

An introduction to the Y-chromosomal data from Phase 3 of the 1000 Genomes Project

Yali Xue

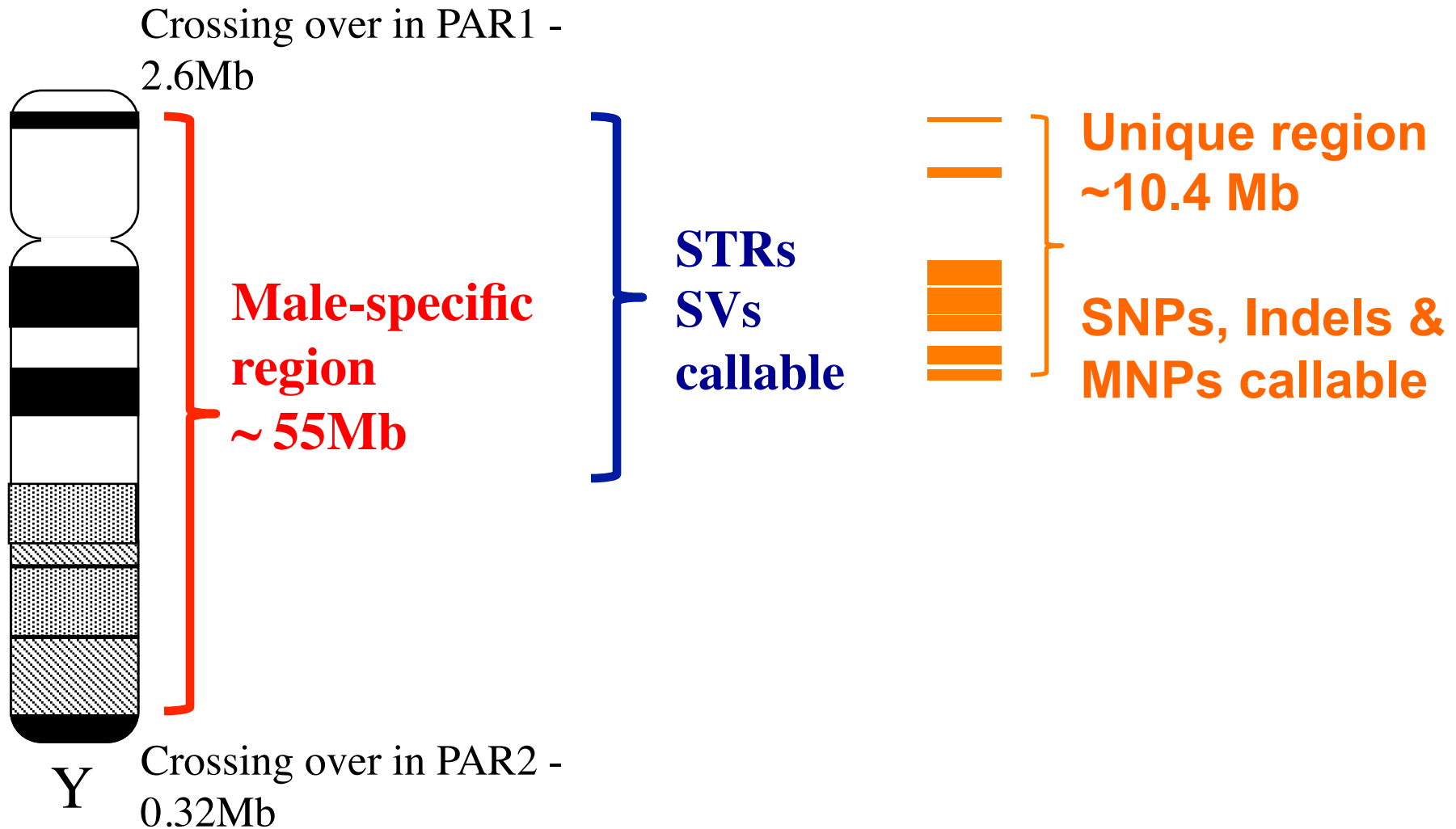
on behalf of The 1000 Genomes Y chromosome group



