

Whole Genome SNP Profiling using GSA + Psych Array data report

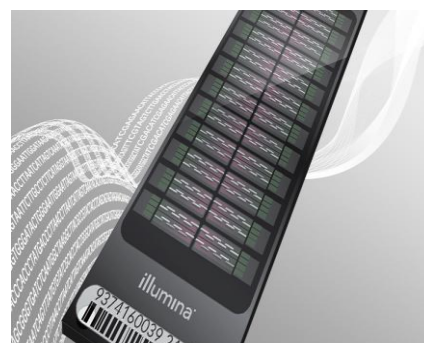


Project Details

Project Name :	Whole genome SNP profiling using GSA + Psych array chip
Institute Name:	NIMHANS
Application:	SNP Genotyping
Platform/Chip:	GSA+Psych array
Number of Samples :	408 samples

Project Introduction:

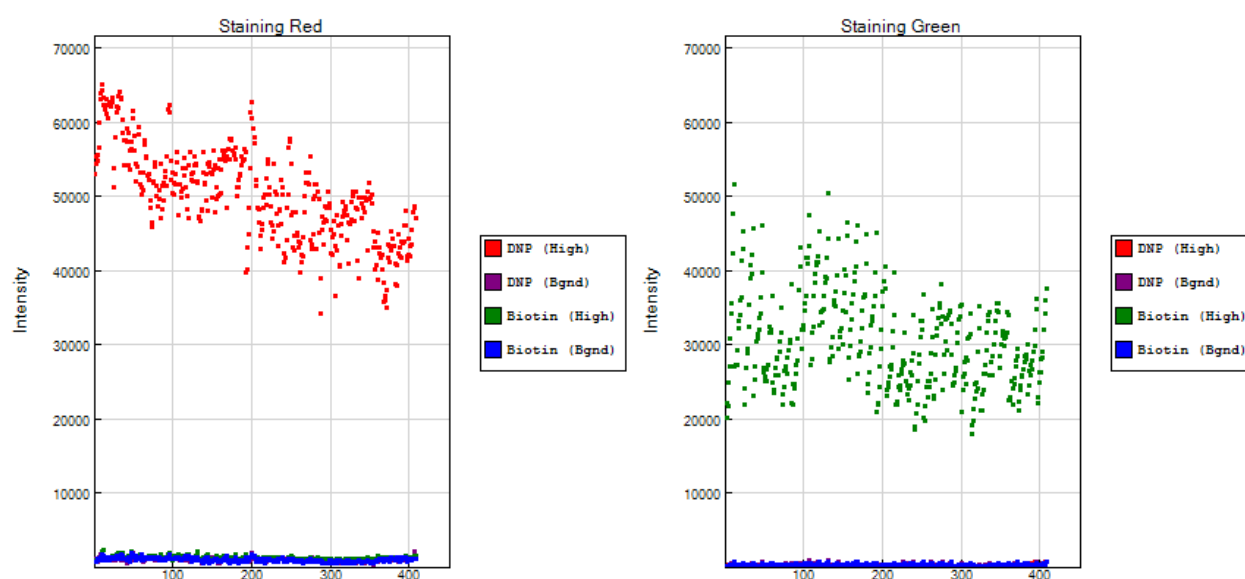
Technological progress in genotyping has opened the possibilities towards genomic screens and novel opportunities to explore polygenetic perspectives, now spanning a wide array of possible analyses falling under the term Genome-Wide SNP profiling. This project aims at Genome wide SNP profiling to identify genetic markers conferring risk and includes sample size of 408 samples. Microarray experiment is performed using Illumina SNP genotyping microarray chip: **Global Screening Array-24 + Psycharray BeadChip**. This chip combines multi-ethnic genome-wide content, curated clinical research variants, and quality control (QC) markers for precision medicine research. The clinical research content includes variants with established disease associations, relevant pharmacogenomics markers, and curated exonic content based on ClinVar, NHGRI, PharmGKB, and ExAC databases. The Infinium GSA-24 BeadChip comprise of highly optimized multiethnic genome-wide content, curated clinical research variants, and QC markers for a broad range of clinical research and variant screening applications. These applications include disease association and risk profiling studies, pharmacogenomics research, disease characterization, lifestyle and wellness characterization, and marker discovery in complex disease research.



