Supporting Tables

Table S1. The genic CNV groups identified CNVnet that differentiate CEU from non-CEU, the overlapping Ensembl and Refseq genes, the p-values, r-values, F_{st} scores, and the sums of phastcons44way scores.

Group	Genic CNV	Ensembl gene	Refseq gene	p-value	r-value	Fst	Phastcons44way sum score
1	chr12:1734273-1734587	ENSG00000006831	*ADIPOR2*	0.007	-0.43	0.504	219.99
	chr2:242154777-242156007	ENSG00000176720	*BOK*		0.23	0.327	257.821
	chr5:112505727-112506265	ENSG00000171444	*MCC*		-0.22	0.067	95.867
	chr5:112694735-112700215	ENSG00000171444	*MCC*		0.089	0	360.154
	chr1:179016392-179022016	ENSG00000143324	*XPR1*		-0.22	0.067	479.722
2	chr12:6777645-6777900	ENSG00000010610	*CD4*	0.002	-0.42	0.406	47.701
	chr2:216866592-216866972	ENSG00000144583	*MARCH4*		-0.409	0.4	425.569
3	chr17:9005941-9006057	ENSG00000065320	*NTN1*	0.037	-0.326	0.216	41.301
	chr4:96601478-96601683	ENSG00000182168	*UNC5C*		0.284	0.277	22.035
	chr4:96620089-96620483	ENSG00000182168	*UNC5C*		0.073	0	170.431
4	chr10:33532647-33532935	ENSG00000099250	*NRP1*	0.203	0.073	0.287	377.15
	chr7:80316526-80316710	ENSG00000075223	*SEMA3C*		0.116	0	154.562
5	chr7:75502568-75505743	ENSG00000127952	*STYXL1*	0.069	-0.366	0.241	133.991
	chr7:75505947-75506596	ENSG00000127952	*STYXL1*		0.116	0	38.606
	chr2:152231858-152231947	ENSG00000183091	*NEB*		-0.128	0.085	33.484
6	chr9:36352882-36354079	ENSG00000137075	*RNF38*	0.103	-0.382	0.665	160.929
	chr3:47465672-47468444	ENSG00000114650	*SCAP*		-0.037	0.51	380.61
	chr4:119463162-119466525	ENSG00000164099	*PRSS12*		-0.179	0.039	190.176
7	chr4:123462021-123462145	ENSG00000138688	*KIAA1109*	<0.001	-0.019	0.084	237.541
	chr3:192547380-192554361	ENSG00000152492	*CCDC50*		-0.619	0.564	244.574
	chr3:192547712-192551630	ENSG00000152492	*CCDC50*		-0.619	0.564	184.755
	chr3:192552252-192554450	ENSG00000152492	*CCDC50*		-0.634	0.578	59.819
8	chr15:74671612-74673430	ENSG00000140386	*SCAPER*	0.045	-0.452	0.502	420.239
	chr15:74671652-74683993	ENSG00000140386	*SCAPER*		-0.427	0.468	2442.521
	chr15:74677967-74682525	ENSG00000140386	*SCAPER*		-0.427	0.468	895.306
	chr5:126699607-126699670	ENSG00000145794	*MEGF10*		0.086	0.024	42.897
9	chr2:216866592-216866972	ENSG00000144583	*MARCH4*	<0.001	-0.409	0.4	425.569
	chr12:6777645-6777900	ENSG00000010610	*CD4*		-0.42	0.406	47.701
	chr15:23482491-23483470	ENSG00000206190	*ATP10A*		0.447	0.622	132.629
	chr15:23483812-23483877	ENSG00000206190	*ATP10A*		0.038	0.02	123.733
10	chr7:2795297-2795627	ENSG00000146535	*GNA12*	<0.001	-0.17	0.039	4.917
	chr15:83750610-83755243	ENSG00000170776	*AKAP13*		0.127	0	258.914
	chr15:83858012-83860202	ENSG00000170776	*AKAP13*		0.163	0.188	81.636
	chr15:83903890-83903984	ENSG00000170776	*AKAP13*		0.408	0.134	446.118
	chr6:152431683-152433926	ENSG00000091831	*ESR1*		0.465	0.23	258.929
	chr6:126224758-126229050	ENSG00000111912	*NCOA7*		0.316	0.268	134.549
11	chr20:30791024-30791695	ENSG00000149600	*COMMD7*	0.143	0.201	0.036	118.253
	chr2:62203864-62204657	ENSG00000173163	*COMMD1*		0.051	0	196.843
12	chr5:82518513-82518818	ENSG00000152422	*XRCC4*	0.127	-0.208	0.511	46.832
	chr2:68563584-68569736	ENSG00000169621	*APLF*		0.051	0	542.145
13	chr16:84339154-84339294	ENSG00000154102	*C16orf74*	<0.001	-0.608	0.602	44.329
	chr4:102179177-102180210	ENSG00000138814	*PPP3CA*		-0.169	0.145	62.71
	chr1:201950932-201952854	ENSG00000058668	*ATP2B4*		0.215	0.066	114.943
	chr3:198418971-198423652	ENSG00000075711	*DLG1*		-0.363	0.338	1054.866
	chr10:89642804-89643702	ENSG00000171862	*PTEN*		-0.255	0.094	227.035
	chr1:241849369-241850386	ENSG00000117020	*AKT3*		-0.531	0.614	158.503
	chr17:52845701-52846506	ENSG00000153944	*MSI2*		0.073	0	109.438
	chr17:53042809-53044913	ENSG00000153944	*MSI2*		-0.261	0.613	260.089
14	chr16:76729167-76729845	ENSG00000186153	*WWOX*	0.004	-0.207	0.096	209.107
	chr16:76929112-76931230	ENSG00000186153	*WWOX*		-0.408	0.45	114.973
	chr16:76929140-76942399	ENSG00000186153	*WWOX*		-0.43	0.46	761.517
	chr16:76931812-76937930	ENSG00000186153	*WWOX*		-0.43	0.46	350.524
	chr16:76932967-76934025	ENSG00000186153	*WWOX*		-0.462	0.478	21.609
	chr16:76939612-76942430	ENSG00000186153	*WWOX*		-0.43	0.46	97.124
	chr16:77257347-77257436	ENSG00000186153	*WWOX*		-0.048	0.047	33.094
	chr16:77319462-77330036	ENSG00000186153	*WWOX*		0.089	0	287.291
	chr16:77434154-77435078	ENSG00000186153	*WWOX*		-0.307	0.205	868.35

chr16:77583705-77596691	ENSG00000186153	*WWOX*	0.051	0	2821.271
chr16:77617288-77620503	ENSG00000186153	*WWOX*	0.149	0.002	266.92
chr22:29067507-29067593	ENSG00000099995	*SF3A1*	-0.119	0.149	439.072
chr19:6338636-6339192	ENSG00000125651	*GTF2F1*	-0.433	0.274	129.152
chr20:60004020-60004338	ENSG00000130699	*TAF4*	0.05	0.759	42.497
chr7:140255056-140255272	ENSG00000133612; ENSG00000157764	*BRAF*	0.029	0.032	68.718
chr15:64493591-64493737	ENSG00000169032	*MAP2K1*	-0.363	0.309	50.768
chr11:12358302-12358925	ENSG00000197702	*PARVA*	0.131	0.007	45.128

Table S2. The genic CNV groups identified by CNVnet that differentiate ASN from non-ASN, the overlapping Ensembl and Refseq genes, the p-values, r-values, F_{st} scores, and the sums of phastcons44way scores.

Group	Genic CNV	Ensembl gene	Refseq gene	p-value	r-value	Fst	Phastcons44way sum score
1	chr1:208001959-208002699	ENSG00000009790	TRAF3IP3	0.106	0.204	0.098	209.709
	chr19:13340551-13340870	ENSG00000141837	CACNA1A		0.143	0.092	30.672
	chr17:61785009-61785204	ENSG00000154229	PRKCA		0.123	0	11.683
	chr1:239252928-239253050	ENSG00000182901	RGS7		0.338	0.166	34.898
		ENSG00000049323;					
2	chr2:33077967-33080796	ENSG00000152683	LTBP1	<0.001	-0.502	0.553	94.35
	chr5:127804627-127806206	ENSG00000138829	FBN2		0.061	0	316.077
3	chr17:9005941-9006057	ENSG00000065320	NTN1	0.016	0.258	0.043	41.301
	chr4:96601478-96601683	ENSG00000182168	UNC5C		-0.182	0.445	22.035
	chr4:96620089-96620483	ENSG00000182168	UNC5C		0.086	0	170.431
4	chr1:210537759-210539239	ENSG00000066027	PPP2R5A	0.001	0.534	0.62	264.294
	chr17:41442561-41442798	ENSG00000186868	MAPT		0.283	0.051	224.887
	chr9:132741961-132743041	ENSG00000097007	ABL1		0.339	0.089	68.664
		ENSG00000183785;					
	chr22:21906857-21907812	ENSG00000186716	BCR		0.214	0.055	11.602
	chr3:186994746-186994971	ENSG00000073792	IGF2BP2		0.286	0.235	27.184
	chr3:161716764-161718359	ENSG00000186432	KPNA4		0.165	0.008	92.363
	chr20:35249024-35249335	ENSG00000118705	RPN2		0.074	0.599	81.271
5	chr7:80316526-80316710	ENSG00000075223	SEMA3C	<0.001	-0.24	0.064	154.562
	chr10:33532647-33532935	ENSG00000099250	NRP1		-0.522	0.479	377.15
6	chr19:50970493-50971468	ENSG00000104936	DMPK	0.116	-0.319	0.634	106.733
	chr11:47482692-47484577	ENSG00000149187	CELF1		0.106	0	325.677
7	chr4:102179177-102180210	ENSG00000138814	PPP3CA	0.149	0.204	0.052	62.71
	chr2:210113466-210114606	ENSG00000078018	MAP2		0.209	0.021	401.74
	chr13:35336405-35336722	ENSG00000133083	DCLK1		0.1	0.35	148.536
	chr13:35519764-35523750	ENSG00000133083	DCLK1		0.061	0	292.107
	chr2:179774912-179783830	ENSG00000187231	SESTD1		0.363	0.222	1214.63
	chr2:179774967-179780525	ENSG00000187231	SESTD1		0.363	0.222	609.093
	chr2:179784064-179785837	ENSG00000187231	SESTD1		0.378	0.235	357.856
8	chr12:11917683-11918420	ENSG00000139083	ETV6	<0.001	-0.595	0.696	44.519
	chr12:11917690-11918167	ENSG00000139083	ETV6		-0.586	0.709	44.519
	chr7:18792619-18792968	ENSG00000048052	HDAC9		0.44	0.198	44.105
	chr2:239666529-239666824	ENSG00000068024	HDAC4		0.123	0	4.986
	chr5:88078895-88079222	ENSG00000081189	MEF2C		-0.051	0.369	357.821
	chr9:138547346-138547616	ENSG00000148400	NOTCH1		-0.426	0.755	47.902
	chr4:96089472-96089789	ENSG00000138696	BMPR1B		0.364	0.238	245.715
	chr6:36178098-36178171	ENSG00000112062	MAPK14		0.135	0.137	137.261
9	chr7:2795297-2795627	ENSG00000146535	GNA12	<0.001	0.082	0	4.917
	chr15:83750610-83755243	ENSG00000170776	AKAP13		0.152	0.004	258.914
	chr15:83858012-83860202	ENSG00000170776	AKAP13		-0.007	0.243	81.636
	chr15:83903890-83903984	ENSG00000170776	AKAP13		-0.538	0.454	446.118
	chr6:152431683-152433926	ENSG00000091831	ESR1		0.006	0.316	258.929
	chr6:126224758-126229050	ENSG00000111912	NCOA7		0.155	0.331	134.549
10	chr20:30791024-30791695	ENSG00000149600	COMMD7	0.082	-0.322	0.202	118.253
	chr2:62203864-62204657	ENSG00000173163	COMMD1		0.061	0	196.843
11	chr13:69299968-69304789	ENSG00000150361	KLHL1	0.112	0.219	0.025	647.864
	chr13:69357060-69357373	ENSG00000150361	KLHL1		0.238	0.032	223.331
	chr4:96089472-96089789	ENSG00000138696	BMPR1B		0.364	0.238	245.715
	chr4:146658257-146659900	ENSG00000170365	SMAD1		0.141	0.163	150.048
	chr15:39805028-39805359	ENSG00000174197	MGA		-0.438	0.888	102.567
12	chr5:82518513-82518818	ENSG00000152422	XRCC4	0.028	0.299	0.4	46.832
	chr2:68563584-68569736	ENSG00000169621	APLF		0.061	0	542.145
13	chr1:54864862-54868554	ENSG00000162390	ACOT11	0.006	-0.284	0.4	521.906
	chr1:54865467-54866525	ENSG00000162390	ACOT11		-0.284	0.4	77.872
	chr20:19960660-19961103	ENSG00000173418	NAA20		0.176	0.011	367.056
		ENSG00000133612;					
	chr7:127407391-127407708	ENSG00000197157	SND1		0.58	0.633	380.01
		ENSG00000164944;					
14	chr8:95627342-95630123	ENSG00000212997	KIAA1429	<0.001	0.202	0.021	128.54
	chr12:11917683-11918420	ENSG00000139083	ETV6		-0.595	0.696	44.519