1000 Genomes

A Deep Catalog of Human Genetic Variation

Workable part of Y chromosome

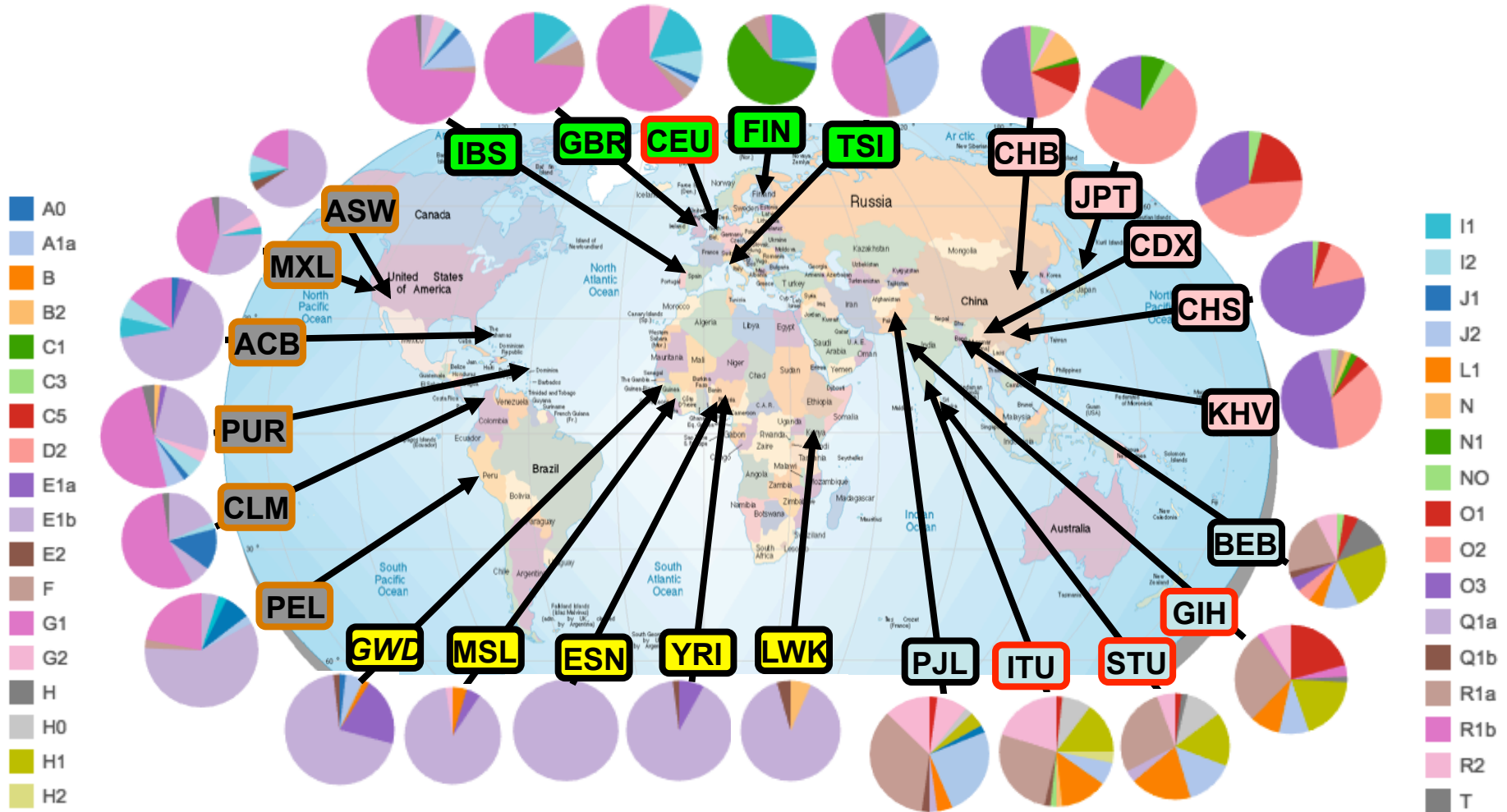


Release dataset

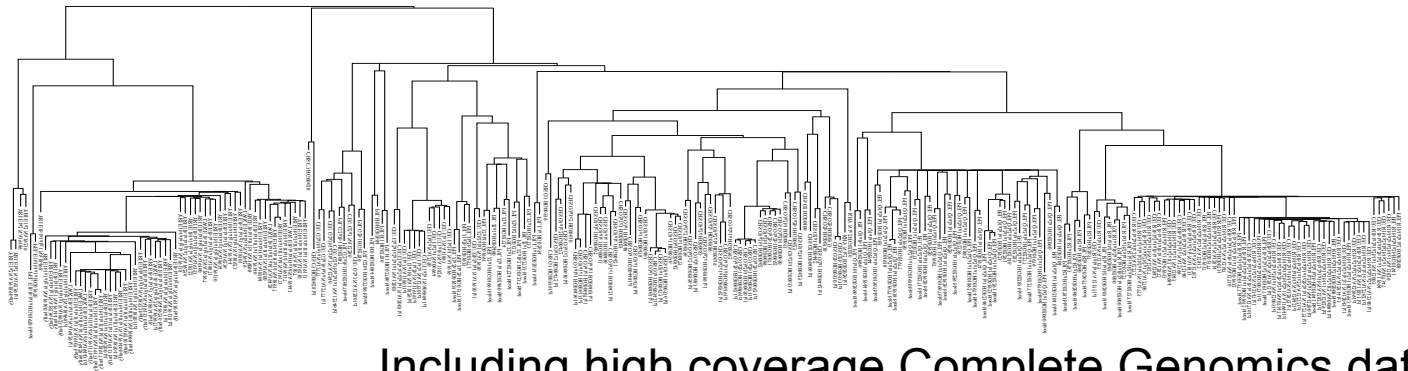
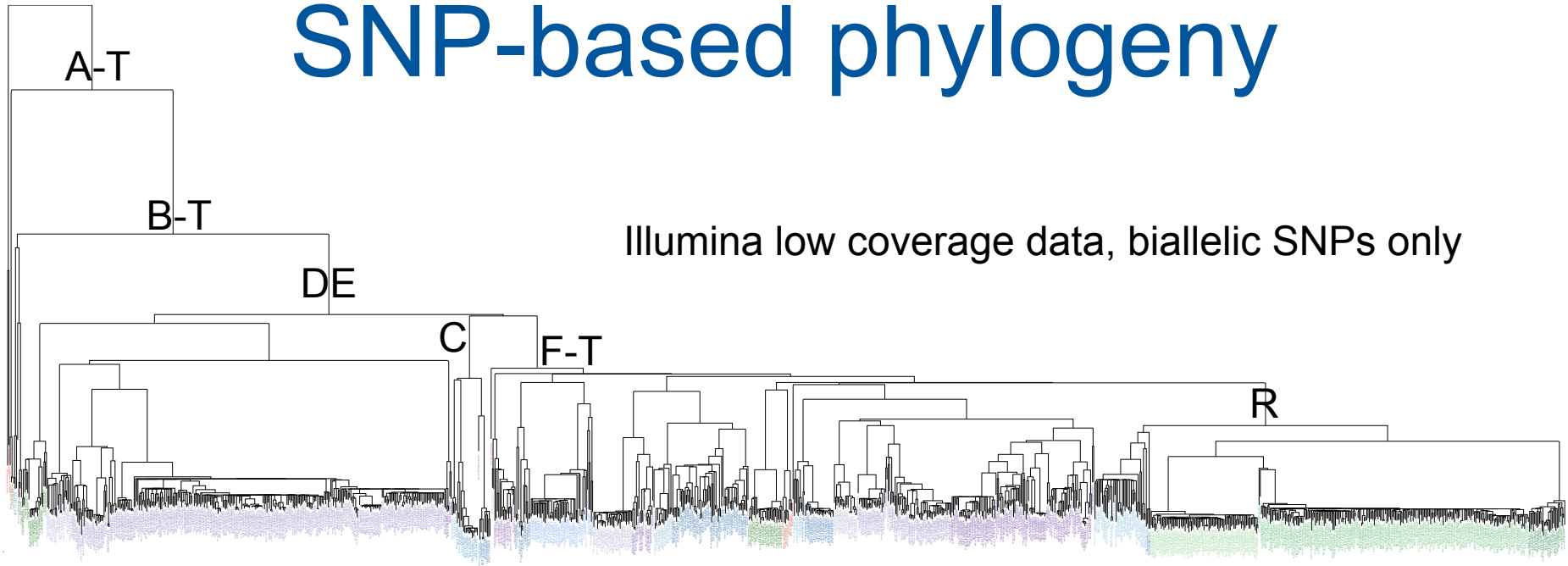
Variants	Total number	Validation dataset
Biallelic SNPs	60,555	CG data and PCR free high coverage genomes
Indels & MNPs	~1,500	PCR free high coverage genomes
STRs*	~3,000	Capillary genotyping data
SVs	~100	aCGH from Charles Lee's group & FISH

