (*Pan troglodytes*, *PTR*) chromosomes using the macaque genome as an outgroup

Human chromosome	Human [<i>PTR</i>] BAC spanning the breakpoint (Acc. number)	Breakpoint position in bp according to NCBI 36, Ensembl, Aug 2006, 41.36c [PanTro 2.1, Mar 2006]	Position in BAC in bp	Macaque chromosome	Number of orthologous contig in the macaque	Position in the macaque contig in bp according to MMUL 1.0, Feb 2006; Ensembl, Jan 2006, 41.10a	Lineage specificity of the inversion
4p	RP11-779N22 (AC110811.4)	44506422-44506421 ^a	147961-147962	5	1099214622713	33141-33140	Chimpanzee-specific
4q	RP11-8N8 (AC108021.3)	86177282 ^a	141261	5	1099214584960	1159	
5p	RP11-35A11 (AC113389.2)	18589078-18589070 ^b	15650-15658	6	1099214179184	1144-1152	Chimpanzee-specific
5q	RP11-432G16 (AC104125.3)	95947182-95947292 ^b	103611-103621	6	1099214724495	38736-38746	
17p	RP1-179H24 (AF503578)	7871629-7871749 ^c	3255-3375	16	1099214672462	12104-12225	Chimpanzee-specific
17q	RP5-1029K10 (AC006487.8)	44975473-44975594 ^c	57448-57569	16	1099214668046, 1099214354398		
12p	RP11-80N2 (AC011604.10)	20940591 ^d	107110	11	1099214625617	43265	Chimpanzee-specific
12q	GSHB-410F4 (AC005294.1)	66667771 ^d	40621	11	1099214698319	19743	
9q	RP11-507D14 (AL137849.13)	88052468 ^e	63632	15	1099214642297	46169	Chimpanzee-specific
16p	CTD-2144E22 (AC135776.3)	34054535 ^f	144663	20	1099214596978	1-3727	unknown ^g
16q	RP11-696P19 (AC106819.3)	45058013 ^f	114711	20	1099214596978	3725-12029	
18p	[RP43056O01] (AG183403)	16516209-16516676 ^h		18	1099214061312	9013-9574	Human-specific
18q	[RP43-056O01] (AG183404)	16702314-16702964 ^h		18	1099214718353	6517-7145	
1p		120485240-120639476 [117357308-117369956, 186597123-186688460] ⁱ		1	1099214667781, 1099214722949	24804-25819, 7073-8076	Human-specific
1q		147158860-147687946 [128363959-128847764, 186545285-186550865] ⁱ		1	1099214720630, 1099214613637	1894-2894, 16107-17105	

a: Kehrer-Sawatzki H, Sandig C, Chuzhanova N, Goidts V, Szamalek JM, Tänzer S, Müller S, Platzer M, Cooper DN, Hameister H (2005) Breakpoint analysis of the pericentric inversion distinguishing human chromosome 4 from the homologous chromosome in the chimpanzee (*Pan troglodytes*). Hum Mut 25:45-55.

b: Szamalek JM, Goidts V, Chuzhanova N, Hameister H, Cooper DN, Kehrer-Sawatzki H (2005) Molecular characterisation of the pericentric inversion that distinguishes human chromosome 5 from the homologous chimpanzee chromosome. Hum Genet 117:168-176.

c: Kehrer-Sawatzki H, Schreiner B, Tänzer S, Platzer M, Müller S, Hameister H (2002) Molecular characterization of the pericentric inversion that causes differences between chimpanzee chromosome 19 and human chromosome 17. Am J Hum Genet 71:375-388.

d: Kehrer-Sawatzki H, Sandig CA, Goidts V, Hameister H (2005) Breakpoint analysis of the pericentric inversion between chimpanzee chromosome 10 and the homologous chromosome 12 in humans. Cytogenet Genome Res 108:91-97.

e: Kehrer-Sawatzki H, Szamalek JM, Tanzer S, Platzer M, Hameister H (2005) Molecular characterization of the pericentric inversion of chimpanzee chromosome 11 homologous to human chromosome 9. Genomics 85:542-550.

f: Goidts V, Szamalek JM, de Jong PJ, Cooper DN, Chuzhanova N, Hameister H, Kehrer-Sawatzki H (2005) Independent intrachromosomal recombination events underlie the pericentric inversions of chimpanzee and gorilla chromosomes homologous to human chromosome 16. Genome Res 15:1232-1242.

g: at the assumed breakpoint region in 16q, the macaque sequence is highly homologous to the corresponding sequence in chimpanzee, which is thus ancestral and not derived. In order to determine the lineage specificity of this rearrangement unequivocally, further studies are needed including the analysis of the corresponding regions of the gorilla genome.

h: Goidts V, Szamalek JM, Hameister H, Kehrer-Sawatzki H (2004) Segmental duplication associated with the human-specific inversion of chromosome 18: a further example of the impact of segmental duplications on karyotype and genome evolution in primates. Hum Genet 115:116-122.

i: Szamalek JM, Cooper DN, Goidts V, Hameister H, Kehrer-Sawatzki H (2006) Characterization of the human-specific pericentric inversion that discriminates human chromosome 1 from the homologous chromosomes in great apes. Hum Genet 120:126-138