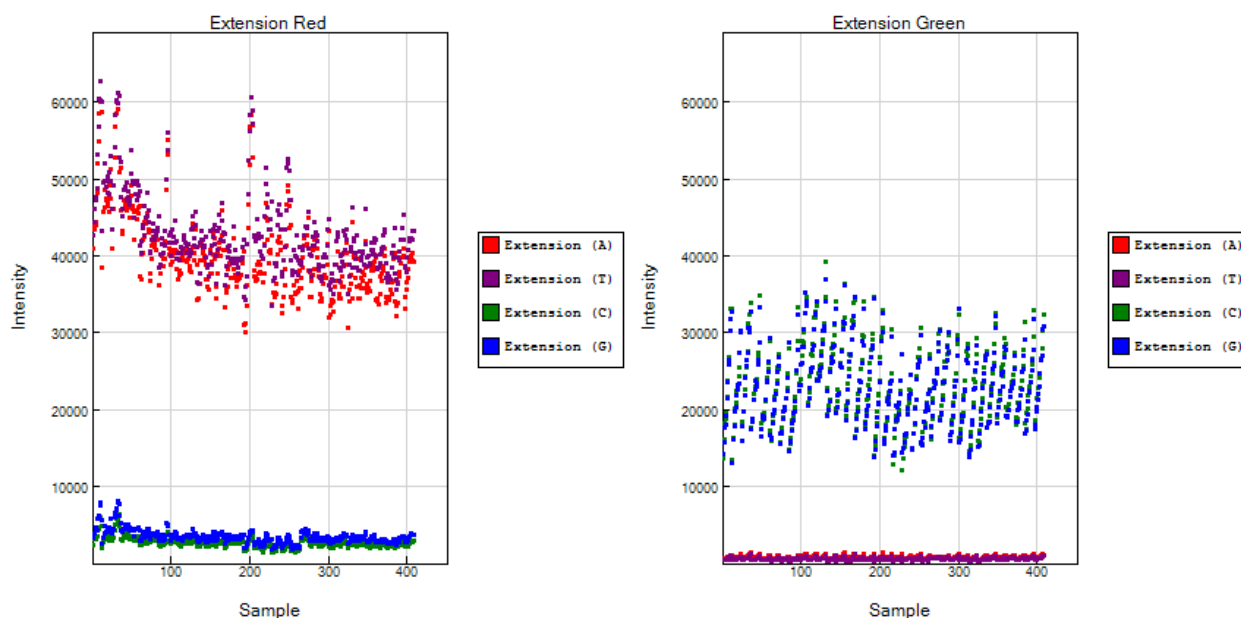
Control Summary

Experimental (Sample independent Controls)

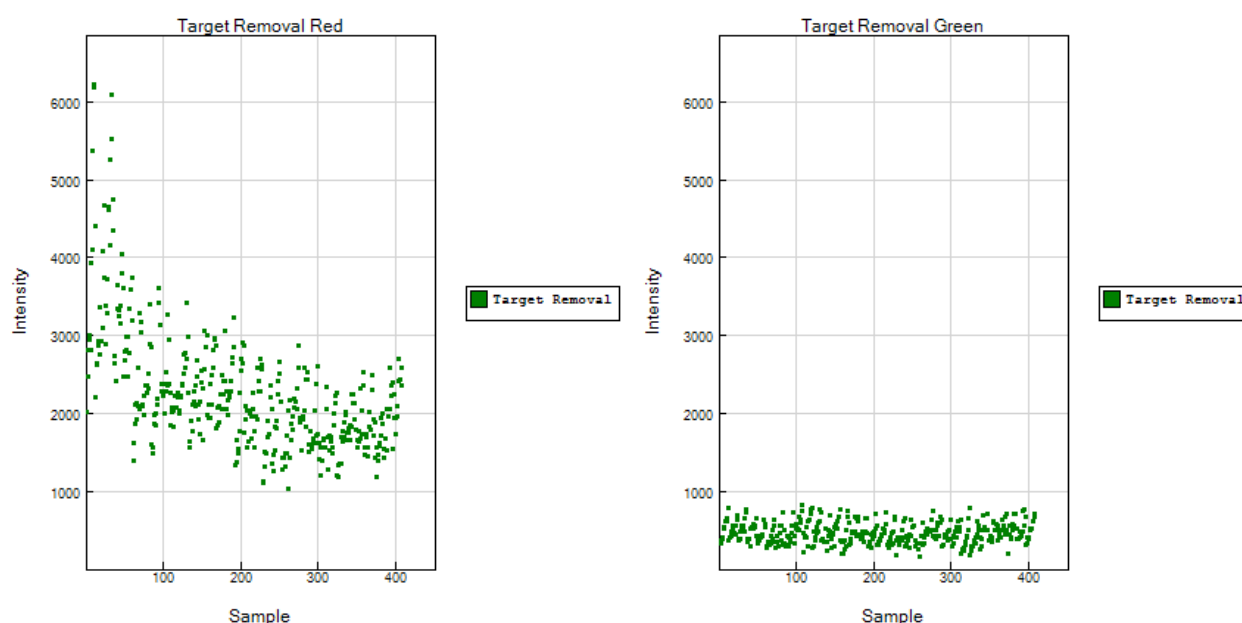
- 1. Staining Controls :** Staining controls are used to examine the efficiency of the staining step in both the red and green channels. Staining controls have various levels of dinitrophenyl (DNP) or biotin attached to the beads. These controls are independent of the hybridization and extension step. Various levels of DNP and biotin monitor the sensitivity and efficiency of the staining step. Both red and green channels can be evaluated using the Staining Controls (*High signal intensity for DNP in red channel and for Biotin in Green channel*).