* Not included in the official release

Sample and haplogroup distribution

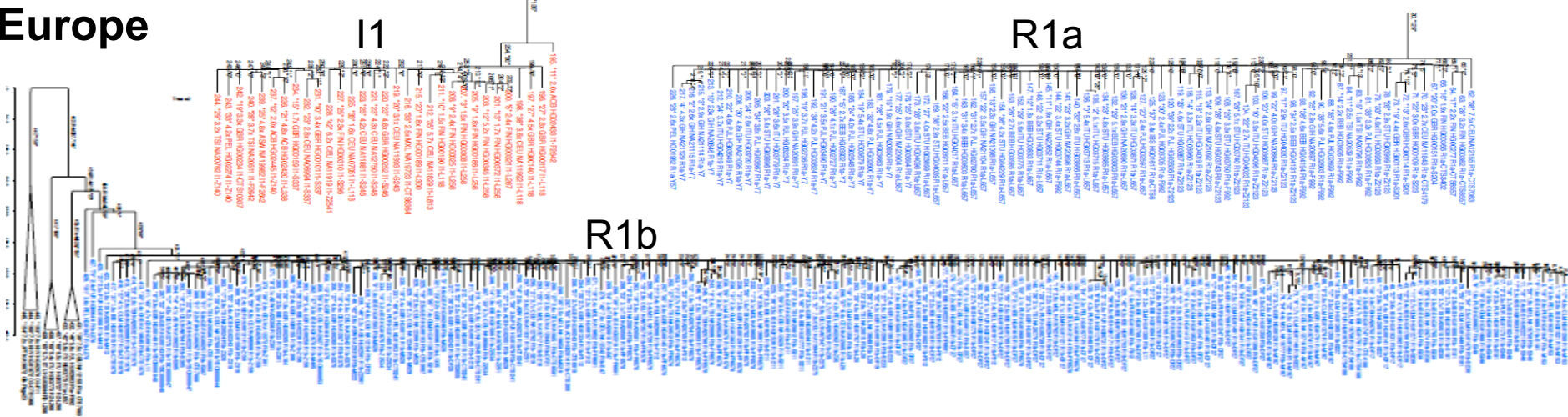


SNP-based phylogeny

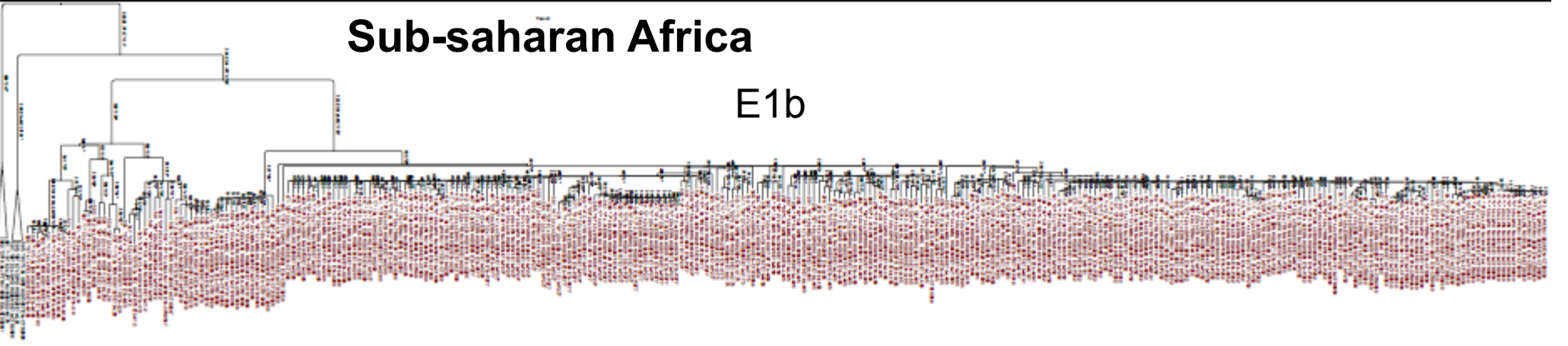


Contrasting recent expansions

Europe