	chr12:11917690-11918167	ENSG00000139083 ENSG00000008128; ENSG00000189229; ENSG00000189339; ENSG00000215790; ENSG00000215914;	ETV6 CDK11A; CDK11B; MMP23A; SLC35E2;		-0.586	0.709	44.519
	chr1:1607171-1662053	ENSG00000219541	SLC35E2B		-0.106	0.001	7087.82
	chr13:113039048-113039117	ENSG00000139835	GRTP1		-0.252	0.888	37.057
	chr3:19986449-19986548	ENSG00000144566	RAB5A		0.249	0.039	124.348
	chr17:5131275-5131369	ENSG00000029725	RABEP1		-0.028	0.061	140.31
	chr17:5218143-5218717	ENSG00000029725	RABEP1		0.086	0	74.972
15	chr2:181634073-181634394	ENSG00000170035	UBE2E3	0.004	-0.318	0.918	10.968
	chr18:53958688-53958856	ENSG00000049759	NEDD4L		0.061	0	70.951
	chr18:54059726-54060325	ENSG00000049759	NEDD4L		0.123	0	6.616
	chr18:54082073-54088115	ENSG00000049759	NEDD4L		0.086	0	397.106
	chr9:97305837-97306191	ENSG00000185920	PTCH1		0.183	0.488	9.877
	chr1:72222209-72222620	ENSG00000172260	NEGR1		-0.482	0.411	8.974
16	chr4:69076627-69093237	ENSG00000197888	UGT2B17	<0.001	-0.629	0.661	2100.33
	chr4:69096467-69108525	ENSG00000197888	UGT2B17		-0.629	0.661	1376.5
	chr4:69096712-69105730	ENSG00000197888	UGT2B17		-0.629	0.661	1116.886
	chr4:69106612-69114230	ENSG00000197888	UGT2B17		-0.62	0.661	490.938
	chr4:69108967-69117025	ENSG00000197888	UGT2B17		-0.62	0.661	770.941
	chr4:69115412-69117030	ENSG00000197888	UGT2B17		-0.617	0.662	398.913
	chr10:5238654-5238756	ENSG00000198610	AKR1C4		0.335	0.085	128.774
	chr10:5239893-5240994	ENSG00000198610	AKR1C4		0.191	0.018	754.025
	chr2:234208051-234209717	ENSG00000167165	UGT1A8 UGT1A10; UGT1A4; UGT1A5; UGT1A6; UGT1A7; UGT1A8;		0.188	0.014	492.352
	chr2:234296217-234298725	ENSG00000167165	UGT1A9		-0.294	0.23	4.157
17	chr8:27718420-27718749	ENSG00000171320		0.025	-0.234	0.238	124.935
	chr1:54864862-54868554	ENSG00000162390	ACOT11		-0.284	0.4	521.906
	chr1:54865467-54866525	ENSG00000162390	ACOT11		-0.284	0.4	77.872
	chr20:19960660-19961103	ENSG00000173418	NAA20		0.176	0.011	367.056
	chr7:127407391-127407708	ENSG00000133612; ENSG00000197157	SND1		0.58	0.633	380.01

Table S3. The genic CNV groups identified by CNVnet that differentiate YRI from non-YRI, the overlapping Ensembl and Refseq genes, the p-values, r-values, F_{st} scores, and the sums of phastcons44way scores.

Group	Genic CNV	Ensembl gene	Refseq gene	p-value	r-value	Fst	Phastcons44way sum score
1	chr12:1734273-1734587	ENSG00000006831	ADIPOR2	0.001	0.499	0.279	219.99
	chr2:242154777-242156007	ENSG00000176720	BOK		-0.071	0.415	257.821
	chr5:112505727-112506265	ENSG00000171444	MCC		0.102	0	95.867
	chr5:112694735-112700215	ENSG00000171444	MCC		-0.193	0.039	360.154
	chr1:179016392-179022016	ENSG00000143324	XPR1		0.102	0	479.722
2	chr12:6777645-6777900	ENSG00000010610	*CD4*	0.001	-0.038	0.157	47.701
	chr2:216866592-216866972	ENSG00000144583	*MARCH4*		0.421	0.144	425.569
		ENSG00000049323;					
3	chr2:33077967-33080796	ENSG00000152683	*LTBP1*	0.124	0.411	0.332	94.35
	chr5:127804627-127806206	ENSG00000138829	*FBN2*		-0.11	0.003	316.077
4	chr1:210537759-210539239	ENSG00000066027	*PPP2R5A*	<0.001	-0.578	0.724	264.294
	chr17:41442561-41442798	ENSG00000186868	*MAPT*		0.271	0.044	224.887
	chr9:132741961-132743041	ENSG00000097007	*ABL1*		-0.311	0.231	68.664
		ENSG00000183785;					
	chr22:21906857-21907812	ENSG00000186716	*BCR*		0.264	0.056	11.602
	chr3:186994746-186994971	ENSG00000073792	*IGF2BP2*		-0.334	0.389	27.184
	chr3:161716764-161718359	ENSG00000186432	*KPNA4*		-0.298	0.111	92.363
	chr20:35249024-35249335	ENSG00000118705	*RPN2*		0.141	0.597	81.271
5	chr7:80316526-80316710	ENSG00000075223	*SEMA3C*	0.011	0.132	0	154.562
	chr10:33532647-33532935	ENSG00000099250	*NRP1*		0.458	0.261	377.15
6	chr8:17896482-17897538	ENSG00000078674	*PCM1*	0.012	-0.21	0.057	446.671
	chr8:17903778-17904702	ENSG00000078674	*PCM1*		-0.592	0.49	106.188
	chr9:113205490-113209552	ENSG00000136813	*KIAA0368*		-0.283	0.284	777.001
	chr11:66468058-66469862	ENSG00000173599	*PC*		0.114	0.439	71.199
	chr11:85963610-85964370	ENSG00000151376	*ME3*		-0.499	0.375	31.632
	chr11:85981879-85984206	ENSG00000151376	*ME3*		0.201	0.128	458.853
7	chr3:1340574-1341706	ENSG00000134115	*CNTN6*	<0.001	-0.452	0.275	4.137
	chr3:157574858-157576385	ENSG00000169282	*KCNAB1*		-0.634	0.592	105.36
	chr3:198418971-198423652	ENSG00000075711	*DLG1*		0.356	0.091	1054.866
	chr4:22111163-22111260	ENSG00000152990	*GPR125*		0.235	0.106	11.256
8	chr4:102179177-102180210	ENSG00000138814	*PPP3CA*	0.039	-0.045	0.077	62.71
	chr2:210113466-210114606	ENSG00000078018	*MAP2*		-0.379	0.183	401.74
			DCLK1;				
	chr13:35336405-35336722	ENSG00000133083	*MIR548F5*		-0.148	0.384	148.536
	chr13:35519764-35523750	ENSG00000133083	*DCLK1*		0.058	0	292.107
	chr2:179774912-179783830	ENSG00000187231	*SESTD1*		-0.418	0.428	1214.63
	chr2:179774967-179780525	ENSG00000187231	*SESTD1*		-0.418	0.428	609.093
	chr2:179784064-179785837	ENSG00000187231	*SESTD1*		-0.446	0.459	357.856
9	chr15:74671612-74673430	ENSG00000140386	*SCAPER*	0.079	0.442	0.26	420.239
	chr15:74671652-74683993	ENSG00000140386	*SCAPER*		0.441	0.245	2442.521
	chr15:74677967-74682525	ENSG00000140386	*SCAPER*		0.441	0.245	895.306
	chr5:126699607-126699670	ENSG00000145794	*MEGF10*		0.038	0.037	42.897
10	chr2:216866592-216866972	ENSG00000144583	*MARCH4*	<0.001	0.421	0.144	425.569
	chr12:6777645-6777900	ENSG00000010610	*CD4*		-0.038	0.157	47.701
	chr15:23482491-23483470	ENSG00000206190	*ATP10A*		-0.523	0.745	132.629
	chr15:23483812-23483877	ENSG00000206190	*ATP10A*		-0.024	0.035	123.733
11	chr13:69299968-69304789	ENSG00000150361	*KLHL1*	0.115	-0.245	0.109	647.864
	chr13:69357060-69357373	ENSG00000150361	*KLHL1*		-0.384	0.203	223.331
	chr4:96089472-96089789	ENSG00000138696	*BMPR1B*		-0.243	0.36	245.715
	chr4:146658257-146659900	ENSG00000170365	*SMAD1*		0.297	0.15	150.048
	chr15:39805028-39805359	ENSG00000174197	*MGA*		0.423	0.876	102.567
12	chr11:106743812-106746730	ENSG00000152404	*CWF19L2*	0.332	-0.262	0.389	203.289
	chr11:106747014-106748737	ENSG00000152404	*CWF19L2*		-0.262	0.389	113.51
	chr17:5279813-5280802	ENSG00000108561	*C1QBP*		-0.138	0.026	58.003
	chr11:82277695-82277812	ENSG00000137509	*PRCP*		-0.565	0.448	96.736
13	chr1:54864862-54868554	ENSG00000162390	*ACOT11*	0.001	0.468	0.261	521.906
	chr1:54865467-54866525	ENSG00000162390	*ACOT11*		0.468	0.261	77.872
	chr20:19960660-19961103	ENSG00000173418	*NAA20*		-0.32	0.129	367.056
	chr7:127407391-127407708	ENSG00000133612;	*SND1*		-0.563	0.713	380.01

		ENSG00000197157					
14	chr3:157574858-157576385	ENSG00000169282	*KCNAB1*	<0.001	-0.634	0.592	105.36
	chr3:1340574-1341706	ENSG00000134115	*CNTN6*		-0.452	0.275	4.137
15	chr4:69076627-69093237	ENSG00000197888	*UGT2B17*	<0.001	0.508	0.468	2100.33
	chr4:69096467-69108525	ENSG00000197888	*UGT2B17*		0.508	0.468	1376.5
	chr4:69096712-69105730	ENSG00000197888	*UGT2B17*		0.508	0.468	1116.886
	chr4:69106612-69114230	ENSG00000197888	*UGT2B17*		0.495	0.468	490.938
	chr4:69108967-69117025	ENSG00000197888	*UGT2B17*		0.495	0.468	770.941
	chr4:69115412-69117030	ENSG00000197888	*UGT2B17*		0.487	0.473	398.913
	chr10:5238654-5238756	ENSG00000198610	*AKR1C4*		-0.351	0.237	128.774
	chr10:5239893-5240994	ENSG00000198610	*AKR1C4*		-0.346	0.165	754.025
	chr2:234208051-234209717	ENSG00000167165	*UGT1A8 UGT1A10; UGT1A4; UGT1A5; UGT1A6; UGT1A7; UGT1A8;*		-0.167	0.06	492.352
	chr2:234296217-234298725	ENSG00000167165	*UGT1A9*		0.165	0.086	4.157
		ENSG00000154611;					
16	chr18:22001804-22005320	ENSG00000188985	*PSMA8*	<0.001	-0.705	0.618	1123.877
	chr2:173597167-173597490	ENSG00000091428	*RAPGEF4*		-0.059	0.194	29.721
	chr11:14304887-14305202	ENSG00000133818	*RRAS2*		0.256	0.606	166.382
	chr12:18614538-18616361	ENSG00000139144	*PIK3C2G*		-0.25	0.075	174.031
	chr10:95843672-95843987	ENSG00000138193	*PLCE1*		-0.157	0.021	24.103
	chr3:187382682-187384899	ENSG00000058866	*DGKG*		0.058	0	117.736
	chr3:187512304-187512639	ENSG00000058866	*DGKG*		-0.496	0.311	36.096
	chr20:9097570-9097622	ENSG00000101333	*PLCB4*		-0.033	0.133	81.563
	chr20:9141752-9142025	ENSG00000101333	*PLCB4*		-0.445	0.276	42.963
17	chr6:123805116-123805420	ENSG00000186439	*TRDN*	0.012	-0.508	0.386	23.35
	chr8:62603331-62603752	ENSG00000198363	*ASPH*		0.058	0	110.77
	chr8:62675414-62677986	ENSG00000198363	*ASPH*		-0.32	0.129	415.948
18	chr8:27718420-27718749	ENSG00000171320		0.009	-0.007	0.161	124.935
	chr1:54864862-54868554	ENSG00000162390	*ACOT11*		0.468	0.261	521.906
	chr1:54865467-54866525	ENSG00000162390	*ACOT11*		0.468	0.261	77.872
	chr20:19960660-19961103	ENSG00000173418	*NAA20*		-0.32	0.129	367.056
	chr7:127407391-127407708	ENSG00000133612; ENSG00000197157	*SND1*		-0.563	0.713	380.01

Table S4. The genic CNV pairs identified by CNVnet that differentiate CEU from non-CEU, the overlapping Ensembl and Refseq genes, the p-values, r-values, F_{st} scores, and the sums of phastcons44way scores.