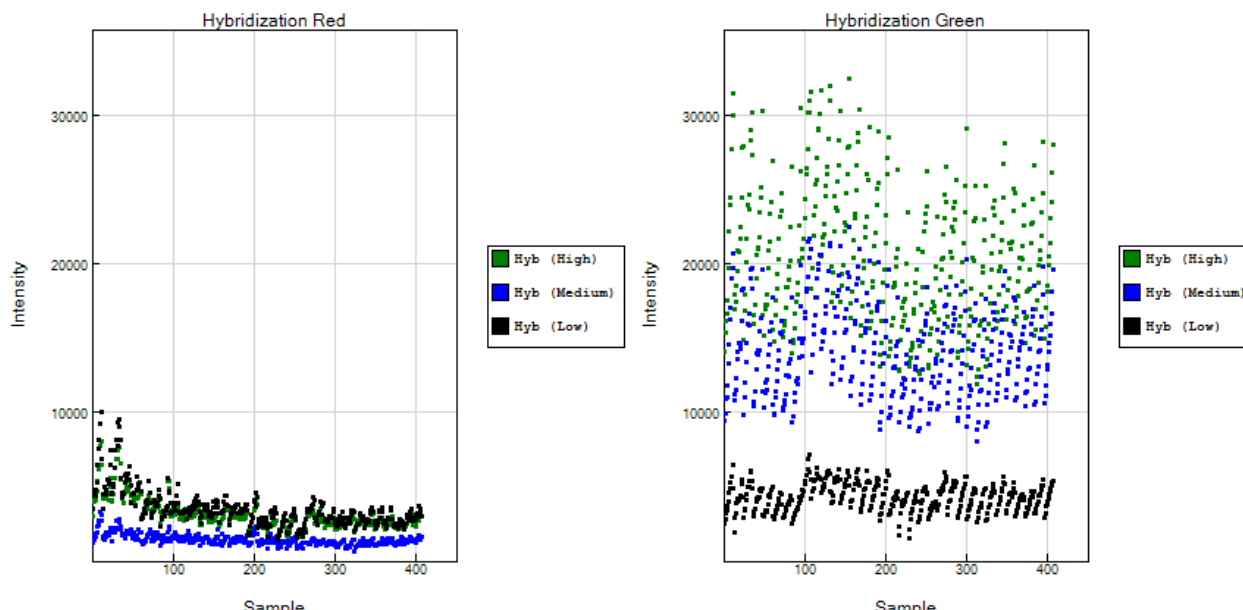
- 2. Extension Controls:** Extension controls test the extension efficiency of A, T, C, and G nucleotides from a hairpin probe, and are therefore sample-independent. Both red (A,T) and green (C,G) channels are monitored (*High signal intensity for A,T nucleotide in Red channel G,C nucleotide for Green channel*).