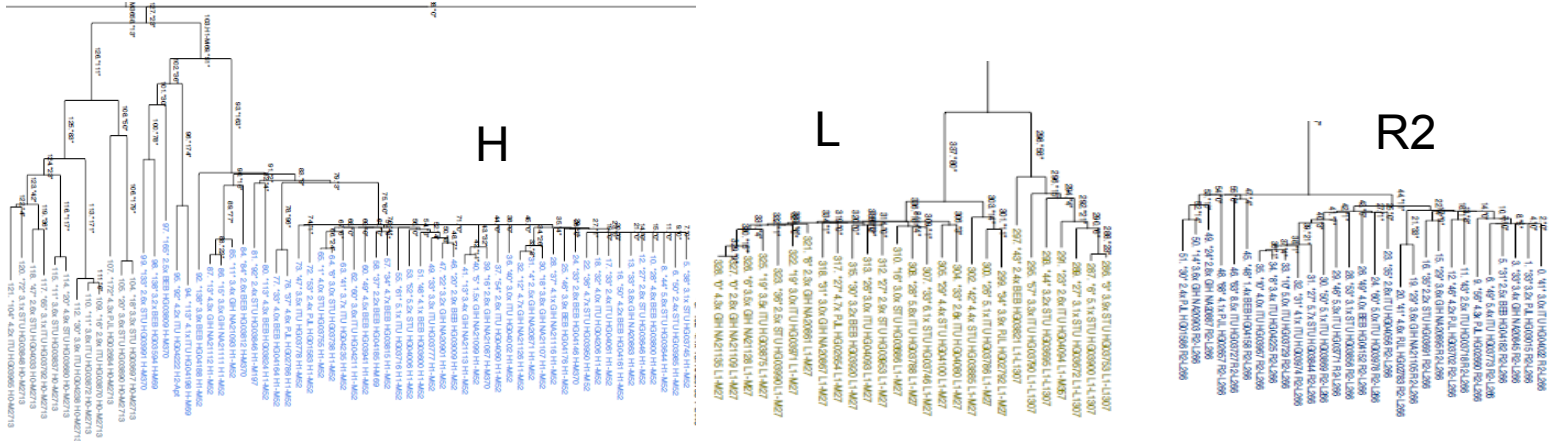


Sub-saharan Africa

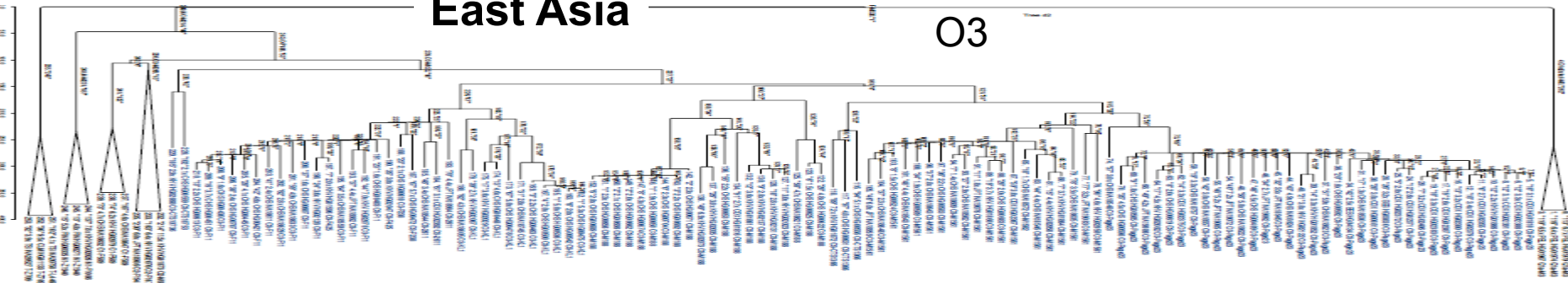


Contrasting recent expansions

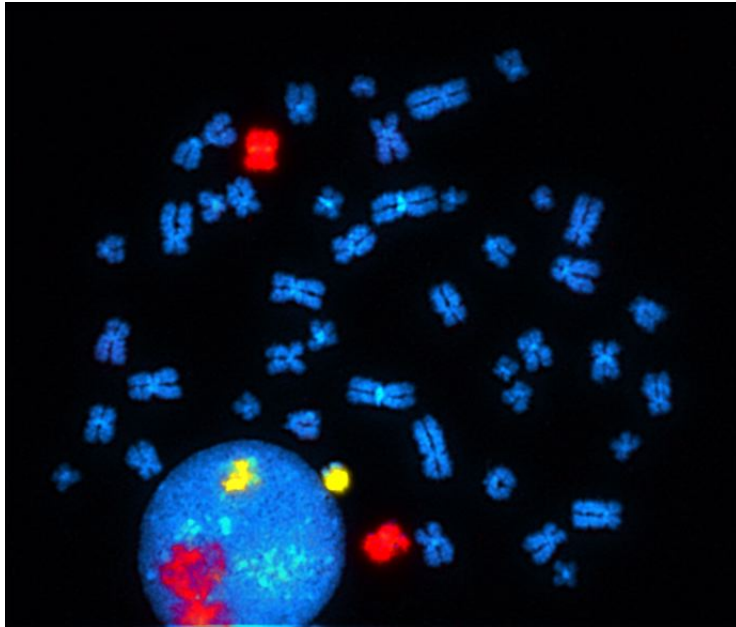
South Asia



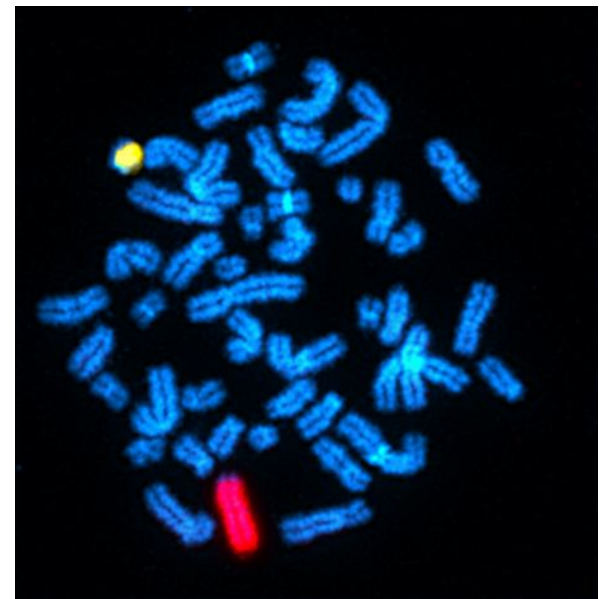