Pair	Genic CNV	Ensembl gene	Refseq gene	p-value	r-value	Fst	Phastcons44way sum score
1	chr12:6777645-6777900	ENSG00000010610	*CD4*	0.016	-0.42	0.406	47.701
	chr15:23482491-23483470	ENSG00000206190	*ATP10A*		0.447	0.622	132.629
2	chr15:66205377-66206224	ENSG00000033800	*PIAS1*	<0.001	0.051	0	226.5
	chr2:200020701-200020924	ENSG00000119042	*SATB2*		-0.568	0.631	377.505
3	chr4:24850148-24850288	ENSG00000038210	*PI4K2B*	0.001	-0.485	0.547	83.98
	chr1:218352078-218352397	ENSG00000067704	*IARS2*		0.151	0.292	8.728
4	chr15:46337554-46337873	ENSG00000074803	*SLC12A1*	0.019	0.452	0.26	57.11
	chr2:168604053-168604372	ENSG00000198648	*STK39*		-0.245	0.69	68.956
5	chr22:29067507-29067593	ENSG00000099995	*SF3A1*	0.002	-0.119	0.149	439.072
	chr16:76932967-76934025	ENSG00000186153	*WWOX*		-0.462	0.478	21.609
6	chr3:193545876-193546185	ENSG00000114279	*FGF12*	<0.001	0.652	0.753	148.21
	chr7:55102446-55103351	ENSG00000146648	*EGFR*		0.208	0.015	50.888
7	chr1:185170173-185170282	ENSG00000116711	*PLA2G4A*	0.002	0.229	0.034	56.17
	chr8:55134403-55134716	ENSG00000120992	*LYPLA1*		0.468	0.287	90.351
8	chr10:24416844-24418447	ENSG00000120549	*KIAA1217*	<0.001	-0.7	0.663	239.388
	chr17:7449181-7449236	ENSG00000129245	*FXR2*		0.198	0.015	129.612
9	chr4:102179177-102180210	ENSG00000138814	*PPP3CA*	<0.001	-0.169	0.145	62.71
	chr16:84339154-84339294	ENSG00000154102	C16orf74		-0.608	0.602	44.329

Table S5. The genic CNV pairs identified by CNVnet that differentiate ASN from non-ASN, the overlapping Ensembl and Refseq genes, the p-values, r-values, Fst scores, and the sums of phastcons44way scores.

Pair	Genic CNV	Ensembl gene	Refseq gene	p-value	r-value	Fst	Phastcons44way sum score
1	chr2:33077967-33080796	ENSG00000049323; ENSG00000152683	*LTBP1*	0.043	-0.502	0.553	94.35
	chr5:127804627-127806206	ENSG00000138829	*FBN2*		0.061	0	316.077
2	chr7:80316526-80316710	ENSG00000075223	*SEMA3C*	0.018	-0.24	0.064	154.562
	chr10:33532647-33532935	ENSG00000099250	*NRP1*		-0.522	0.479	377.15
3	chr2:173010737-173012713	ENSG00000091409	*ITGA6*	<0.001	0.061	0	137.912
	chr6:129361209-129367267	ENSG00000196569	*LAMA2*		0.617	0.403	31.39
4	chr19:18103159-18103478	ENSG00000099308	*MAST3*	0.008	0.28	0.1	288.095
	chr16:75096635-75101526	ENSG00000152910	*CNTNAP4*		-0.499	0.686	316.337
5	chr4:154451921-154454465	ENSG00000109654	*TRIM2*	0.001	0.086	0	217.314
	chr21:14251883-14251952	ENSG00000215559	*C21orf81*		0.411	0.152	25.601
6	chr1:185170173-185170282	ENSG00000116711	*PLA2G4A*	0.002	-0.03	0.072	56.17
	chr8:55134403-55134716	ENSG00000120992	*LYPLA1*		-0.4	0.51	90.351
7	chr9:100348865-100351485	ENSG00000136928	*GABBR2*	0.003	-0.563	0.676	202.21
	chr15:82071624-82071832	ENSG00000140600	*SH3GL3*		-0.283	0.2	96.863
8	chr9:100387807-100388137	ENSG00000136928	*GABBR2*	0.061	0.479	0.502	128.096
	chr15:82071624-82071832	ENSG00000140600	*SH3GL3*		-0.283	0.2	96.863
9	chr4:106589529-106589866	ENSG00000138777	*PPA2*	<0.001	0.274	0.047	81.401
	chr12:122803585-122803664	ENSG00000185344	*ATP6V0A2*		-0.86	0.904	107.905
10	chr1:72222209-72222620	ENSG00000172260	*NEGR1*	0.039	-0.482	0.411	8.974
	chr9:97305837-97306191	ENSG00000185920	*PTCH1*		0.183	0.488	9.877
11	chr20:19960660-19961103	ENSG00000173418	*NAA20*	<0.001	0.176	0.011	367.056
	chr7:127407391-127407708	ENSG00000133612; ENSG00000197157	*SND1*		0.58	0.633	380.01

Table S6. The genic CNV pairs identified by CNVnet that differentiate YRI from non-YRI, the overlapping Ensembl and Refseq genes, the p-values, r-values, F_{st} scores, and the sums of phastcons44way scores.

Pair	Genic CNV	Ensembl gene	Refseq gene	p-value	r-value	Fst	Phastcons44way sum score
1	chr12:1734273-1734587	ENSG00000006831	*ADIPOR2*	0.032	0.499	0.279	219.99
	chr2:242154777-242156007	ENSG00000176720	*BOK*		-0.071	0.415	257.821
2	chr12:6777645-6777900	ENSG00000010610	*CD4*	0.013	-0.038	0.157	47.701
	chr15:23482491-23483470	ENSG00000206190	*ATP10A*		-0.523	0.745	132.629
3	chr14:101642858-101644890	ENSG00000080824	*HSP90AA1*	<0.001	-0.193	0.039	256.175
	chr1:153926384-153931088	ENSG00000132676	*DAP3*		-0.807	0.823	1545.861
4	chr14:101642858-101644890	ENSG00000080824	*HSP90AA1*	<0.001	-0.193	0.039	256.175
	chr1:153927714-153930237	ENSG00000132676	*DAP3*		-0.807	0.823	929.535
5	chr1:195058412-195060030	ENSG00000080910	*CFHR1*	0.014	-0.531	0.536	85.351
		ENSG00000110244;	*APOA1;*				
		ENSG00000110245;	*APOA4;*				
		ENSG00000118137;	*APOC3;*				
	chr11:116188112-116226985	ENSG00000160584	*SIK3*		0.317	0.078	4014.147
6	chr19:18103159-18103478	ENSG00000099308	*MAST3*	<0.001	0.023	0.122	288.095
	chr16:75096635-75101526	ENSG00000152910	*CNTNAP4*		0.78	0.691	316.337
7	chr1:205358986-205359816	ENSG00000123838	*C4BPA*	0.058	0.444	0.609	434.753
	chr2:203003671-203021160	ENSG00000204217	*BMPR2*		-0.527	0.348	2055.341
8	chr9:100348865-100351485	ENSG00000136928	*GABBR2*	0.001	0.583	0.519	202.21
	chr15:82071624-82071832	ENSG00000140600	*SH3GL3*		0.185	0.065	96.863
9	chr7:55102446-55103351	ENSG00000146648	*EGFR*	0.001	-0.448	0.256	50.888
	chr11:48076638-48076946	ENSG00000149177	*PTPRJ*		0.56	0.652	42.008
10	chr20:19960660-19961103	ENSG00000173418	*NAA20*	0.012	-0.32	0.129	367.056
		ENSG00000133612;					
	chr7:127407391-127407708	ENSG00000197157	*SND1*		-0.563	0.713	380.01

Table S7. A list of genic CNV pairs (CNV_1, CNV_2), that might differentiate CEU from YRI identified by CNVnet, affect the gene expression differently in the two populations while each individual CNV doesn't affect gene expression differently in these two populations. The p-values (2-way ANOVA, with significant level α=0.01) indicate their statistical power of affecting the expression of corresponding gene (Gene) individually (CNV_1-gene or CNV_2-gene) and in pair (CNV_1-CNV_2-Gene) in CEU versus YRI.

			CEU	CEU	CEU	YRI	YRI	YRI
CNV_1	CNV_2	Gene	CNV_1-Gene	CNV_2-Gene	CNV_1-CNV_2-Gene	CNV1-Gene	CNV2-Gene	CNV1-CNV2-Gene
chr22:29067507-29067593	chr16:76932967-76934025	*AKR7A2*	0.31325	0.35339	0.0070477	0.28556	0.48287	0.28886
chr22:29067507-29067593	chr16:76932967-76934025	*ARHGAP15*	0.2879	0.39565	0.22673	0.28713	0.47481	0.0080651
chr22:29067507-29067593	chr16:76932967-76934025	*ARHGEF17*	0.84546	0.83962	0.48232	0.81155	0.18722	0.0035282
chr22:29067507-29067593	chr16:76932967-76934025	*ARTN*	0.77715	0.02811	0.34619	0.66534	0.021978	0.0048516
chr22:29067507-29067593	chr16:76932967-76934025	*ATP8A2*	0.83113	0.092942	0.0075311	0.60064	0.42088	0.78982
chr22:29067507-29067593	chr16:76932967-76934025	*BCL3*	0.32051	0.078625	0.0070371	0.59593	0.32456	0.032243
chr22:29067507-29067593	chr16:76932967-76934025	*C14orf118*	0.10627	0.36516	0.0089385	0.75918	0.46609	0.70639
chr22:29067507-29067593	chr16:76932967-76934025	*C3orf21*	0.14372	0.01191	0.0072503	0.90738	0.90497	0.24703
chr22:29067507-29067593	chr16:76932967-76934025	*C9orf167*	0.4587	0.05255	0.039582	0.83013	0.00068151	0.0035105
chr22:29067507-29067593	chr16:76932967-76934025	*CAPS2*	0.78145	0.003124	0.0096157	0.87399	0.4637	0.63417
chr22:29067507-29067593	chr16:76932967-76934025	*CCDC50*	0.15588	0.059673	0.0044298	0.58875	0.66629	0.5653
chr22:29067507-29067593	chr16:76932967-76934025	*OCIAD1*	0.94378	0.066773	0.0087412	0.27079	0.37725	0.68709
chr22:29067507-29067593	chr16:76932967-76934025	*OXCT2*	0.7382	0.28911	0.17998	0.031627	0.0021807	0.0021187
chr22:29067507-29067593	chr16:76932967-76934025	*PGRMC2*	0.41405	0.29136	0.0058639	0.44678	0.40473	0.51166
chr22:29067507-29067593	chr16:76932967-76934025	*PITX1*	0.60574	0.00807	0.006391	0.35675	0.20609	0.25849
chr22:29067507-29067593	chr16:76932967-76934025	*PPP1R13L*	0.19674	0.47367	0.16915	0.92742	0.61539	0.007449
chr22:29067507-29067593	chr16:76932967-76934025	*PRPF39*	0.34427	0.35639	0.0081931	0.55223	0.93767	0.28285
chr22:29067507-29067593	chr16:76932967-76934025	*SNRPB2*	0.031663	0.55292	0.0079794	0.18339	0.16008	0.14228
chr22:29067507-29067593	chr16:76932967-76934025	*SPRED2*	0.82113	0.054554	0.25837	0.90777	0.029083	0.0011893
chr22:29067507-29067593	chr16:76932967-76934025	*SYCP3*	0.88854	0.11009	0.0084489	0.40338	0.2178	0.10767
chr22:29067507-29067593	chr16:76932967-76934025	*TNFAIP8L1*	0.57875	0.25587	0.21573	0.31993	0.0053747	0.007449
chr22:29067507-29067593	chr16:76932967-76934025	*TNPO2*	0.17053	0.20572	0.0067377	0.26465	0.040881	0.20443
chr22:29067507-29067593	chr16:76932967-76934025	*TSPAN17*	0.83122	0.23434	0.0026296	0.67669	0.69411	0.71987
chr22:29067507-29067593	chr16:76932967-76934025	*USP20*	0.58536	0.042126	0.0053286	0.66797	0.75286	0.87334
chr22:29067507-29067593	chr16:76932967-76934025	*ZBED2*	0.11251	0.0003555	0.0073561	0.52778	0.90085	0.71183
chr22:29067507-29067593	chr16:76932967-76934025	*ZIK1*	0.101	0.17668	0.0095188	0.59334	0.30765	0.48712
chr22:29067507-	chr16:76932967-	*ZNF575*	0.13184	0.99685	0.002585	0.4906	0.24223	0.36515

