

## 9 Tables

A105 Chr Y	SVs (InDels+SNPs)	SNPs	%age representation SVs	%age representation SNPs
<b>A105A</b>	309158	2208	100	100
<b>A105B</b>	1376	525	0.4450798621	23.77717391
<b>A105C</b>	1600	534	0.5175347233	24.18478261
<b>A105D</b>	1195	502	0.3865337465	22.73550725
	Average =		<b>0.4497161106</b>	<b>23.56582126</b>

**Table A:** SVs highly conserved in terms of number for Y-chromosome with mean 0.45% than SNPs 23.57%. Percentage is calculated keeping the number for A105A (father) as the reference.

	Genome	Break		Plot	Data		
Stringent	parameters			Lenient	parameters		
A105A	A105B	A105C	A105D	A105A	A105B	A105C	A105D
331 A	331 A	131 T	339 A	331 A	331 A	131 T	339 A
493 A	1476 G	750 A	6474 A	493 A	15380 A	750 A	6474 A
16496 G	1518 C	4769 A	6497 T	1476 G	15408 A	4769 A	6497 T
<b>16519 T</b>	15380 A	<b>16519 T</b>	15476 C	<b>16519 T</b>	16220 A	<b>16519 T</b>	15476 C
16527 C	15408 A			16496 G	16249 T		
	16220 A			1518 C	16437 T		
	16249 T			16527 C	16469 T		
	16437 T						
	16469 T						

**Table B: GenomeBreak findings:** SNP of mother mtDNA not inherited by a female offspring, but found similar to that of father mtDNA

	Genome	Break			Plot	Data		
Stringent	parameters				Lenient		parameters	
A105A	A105B	A105C	A10 5D		A105A	A105 B	A105C	A10 5D
3107 N .	3107 N .	3107 N .	310 7 N .		3107 N .	3107 N .	3107 N .	310 7 N .
314 . C	314 . C	Missing	314 . C		314 . C	314 . C	Missing	314 . C
522 C .		4824 . N			522 C . .			
523 A .					523 A . .			

**Table C: GenomeBreak findings - a SV of mother mtDNA not present in a female offspring**

A105				
	SVs (InDels+SNPs)	SNPs	Statistics	Value
<b>A105A</b>	47083259	2001074	SD SVs	4018934.7
<b>A105B</b>	49139752	2022063	SD SNPs	24543.9
<b>A105C</b>	56138900	2021242	Mean SVs	51318148.3
<b>A105D</b>	52910682	2059879	Mean SNPs	2026064.5

**Table D:** The Standard Deviation of SNPs being far smaller than that of SVs for whole genome

## 10 Figure Legends

**Figure A:** Top to bottom: A105A, A105B, A105C, A105D. The large breaks and differences between the graphs are visible to naked eye, though much of the variations can be found when zoomed in.

