

# EARLY INTERBREEDING BETWEEN ANCESTORS OF HUMANS, NEANDERTALS, AND DENISOVANS WITHIN AFRICA



Gundula Povysil, Sepp Hochreiter

Institute of Bioinformatics, Johannes Kepler University, Linz, Austria

## ABSTRACT

Since the release of high-coverage whole genome sequences of a Neandertal and a Denisovan, the interbreeding of ancestors of humans and other hominins outside of Africa has been intensively studied. In contrast to that, their interbreeding within Africa still lacks proper attention although hominins lived longer side by side within than outside of Africa and therefore had plentiful opportunities for admixture.

We investigate the genetic relationships of humans, Neandertals, and Denisovans by identifying very short DNA segments that these hominins share **identical by descent (IBD)**. By focusing on rare variants, our method **HapFABIA** reliably identifies very short IBD segments that reveal events from a very distant past because shorter segments are presumably older than longer ones. Using the **1000 Genomes Phase 3** whole genome sequencing data, we extracted two types of very old IBD segments that are shared with **Neandertals/Denisovans**: (1) longer segments primarily found in Asians and Europeans that indicate introgression events outside of Africa; (2) shorter segments mainly shared by **Africans** that indicate interbreeding of ancestors of humans and other ancient hominins within Africa. These segments are not restricted to a single population of African descent, but are common in all African populations of the 1000 Genomes Project.

Our findings can help to shed light on selection processes that were initiated by interbreeding between hominin groups at different points in time. Furthermore, our results suggest that interbreeding with other hominins was a common feature of human evolution starting already long before ancestors of modern humans left Africa.

## IBD SEGMENTS PER INDIVIDUAL

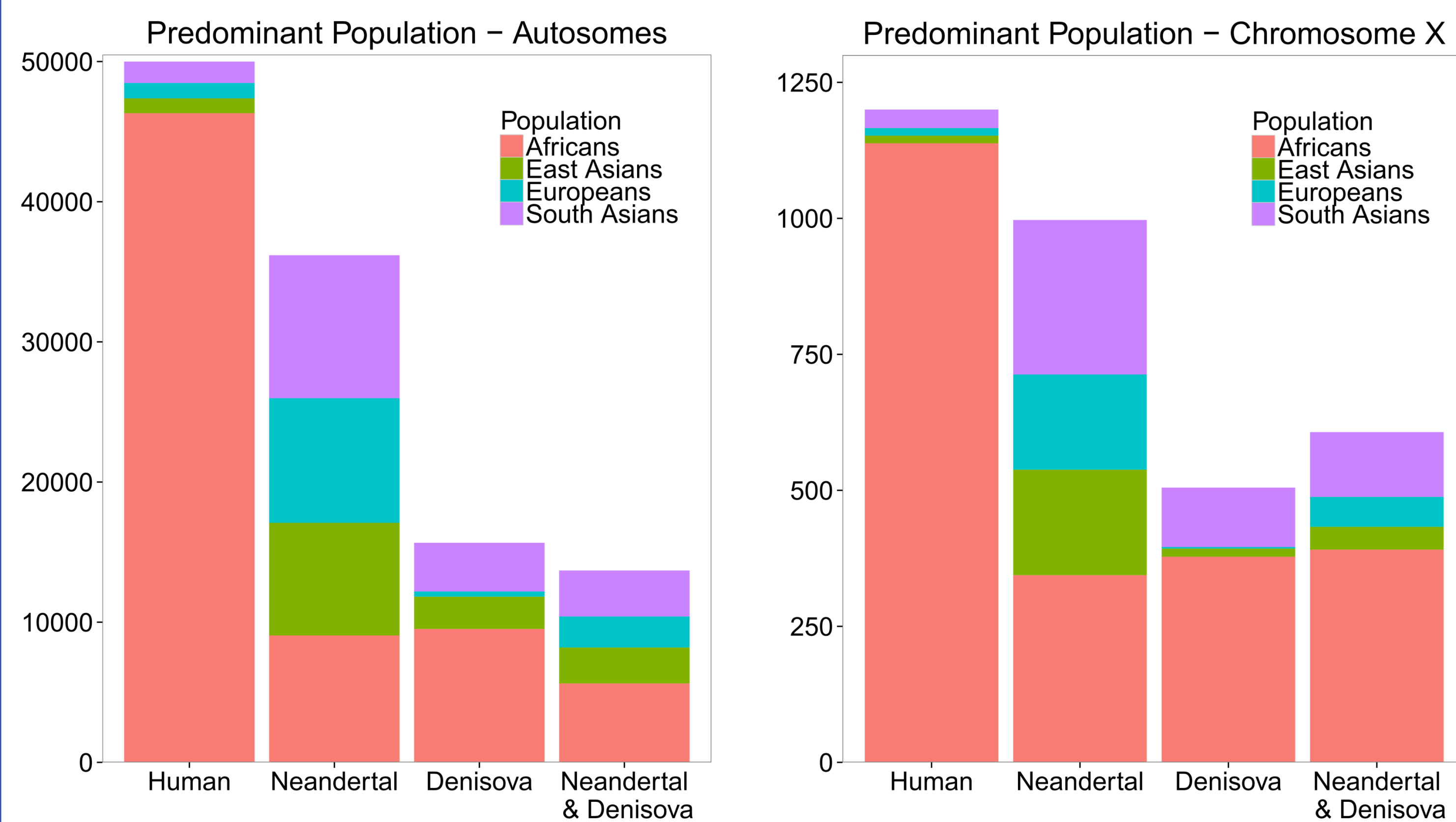
	Minimum	Median	Mean	Maximum
ALL	748	1,836	7,168	26,950
AFR	951	22,440	22,210	26,950
ACB	12,460	21,240	20,900	23,740
ASW	951	18,920	17,850	22,280
ESN	21,960	22,940	22,950	24,460
GWD	20,210	22,280	22,280	24,170
LWK	20,410	21,950	21,970	23,310
MSL	23,140	25,590	25,550	26,950
YRI	21,630	22,580	22,660	24,290
AMR	748	2,308	2,767	16,350
EAS	1,021	1,292	1,293	1,681
SAS	1,436	1,935	1,941	2,520
EUR	985	1,392	1,409	2,368