29067593	76934025							
chr22:29067507-29067593	chr16:76932967-76934025	*ZNF85*	0.74943	0.29102	0.0091029	0.77798	0.25623	0.4999
chr3:193545876-193546185	chr7:55102446-55103351	*FAM102B*	0.38109	0.18095	0.0041196	0.62042	0.27257	0.23521
chr3:193545876-193546185	chr7:55102446-55103351	*NOTUM*	0.81366	0.0014054	0.0010704	0.76562	0.48388	0.11558
chr3:193545876-193546185	chr7:55102446-55103351	*USP34*	0.76651	0.028445	0.00085753	0.29445	0.96841	0.88476
chr15:46337554-46337873	chr2:168604053-168604372	*AAK1*	0.094001	0.51409	0.18689	0.70184	0.0087403	0.0042812
chr15:46337554-46337873	chr2:168604053-168604372	*ACACB*	0.43321	0.84798	0.0077101	0.78054	0.85029	0.0092735
chr15:46337554-46337873	chr2:168604053-168604372	*AGR3*	0.096759	0.62349	0.054915	0.23102	0.2742	0.0032191
chr15:46337554-46337873	chr2:168604053-168604372	*ARTN*	0.83015	0.086575	0.30902	0.50562	0.033469	0.0042738
chr15:46337554-46337873	chr2:168604053-168604372	*ATP8A2*	0.56355	0.42269	0.0040994	0.85538	0.93976	0.97764
chr15:46337554-46337873	chr2:168604053-168604372	*BCL3*	0.049233	0.47029	0.0092026	0.80031	0.31595	0.23397
chr15:46337554-46337873	chr2:168604053-168604372	*BMP2K*	0.65458	0.31427	0.58739	0.20397	0.71612	0.0086869
chr15:46337554-46337873	chr2:168604053-168604372	*C11orf49*	0.001703	0.64434	0.003193	0.49158	0.13337	0.099584
chr15:46337554-46337873	chr2:168604053-168604372	*C1orf106*	0.070832	0.85359	0.33869	0.12552	0.76458	0.0091734
chr15:46337554-46337873	chr2:168604053-168604372	*C2orf34*	0.19484	0.61417	0.62354	0.83852	0.10978	0.0045436
chr15:46337554-46337873	chr2:168604053-168604372	*C7orf41*	0.71963	0.61727	0.37806	0.88464	0.95472	0.0017459
chr15:46337554-46337873	chr2:168604053-168604372	*CCDC132*	0.54611	0.89854	0.99032	0.029802	0.57722	0.0095261
chr15:46337554-46337873	chr2:168604053-168604372	*CD247*	0.56645	0.6623	0.33509	0.28562	0.53498	0.0006917
chr15:46337554-46337873	chr2:168604053-168604372	*CDCP1*	0.0489	0.58409	0.0087262	0.079781	0.8088	0.62262
chr15:46337554-46337873	chr2:168604053-168604372	*CNR2*	0.24445	0.038118	0.0053466	0.31911	0.71524	0.44594
chr15:46337554-46337873	chr2:168604053-168604372	*COL4A2*	0.015519	0.62453	0.0059815	0.37543	0.67427	0.6396
chr15:46337554-46337873	chr2:168604053-168604372	*COQ10A*	0.29788	0.25006	0.0099043	0.71167	0.67334	0.037737
chr15:46337554-46337873	chr2:168604053-168604372	*CPNE2*	0.49779	0.32645	0.0077304	0.91434	0.25225	0.61149
chr15:46337554-46337873	chr2:168604053-168604372	*CXCL9*	0.43256	0.0006319	8.9661e-05	0.21463	0.032377	0.0067837
chr15:46337554-46337873	chr2:168604053-168604372	*DEAF1*	0.049575	0.40383	0.0099401	0.65447	0.25899	0.72458
chr15:46337554-46337873	chr2:168604053-168604372	*ENTHD1*	0.95998	0.062357	0.00060175	0.54193	0.97625	0.9131
chr15:46337554-46337873	chr2:168604053-168604372	*ETFDH*	0.62933	0.64109	0.6993	0.33191	0.60746	0.0032435
chr15:46337554-46337873	chr2:168604053-168604372	*GLUL*	0.018979	0.96162	0.0066271	0.81329	0.5782	0.33024
chr15:46337554-46337873	chr2:168604053-168604372	*HBG2*	0.14512	0.21223	0.60148	0.40959	0.070392	0.0009665
chr15:46337554-46337873	chr2:168604053-168604372	*HSPB1*	0.39669	0.26675	0.36533	0.49202	0.080158	0.0033668
chr15:46337554-46337873	chr2:168604053-168604372	*IFT57*	0.62248	0.67948	0.83616	0.7991	0.095875	0.0093959
chr15:46337554-46337873	chr2:168604053-168604372	*IL1R2*	0.98752	0.56937	0.34766	0.093765	0.95401	0.0049754
chr15:46337554-46337873	chr2:168604053-168604372	*ITGA11*	0.29532	0.31022	0.88274	0.26377	0.8486	0.008601
chr15:46337554-	chr2:168604053-	*JUB*	0.23751	0.051984	0.0072655	0.71088	0.76155	0.94662

46337873	168604372							
chr15:46337554-46337873	chr2:168604053-168604372	*KCNIP2*	0.023761	0.49808	0.00059484	0.511	0.521	0.89492
chr15:46337554-46337873	chr2:168604053-168604372	*KLF2*	0.67793	0.53142	0.90479	0.17069	0.51975	0.0068055
chr15:46337554-46337873	chr2:168604053-168604372	*LRCH1*	0.92983	0.32606	0.16558	0.36068	0.3221	0.0020317
chr15:46337554-46337873	chr2:168604053-168604372	*MAP1A*	0.57558	0.12909	0.0082707	0.28738	0.90018	0.28419
chr15:46337554-46337873	chr2:168604053-168604372	*MCM3*	0.92266	0.56764	0.64403	0.052121	0.042817	0.0092141
chr15:46337554-46337873	chr2:168604053-168604372	*MCM9*	0.48086	0.86448	0.35917	0.75961	0.39607	0.0044612
chr15:46337554-46337873	chr2:168604053-168604372	*MLF2*	0.14242	0.24662	0.845	0.70863	0.056522	0.0037686
chr15:46337554-46337873	chr2:168604053-168604372	*MT1E*	0.0005403	0.22478	0.0019474	0.63252	0.53306	0.70845
chr15:46337554-46337873	chr2:168604053-168604372	*NCOR2*	0.17866	0.25663	0.41346	0.36037	0.42694	0.0091836
chr15:46337554-46337873	chr2:168604053-168604372	*NDUFA13*	0.012654	0.47877	0.0087411	0.62253	0.75821	0.25433
chr15:46337554-46337873	chr2:168604053-168604372	*NOTUM*	0.83447	0.0073426	0.052276	0.4486	0.14052	0.0020834
chr15:46337554-46337873	chr2:168604053-168604372	*OIT3*	0.0002114	0.30548	0.004176	0.10927	0.90807	0.72436
chr15:46337554-46337873	chr2:168604053-168604372	*PCID2*	0.82687	0.21914	0.83456	0.81428	0.13335	0.0028534
chr15:46337554-46337873	chr2:168604053-168604372	*PPFIBP2*	0.99849	0.062512	0.0034914	0.53025	0.47645	0.4779
chr15:46337554-46337873	chr2:168604053-168604372	*PPP1R1B*	0.95603	0.39664	0.89062	0.017839	0.4519	0.0074723
chr15:46337554-46337873	chr2:168604053-168604372	*PPP1R7*	0.19418	0.75154	0.64793	0.63629	0.10857	0.0012711
chr15:46337554-46337873	chr2:168604053-168604372	*PTPRS*	0.32505	0.51472	0.0084256	0.27613	0.63943	0.93387
chr15:46337554-46337873	chr2:168604053-168604372	*RREB1*	0.068594	0.65402	0.0032866	0.2393	0.73045	0.67297
chr15:46337554-46337873	chr2:168604053-168604372	*SCML4*	0.42809	0.033155	0.0080316	0.34761	0.11622	0.16549
chr15:46337554-46337873	chr2:168604053-168604372	*SIGLEC5*	0.37776	0.027646	0.06105	0.0073026	0.58688	0.0010725
chr15:46337554-46337873	chr2:168604053-168604372	*SLC30A3*	0.83811	0.20355	0.77771	0.84142	0.45638	0.0058991
chr15:46337554-46337873	chr2:168604053-168604372	*SPRED2*	0.26779	0.12486	0.22436	0.3879	0.067015	0.0013095
chr15:46337554-46337873	chr2:168604053-168604372	*TGIF1*	0.57396	0.86242	0.65677	0.99732	0.17802	0.0080522
chr15:46337554-46337873	chr2:168604053-168604372	*TMEM2*	0.048447	0.74394	0.00085767	0.60567	0.28012	0.76524
chr15:46337554-46337873	chr2:168604053-168604372	*USP48*	0.08336	0.0051876	0.0020981	0.49383	0.49381	0.2452
chr15:46337554-46337873	chr2:168604053-168604372	*ZNF501*	0.010334	0.12006	0.007627	0.94937	0.99	0.82138
chr15:46337554-46337873	chr2:168604053-168604372	*ZNF577*	0.21038	0.084839	0.0013047	0.42346	0.70526	0.76164
chr15:46337554-46337873	chr2:168604053-168604372	*ZYG11A*	0.0015788	0.83397	0.0050546	0.090605	0.09169	0.37076

TABLE S8. A table showing CNV genes found in the ASN group matching genes linked to disease phenotypes from the OMIM database.