**Figure B:** SVs & SNPs from only Chr 20

**Figure C:** Genome Wide SVs and SNPs: SNPs are more conserved genome wide

**Figure D:** SVs at Y-Chr homology exhibit higher selection pressure

**Figure E:** SNPs at Y-Chr homology exhibit lower selection pressure, yet good enough to distinguish gender

**Figure F:** Variations in Genome Architecture

**Figure G:** Snippet of SNP OMIM database match

**Figure H:** Snippet of Clinically relevant SNPs using OMIM

**Figure I:** ClinVar matches A105A

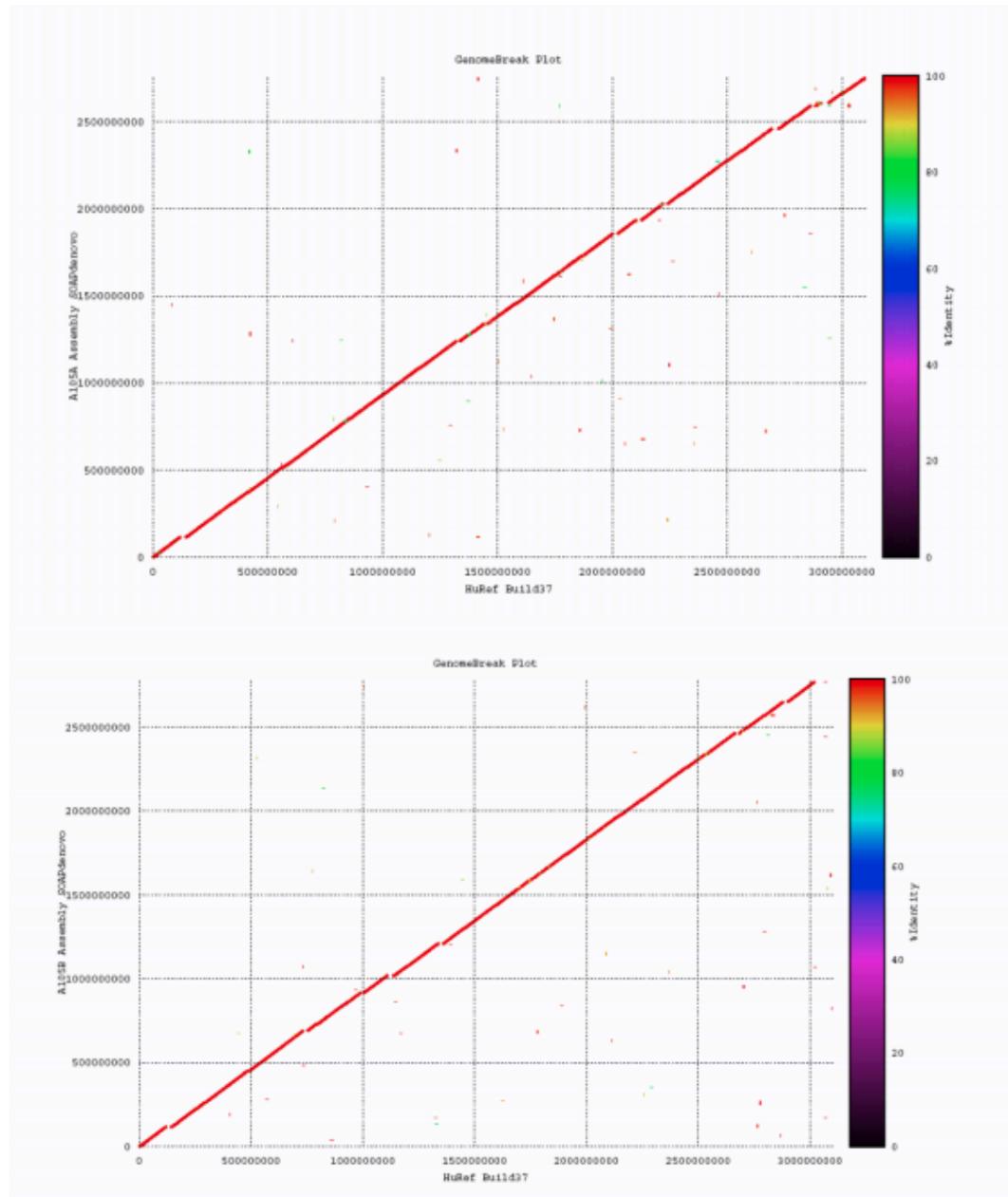
**Figure J:** ClinVar SNPs for A105A

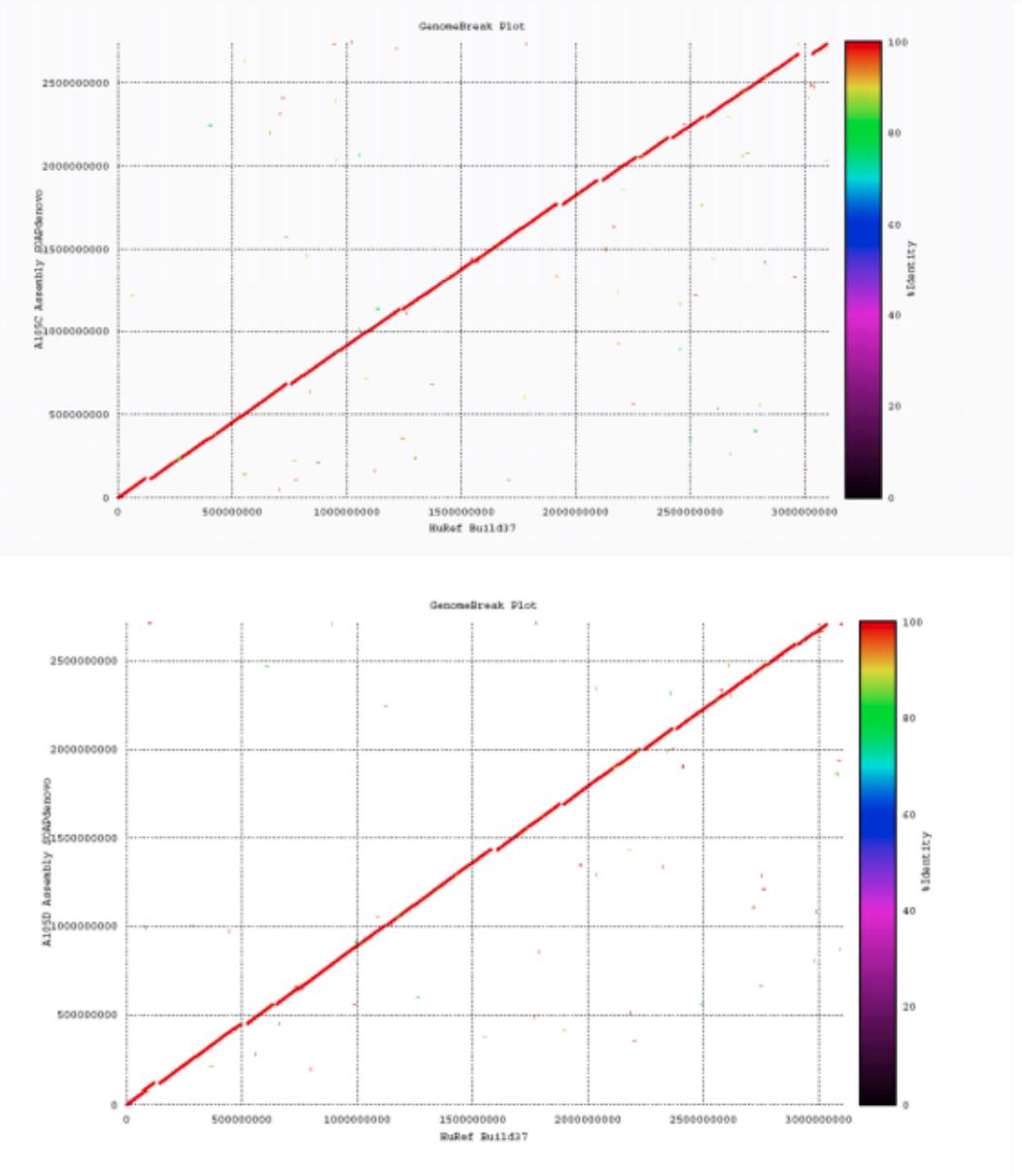
**Figure K:** ClinVar matches A105B

**Figure L:** ClinVar matches A105C

**Figure M:** Novel ClinVar SNPs in A105C

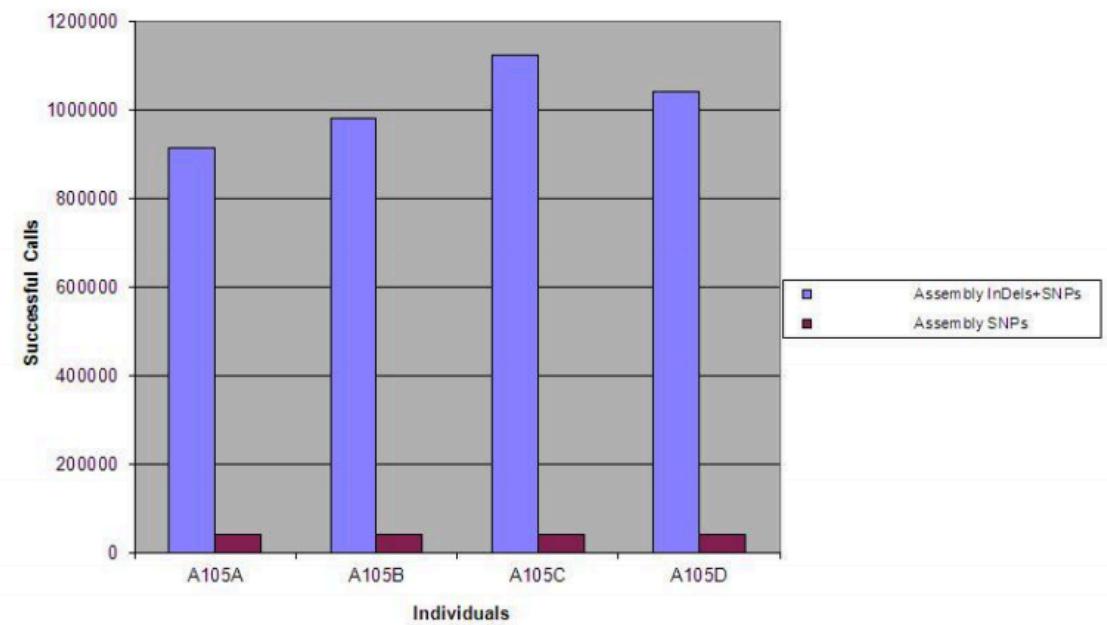
**Figure N:** Novel ClinVar SNPs in A105D



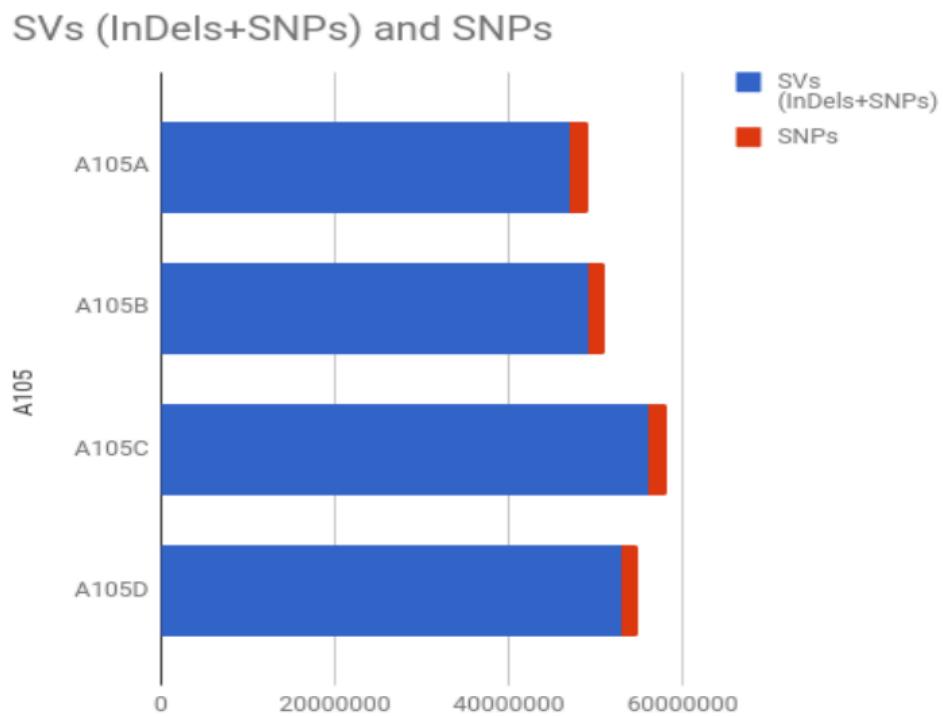


**Figure A: Top to bottom: A105A, A105B, A105C, A105D.** The large breaks and differences between the graphs are visible to naked eye, though much of the variations can be found when zoomed in.