AFR = Africans (654), AMR = Americans (347), EAS = East Asians (504), SAS = South Asians (485), EUR = Europeans (503)

ASW = African Ancestry in Southwest US, ACB = African Caribbean in Barbados, ESN = Esan in Nigeria, GWD = Gambian in Western Division, The Gambia, LWK = Luhya in Webuye, Kenya, MSL = Mende in Sierra Leone, YRI = Yoruba in Ibadan, Nigeria

**each African individual has about ten times more IBD segments than any East Asian, South Asian, or European individual**

## PREDOMINANT POPULATION PER GENOME



IBD segments are given for each matching genome, where the color indicates the population to which the majority of individuals sharing a segment belong. For the human genome 50,000 random IBD segments were chosen for the autosomes and 1,200 for chromosome X.

## EXCLUSIVELY SHARED IBD SEGMENTS PER CHROMOSOME

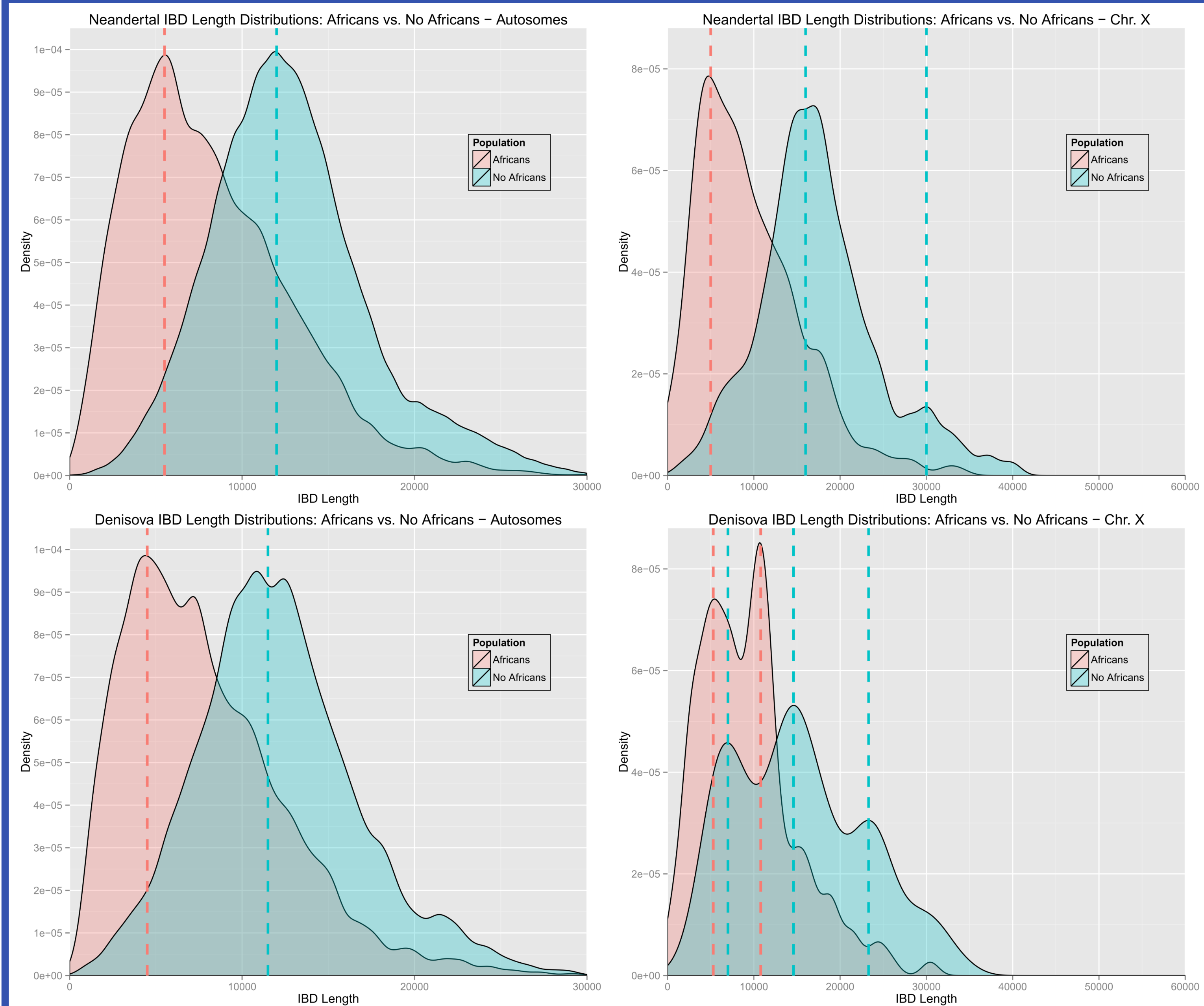
Chr.	Neandertal					Denisova				
	ALL	EAS	SAS	EUR	AFR	ALL	EAS	SAS	EUR	AFR
1	2954	220 (7.4%)	277 (9.4%)	115 (3.9%)	526 (17.8%)	1359	131 (9.6%)	152 (11.2%)	3 (0.2%)	643 (47.3%)
2	2940	214 (7.3%)	276 (9.4%)	110 (3.7%)	574 (19.5%)	1261	79 (6.3%)	205 (16.3%)	6 (0.5%)	644 (51.1%)
3	2452	165 (6.7%)	349 (14.2%)	73 (3.0%)	497 (20.3%)	1011	53 (5.2%)	194 (19.2%)	1 (0.1%)	553 (54.7%)
4	2765	197 (7.1%)	293 (10.6%)	84 (3.0%)	566 (20.5%)	1317	170 (12.9%)	257 (19.5%)	2 (0.2%)	588 (44.6%)
5	2293	188 (8.2%)	259 (11.3%)	113 (4.9%)	388 (16.9%)	1108	114 (10.3%)	178 (16.1%)	3 (0.3%)	458 (41.3%)
6	2554	187 (7.3%)	204 (8.0%)	98 (3.8%)	340 (13.3%)	1087	88 (8.1%)	139 (12.8%)	5 (0.5%)	532 (48.9%)
7	1990	140 (7.0%)	192 (9.6%)	83 (4.2%)	383 (19.2%)	764	40 (5.2%)	76 (9.9%)	2 (0.3%)	458 (59.9%)