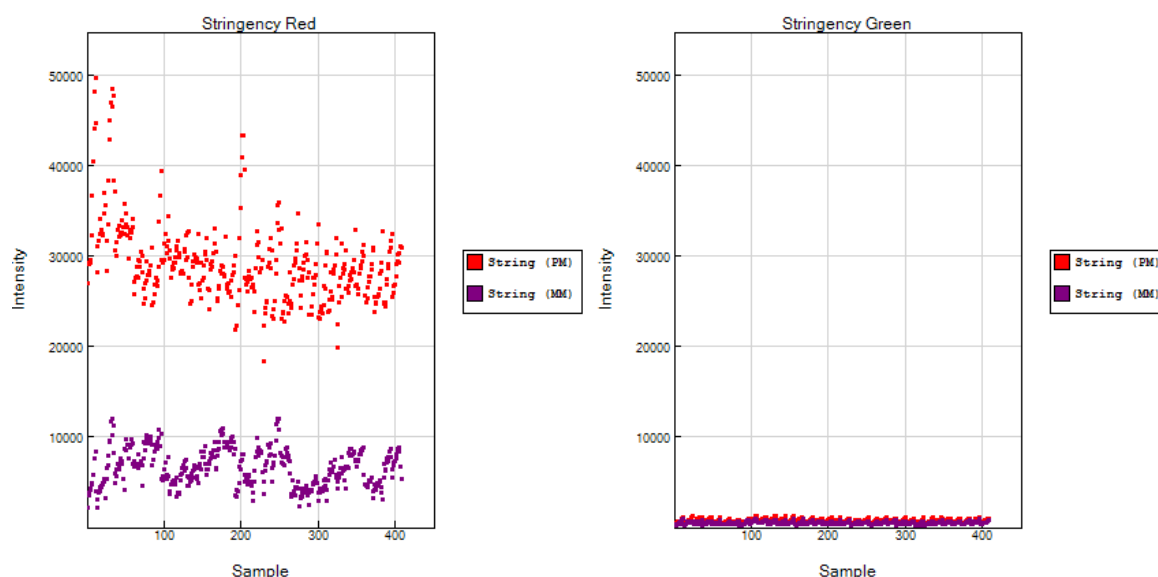
- 3. Target Removal Controls:** Target removal controls test the efficiency of the stripping step after the extension reaction. In contrast to allele-specific extension, the control oligos are extended using the probe sequence as template. This process generates labeled targets. The probe sequences are designed such that extension from the probe does not occur. All target removal controls should result in low signal compared to the hybridization controls, indicating that the targets were removed efficiently after extension. The target removal controls are present in the hybridization buffer RA1. Performance of target removal controls should only be monitored in the green channel (*lowering of signal intensity in Green channel plot*).



- 4. Hybridization Controls:** Hybridization controls test the overall performance of the Infinium Assay using synthetic targets. These synthetic targets complement the sequence on the array perfectly, allowing the probe to extend on the synthetic target as a template. Synthetic targets are present in the hybridization buffer at three levels, monitoring the response from high-concentration (5pM)-*High signal*, medium concentration (1pM)-*Moderate signal*, and low-concentration (0.2pM) targets-*Low signal*.