A105 Family Chromosome 20 Decoded

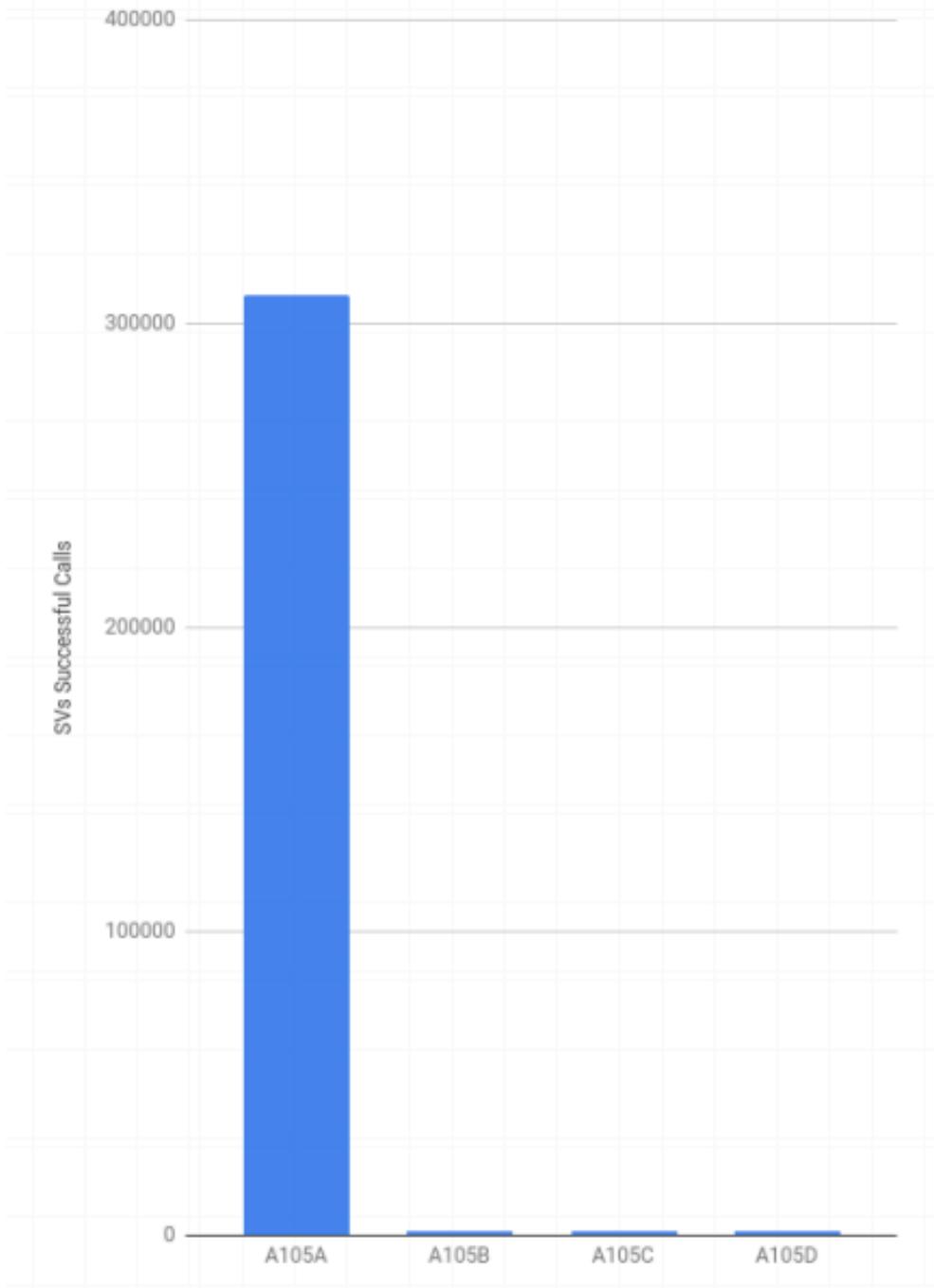


**Figure B:** SVs & SNPs from only Chr 20



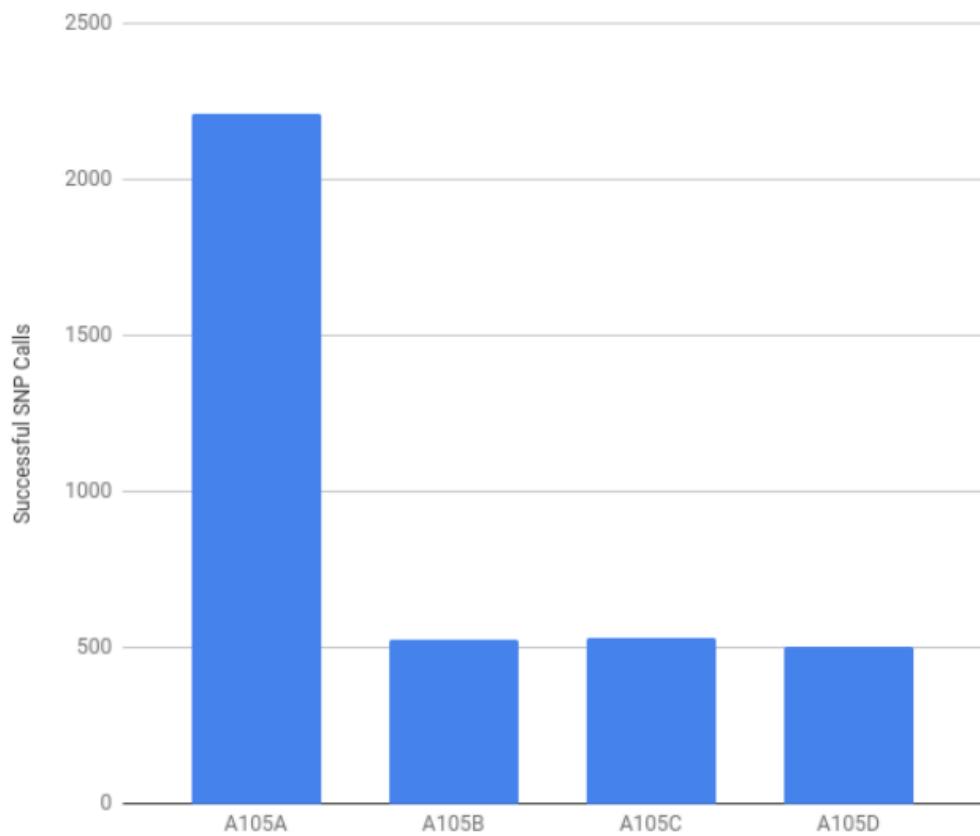
**Figure C: Genome Wide SVs and SNPs: SNPs are more conserved genome wide**