East Asia



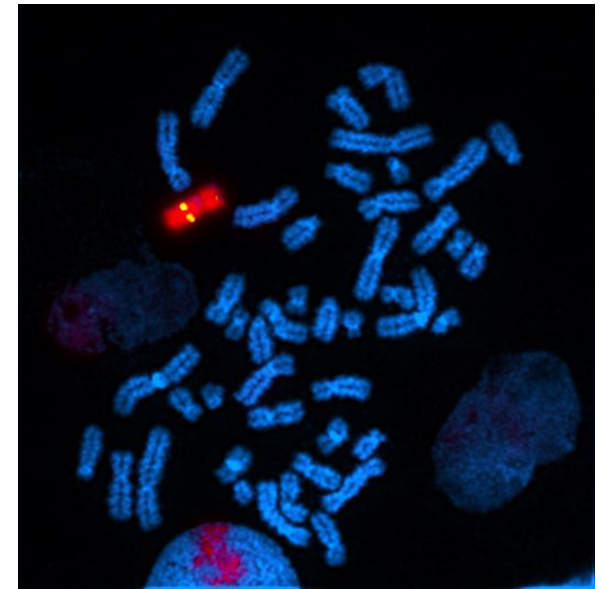
Chromosomal anomalies



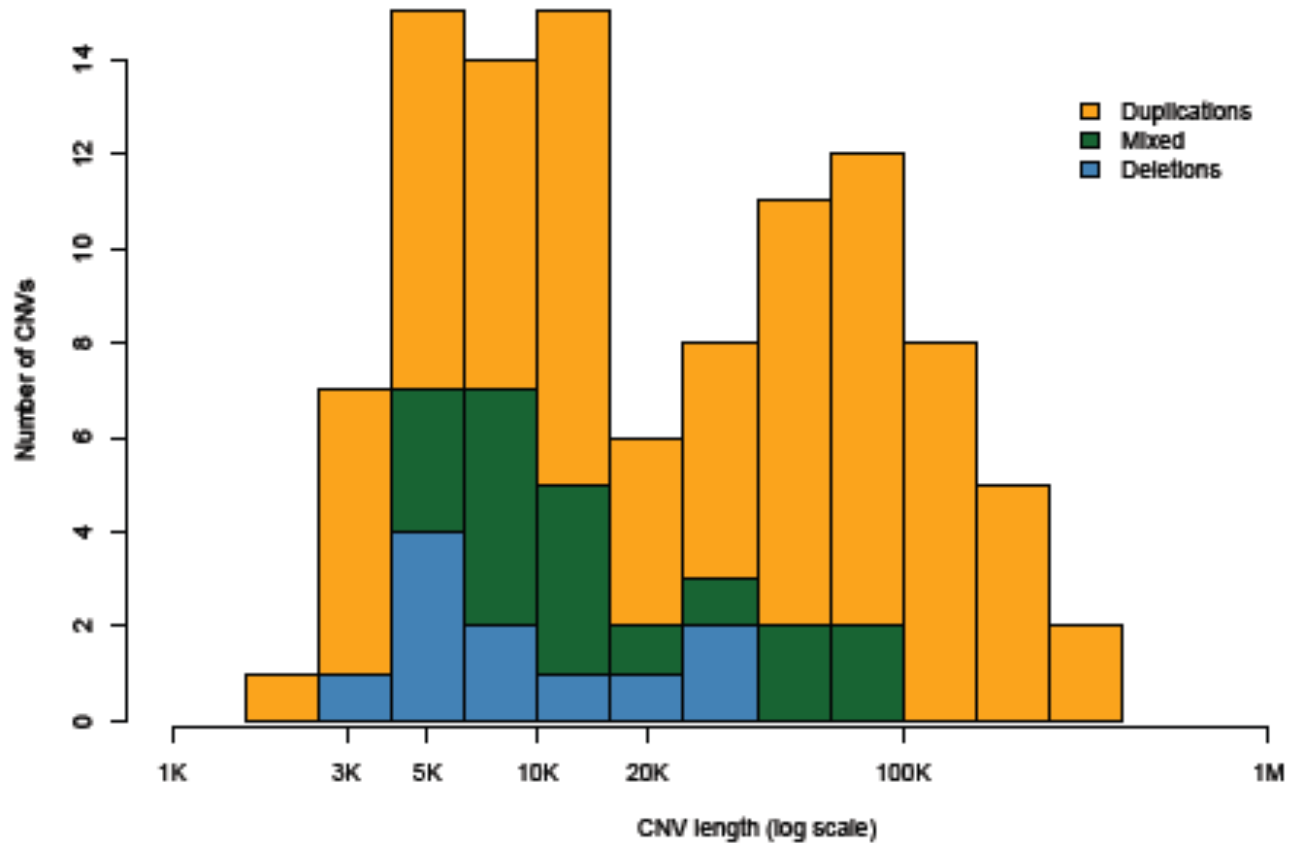
HG02372: 47,XXY (100%)
Klinefelter syndrome,
~1 in 1000 live male births,
often undiagnosed