ASN matches in OMIM		
Gene/Locus	**Gene/Locus name**	**Phenotype**
ABL2, ABLL, ARG	Abelson murine leukemia viral (v-abl) oncogene homolog 2 (arg, Abelson-related gene)	Leukemia, acute myeloid, with eosinophilia
HDAC4, HDACA, BDMR, AHO3	Histone deacetylase 4	Brachydactyly-mental retardation syndrome
IGF2BP2, IMP2	Insulin-like growth factor 2 mRNA-binding protein 2	{Diabetes mellitus, noninsulin-dependent, susceptibility to}
PDGFRA	Platelet-derived growth factor receptor, alpha polypeptide	Hypereosinophilic syndrome, idiopathic, resistant to imatinib
PDGFRA	Platelet-derived growth factor receptor, alpha polypeptide	Gastrointestinal stromal tumor, somatic
UGT2B17, BMND12	UDP-glucuronyltransferase, family 2, beta-17	{Bone mineral density QTL 12, osteoporosis}
BMPR1B, ALK6	Bone morphogenetic protein receptor, type IB	Chrondrodysplasia, acromesomelic, with genital anomalies
BMPR1B, ALK6	Bone morphogenetic protein receptor, type IB	Brachydactyly, type A2
MEF2C, C5DELq14.3, DEL5q14.3	MADS box transcription enhancer factor 2, polypeptide C (myocyte enhancer factor 2C)	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations
MEF2C, C5DELq14.3, DEL5q14.3	MADS box transcription enhancer factor 2, polypeptide C (myocyte enhancer factor 2C)	Chromosome 5q14.3 deletion syndrome
FBN2, CCA, EOMD	Fibrillin-2	Macular degeneration, early-onset
FBN2, CCA, EOMD	Fibrillin-2	Contractural arachnodactyly, congenital
PDGFRB, PDGFR, IBGC4, IMF1	Platelet-derived growth factor receptor, beta polypeptide	Myofibromatosis, infantile, 1
PDGFRB, PDGFR, IBGC4, IMF1	Platelet-derived growth factor receptor, beta polypeptide	Myeloproliferative disorder with eosinophilia
PDGFRB, PDGFR, IBGC4, IMF1	Platelet-derived growth factor receptor, beta polypeptide	Basal ganglia calcification, idiopathic, 4
ESR1, ESR, ESTRR	Estrogen receptor 1	{Migraine, susceptibility to}
ESR1, ESR, ESTRR	Estrogen receptor 1	{HDL response to hormone replacement, augmented}
ESR1, ESR, ESTRR	Estrogen receptor 1	{Breast cancer}
ESR1, ESR, ESTRR	Estrogen receptor 1	{Atherosclerosis, susceptibility to}
ESR1, ESR, ESTRR	Estrogen receptor 1	Estrogen resistance
ESR1, ESR, ESTRR	Estrogen receptor 1	{Myocardial infarction, susceptibility to}
GARS, SMAD1, CMT2D, HMN5	Glycyl-tRNA synthetase	Neuropathy, distal hereditary motor, type VA
GARS, SMAD1, CMT2D, HMN5	Glycyl-tRNA synthetase	Charcot-Marie-Tooth disease, type 2D
FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS	Fibroblast growth factor receptor-1 (fms-related tyrosine kinase-2)	Pfeiffer syndrome
FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS	Fibroblast growth factor receptor-1 (fms-related tyrosine kinase-2)	Osteoglophonic dysplasia
FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS	Fibroblast growth factor receptor-1 (fms-related tyrosine kinase-2)	Jackson-Weiss syndrome
FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS	Fibroblast growth factor receptor-1 (fms-related tyrosine kinase-2)	Hypogonadotropic hypogonadism 2 with or without anosmia
FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS	Fibroblast growth factor receptor-1 (fms-related tyrosine kinase-2)	Hartsfield syndrome
FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS	Fibroblast growth factor receptor-1 (fms-related tyrosine kinase-2)	Trigonocephaly 1
PTCH1, NBCCS, BCNS, HPE7	Patched, Drosophila, homolog of	Basal cell nevus syndrome
PTCH1, NBCCS, BCNS, HPE7	Patched, Drosophila, homolog of	Basal cell carcinoma, somatic
PTCH1, NBCCS, BCNS, HPE7	Patched, Drosophila, homolog of	Holoprosencephaly-7
ABL1	Abelson murine leukemia viral (v-abl) oncogene homolog 1	Leukemia, Philadelphia chromosome-positive, resistant to imatinib
NUP214, D9S46E, CAN, CAIN	Nucleoporin, 214kD	Leukemia, T-cell acute lymphoblastic
NUP214, D9S46E, CAN, CAIN	Nucleoporin, 214kD	Leukemia, acute myeloid
LHX3, CPHD3	LIM/homeodomain protein LHX3	Pituitary hormone deficiency, combined, 3
NOTCH1, TAN1, AOS5, AOVD1	Notch, Drosophila, homolog of, 1, translocation-associated	Aortic valve disease 1
NOTCH1, TAN1, AOS5, AOVD1	Notch, Drosophila, homolog of, 1, translocation-associated	Adams-Oliver syndrome 5
AKR1C4, CHDR, CDR, HAKRA, DD4	Aldo-keto reductase family 1, member C4 (chlordecone reductase)	{46XY sex reversal 8, modifier of}

ETV6, TEL	ETS variant gene-6 (TEL oncogene)	Leukemia, acute myeloid, somatic
MAPT, MTBT1, DDPAC, MSTD	Microtubule-associated protein tau	Dementia, frontotemporal, with or without parkinsonism
MAPT, MTBT1, DDPAC, MSTD	Microtubule-associated protein tau	{Parkinson disease, susceptibility to}
MAPT, MTBT1, DDPAC, MSTD	Microtubule-associated protein tau	Supranuclear palsy, progressive atypical
MAPT, MTBT1, DDPAC, MSTD	Microtubule-associated protein tau	Supranuclear palsy, progressive
MAPT, MTBT1, DDPAC, MSTD	Microtubule-associated protein tau	Pick disease
PRKCA, PKCA	Protein kinase C, alpha polypeptide	Pituitary tumor, invasive
CACNA1A, CACNL1A4, SCA6	Calcium channel, voltage-dependent, P/Q type, alpha 1A subunit	Spinocerebellar ataxia 6
CACNA1A, CACNL1A4, SCA6	Calcium channel, voltage-dependent, P/Q type, alpha 1A subunit	Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia
CACNA1A, CACNL1A4, SCA6	Calcium channel, voltage-dependent, P/Q type, alpha 1A subunit	Migraine, familial hemiplegic, 1
CACNA1A, CACNL1A4, SCA6	Calcium channel, voltage-dependent, P/Q type, alpha 1A subunit	Episodic ataxia, type 2
DMPK, DM, DMK	Dystrophia myotonica-protein kinase	Myotonic dystrophy 1
HCF2, HC2, SERPIND1, THPH10	Heparin cofactor II	Thrombophilia due to heparin cofactor II deficiency
BCR, CML, PHL, ALL	Breakpoint cluster region	Leukemia, chronic myeloid
BCR, CML, PHL, ALL	Breakpoint cluster region	Leukemia, acute lymphocytic

Table S9. A table showing CNV genes found in the CEU group matching genes linked to specific disease phenotypes from the OMIM database.

CEU matches in OMIM		
Gene/Locus	**Gene/Locus name**	**Phenotype**
PINK1, PARK6	PTEN-induced putative kinase 1	Parkinson disease 6, early onset
XPR1, SYG1	Xenotropic and polytropic retrovirus receptor	N/A
ATP2B4, ATP2B2, PMCA4	ATPase, Ca++ transporting, plasma membrane, 4	N/A
AKT3, PKBG, MPPH2	v-Akt murine thymoma viral oncogene homolog 3	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2
COMMD1, MURR1	Copper metabolism Murr1 domain-containing 1	N/A
NEB, NEM2	Nebulin	Nemaline myopathy 2, autosomal recessive
SCAP	SREBP cleavage-activating protein	N/A
CCDC50, C3orf6, DFNA44	Coiled-coil domain-containing protein 50	Deafness, autosomal dominant 44
DLG1	Discs, large, Drosophila, homolog of, 1	N/A
UNC5C, UNC5H3	UNC5, C. elegans, homolog of, C	N/A
PPP3CA, PPP2B, CALNA, CNA1	Protein phosphatase-3 (formerly 2B), catalytic subunit, alpha isoform (calcineurin A alpha)	N/A
PRSS12, BSSP3, MRT1	Protease, serine, 12	Mental retardation, autosomal recessive 1
KIAA1109	KIAA1109 gene	N/A
XRCC4	X-ray repair, complementing defective, repair in Chinese hamster cells-4	N/A
APC, GS, FPC, BTPS2	Adenomatous polyposis coli	Brain tumor-polyposis syndrome 2
APC, GS, FPC, BTPS2	Adenomatous polyposis coli	Hepatoblastoma, somatic
APC, GS, FPC, BTPS2	Adenomatous polyposis coli	Adenomatous polyposis coli
APC, GS, FPC, BTPS2	Adenomatous polyposis coli	Gastric cancer, somatic
APC, GS, FPC, BTPS2	Adenomatous polyposis coli	Adenoma, periampullary, somatic
APC, GS, FPC, BTPS2	Adenomatous polyposis coli	Gardner syndrome
APC, GS, FPC, BTPS2	Adenomatous polyposis coli	Desmoid disease, hereditary

APC, GS, FPC, BTPS2	Adenomatous polyposis coli	Colorectal cancer, somatic
MCC	Mutated in colorectal cancers	Colorectal cancer, somatic
MEGF10, KIAA1780, EMARDD	Multiple epidermal growth factor-like domains 10	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant
MEGF10, KIAA1780, EMARDD	Multiple epidermal growth factor-like domains 10	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset
NCOA7, ERAP140	Nuclear receptor coactivator 7	N/A
ESR1, ESR, ESTRR	Estrogen receptor 1	{Atherosclerosis, susceptibility to}
ESR1, ESR, ESTRR	Estrogen receptor 1	Estrogen resistance
ESR1, ESR, ESTRR	Estrogen receptor 1	{Myocardial infarction, susceptibility to}
ESR1, ESR, ESTRR	Estrogen receptor 1	{Migraine, susceptibility to}
ESR1, ESR, ESTRR	Estrogen receptor 1	{HDL response to hormone replacement, augmented}
ESR1, ESR, ESTRR	Estrogen receptor 1	{Breast cancer}
GNA12	Guanine nucleotide-binding protein, alpha-12	N/A
BRAF, NS7	Murine sarcoma viral (v-raf) oncogene homolog B1	LEOPARD syndrome 3
BRAF, NS7	Murine sarcoma viral (v-raf) oncogene homolog B1	Colorectal cancer, somatic
BRAF, NS7	Murine sarcoma viral (v-raf) oncogene homolog B1	Cardiofaciocutaneous syndrome
BRAF, NS7	Murine sarcoma viral (v-raf) oncogene homolog B1	Noonan syndrome 7
BRAF, NS7	Murine sarcoma viral (v-raf) oncogene homolog B1	Adenocarcinoma of lung, somatic
BRAF, NS7	Murine sarcoma viral (v-raf) oncogene homolog B1	Nonsmall cell lung cancer, somatic
BRAF, NS7	Murine sarcoma viral (v-raf) oncogene homolog B1	Melanoma, malignant, somatic
RNF38	Ring finger protein 38	N/A
NRP1, NRP, VEGF165R	Neuropilin 1	N/A
PTEN, MMAC1, GLM2, CWS1	Phosphatase and tensin homolog (mutated in multiple advanced cancers 1)	Bannayan-Riley-Ruvalcaba syndrome
PTEN, MMAC1, GLM2, CWS1	Phosphatase and tensin homolog (mutated in multiple advanced cancers 1)	Malignant melanoma, somatic
PTEN, MMAC1, GLM2, CWS1	Phosphatase and tensin homolog (mutated in multiple advanced cancers 1)	{Glioma susceptibility 2}
PTEN, MMAC1, GLM2, CWS1	Phosphatase and tensin homolog (mutated in multiple advanced cancers 1)	Macrocephaly/autism syndrome
PTEN, MMAC1, GLM2, CWS1	Phosphatase and tensin homolog (mutated in multiple advanced cancers 1)	VATER association with macrocephaly and ventriculomegaly
PTEN, MMAC1, GLM2, CWS1	Phosphatase and tensin homolog (mutated in multiple advanced cancers 1)	Lhermitte-Duclos syndrome
PTEN, MMAC1, GLM2, CWS1	Phosphatase and tensin homolog (mutated in multiple advanced cancers 1)	Thyroid carcinoma, follicular, somatic
PTEN, MMAC1, GLM2, CWS1	Phosphatase and tensin homolog (mutated in multiple advanced cancers 1)	Endometrial carcinoma, somatic
PTEN, MMAC1, GLM2, CWS1	Phosphatase and tensin homolog (mutated in multiple advanced cancers 1)	Squamous cell carcinoma, head and neck, somatic
PTEN, MMAC1, GLM2, CWS1	Phosphatase and tensin homolog (mutated in multiple advanced cancers 1)	{Prostate cancer, somatic}
PTEN, MMAC1, GLM2, CWS1	Phosphatase and tensin homolog (mutated in multiple advanced cancers 1)	Cowden syndrome 1
PTEN, MMAC1, GLM2, CWS1	Phosphatase and tensin homolog (mutated in multiple advanced cancers 1)	PTEN hamartoma tumor syndrome
PTEN, MMAC1, GLM2, CWS1	Phosphatase and tensin homolog (mutated in multiple advanced cancers 1)	{Meningioma}
PARVA	Parvin, alpha	N/A
ADIPOR2, FLJ21432	Adiponectin receptor 2	N/A

CD4	CD4 antigen (p55)	OKT4 epitope deficiency
ATP10A, ATP10C, ATPVC	ATPase, class V, type 10A	N/A
MAP2K1, PRKMK1, MKK1, MEK1, CFC3	Mitogen-activated protein kinase kinase 1	Cardiofaciocutaneous syndrome 3
SCAPER, KIAA1454	S-phase cyclin A-associated protein in the endoplasmic reticulum	N/A
AKAP13, HT31, LBC, BRX	A-kinase anchor protein 13	N/A
WWOX, FOR, SCAR12	WW domain-containing oxidoreductase	Spinocrebellar ataxia, autosomal recessive 12
WWOX, FOR, SCAR12	WW domain-containing oxidoreductase	Esophageal squamous cell carcinoma, somatic
NTN1, NTN1L	Netrin 1, mouse, homolog of	N/A
MSI2	Musashi, Drosophila, homolog of, 2	N/A
GTF2F1, RAP74	General transcription factor IIF, polypeptide 1 (74kD subunit)	N/A
TAF4, TAF2C1, TAFII130, TAF2C	TAF4 RNA polymerase II, TATA box-binding protein-associated factor, 135kD	N/A
SF3A1, SF3A120, SAP114, PRP21	Splicing factor 3A, subunit 1	N/A

Table S10. A table showing CNV genes found in the YRI group matching genes linked to specific disease phenotypes from the OMIM database.