## Sex Determination by Y chromosome

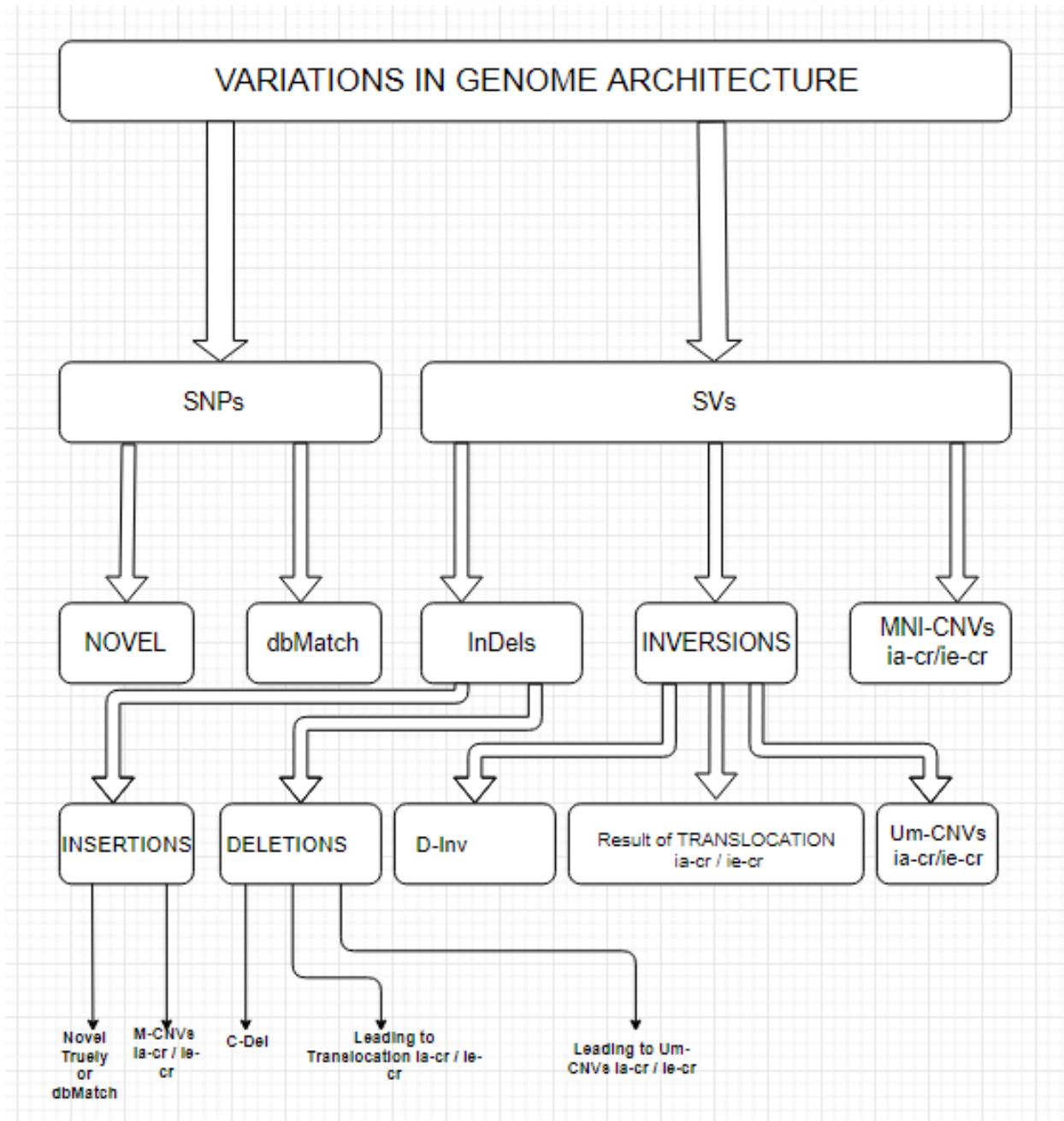


**Figure D: SVs at Y-Chr homology exhibit higher selection pressure**

## Sex Determination by Y chromosome



**Figure E: SNPs at Y-Chr homology exhibit lower selection pressure, yet good enough to distinguish gender**



**Figure F:** Variations in Genome Architecture. Um= Un-matching, M= Matching, MNI = Matching Non-Insertion, ia-cr = intra-chromosomal = tandem duplication, ie-cr = inter-chromosomal, SNPs = Single Nucleotide Polymorphism = SNV = Single Nucleotide Variations, InDels = Insertions and Deletions, CNVs= Copy Number Variations, Translocation = Single copy match elsewhere in genome, Tandem Duplication & Multiplication is a subset of various types of CNVs, Mobile element insertion is a subset of M-CNVs, C-Del = Complete Deletion, D-Inv = Direct Inversion. The classification is done based on the characteristic of variation and not on basis of sizes of variations. Figure plotted using [www.draw.io](http://www.draw.io).

#	A	B	C	D	E	F	G	H	I	J	K	L	M	N	O	P	Q	R	S	
	#Individu	inDBSNP	chromo	position	reference	sampleGe	sampleAll	alleles	DBSNP	accession	functionG	functionD	rS10	aminoAcid	proteinPosition	geneList	hasGenotypes	dbSNPValidation	clinicalAssociation	proteinSequ
2	1:10321	none	1	10321 C	T	T/T	NA	none	upstream-unknown	0	none	NA	none	no	NA	unknown	NA	unknown	NA	
3	1:14937	none	1	14937 T	C	C/C	NA	NR_02454	intron- unknown	0	none	NA	WASH7P	no	NA	unknown	NA	unknown	NA	
4	1:126113	dbSNP_13	1	126113 C	A	A/A	C/A	none	intergenic-unknown	79114531	none	NA	none	yes	by-frequency,by-cluster,by-	unknown	NA	unknown	NA	
5	1:158006	dbSNP_13	1	158006 A	T	T/T	A/T	none	intergenic-unknown	3.71E+08	none	NA	none	no	NA	unknown	NA	unknown	NA	
6	1:165163	none	1	165163 G	T	T/T	NA	none	intergenic-unknown	0	none	NA	none	no	NA	unknown	NA	unknown	NA	
7	1:168964	dbSNP_13	1	168964 A	G	G/G	A/G	none	intergenic-upstream	1.87E+08	none	NA	none	no	NA	unknown	NA	unknown	NA	
8	1:172595	dbSNP_13	1	172595 G	A	A/A	G/A	none	intergenic-unknown	1.87E+08	none	NA	none	no	NA	unknown	NA	unknown	NA	
9	1:173173	dbSNP_13	1	173173 A	G	G/G	A/G	none	intergenic-unknown	2.02E+08	none	NA	none	no	NA	unknown	NA	unknown	NA	
10	1:230058	dbSNP_12	1	230058 T	G	G/G	T/G	none	intergenic-unknown	62635260	none	NA	none	yes	by-frequency	unknown	NA	unknown	NA	
11	1:235844	dbSNP_13	1	235844 C	G	G/G	C/G	none	intergenic-unknown	1.88E+08	none	NA	none	no	NA	unknown	NA	unknown	NA	
12	1:235976	dbSNP_13	1	235976 C	A	A/A	C/A	none	intergenic-unknown	2.02E+08	none	NA	none	no	NA	unknown	NA	unknown	NA	
13	1:236055	none	1	236055 T	G	G/G	NA	none	intergenic-unknown	0	none	NA	none	no	NA	unknown	NA	unknown	NA	
14	1:918463	dbSNP_13	1	918463 G	A	A/A	G/A	none	downstre-unknown	1.14E+08	none	NA	none	no	NA	unknown	NA	unknown	NA	
15	1:967627	dbSNP_13	1	967627 T	C	C/C	T/C	none	upstream-upstream	1.87E+08	none	NA	none	no	NA	unknown	NA	unknown	NA	
16	1:967631	dbSNP_13	1	967631 A	G	G/G	A/G	none	upstream-upstream	3.77E+08	none	NA	none	no	NA	unknown	NA	unknown	NA	
17	1:70631	dbSNP_13	1	70631 G	A	A/A	G/A	NR_03390	intron-intron-var	6.99E+08	none	NA	LOC100288069	no	NA	unknown	NA	unknown	NA	
18	1:75348	dbSNP_10	1	75348 T	G	G/G	T/A/G	none	upstream-upstream	3131984	none	NA	none	yes	by-cluster,by-2hit-2allele,by	unknown	NA	unknown	NA	
19	1:752092	dbSNP_13	1	752092 G	A	A/A	G/A	none	intergenic-unknown	3.73E+08	none	NA	none	no	NA	unknown	NA	unknown	NA	
20	1:725094	none	1	725094 A	C	C/C	NA	none	intergenic-unknown	0	none	NA	none	no	NA	unknown	NA	unknown	NA	
21	1:725096	none	1	725096 G	C	C/C	NA	none	intergenic-unknown	0	none	NA	none	no	NA	unknown	NA	unknown	NA	
22	1:725097	dbSNP_13	1	725097 G	A	A/A	G/A	none	intergenic-unknown	3.75E+08	none	NA	none	no	NA	unknown	NA	unknown	NA	
23	1:725104	dbSNP_13	1	725104 C	T	T/T	C/A	none	intergenic-unknown	2.01E+08	none	NA	none	no	NA	unknown	NA	unknown	NA	
24	1:725106	dbSNP_13	1	725106 C	A	A/A	C/G	none	intergenic-unknown	2.01E+08	none	NA	none	no	NA	unknown	NA	unknown	NA	
25	1:725108	none	1	725108 A	C	C/C	NA	none	intergenic-unknown	0	none	NA	none	no	NA	unknown	NA	unknown	NA	
26	1:725117	none	1	725117 G	A	A/A	NA	none	intergenic-unknown	0	none	NA	none	no	NA	unknown	NA	unknown	NA	
27	1:725119	none	1	725119 A	G	G/G	NA	none	intergenic-unknown	0	none	NA	none	no	NA	unknown	NA	unknown	NA	
28	1:725124	none	1	725124 A	C	C/C	NA	none	intergenic-unknown	0	none	NA	none	no	NA	unknown	NA	unknown	NA	
29	1:725131	none	1	725131 C	A	A/A	NA	none	intergenic-unknown	0	none	NA	none	no	NA	unknown	NA	unknown	NA	
30	1:226481	dbSNP_10	1	226481 T	G	G/G	T/G	none	intergenic-unknown	7343980	none	NA	none	no	NA	unknown	NA	unknown	NA	
31	1:741579	dbSNP_12	1	741579 T	C	C/C	T/C	none	intergenic-unknown	6170168	none	NA	none	no	NA	unknown	NA	unknown	NA	
32	1:927221	dbSNP_10	1	927221 A	G	G/G	A/G	none	upstream-upstream	3131972	none	NA	none	yes	by-frequency,by-cluster,by-	unknown	NA	unknown	NA	