8	1629	72 (4.4%)	177 (10.9%)	98 (6.0%)	434 (26.6%)	740	33 (4.5%)	119 (16.1%)	1 (0.1%)	421 (56.9%)
9	1473	99 (6.7%)	135 (9.2%)	77 (5.2%)	247 (16.8%)	777	17 (2.2%)	170 (21.9%)	1 (0.1%)	413 (53.2%)
10	1798	138 (7.7%)	181 (10.1%)	81 (4.5%)	238 (13.2%)	810	52 (6.4%)	196 (24.2%)	1 (0.1%)	369 (45.6%)
11	2149	98 (4.6%)	237 (11.0%)	143 (6.7%)	270 (12.6%)	883	60 (6.8%)	138 (15.6%)	7 (0.8%)	433 (49.0%)
12	1962	115 (5.9%)	213 (10.9%)	67 (3.4%)	240 (12.2%)	534	34 (6.4%)	92 (17.2%)	1 (0.2%)	298 (55.8%)
13	1442	99 (6.9%)	190 (13.2%)	60 (4.2%)	231 (16.0%)	619	63 (10.2%)	114 (18.4%)	1 (0.2%)	242 (39.1%)
14	1379	153 (11.1%)	139 (10.1%)	54 (3.9%)	171 (12.4%)	451	33 (7.3%)	59 (13.1%)	0 (0.0%)	249 (55.2%)
15	1083	121 (11.2%)	112 (10.3%)	45 (4.2%)	166 (15.3%)	490	17 (3.5%)	101 (20.6%)	0 (0.0%)	249 (50.8%)
16	880	34 (3.9%)	91 (10.3%)	36 (4.1%)	197 (22.4%)	453	8 (1.8%)	78 (17.2%)	1 (0.2%)	264 (58.3%)
17	821	45 (5.5%)	137 (16.7%)	39 (4.8%)	172 (21.0%)	367	19 (5.2%)	60 (16.3%)	1 (0.3%)	200 (54.5%)
18	1152	68 (5.9%)	115 (10.0%)	57 (4.9%)	257 (22.3%)	448	26 (5.8%)	90 (20.1%)	2 (0.4%)	223 (49.8%)
19	793	54 (6.8%)	79 (10.0%)	37 (4.7%)	161 (20.3%)	396	33 (8.3%)	68 (17.2%)	0 (0.0%)	184 (46.5%)
20	823	34 (4.1%)	99 (12.0%)	43 (5.2%)	172 (20.9%)	312	6 (1.9%)	59 (18.9%)	3 (1.0%)	165 (52.9%)
21	461	16 (3.5%)	65 (14.1%)	15 (3.3%)	105 (22.8%)	217	20 (9.2%)	47 (21.7%)	0 (0.0%)	93 (42.9%)
22	390	25 (6.4%)	34 (8.7%)	8 (2.1%)	58 (14.9%)	260	29 (11.2%)	81 (31.2%)	1 (0.4%)	85 (32.7%)
1-22	36183	2482 (6.9%)	3854 (10.7%)	1536 (4.2%)	6393 (17.7%)	15664	1125 (7.2%)	2673 (17.1%)	42 (0.3%)	7764 (49.6%)
X	997	93 (9.3%)	161 (16.1%)	51 (5.1%)	300 (30.1%)	505	8 (1.6%)	90 (17.8%)	0 (0.0%)	327 (64.8%)