YRI matches in OMIM		
Gene/Locus	**Gene/Locus name**	**Phenotype**
ACOT11, THEA, BFIT, BFIT1, BFIT2, KIAA0707	Acyl-CoA thioesterase 11	N/A
XPR1, SYG1	Xenotropic and polytropic retrovirus receptor	N/A
PPP2R5A	Protein phosphatase-2, regulatory subunit B (B56), alpha isoform	N/A
LTBP1	Latent transforming growth factor beta binding protein 1	N/A
PROC, PC, THPH3, THPH4	Protein C (inactivator of coagulation factors Va and VIIIa)	Thrombophilia due to protein C deficiency, autosomal recessive
PROC, PC, THPH3, THPH4	Protein C (inactivator of coagulation factors Va and VIIIa)	Thrombophilia due to protein C deficiency, autosomal dominant
MAP2	Microtubule-associated protein-2	N/A
UGT1A8	UDP-glycosyltransferase 1 family, polypeptide A8	N/A
UGT1A10	UDP-glycosyltransferase 1 family, polypeptide A10	N/A
UGT1A9	UDP-glycosyltransferase 1 family, polypeptide A9	N/A
UGT1A7	UDP-glycosyltransferase 1 family, polypeptide A7	N/A
UGT1A6	UDP-glycosyltransferase 1 family, polypeptide A6	N/A
UGT1A5	UDP-glycosyltransferase 1 family, polypeptide A5	N/A
UGT1A4, UGT1D	UDP-glycosyltransferase 1 family, polypeptide A4	N/A
CNTN6, NB3	Contactin 6	N/A
KCNAB1, KCNA1B	Potassium voltage-gated channel, shaker-related subfamily, beta member 1	N/A
KPNA4, QIP1	Karyopherin alpha-4	N/A
IGF2BP2, IMP2	Insulin-like growth factor 2 mRNA-binding protein 2	{Diabetes mellitus, noninsulin-dependent, susceptibility to}
DGKG, DAGK3	Diacylglycerol kinase, gamma, 90-kD	N/A

DLG1	Discs, large, Drosophila, homolog of, 1	N/A
GPR125	G protein-coupled receptor 125	N/A
PDGFRA	Platelet-derived growth factor receptor, alpha polypeptide	Gastrointestinal stromal tumor, somatic
PDGFRA	Platelet-derived growth factor receptor, alpha polypeptide	Hypereosinophilic syndrome, idiopathic, resistant to imatinib
UGT2B17, BMND12	UDP-glucuronyltransferase, family 2, beta-17	{Bone mineral density QTL 12, osteoporosis}
BMPR1B, ALK6	Bone morphogenetic protein receptor, type IB	Chrondrodysplasia, acromesomelic, with genital anomalies
BMPR1B, ALK6	Bone morphogenetic protein receptor, type IB	Brachydactyly, type A2
PPP3CA, PPP2B, CALNA, CNA1	Protein phosphatase-3 (formerly 2B), catalytic subunit, alpha isoform (calcineurin A alpha)	N/A
SMAD1, MADH1, MADR1, BSP1	Mothers against decapentaplegic, Drosophila, homolog of, 1	N/A
APC, GS, FPC, BTPS2	Adenomatous polyposis coli	Colorectal cancer, somatic
APC, GS, FPC, BTPS2	Adenomatous polyposis coli	Brain tumor-polyposis syndrome 2
APC, GS, FPC, BTPS2	Adenomatous polyposis coli	Hepatoblastoma, somatic
APC, GS, FPC, BTPS2	Adenomatous polyposis coli	Adenomatous polyposis coli
APC, GS, FPC, BTPS2	Adenomatous polyposis coli	Gastric cancer, somatic
APC, GS, FPC, BTPS2	Adenomatous polyposis coli	Adenoma, periampullary, somatic
APC, GS, FPC, BTPS2	Adenomatous polyposis coli	Gardner syndrome
APC, GS, FPC, BTPS2	Adenomatous polyposis coli	Desmoid disease, hereditary
MCC	Mutated in colorectal cancers	Colorectal cancer, somatic
MEGF10, KIAA1780, EMARDD	Multiple epidermal growth factor-like domains 10	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant
MEGF10, KIAA1780, EMARDD	Multiple epidermal growth factor-like domains 10	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset
FBN2, CCA, EOMD	Fibrillin-2	Macular degeneration, early-onset
FBN2, CCA, EOMD	Fibrillin-2	Contractural arachnodactyly, congenital
IRF1, MAR	Interferon regulatory factor-1	Myelodysplastic syndrome, preleukemic
IRF1, MAR	Interferon regulatory factor-1	Gastric cancer, somatic
IRF1, MAR	Interferon regulatory factor-1	Nonsmall cell lung cancer, somatic
IRF1, MAR	Interferon regulatory factor-1	Myelogenous leukemia, acute
TRDN, TDN, CPVT5	Triadin	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness
GARS, SMAD1, CMT2D, HMN5	Glycyl-tRNA synthetase	Charcot-Marie-Tooth disease, type 2D
GARS, SMAD1, CMT2D, HMN5	Glycyl-tRNA synthetase	Neuropathy, distal hereditary motor, type VA
P100, SND1	EBNA-2 coactivator p100	N/A
MGAM, MGA	Maltase-glucoamylase	N/A
PCM1, PTC4	Pericentriolar material 1	Thyroid carcinoma, papillary
FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS	Fibroblast growth factor receptor-1 (fms-related tyrosine kinase-2)	Osteoglophonic dysplasia
FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS	Fibroblast growth factor receptor-1 (fms-related tyrosine kinase-2)	Jackson-Weiss syndrome
FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS	Fibroblast growth factor receptor-1 (fms-related tyrosine kinase-2)	Hypogonadotropic hypogonadism 2 with or without anosmia
FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS	Fibroblast growth factor receptor-1 (fms-related tyrosine kinase-2)	Hartsfield syndrome

FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS	Fibroblast growth factor receptor-1 (fms-related tyrosine kinase-2)	Trigonocephaly 1
FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS	Fibroblast growth factor receptor-1 (fms-related tyrosine kinase-2)	Pfeiffer syndrome
ASPH, HAAH, FDLAB	Aspartate beta-hydroxylase (junctin; junctate)	Traboulsi syndrome
ABL1	Abelson murine leukemia viral (v-abl) oncogene homolog 1	Leukemia, Philadelphia chromosome-positive, resistant to imatinib
NUP214, D9S46E, CAN, CAIN	Nucleoporin, 214kD	Leukemia, acute myeloid
NUP214, D9S46E, CAN, CAIN	Nucleoporin, 214kD	Leukemia, T-cell acute lymphoblastic
AKR1C4, CHDR, CDR, HAKRA, DD4	Aldo-keto reductase family 1, member C4 (chlordecone reductase)	{46XY sex reversal 8, modifier of}
NRP1, NRP, VEGF165R	Neuropilin 1	N/A
PLCE1, KIAA1516, NPHS3	Phospholipase C, epsilon-1	Nephrotic syndrome, type 3
RRAS2, TC21	Related Ras viral oncogene homolog 2	Ovarian carcinoma
PC	Pyruvate carboxylase	Pyruvate carboxylase deficiency
PRCP, PCP	Prolylcarboxypeptidase (angiotensinase C)	N/A
ME3	Malic enzyme 3	N/A
ADIPOR2, FLJ21432	Adiponectin receptor 2	N/A
CD4	CD4 antigen (p55)	OKT4 epitope deficiency
PCS	Parotid proline-rich salivary protein Pc	N/A
PIK3C2G	Phosphatidylinositol 3-kinase, class 2, gamma	N/A
DCLK1, DCAMKL1, CLICK1, CL1, KIAA0369	Doublecortin-like kinase 1	N/A
KLHL1	Kelch-like 1	N/A
ATP10A, ATP10C, ATPVC	ATPase, class V, type 10A	N/A
MGA, KIAA0518	MAX dimerization protein MGA	N/A
SCAPER, KIAA1454	S-phase cyclin A-associated protein in the endoplasmic reticulum	N/A
ABR	Active BCR-related gene	N/A
C1QBP, HABP1	Complement component C1q binding protein (hyaluronic acid-binding protein 1)	N/A
MAPT, MTBT1, DDPAC, MSTD	Microtubule-associated protein tau	Pick disease
MAPT, MTBT1, DDPAC, MSTD	Microtubule-associated protein tau	Dementia, frontotemporal, with or without parkinsonism
MAPT, MTBT1, DDPAC, MSTD	Microtubule-associated protein tau	{Parkinson disease, susceptibility to}
MAPT, MTBT1, DDPAC, MSTD	Microtubule-associated protein tau	Supranuclear palsy, progressive atypical
MAPT, MTBT1, DDPAC, MSTD	Microtubule-associated protein tau	Supranuclear palsy, progressive
MBD1, PCM1	Methyl=CpG binding domain protein 1 (protein containing methyl-CpG binding domain 1)	N/A
PLCB4, ARCND2	Phospholipase C, beta 4	Auriculocondylar syndrome 2
RPN2	Ribophorin II	N/A
HCF2, HC2, SERPIND1, THPH10	Heparin cofactor II	Thrombophilia due to heparin cofactor II deficiency
BCR, CML, PHL, ALL	Breakpoint cluster region	Leukemia, chronic myeloid
BCR, CML, PHL, ALL	Breakpoint cluster region	Leukemia, acute lymphocytic

Table S11. A table showing CNV genes found in the ASN CNV pairs matching genes linked to specific disease phenotypes from the OMIM database.

Gene/Locus	Gene/Locus name	Phenotype
NEGR1, KILON	Neuronal growth regulator 1	N/A
PLA2G4A, PLA2G4	Phospholipase A2, group IVA, cytosolic	Phospholipase A2, group IV A, deficiency of
LTBP1	Latent transforming growth factor beta binding protein 1	N/A
ITGA6	Integrin, alpha-6	Epidermolysis bullosa, junctional, with pyloric stenosis
PPA2	Pyrophosphatase, inorganic, 2	N/A
TRIM2, KIAA0517, CMT2R	Tripartite motif-containing protein 2	Charcot-Marie-Tooth disease, type 2R
FBN2, CCA, EOMD	Fibrillin-2	Macular degeneration, early-onset
FBN2, CCA, EOMD	Fibrillin-2	Contractural arachnodactyly, congenital
LAMA2, LAMM	Laminin, alpha-2 (merosin)	Muscular dystrophy, congenital, due to partial LAMA2 deficiency
LAMA2, LAMM	Laminin, alpha-2 (merosin)	Muscular dystrophy, congenital merosin-deficient
P100, SND1	EBNA-2 coactivator p100	N/A
LYPLA1	Lysophospholipase I	N/A
PTCH1, NBCCS, BCNS, HPE7	Patched, Drosophila, homolog of	Basal cell carcinoma, somatic
PTCH1, NBCCS, BCNS, HPE7	Patched, Drosophila, homolog of	Holoprosencephaly-7
PTCH1, NBCCS, BCNS, HPE7	Patched, Drosophila, homolog of	Basal cell nevus syndrome
GPR51, GABBR2	G protein-coupled receptor 51	{Nicotine dependence, susceptibility to}
GPR51, GABBR2	G protein-coupled receptor 51	{Nicotine dependence, protection against}
NRP1, NRP, VEGF165R	Neuropilin 1	N/A
ATP6V0A2, WSS, ARCL2A	ATPase, H+ transporting, lysosomal, V0 subunit A2	Wrinkly skin syndrome
ATP6V0A2, WSS, ARCL2A	ATPase, H+ transporting, lysosomal, V0 subunit A2	Cutis laxa, autosomal recessive, type IIA
SH3GL3	SH3-domain GRB2-like 3	N/A
CNTNAP4, CASPR4, KIAA1763	Contactin-associated protein-like 4	N/A
MAST3, KIAA0561	Microtubule-associated serine/threonine kinase 3	N/A

Table S12. A table showing CNV genes found in the CEU CNV pairs matching genes linked to specific disease phenotypes from the OMIM database.