Figure G: Snippet of SNP OMIM database match

#	A	B	C	D	E	F	G	H	I	J	K	L	M	N	O	P	Q	R	S	T	
	#Indivi	inDBSNP	chromo	positio	referen	sample	sample	alle	DBSNP	accessi	functio	functio	rS10	amino	protein	geneList	hasG	dbSNPn	clinica	protei	quenc
7	1:107198	T	1	107198 C	C	C/C	T/C	NR_03886	non-codin nc-transcr	11260603	none	NA	LOC254099	yes	by-freqe	http://www.ncbi.nlm.nih.gov/pubmed?term=233826	NA	NA	NA		
9	1:1265154	dbSNP_79	1	1265154 T	C	C/C	T/C	none	upstream- unknown	307355	none	NA	none	yes	by-cluster,	http://omim.org/entry/605865	NA	NA	NA		
0	1:1265402	dbSNP_10	1	1265402 T	C	C/C	T/C	none	upstream-unknown	35744813	none	NA	none	yes	by-cluster,	http://omim.org/entry/605865	NA	NA	NA		
1	1:2513216	dbSNP_86	1	2513216 C	T	T/T	C/T	none	upstream-intron-var	7343999	none	NA	none	yes	by-freqe	http://www.ncbi.nlm.nih.gov/pubmed?term=212976	NA	NA	NA		
3	1:252746	dbSNP_10	1	252746 A	G	G/G	A/G	XM_0334	missense missense	3748816	MET,THR	518/780	MME1	yes	by-freqe	http://www.ncbi.nlm.nih.gov/pubmed?term=201907_XP_005244853.2	NA	NA	NA		
4	1:252746	dbSNP_10	1	252746 A	G	G/G	A/G	XM_00524	missense unknown	3748816	MET,THR	592/854	MME1	yes	by-freqe	http://www.ncbi.nlm.nih.gov/pubmed?term=201907_XP_005244853.1	NA	NA	NA		
9	1:2553624	dbSNP_10	1	2553624 T	C	C/C	T/C	XM_00524	intron unknown	3890745	none	NA	MME1	yes	by-freqe	http://omim.org/entry/180300 http://www.ncbi.nlm	NA	NA	NA		
0	1:2553624	dbSNP_10	1	2553624 T	C	C/C	T/C	XM_00524	intron unknown	3890745	none	NA	MME1	yes	by-freqe	http://omim.org/entry/180300 http://www.ncbi.nlm	NA	NA	NA		
8	1:2709164	dbSNP_11	1	2709164 C	A	A/A	C/A	none	upstream-unknown	4648356	none	NA	none	yes	by-freqe	http://www.ncbi.nlm.nih.gov/pubmed?term=21830	NA	NA	NA		
0	1:2878366	dbSNP_12	1	2878366 G	A	A/A	G/A	none	intergenic-unknown	11583804	none	NA	none	yes	by-freqe	http://omim.org/entry/613238	NA	NA	NA		
7	1:4315204	dbSNP_86	1	4315204 G	T	T/T	G/T	none	intergenic-unknown	9663321	none	NA	none	yes	by-freqe	http://www.ncbi.nlm.nih.gov/pubmed?term=217902	NA	NA	NA		
3	1:4444539	dbSNP_79	1	4444539 G	T	T/T	G/T	none	intergenic-unknown	2339339	none	NA	none	yes	by-freqe	http://www.ncbi.nlm.nih.gov/pubmed?term=233776	NA	NA	NA		
5	1:5354844	dbSNP_86	1	5354844 C	T	T/T	C/T	none	intergenic-unknown	912888	none	NA	none	yes	by-freqe	http://www.ncbi.nlm.nih.gov/pubmed?term=233776	NA	NA	NA		
2	1:5661009	dbSNP_11	1	5661009 T	A	A/A	T/A	none	intergenic-unknown	7528859	none	NA	none	yes	by-freqe	http://www.ncbi.nlm.nih.gov/pubmed?term=237930	NA	NA	NA		
8	1:5707816	dbSNP_11	1	5707816 C	T	T/T	C/T	none	intergenic-unknown	4845812	none	NA	none	yes	by-freqe	http://www.ncbi.nlm.nih.gov/pubmed?term=233190	NA	NA	NA		
1	1:5913621	dbSNP_83	1	5913621 T	C	C/C	T/C	none	intergenic-unknown	548726	none	NA	none	yes	by-freqe	http://www.ncbi.nlm.nih.gov/pubmed?term=233064	NA	NA	NA		
6	1:6631431	dbSNP_12	1	6631431 C	T	T/T	C/T	NM_13865	intron-intron-var	11587438	none	NA	TAS1R1	yes	by-freqe	http://www.ncbi.nlm.nih.gov/pubmed?term=217384	NA	NA	NA		
7	1:6631431	dbSNP_12	1	6631431 C	T	T/T	C/T	NM_1775	intron-intron-var	11587438	none	NA	TAS1R1	yes	by-freqe	http://www.ncbi.nlm.nih.gov/pubmed?term=217384	NA	NA	NA		
28	1:6631431	dbSNP_12	1	6631431 C	T	T/T	C/T	NM_00526	intron-unknown	11587438	none	NA	TAS1R1	yes	by-freqe	http://www.ncbi.nlm.nih.gov/pubmed?term=217384	NA	NA	NA		
25	1:7825311	dbSNP_86	1	7825311 G	A	A/A	G/A	XM_01521	intron-var	70455	none	NA	CAMTA1	yes	by-freqe	http://omim.org/entry/116600	NA	NA	NA		
50	1:8338355	dbSNP_80	1	8338355 G	A	A/A	G/A	none	intergenic-unknown	438895	none	NA	none	yes	by-freqe	http://www.ncbi.nlm.nih.gov/pubmed?term=225666	NA	NA	NA		
88	1:8447722	dbSNP_96	1	8447722 C	T	T/T	C/T	NM_00101	intron-var	2120461	none	NA	RERE	yes	by-freqe	http://www.ncbi.nlm.nih.gov/pubmed?term=225044	NA	NA	NA		
99	1:8447722	dbSNP_96	1	8447722 C	T	T/T	C/T	NM_00101	intron-var	2120461	none	NA	RERE	yes	by-freqe	http://www.ncbi.nlm.nih.gov/pubmed?term=225044	NA	NA	NA		
0	1:8447722	dbSNP_96	1	8447722 C	T	T/T	C/T	NM_01211	intron-var	2120461	none	NA	RERE	yes	by-freqe	http://www.ncbi.nlm.nih.gov/pubmed?term=225044	NA	NA	NA		
01	1:8447722	dbSNP_96	1	8447722 C	T	T/T	C/T	XM_00526	intron-unknown	2120461	none	NA	RERE	yes	by-freqe	http://www.ncbi.nlm.nih.gov/pubmed?term=225044	NA	NA	NA		
02	1:8447722	dbSNP_96	1	8447722 C	T	T/T	C/T	XM_00526	intron-unknown	2120461	none	NA	RERE	yes	by-freqe	http://www.ncbi.nlm.nih.gov/pubmed?term=225044	NA	NA	NA		
03	1:8447722	dbSNP_96	1	8447722 C	T	T/T	C/T	XM_00526	intron-unknown	2120461	none	NA	RERE	yes	by-freqe	http://www.ncbi.nlm.nih.gov/pubmed?term=225044	NA	NA	NA		
20	1:8526142	dbSNP_11	1	8526142 G	A	A/A	G/A	NM_00104	intron	4908760	none	NA	RERE	yes	by-freqe	http://www.ncbi.nlm.nih.gov/pubmed?term=204105	NA	NA	NA		
21	1:8526142	dbSNP_11	1	8526142 G	A	A/A	G/A	NM_01211	intron	4908760	none	NA	RERE	yes	by-freqe						