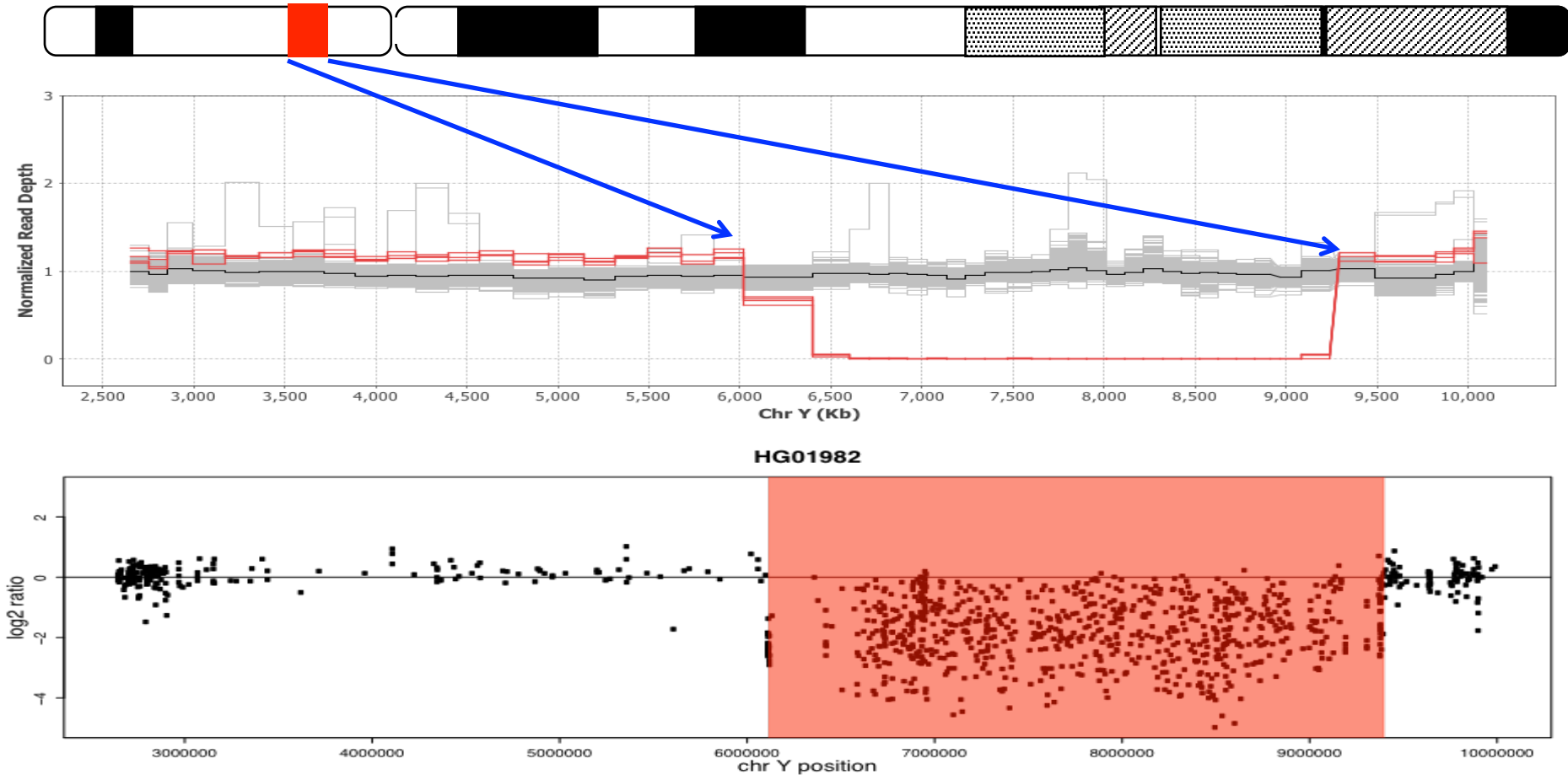
HG00246
46,XY (38%)
45,XO (62%)