Gene/Locus	Gene/Locus name	Phenotype
PLA2G4A, PLA2G4	Phospholipase A2, group IVA, cytosolic	Phospholipase A2, group IV A, deficiency of
SATB2, KIAA1034, GLSS	Special AT-rich sequence-binding protein 2	Glass syndrome
EGFR, NISBD2	Epidermal growth factor receptor	?Inflammatory skin and bowel disease, neonatal, 2
EGFR, NISBD2	Epidermal growth factor receptor	Adenocarcinoma of lung, response to tyrosine kinase inhibitor in
EGFR, NISBD2	Epidermal growth factor receptor	Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in
EGFR, NISBD2	Epidermal growth factor receptor	{Nonsmall cell lung cancer, susceptibility to}
CD4	CD4 antigen (p55)	OKT4 epitope deficiency
SLC12A1, NKCC2	Solute carrier family 12 (sodium/potassium/chloride transporters), member 1	Bartter syndrome, type 1

WWOX, FOR, SCAR12	WW domain-containing oxidoreductase	Esophageal squamous cell carcinoma, somatic
WWOX, FOR, SCAR12	WW domain-containing oxidoreductase	Spinocrebellar ataxia, autosomal recessive 12

Table S13. A table showing CNV genes found in the YRI CNV pairs matching genes linked to specific disease phenotypes from the OMIM database.

Gene/Locus	Gene/Locus name	Phenotype
DAP3	Death associated protein 3	N/A
CFHR1, FHR1, HFL1, CFHL1	Complement factor H-related 1	{Hemolytic uremic syndrome, atypical, susceptibility to}
CFHR1, FHR1, HFL1, CFHL1	Complement factor H-related 1	{Macular degeneration, age-related, reduced risk of}
C4BPA	Complement component 4-binding protein, alpha polypeptide	N/A
BMPR2, PPH1, POVD1	Bone morphogenetic receptor, type II	Pulmonary venoocclusive disease 1
BMPR2, PPH1, POVD1	Bone morphogenetic receptor, type II	Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated
BMPR2, PPH1, POVD1	Bone morphogenetic receptor, type II	Pulmonary hypertension, familial primary, 1, with or without HHT
EGFR, NISBD2	Epidermal growth factor receptor	{Nonsmall cell lung cancer, susceptibility to}
EGFR, NISBD2	Epidermal growth factor receptor	Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in
EGFR, NISBD2	Epidermal growth factor receptor	Adenocarcinoma of lung, response to tyrosine kinase inhibitor in
EGFR, NISBD2	Epidermal growth factor receptor	?Inflammatory skin and bowel disease, neonatal, 2
P100, SND1	EBNA-2 coactivator p100	N/A
GPR51, GABBR2	G protein-coupled receptor 51	{Nicotine dependence, susceptibility to}
GPR51, GABBR2	G protein-coupled receptor 51	{Nicotine dependence, protection against}
PTPRJ, DEP1	Protein tyrosine phosphatase, receptor type, J polypeptide	Colon cancer, somatic
DRD2	Dopamine receptor D2	Dystonia, myoclonic
APOA5	Apolipoprotein A-V	{Hypertriglyceridemia, susceptibility to}
APOA5	Apolipoprotein A-V	Hyperchylomicronemia, late-onset
APOA4	Apolipoprotein A-IV	N/A
APOC3, HALP2	Apolipoprotein C-III	Apolipoprotein C-III deficiency
APOA1	Apolipoprotein A-I	Hypoalphalipoproteinemia
APOA1	Apolipoprotein A-I	Corneal clouding, autosomal recessive
APOA1	Apolipoprotein A-I	ApoA-I and apoC-III deficiency, combined
APOA1	Apolipoprotein A-I	Amyloidosis, 3 or more types
SIK3, KIAA0999	Salt-inducible kinase 3	N/A
ADIPOR2, FLJ21432	Adiponectin receptor 2	N/A
CD4	CD4 antigen (p55)	OKT4 epitope deficiency
HSP90AA1, HSPCA, HSPC1, HSP90A, HSP89A, HSPCAL4, LAP2	Heat-shock protein, 90kD, alpha, class A, member 1	N/A
ATP10A, ATP10C, ATPVC	ATPase, class V, type 10A	N/A
SH3GL3	SH3-domain GRB2-like 3	N/A

CNTNAP4, CASPR4, KIAA1763	Contactin-associated protein-like 4	N/A
MAST3, KIAA0561	Microtubule-associated serine/threonine kinase 3	N/A

Table S14. A table showing biological GO terms of identified CNV genes with a cutoff p-value of 0.01 in the CEU group.

Term	P-value
organ development (GO:0048513)	0.0000008
positive regulation of macromolecule metabolic process (GO:0010604)	0.0000231
positive regulation of metabolic process (GO:0009893)	0.0000492
positive regulation of cellular metabolic process (GO:0031325)	0.0000898
regulation of cellular process (GO:0050794)	0.0001038
system development (GO:0048731)	0.0002277
regulation of multicellular organismal process (GO:0051239)	0.0002951
regulation of biological process (GO:0050789)	0.0003105
regulation of developmental process (GO:0050793)	0.0003360
positive regulation of cellular process (GO:0048522)	0.0003914
anatomical structure development (GO:0048856)	0.0004357
multicellular organismal development (GO:0007275)	0.0004461
response to chemical (GO:0042221)	0.0004502
positive regulation of biological process (GO:0048518)	0.0005009
regulation of cell migration (GO:0030334)	0.0005106
single-organism developmental process (GO:0044767)	0.0007314
regulation of cell motility (GO:2000145)	0.0007363
cell development (GO:0048468)	0.0008301
developmental process (GO:0032502)	0.0008817
cell differentiation (GO:0030154)	0.0008912
reproductive structure development (GO:0048608)	0.0009907
regulation of cell morphogenesis (GO:0022604)	0.0010060
biological regulation (GO:0065007)	0.0010370
reproductive system development (GO:0061458)	0.0010550
response to stimulus (GO:0050896)	0.0011750
regulation of locomotion (GO:0040012)	0.0014300
cellular developmental process (GO:0048869)	0.0016350
regulation of cellular component movement (GO:0051270)	0.0016660
cell surface receptor signaling pathway (GO:0007166)	0.0016930
cell morphogenesis involved in differentiation (GO:0000904)	0.0018270
positive regulation of molecular function (GO:0044093)	0.0018670
organ morphogenesis (GO:0009887)	0.0021500
axonogenesis (GO:0007409)	0.0022350

positive regulation of catalytic activity (GO:0043085)	0.0023490
axon development (GO:0061564)	0.0029270
cellular response to stimulus (GO:0051716)	0.0030500
cell projection morphogenesis (GO:0048858)	0.0030960
cell morphogenesis involved in neuron differentiation (GO:0048667)	0.0036930
cell part morphogenesis (GO:0032990)	0.0037320
regulation of cellular component organization (GO:0051128)	0.0039280
anterior/posterior axon guidance (GO:0033564)	0.0042440
dichotomous subdivision of terminal units involved in salivary gland branching (GO:0060666)	0.0042440
neuron projection morphogenesis (GO:0048812)	0.0044510
response to external stimulus (GO:0009605)	0.0045430
gland morphogenesis (GO:0022612)	0.0046760
T cell activation (GO:0042110)	0.0048160
regulation of cellular component size (GO:0032535)	0.0052590
axon guidance (GO:0007411)	0.0057470
neuron projection guidance (GO:0097485)	0.0057470
cellular response to chemical stimulus (GO:0070887)	0.0058460
regulation of anatomical structure morphogenesis (GO:0022603)	0.0068880
regulation of muscle adaptation (GO:0043502)	0.0076530
negative regulation of signaling (GO:0023057)	0.0077950
negative regulation of cell communication (GO:0010648)	0.0079770
regulation of molecular function (GO:0065009)	0.0086230
developmental process involved in reproduction (GO:0003006)	0.0091680
chemotaxis (GO:0006935)	0.0097760
taxis (GO:0042330)	0.0097760

Table S15. A table showing molecular GO terms of identified CNV genes with a cutoff p-value of 0.01 in the CEU group.

Term	**P-value**
protein binding (GO:0005515)	0.0000111
phosphoprotein phosphatase activity (GO:0004721)	0.0006978
enzyme binding (GO:0019899)	0.0013290
nitric-oxide synthase regulator activity (GO:0030235)	0.0045920
binding (GO:0005488)	0.0046730
phosphatase activity (GO:0016791)	0.0051200
molecular_function (GO:0003674)	0.0052580

Table S16. A table showing cellular GO terms of identified CNV genes with a cutoff p-value of 0.01 in the CEU group.

Term	P-value
cell projection (GO:0042995)	0.0004382
Z disc (GO:0030018)	0.0015260
plasma membrane (GO:0005886)	0.0020200
I band (GO:0031674)	0.0025290
cell periphery (GO:0071944)	0.0028250

Table S17. A table showing biological GO terms of identified CNV genes with a cutoff p-value of 0.01 in the ASN group.

Term	P-value
regulation of multicellular organismal process (GO:0051239)	0.0000000
regulation of developmental process (GO:0050793)	0.0000001
cell differentiation (GO:0030154)	0.0000001
cellular developmental process (GO:0048869)	0.0000004
regulation of cell differentiation (GO:0045595)	0.0000004
negative regulation of biological process (GO:0048519)	0.0000007
response to chemical (GO:0042221)	0.0000011
regulation of biological process (GO:0050789)	0.0000011
regulation of cellular component movement (GO:0051270)	0.0000012
cell morphogenesis involved in neuron differentiation (GO:0048667)	0.0000012
response to stimulus (GO:0050896)	0.0000013
regulation of cellular process (GO:0050794)	0.0000013
regulation of myotube differentiation (GO:0010830)	0.0000017
positive regulation of biological process (GO:0048518)	0.0000018
generation of neurons (GO:0048699)	0.0000019
positive regulation of developmental process (GO:0051094)	0.0000020
regulation of osteoblast differentiation (GO:0045667)	0.0000023
anatomical structure morphogenesis (GO:0009653)	0.0000027
positive regulation of cellular process (GO:0048522)	0.0000033
positive regulation of cell differentiation (GO:0045597)	0.0000033
regulation of metabolic process (GO:0019222)	0.0000045
neurogenesis (GO:0022008)	0.0000046
regulation of multicellular organismal development (GO:2000026)	0.0000052
regulation of muscle cell differentiation (GO:0051147)	0.0000059
regulation of skeletal muscle fiber development (GO:0048742)	0.0000064

biological regulation (GO:0065007)	0.0000067
negative regulation of cellular process (GO:0048523)	0.0000109
neuron development (GO:0048666)	0.0000110
cellular response to stimulus (GO:0051716)	0.0000111
regulation of cellular metabolic process (GO:0031323)	0.0000114
neuron projection development (GO:0031175)	0.0000120
cell morphogenesis involved in differentiation (GO:0000904)	0.0000133
neuron differentiation (GO:0030182)	0.0000149
regulation of molecular function (GO:0065009)	0.0000161
regulation of striated muscle cell differentiation (GO:0051153)	0.0000168
neuron projection morphogenesis (GO:0048812)	0.0000175
cell development (GO:0048468)	0.0000178
developmental process (GO:0032502)	0.0000189
regulation of cell development (GO:0060284)	0.0000210
cellular response to chemical stimulus (GO:0070887)	0.0000211
cell surface receptor signaling pathway (GO:0007166)	0.0000265
regulation of localization (GO:0032879)	0.0000344
regulation of cell motility (GO:2000145)	0.0000344
nervous system development (GO:0007399)	0.0000395
signal transduction (GO:0007165)	0.0000460
cell morphogenesis (GO:0000902)	0.0000517
single-organism developmental process (GO:0044767)	0.0000614
axonogenesis (GO:0007409)	0.0000643
regulation of ossification (GO:0030278)	0.0000655
anatomical structure development (GO:0048856)	0.0000746
gland morphogenesis (GO:0022612)	0.0000759
regulation of locomotion (GO:0040012)	0.0000853
axon development (GO:0061564)	0.0000945
regulation of primary metabolic process (GO:0080090)	0.0000953
regulation of anatomical structure morphogenesis (GO:0022603)	0.0000987
cellular response to organic substance (GO:0071310)	0.0001033
positive regulation of developmental growth (GO:0048639)	0.0001047
growth (GO:0040007)	0.0001064
regulation of biological quality (GO:0065008)	0.0001067
regulation of skeletal muscle tissue development (GO:0048641)	0.0001214
cellular component morphogenesis (GO:0032989)	0.0001273
cellular component organization (GO:0016043)	0.0001329
positive regulation of neurogenesis (GO:0050769)	0.0001406
single-organism cellular process (GO:0044763)	0.0001520
regulation of cell migration (GO:0030334)	0.0002009
positive regulation of multicellular organismal process (GO:0051240)	0.0002030
negative regulation of metabolic process (GO:0009892)	0.0002178
cellular component organization or biogenesis (GO:0071840)	0.0002275
developmental growth (GO:0048589)	0.0002280
cell projection morphogenesis (GO:0048858)	0.0002452