	B	C	D	E	F	G	H	I
1	Chromosome	Co-ordinates	Ref	Var	Clinical Aspect			
2	1	100672060	T	C	CLNSIG=pathogenic;CLNDBN=Intermediate_maple_syrup_urine_disease_type_2;CLNACC=RCV000012727.21			
3	1	114377568	A	G	CLNSIG=other other other other other other;CLNDBN=Diabetes_mellitus\x2c_insulin-dependent\x2c_susceptibility_to Rheumatoid_arthritis			
4	1	169519049	T	C	CLNSIG=pathogenic untested other other;CLNDBN=Thrombophilia_due_to_factor_V_Leiden Ischemic_stroke\x2c_susceptibility_to Budd-Chiari_syndrome			
5	1	193218843	C	G	CLNSIG=non-pathogenic;CLNDBN=Parathyroid_carcinoma;CLNACC=RCV000020880.2			
6	1	197031021	C	T	CLNSIG=other;CLNDBN=Venous_thrombosis\x2c_susceptibility_to;CLNACC=RCV000017984.1			
7	1	197070815	T	C	CLNSIG=non-pathogenic;CLNDBN=Primary_autosomal_recessive_microcephaly_5;CLNACC=RCV000020792.1			
8	1	197070901	A	G	CLNSIG=non-pathogenic;CLNDBN=Primary_autosomal_recessive_microcephaly_5;CLNACC=RCV000020789.1			
9	1	197091537	A	T	CLNSIG=non-pathogenic;CLNDBN=Primary_autosomal_recessive_microcephaly_5;CLNACC=RCV000020768.1			
10	1	197094030	C	T	CLNSIG=non-pathogenic;CLNDBN=Primary_autosomal_recessive_microcephaly_5;CLNACC=RCV000020763.1			
11	1	203194186	C	T	CLNSIG=pathogenic;CLNDBN=Chitotriosidase_deficiency;CLNACC=RCV000010134.1			
12	1	216172380	A	G	CLNSIG=non-pathogenic;CLNDBN=AllHighlyPenetrant;CLNACC=RCV000041888.1			
13	1	216371934	A	C	CLNSIG=non-pathogenic;CLNDBN=AllHighlyPenetrant;CLNACC=RCV000041834.1			
14	1	216595306	C	T	CLNSIG=non-pathogenic;CLNDBN=AllHighlyPenetrant;CLNACC=RCV000041833.1			
15	1	223285200	G	A	CLNSIG=other pathogenic pathogenic;CLNDBN=Legionellosis\Systemic_lupus erythematosus\x2c_resistance_to\x2c_1 Melioidosis\x2c_septicemia			
16	1	46870761	C	A	CLNSIG=other;CLNDBN=Drug_addiction\x2c_susceptibility_to;CLNACC=RCV000007116.2			
17	1	47767914	G	A	CLNSIG=non-pathogenic;CLNDBN=Primary_autosomal_recessive_microcephaly_7;CLNACC=RCV000020526.1			
18	1	55518316	C	T	CLNSIG=non-pathogenic;CLNDBN=Familial_hypercholesterolemia;CLNACC=RCV000030351.1			
19	1	63872032	T	C	CLNSIG=pathogenic non-pathogenic;CLNDBN=Congenital_disorder_of_glycosylation_type_1C AllHighlyPenetrant;CLNACC=RCV000023375			
20	1	66058513	A	G	CLNSIG=non-pathogenic;CLNDBN=LEPTIN_RECECTOR_POLYMORPHISM;CLNACC=RCV000009047.1			
21	1	66075952	G	C	CLNSIG=non-pathogenic;CLNDBN=LEPTIN_RECECTOR_POLYMORPHISM;CLNACC=RCV000009050.1			
22	1	98348885	G	A	CLNSIG=pathogenic;CLNDBN=Dihydropyrimidine_dehydrogenase_deficiency;CLNACC=RCV000000464.1			
23	19	57328825	C	T	CLNSIG=untested;CLNDBN=Malignant_melanoma;CLNACC=RCV000063653.2			
24	3	12645699	G	A	CLNSIG=pathogenic pathogenic pathogenic;CLNDBN=Noonan_syndrome_5 LEOPARD_syndrome_2 LEOPARD_syndrome;CLNACC=RCV000007985.1,RCV000007985.2			
25	4	2833309	T	A	CLNSIG=other;CLNDBN=Fibrous_dysplasia_of_jaw,Fibrous_dysplasia_of_jaw,Fibrous_dysplasia_of_jaw;CLNACC=RCV0000074005.1			
26	5	112130448	G	A	CLNSIG=other;CLNDBN=Familial_colorectal_cancer;CLNACC=RCV0000074005.1			
27	9	71836364	G	T	CLNSIG=unknown;CLNDBN=AllHighlyPenetrant;CLNACC=RCV000037077.1			