## POPULATIONS SHARING EXCLUSIVELY AFRICAN IBD SEGMENTS

	AFR	ACB	ASW	ESN	GWD	LWK	MSL	YRI
Neandertal 1-22	6393	2116	1432	1956	2124	1910	2110	2006
Neandertal X	300	64	45	71	65	70	63	65
Denisova 1-22	7764	2526	1862	2444	2595	2502	2665	2542
Denisova X	327	81	76	89	90	99	96	89

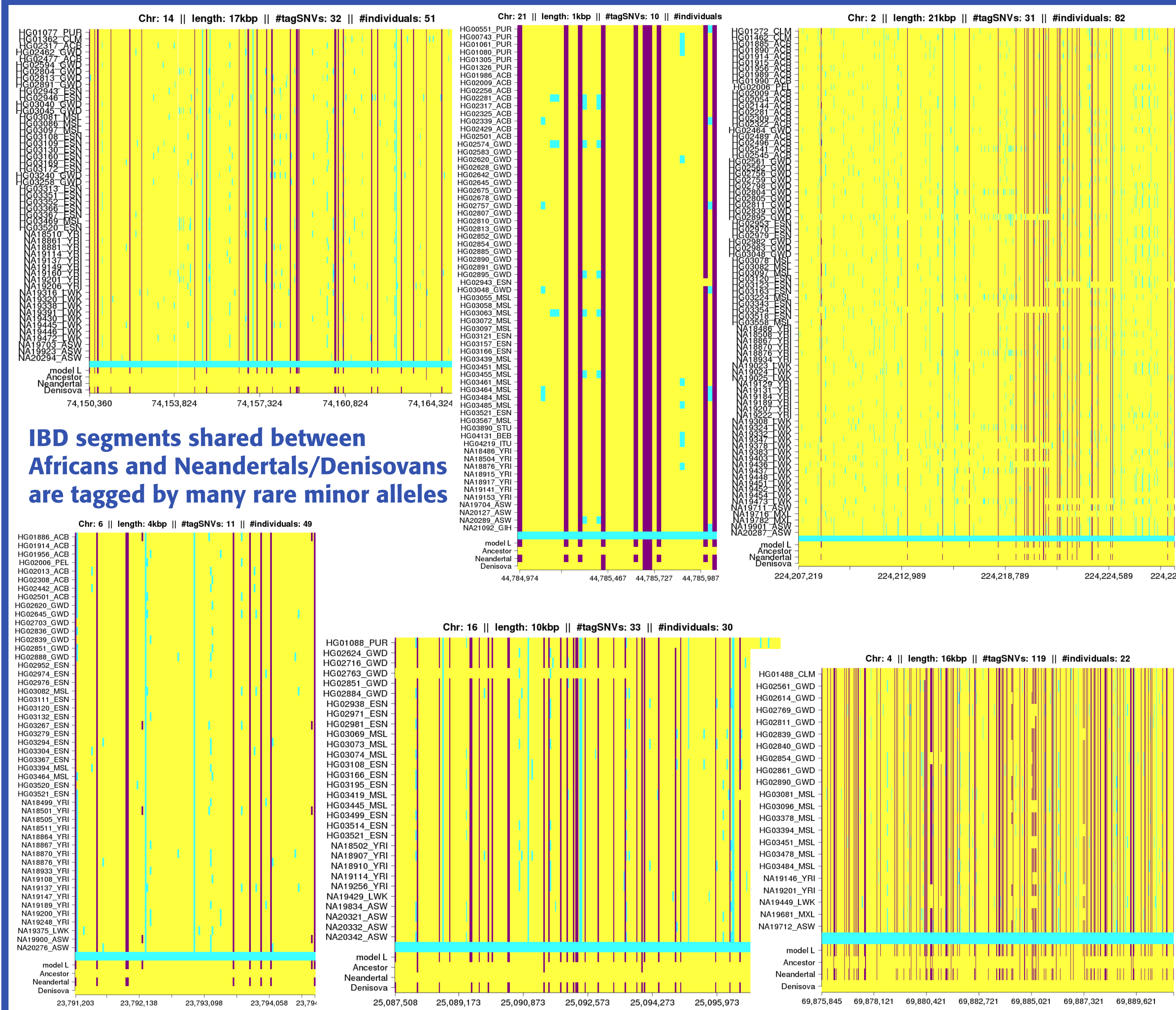
**all African populations (except ASW) share similar numbers of IBD segments that are exclusively shared between Africans and Neandertals or Denisovans**

## IBD SEGMENT LENGTH DENSITIES



**IBD segments shared between Africans and Neandertals/Denisovans are clearly shorter than IBD segments nor shared by Africans**

## IBD SEGMENTS



## CONCLUSION

- African individuals share clearly more IBD segments than Asians or Europeans
- longer IBD segments stem from introgression of Neandertals/Denisovans into ancestors of Asians and Europeans after they left Africa (to a larger extent into ancestors of Asians especially South Asians)
- many Neandertal- and/or Denisova-matching IBD segments predominately shared by Africans (some exclusively)
- no clear differences between African populations
- IBD segments shared between Africans and Neandertals or Denisovans are strikingly short → very old → very early introgression within Africa?