organ development (GO:0048513)	0.0002734
multicellular organismal development (GO:0007275)	0.0003073
dendrite development (GO:0016358)	0.0003154
cell part morphogenesis (GO:0032990)	0.0003165
positive regulation of nervous system development (GO:0051962)	0.0003219
system development (GO:0048731)	0.0003308
single organism signaling (GO:0044700)	0.0003477
signaling (GO:0023052)	0.0003477
response to organic substance (GO:0010033)	0.0003544
regulation of developmental growth (GO:0048638)	0.0004215
limb morphogenesis (GO:0035108)	0.0004488
appendage morphogenesis (GO:0035107)	0.0004488
cell communication (GO:0007154)	0.0005154
positive regulation of metabolic process (GO:0009893)	0.0006064
cell projection organization (GO:0030030)	0.0006665
negative regulation of molecular function (GO:0044092)	0.0006741
positive regulation of cell development (GO:0010720)	0.0008126
regulation of cellular component organization (GO:0051128)	0.0008159
limb development (GO:0060173)	0.0009132
appendage development (GO:0048736)	0.0009132
enzyme linked receptor protein signaling pathway (GO:0007167)	0.0009488
regulation of nucleobase-containing compound metabolic process (GO:0019219)	0.0009658
intracellular signal transduction (GO:0035556)	0.0010260
regulation of endothelial cell migration (GO:0010594)	0.0010710
regulation of muscle adaptation (GO:0043502)	0.0011380
single-multicellular organism process (GO:0044707)	0.0011850
regulation of neurogenesis (GO:0050767)	0.0012260
regulation of muscle system process (GO:0090257)	0.0012410
response to drug (GO:0042493)	0.0012590
central nervous system development (GO:0007417)	0.0013270
regulation of blood vessel endothelial cell migration (GO:0043535)	0.0013590
developmental growth involved in morphogenesis (GO:0060560)	0.0013740
regulation of nitrogen compound metabolic process (GO:0051171)	0.0014900
regulation of neuron projection development (GO:0010975)	0.0015260
regulation of cell projection organization (GO:0031344)	0.0016100
membrane depolarization (GO:0051899)	0.0017410
cellular process (GO:0009987)	0.0017610
positive regulation of growth (GO:0045927)	0.0018190
regulation of neuron differentiation (GO:0045664)	0.0018990
organ morphogenesis (GO:0009887)	0.0020940
regulation of striated muscle tissue development (GO:0016202)	0.0021790
regulation of muscle tissue development (GO:1901861)	0.0022770
neuron migration (GO:0001764)	0.0023770
regulation of muscle organ development (GO:0048634)	0.0023770
connective tissue development (GO:0061448)	0.0023830

regulation of transport (GO:0051049)	0.0025360
cellular glucuronidation (GO:0052695)	0.0027820
positive regulation of cellular metabolic process (GO:0031325)	0.0029910
multicellular organismal process (GO:0032501)	0.0029940
single-organism process (GO:0044699)	0.0030770
regulation of nervous system development (GO:0051960)	0.0030950
positive regulation of endothelial cell migration (GO:0010595)	0.0031910
uronic acid metabolic process (GO:0006063)	0.0032960
glucuronate metabolic process (GO:0019585)	0.0032960
positive regulation of osteoblast differentiation (GO:0045669)	0.0034190
chondrocyte differentiation (GO:0002062)	0.0034190
positive regulation of nucleobase-containing compound metabolic process (GO:0045935)	0.0037870
regulation of vesicle-mediated transport (GO:0060627)	0.0038180
cell death (GO:0008219)	0.0038730
death (GO:0016265)	0.0040540
locomotion (GO:0040011)	0.0042420
negative regulation of sequence-specific DNA binding transcription factor activity (GO:0043433)	0.0043490
positive regulation of neuron differentiation (GO:0045666)	0.0044880
positive regulation of cell morphogenesis involved in differentiation (GO:0010770)	0.0045160
positive regulation of nitrogen compound metabolic process (GO:0051173)	0.0045440
response to external stimulus (GO:0009605)	0.0045490
regulation of signaling (GO:0023051)	0.0047390
embryo development (GO:0009790)	0.0047560
regulation of cell communication (GO:0010646)	0.0048940
negative regulation of cellular metabolic process (GO:0031324)	0.0049080
biological_process (GO:0008150)	0.0051040
regulation of cellular localization (GO:0060341)	0.0051490
negative regulation of ossification (GO:0030279)	0.0053600
regulation of epithelial cell migration (GO:0010632)	0.0058240
brain development (GO:0007420)	0.0059450
sequestering of TGFbeta in extracellular matrix (GO:0035583)	0.0062230
positive regulation of blood vessel endothelial cell migration (GO:0043536)	0.0068120
positive regulation of molecular function (GO:0044093)	0.0072660
regulation of catabolic process (GO:0009894)	0.0072870
positive regulation of neuron projection development (GO:0010976)	0.0084540
striated muscle tissue development (GO:0014706)	0.0086450
cellular response to vascular endothelial growth factor stimulus (GO:0035924)	0.0087160
regulation of catalytic activity (GO:0050790)	0.0092190
head development (GO:0060322)	0.0092510
cartilage development (GO:0051216)	0.0093100
negative regulation of cellular biosynthetic process (GO:0031327)	0.0094520
negative regulation of signaling (GO:0023057)	0.0096180
regulation of myoblast differentiation (GO:0045661)	0.0097860
negative regulation of cell communication (GO:0010648)	0.0098830

Table S18. A table showing molecular GO terms of identified CNV genes with a cutoff p-value of 0.01 in the ASN group.

Term	P-value
protein binding (GO:0005515)	0.0000000
binding (GO:0005488)	0.0000013
molecular_function (GO:0003674)	0.0000065
enzyme binding (GO:0019899)	0.0001235
protein kinase activity (GO:0004672)	0.0002179
core promoter binding (GO:0001047)	0.0008579
phosphotransferase activity, alcohol group as acceptor (GO:0016773)	0.0008896
transferase activity (GO:0016740)	0.0009872
protein serine/threonine kinase activity (GO:0004674)	0.0013090
kinase activity (GO:0016301)	0.0018540
retinoic acid binding (GO:0001972)	0.0022510
small molecule binding (GO:0036094)	0.0039500
catalytic activity (GO:0003824)	0.0040540
enzyme regulator activity (GO:0030234)	0.0050260
glucuronosyltransferase activity (GO:0015020)	0.0056230
transferase activity, transferring phosphorus-containing groups (GO:0016772)	0.0082070

Table S19. A table showing cellular GO terms of identified CNV genes with a cutoff p-value of 0.01 in the ASN group.

Term	P-value
cytosol (GO:0005829)	0.0000835
cellular_component (GO:0005575)	0.0004518
membrane-bounded organelle (GO:0043227)	0.0005780
nucleus (GO:0005634)	0.0006384
cytoplasm (GO:0005737)	0.0010120
organelle (GO:0043226)	0.0019970
dendrite (GO:0030425)	0.0022990
nuclear lumen (GO:0031981)	0.0024160
nucleoplasm (GO:0005654)	0.0034200
somatodendritic compartment (GO:0036477)	0.0035710
nuclear part (GO:0044428)	0.0036080
neuron projection (GO:0043005)	0.0061360
macromolecular complex (GO:0032991)	0.0075980

Table S20. A table showing biological GO terms of identified CNV genes with a cutoff p-value of 0.01 in the YRI group.

Term	P-value
response to stimulus (GO:0050896)	0.0000001
single-organism cellular process (GO:0044763)	0.0000009
single-organism process (GO:0044699)	0.0000028
single organism signaling (GO:0044700)	0.0000053
signaling (GO:0023052)	0.0000053
cell communication (GO:0007154)	0.0000082
regulation of cellular process (GO:0050794)	0.0000085
signal transduction (GO:0007165)	0.0000136
regulation of biological process (GO:0050789)	0.0000334
intracellular signal transduction (GO:0035556)	0.0000337
cellular response to stimulus (GO:0051716)	0.0000409
regulation of biological quality (GO:0065008)	0.0000595
developmental process (GO:0032502)	0.0000894
regulation of localization (GO:0032879)	0.0001452
biological regulation (GO:0065007)	0.0001499
neuron development (GO:0048666)	0.0001641
axonogenesis (GO:0007409)	0.0001726
neurogenesis (GO:0022008)	0.0002076
neuron projection development (GO:0031175)	0.0002285
axon development (GO:0061564)	0.0002438
single-organism developmental process (GO:0044767)	0.0002818
single-multicellular organism process (GO:0044707)	0.0002870
cell morphogenesis involved in neuron differentiation (GO:0048667)	0.0003283
anatomical structure development (GO:0048856)	0.0004125
neuron projection morphogenesis (GO:0048812)	0.0004169
cell projection morphogenesis (GO:0048858)	0.0004785
regulation of multicellular organismal process (GO:0051239)	0.0005721
cell part morphogenesis (GO:0032990)	0.0006032
cell development (GO:0048468)	0.0006156
generation of neurons (GO:0048699)	0.0006158
multicellular organismal process (GO:0032501)	0.0007262
cellular process (GO:0009987)	0.0008723
cell differentiation (GO:0030154)	0.0008913
cell projection organization (GO:0030030)	0.0010180
neuron differentiation (GO:0030182)	0.0011160

biological_process (GO:0008150)	0.0011300
cellular glucuronidation (GO:0052695)	0.0011540
uronic acid metabolic process (GO:0006063)	0.0013670
glucuronate metabolic process (GO:0019585)	0.0013670
anatomical structure morphogenesis (GO:0009653)	0.0014550
monocarboxylic acid metabolic process (GO:0032787)	0.0017230
cellular developmental process (GO:0048869)	0.0017610
cell morphogenesis involved in differentiation (GO:0000904)	0.0019510
system development (GO:0048731)	0.0020730
sequestering of TGFbeta in extracellular matrix (GO:0035583)	0.0027780
dendrite development (GO:0016358)	0.0034530
nervous system development (GO:0007399)	0.0036030
cell morphogenesis (GO:0000902)	0.0038070
negative regulation of cellular glucuronidation (GO:2001030)	0.0049310
cell surface receptor signaling pathway (GO:0007166)	0.0050510
multicellular organismal development (GO:0007275)	0.0054140
axon extension (GO:0048675)	0.0062010
platelet-derived growth factor receptor signaling pathway (GO:0048008)	0.0062010
cytoskeleton organization (GO:0007010)	0.0063150
cellular component morphogenesis (GO:0032989)	0.0074230
xenobiotic glucuronidation (GO:0052697)	0.0076930
flavonoid glucuronidation (GO:0052696)	0.0076930
maintenance of protein location in extracellular region (GO:0071694)	0.0076930
protein localization to extracellular region (GO:0071692)	0.0076930
regulation of cellular glucuronidation (GO:2001029)	0.0076930
dichotomous subdivision of terminal units involved in salivary gland branching (GO:0060666)	0.0076930

Table S21. A table showing molecular GO terms of identified CNV genes with a cutoff p-value of 0.01 in the YRI group.

Term	P-value
catalytic activity (GO:0003824)	0.0001439
molecular_function (GO:0003674)	0.0001958
monocarboxylic acid binding (GO:0033293)	0.0007160
enzyme binding (GO:0019899)	0.0010340
retinoic acid binding (GO:0001972)	0.0013440
protein binding (GO:0005515)	0.0024480
glucuronosyltransferase activity (GO:0015020)	0.0033610
binding (GO:0005488)	0.0034500
kinase activity (GO:0016301)	0.0045700
retinoid binding (GO:0005501)	0.0062310
carbohydrate derivative binding (GO:0097367)	0.0069590
carboxylic acid binding (GO:0031406)	0.0071510
isoprenoid binding (GO:0019840)	0.0072720
organic acid binding (GO:0043177)	0.0073200
kinase binding (GO:0019900)	0.0088710

Table S22. A table showing cellular GO terms of identified CNV genes with a cutoff p-value of 0.01 in the YRI group.

Term	P-value
membrane (GO:0016020)	0.0000108
plasma membrane (GO:0005886)	0.0001778
cytoplasm (GO:0005737)	0.0001790
cell periphery (GO:0071944)	0.0002748
cytoplasmic part (GO:0044444)	0.0005164
cellular_component (GO:0005575)	0.0011650
cytosol (GO:0005829)	0.0016410
cell part (GO:0044464)	0.0052180
cell (GO:0005623)	0.0052310
endoplasmic reticulum (GO:0005783)	0.0070360

endoplasmic reticulum membrane (GO:0005789)	0.0091240
sarcoplasmic reticulum lumen (GO:0033018)	0.0091600