**Figure I:** ClinVar matches A105A

	A	B	C	D	E	F	G	H	I	J	K	L	M	N	O	P	Q	R	S	T	U	V
1	Chromosome	Coordinates	Ref Seq	Var Seq	Clinical Aspect																	
2	1	100672060	T	C	CLNSIG=pathogenic;CLNDBN=Intermediate_maple_syrup_urine_disease_type_2;CLNACC=RCV000012727.21																	
3	1	11854476	T	G	CLNSIG=non-pathogenic other;CLNDBN=MTHFR_deficiency\x2c_thermolabile_type Schizophrenia\x2c_susceptibility_to;CLNACC=RCV000003698.1 RCV000003699.1																	
4	1	159175354	G	A	CLNSIG=non-pathogenic;CLNDBN=DUFFY_BLOOD_GROUP_SYSTEM\x2c_FYA/FYB_POLYMORPHISM;CLNACC=RCV000000005.1																	
5	1	161479745	A	G	CLNSIG=other other other;CLNDBN=Lupus_nephritis\x2c_susceptibility_to Pseudomonas_aeruginosa\x2c_susceptibility_to_chronic_infection_by\x2c_in_cystic_fibrosis Malaria\x2c_septicemia																	
6	1	169519049	T	C	CLNSIG=pathogenic untested other other;CLNDBN=Thrombophilia_due_to_factor_V_Leiden Ischemic_stroke\x2c_susceptibility_to Budd-Chiari_syndrome\x2c_susceptibility_to Recurrent_thrombosis																	
7	1	197031021	C	T	CLNSIG=other;CLNDBN=Venous_thrombosis\x2c_susceptibility_to;CLNACC=RCV000017984.1																	
8	1	197070815	T	C	CLNSIG=non-pathogenic;CLNDBN=Primary_autosomal_recessive_microcephaly_5;CLNACC=RCV000020792.1																	
9	1	197070901	A	G	CLNSIG=non-pathogenic;CLNDBN=Primary_autosomal_recessive_microcephaly_5;CLNACC=RCV000020789.1																	
10	1	197091537	A	T	CLNSIG=non-pathogenic;CLNDBN=Primary_autosomal_recessive_microcephaly_5;CLNACC=RCV000020768.1																	
11	1	197094030	C	T	CLNSIG=non-pathogenic;CLNDBN=Primary_autosomal_recessive_microcephaly_5;CLNACC=RCV000020763.1																	
12	1	203194186	C	T	CLNSIG=pathogenic;CLNDBN=Chitotriosidase_deficiency;CLNACC=RCV000010134.1																	
13	1	215848062	C	T	CLNSIG=non-pathogenic;CLNDBN=AllHighlyPenetrant;CLNACC=RCV0000041736.1																	
14	1	215901492	C	T	CLNSIG=non-pathogenic;CLNDBN=AllHighlyPenetrant;CLNACC=RCV0000041709.1																	
15	1	216172380	A	G	CLNSIG=non-pathogenic;CLNDBN=AllHighlyPenetrant;CLNACC=RCV0000041888.1																	
16	1	216371934	A	C	CLNSIG=non-pathogenic;CLNDBN=AllHighlyPenetrant;CLNACC=RCV0000041834.1																	
17	1	24180962	T	C	CLNSIG=non-pathogenic;CLNDBN=F1/F2_POLYMORPHISM;CLNACC=RCV00000726.1																	
18	1	46870761	C	A	CLNSIG=other;CLNDBN=Drug_addiction\x2c_susceptibility_to;CLNACC=RCV000007116.2																	
19	1	47767914	G	A	CLNSIG=non-pathogenic;CLNDBN=Primary_autosomal_recessive_microcephaly_7;CLNACC=RCV000020526.1																	
20	1	66058513	A	G	CLNSIG=non-pathogenic;CLNDBN=LEPTIN_RECECTOR_POLYMORPHISM;CLNACC=RCV000009047.1																	
21	1	98348885	G	A	CLNSIG=pathogenic;CLNDBN=Dihydropyrimidine_dehydrogenase_deficiency;CLNACC=RCV000000464.1																	
22	10	76285162	T	C	CLNSIG=pathogenic;CLNDBN=Hypermethioninemia_due_to_adenosine_kinase_deficiency;CLNACC=RCV000022444.26																	
23	12	52913630	A	G	CLNSIG=untested;CLNDBN=not_provided;CLNACC=RCV0000056594.1																	
24	17	56292101	A	T	CLNSIG=probable-pathogenic;CLNDBN=Meckel_syndrome_type_1;CLNACC=RCV000050037.1																	
25	3	12645699	G	A	CLNSIG=pathogenic pathogenic pathogenic;CLNDBN=Noonan_syndrome_5 LEOPARD_syndrome_2 LEOPARD_syndrome;CLNACC=RCV000014985.20 RCV000020509.1																	

**Figure J:** ClinVar SNPs for A105A

#	A	B	C	D	E	F	G	H	I	J	K	L	M	N	O	P	Q	R	S	T	U	V
1	Chromosome Coordinates	Ref Seq	Var Seq	Clinical Aspect																		
2	1	114377568	A	G	CLNSIG=other other other other other;CLNDBN=Diabetes_mellitus x2c_insulin-dependent x2c_susceptibility_to Rheumatoid_arthritis Systemic_lupus_erythematosus x2c_susceptibility_to Reactive_arthritis Sjogren_syndrome Vasculitis Wegener_granulomatosis																	
3	1	11854476	T	G	CLNSIG=non-pathogenic other;CLNDBN=MTHFR_deficiency x2c_thermolabile_type Schizophrenia x2c_susceptibility_to;CLNACC=RCV000003698.1 RCV000003699.1																	
4	1	169519049	T	C	CLNSIG=pathogenic untested other other;CLNDBN=Thrombophilia_due_to_factor_V_Leiden Ischemic_stroke x2c_susceptibility_to Budd-Chiari_syndrome x2c_susceptibility_to Reactive_arthritis Sjogren_syndrome Vasculitis Wegener_granulomatosis																	
5	1	171076966	G	A	CLNSIG=other pathogenic;CLNDBN=Trimethylaminuria x2c_mild Trimethylaminuria;CLNACC=RCV000017711.1 RCV000020653.1																	
6	1	197031021	C	T	CLNSIG=other;CLNDBN=Venous_thrombosis x2c_susceptibility_to;CLNACC=RCV000017984.1																	
7	1	197070815	T	C	CLNSIG=non-pathogenic;CLNDBN=Primary_autosomal_recessive_microcephaly_5;CLNACC=RCV000020792.1																	
8	1	197070901	A	G	CLNSIG=non-pathogenic;CLNDBN=Primary_autosomal_recessive_microcephaly_5;CLNACC=RCV000020789.1																	
9	1	197091537	A	T	CLNSIG=other pathogenic;CLNDBN=Primary_autosomal_recessive_microcephaly_5;CLNACC=RCV000020768.1																	
10	1	203194186	C	T	CLNSIG=pathogenic;CLNDBN=Chitotriosidase_deficiency;CLNACC=RCV000010134.1																	
11	1	215901492	C	T	CLNSIG=non-pathogenic;CLNDBN=All HighlyPenetrant;CLNACC=RCV000041709.1																	
12	1	216144049	C	T	CLNSIG=non-pathogenic;CLNDBN=All HighlyPenetrant;CLNACC=RCV000041897.1																	
13	1	216172380	A	G	CLNSIG=non-pathogenic;CLNDBN=All HighlyPenetrant;CLNACC=RCV000041888.1																	
14	1	216592003	T	C	CLNSIG=non-pathogenic;CLNDBN=All HighlyPenetrant;CLNACC=RCV000041854.1																	
15	1	216595306	C	T	CLNSIG=non-pathogenic;CLNDBN=All HighlyPenetrant;CLNACC=RCV000041833.1																	
16	1	47767914	G	A	CLNSIG=non-pathogenic;CLNDBN=Primary_autosomal_recessive_microcephaly_7;CLNACC=RCV000020526.1																	
17	1	66036441	A	G	CLNSIG=non-pathogenic;CLNDBN=LEPTIN_Receptor_POLYMORPHISM;CLNACC=RCV000009049.1																	
18	1	66058513	A	G	CLNSIG=non-pathogenic;CLNDBN=LEPTIN_Receptor_POLYMORPHISM;CLNACC=RCV000009047.1																	
19	1	98348885	G	A	CLNSIG=pathogenic;CLNDBN=Dihydropyrimidine_dehydrogenase_deficiency;CLNACC=RCV000000464.1																	
20	10	76285162	T	C	CLNSIG=pathogenic;CLNDBN=Hypermethioninemia_due_to_adenosine_kinase_deficiency;CLNACC=RCV000022444.26																	
21	16	28950223	C	T	CLNSIG=untested;CLNDBN=Malignant_melanoma;CLNACC=RCV000071079.2																	
22	17	56292101	A	T	CLNSIG=probable-pathogenic;CLNDBN=Meckel_syndrome_type_1;CLNACC=RCV000050037.1																	
23	2	71360250	C	T	CLNSIG=untested;CLNDBN=Malignant_melanoma;CLNACC=RCV000060600.2																	
24	3	12645699	G	A	CLNSIG=pathogenic pathogenic pathogenic;CLNDBN=Noonan_syndrome_3 LEOPARD_syndrome_2 LEOPARD_syndrome_3;CLNACC=RCV000014985.20 RCV000014986.20 RCV000020509.1																	
25	4	2833309	T	A	CLNSIG=pathogenic;CLNDBN=Fibrous_dysplasia_of_jaw,Fibrous_dysplasia_of_jaw,Fibrous_dysplasia_of_jaw;CLNACC=RCV000007985.1,RCV000007984.1,RCV000007983.1																	
26	9	71836364	G	T	CLNSIG=unknown;CLNDBN=All HighlyPenetrant;CLNACC=RCV000037077.1																	