SVs

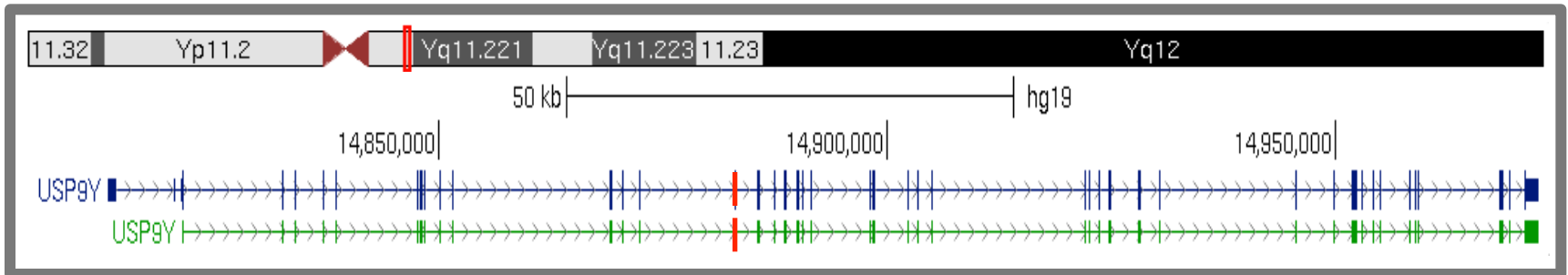
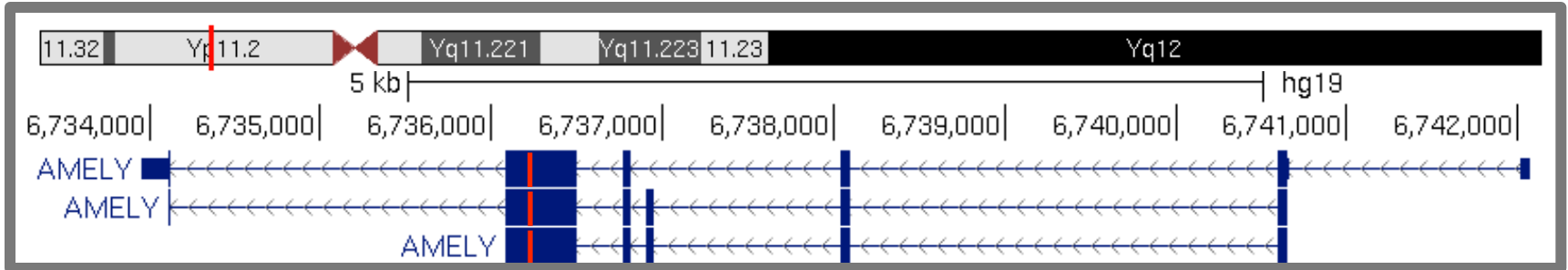


SVs



2.5 Mb deletion removing *AMELY* and other genes in 4 males (3 hg J2; 1 hg R1a)

Functional Variants



Two stop-gained SNPs, both singletons, were found

Conclusions

- Diverse high-quality variant set
- Robust phylogeny
- New insights into male history
- Data available:
 - [Link](#)

Acknowledgements: 1000Y group

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Additional members of
several of these labs

Haplogroups

	A0	A1a	B	B2	C1	C3	C5	D2	E1a	E1b	E2	F	G1	G2	H	H0	H1	H2	I1	I2	J1	J2	L1	N	N1	NO	O1	O2	O3	Q1a	Q1b	R1a	R1b	R2	T
###	2	3	3	4	4	10	17	20	20	298	5	1	2	17	7	12	41	2	31	17	15	61	27	7	25	1	19	75	114	42	3	86	216	29	8
ACB	47	1							2	31									3	3														7	
ASW	26									17	1								1	2														5	
BEB	42					1	2								5		10					5	2				2	2		1	9			3	
CDX	50					2																				10	22	16							
CEU	49													3					8	4	1	1									2	30			
CHB	46					3								1										5	1	5	7	23				1			
CHS	52					1																				2	8	41							
CLM	43										8									1	6								3				24	1	
ESN	53										53																								
FIN	38																		9	1	1				23					3	1				
GBR	46																		6	1		1								4	34				
GIH	58						12						2		1		11					5	5							16	1	5			
GWD	55	1	3	1					11	38	1																								
IBS	54									2			2							2	1	6								1	39		1		
ITU	60						1									5	9	2								1				1	16		12		
JPT	56				4	2		20																			20	10							
KHV	46					1						1													1	1	2	16	22	2					
LWK	45			3						40	2																								
MSL	42		2						2	37				1																					
MXL	33									5			2						1									10				14		1	
PEL	41									2									1		3	1						24		1	9				
PJL	48						1						4		1	2					1	12	2					1	1	17			6		
PUR	54			1					1	14			2							3	1	3										27		2	
STU	55					1									1	6	9											2			15		3		
TSI	53									4				2					2		1	15								2	24			3	
YRI	52								4	47	1																								