**Figure K: ClinVar matches A105B**

	A	B	C	D	E	F	G	H	I	J	K	L	M	N	O	P	Q	R	S	T	U	V
1	Chromosome Coordinates	Ref Seq	Var Seq	Clinical Aspect																		
2	1	100672060	T	C	CLNSIG=pathogenic;CLNDBN=Intermediate_maple_syrup_urine_disease_type_2;CLNACC=RCV000012727.21																	
3	1	114377568	A	G	CLNSIG=other other other other other other;CLNDBN=Diabetes_mellitus x2c_insulin-dependent x2c_susceptibility_to Rheumatoid_arthritis Systemic_lupus erythematosus x2c_susceptibility_to																	
4	1	11854476	T	G	CLNSIG=non-pathogenic;other;CLNDBN=MTHFR_deficiency x2c_thermolabile_type Schizophrenia x2c_susceptibility_to;CLNACC=RCV000003698.1 RCV000003699.1																	
5	1	154426970	A	C	CLNSIG=other other;CLNDBN=Sерum_level_of_interleukin-6 soluble_receptor Serum_level_of_interleukin_6;CLNACC=RCV000015767.1 RCV000029243.1																	
6	1	161479745	A	G	CLNSIG=other other other;CLNDBN=Lupus_nephritis x2c_susceptibility_to Pseudomonas_aeruginosa x2c_susceptibility_to_chronic_infection_by x2c_in_cystic_fibrosis Malaria x2c_sever																	
7	1	169519049	T	C	CLNSIG=pathogenic untested other other;CLNDBN=Thrombophilia_due_to_factor_V_Leiden Ischemic_stroke x2c_susceptibility_to Budd-Chiari_syndrome x2c_susceptibility_to Recurre																	
8	1	171076966	G	A	CLNSIG=other pathogenic;CLNDBN=Trimethylaminuria x2c_mild Trimethylaminuria;CLNACC=RCV000017711.1 RCV000020653.1																	
9	1	182554557	C	T	CLNSIG=other pathogenic;CLNDBN=Prostate_cancer x2c_susceptibility_to;CLNACC=RCV000013880.2																	
10	1	193218843	C	G	CLNSIG=non-pathogenic;CLNDBN=Parathyroid_carcinoma;CLNACC=RCV000020880.2																	
11	1	197031021	C	T	CLNSIG=other;CLNDBN=Venous_thrombosis x2c_susceptibility_to;CLNACC=RCV000017984.1																	
12	1	197070815	T	C	CLNSIG=non-pathogenic;CLNDBN=Primary_autosomal_recessive_microcephaly_5;CLNACC=RCV000020792.1																	
13	1	197070901	A	G	CLNSIG=non-pathogenic;CLNDBN=Primary_autosomal_recessive_microcephaly_5;CLNACC=RCV000020789.1																	
14	1	197091537	A	T	CLNSIG=non-pathogenic;CLNDBN=Primary_autosomal_recessive_microcephaly_5;CLNACC=RCV000020768.1																	
15	1	197094030	C	T	CLNSIG=non-pathogenic;CLNDBN=Primary_autosomal_recessive_microcephaly_5;CLNACC=RCV000020763.1																	
16	1	203194186	C	T	CLNSIG=pathogenic;CLNDBN=Chitotriosidase_deficiency;CLNACC=RCV000010134.1																	
17	1	215848062	C	T	CLNSIG=non-pathogenic;CLNDBN=AllHighlyPenetrant;CLNACC=RCV000041736.1																	
18	1	215901492	C	T	CLNSIG=non-pathogenic;CLNDBN=AllHighlyPenetrant;CLNACC=RCV000041709.1																	
19	1	216172380	A	G	CLNSIG=non-pathogenic;CLNDBN=AllHighlyPenetrant;CLNACC=RCV000041888.1																	
20	1	216371934	A	C	CLNSIG=non-pathogenic;CLNDBN=AllHighlyPenetrant;CLNACC=RCV000041834.1																	
21	1	223285200	G	A	CLNSIG=other pathogenic pathogenic pathogenic;CLNDBN=Legionellosis Systemic_lupus erythematosus x2c_resistance_to x2c_1 Melioidosis x2c_resistance_to;CLNACC=RCV000007037.2 RCV0000																	
22	1	47767914	G	A	CLNSIG=non-pathogenic;CLNDBN=Primary_autosomal_recessive_microcephaly_7;CLNACC=RCV000020526.1																	
23	1	55518316	C	T	CLNSIG=non-pathogenic;CLNDBN=Familial_hypercholesterolemia;CLNACC=RCV000030315.1																	
24	1	55529187	G	A	CLNSIG=non-pathogenic;CLNDBN=Familial_hypercholesterolemia;CLNACC=RCV000030349.1																	
25	1	63872032	T	C	CLNSIG=pathogenic non-pathogenic;CLNDBN=Congenital_disorder_of_glycosylation_type_1C AllHighlyPenetrant;CLNACC=RCV000023375.1 RCV000081558.1																	
26	1	98348885	G	A	CLNSIG=pathogenic;CLNDBN=Dihydropyrimidine_dehydrogenase_deficiency;CLNACC=RCV00000464.1																	
27	10	76285162	T	C	CLNSIG=pathogenic;CLNDBN=Hypermethioninemia_due_to_adenosine_kinase_deficiency;CLNACC=RCV000022444.26																	
28	12	52913630	A	G	CLNSIG=untested;CLNDBN=not_provided;CLNACC=RCV000056594.1																	
29	17	26110113	G	A	CLNSIG=untested;CLNDBN=Malignant_melanoma;CLNACC=RCV000063169.2																	
30	17	56292101	A	T	CLNSIG=probable-pathogenic;CLNDBN=Meckel_syndrome_type_1;CLNACC=RCV000050037.1																	

**Figure L:** ClinVar matches A105C

	A	B	C	D	E	F	G	H	I	J	K	L	M	N	O	P	Q	R
1	Chromosome Locus	Ref SNP	Var SNP	Clinical Aspect														
2	1	215848062	C	T	CLNSIG=non-pathogenic;CLNDBN=AllHighlyPenetrant;CLNACC=RCV000041736.1													
3	1	55529187	G	A	CLNSIG=non-pathogenic;CLNDBN=Familial_hypercholesterolemia;CLNACC=RCV000030349.1													
4	1	182554557	C	T	CLNSIG=other;CLNDBN=Prostate_cancer x2c_susceptibility_to;CLNACC=RCV000013880.2													
5	1	154426970	A	C	CLNSIG=other other;CLNDBN=Sерум_level_of_interleukin-6 soluble_receptor Serum_level_of_interleukin_6;CLNACC=RCV000015767.1 RCV000029243.1													
6	1	161479745	A	G	CLNSIG=other other other;CLNDBN=Lupus_nephritis x2c_susceptibility_to Pseudomonas_aeruginosa x2c_susceptibility_to_chronic_infection_by x2c_in_cystic_fibrosis Malaria x2c_sever													
7	17	26110113	G	A	CLNSIG=untested;CLNDBN=Malignant_melanoma;CLNACC=RCV000063169.2													
8	12	52913630	A	G	CLNSIG=untested;CLNDBN=not_provided;CLNACC=RCV000056594.1													

**Figure M:** Novel ClinVar SNPs in A105C

	A	B	C	D	E	F	G	H	I	J	K	L	M	N	O	P	
1	Chromosome	Locus	Ref SNP	Var SNP	Clinical Aspect												
2	1	215848062	C	T	CLINSIG=non-pathogenic;CLNDBN=All HighlyPenetrant;CLNACC=RCV000041736.1												
3	1	159175354	G	A	CLINSIG=non-pathogenic;CLNDBN=DUFFY_BLOOD_GROUP_SYSTEM x2c_FYA/FYB_POLYMORPHISM;CLNACC=RCV000000005.1												
4	1	24180962	T	C	CLINSIG=non-pathogenic;CLNDBN=FU1/FU2_POLYMORPHISM;CLNACC=RCV000000726.1												
5	1	161479745	A	G	CLINSIG=other other other;CLNDBN=Lupus_nephritis x2c_susceptibility_to Pseudomonas_aeruginosa x2c_susceptibility_to Salmonella_senftenbergii												
6	12	52913630	A	G	CLINSIG=untested;CLNDBN=not_provided;CLNACC=RCV000056594.1												

**Figure N:** Novel ClinVar SNPs in A105D