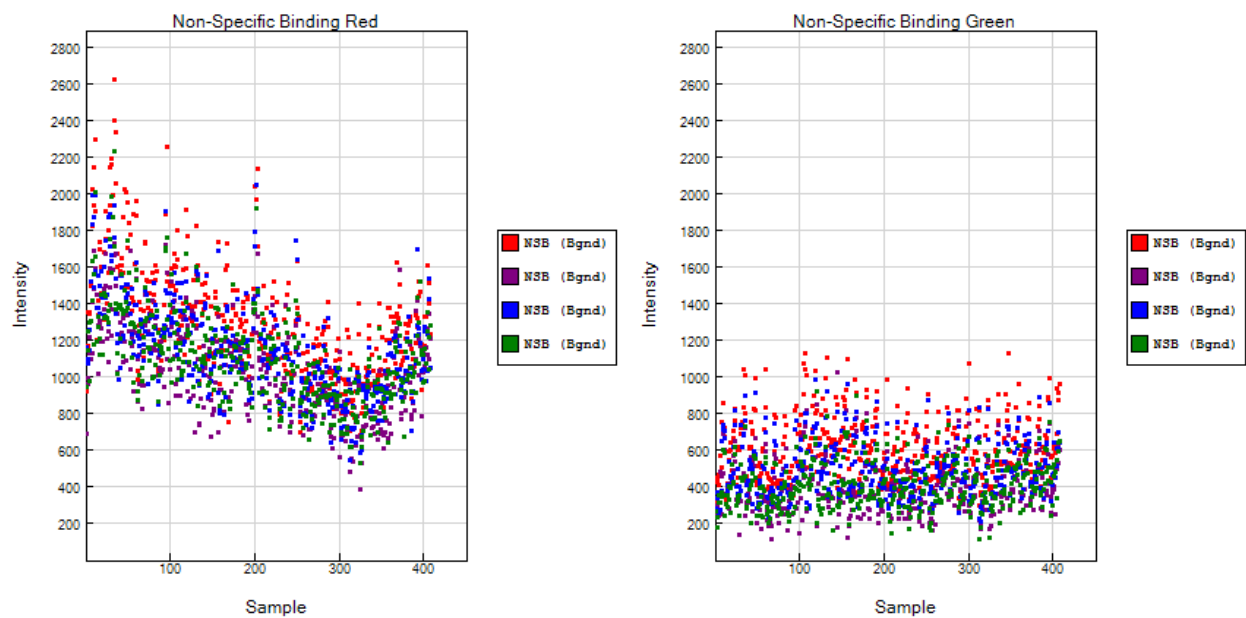


- 5. Stringency Controls:** These controls test the stringency of the hybridization process (to check sample contamination & quality). The same target is used for each stringency control; the only difference between the stringency controls is the number of nucleotides hybridized between the target and the probe sequence on the bead. The probes are designed such that the 3' end of the probe is available for extension. Mismatches are introduced into the body of the probe sequence to affect hybrid stability. The controls have 0 to 12 mismatched nucleotides between target and probe. Performance of stringency controls should only be monitored in the red channel (*High PM-perfect match signal and Low MM-Mismatch signal*).

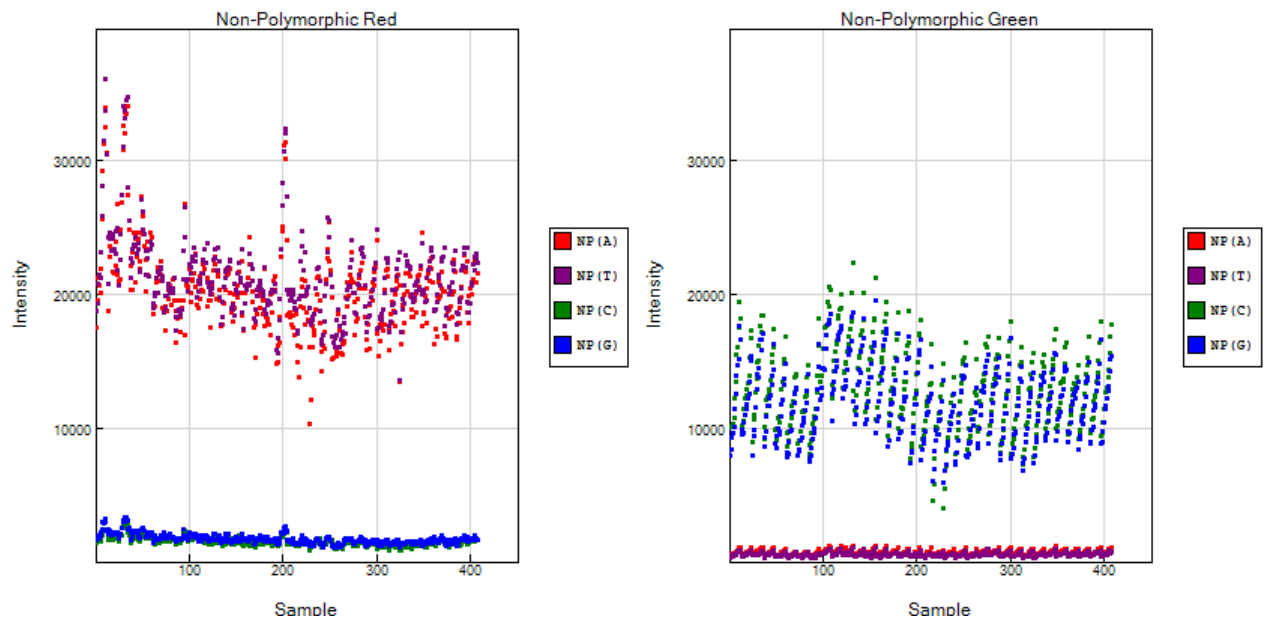


- 6. Non specific binding Controls :** Non-specific binding controls are included to monitor the specificity of the hybridization of the amplified DNA (Check sample contamination and quality). The probe sequences for non-specific binding controls are complementary to bacterial sequences and should

not hybridize to human sequences under standard hybridization stringency conditions. These controls should show low intensities, indicating there is minimal cross hybridization of human DNA to bacterial sequences. Performance of nonspecific binding controls should be monitored in both green and red channels (*low intensity signal in both channels*).



7. Non Polymorphic Controls : Non-polymorphic controls test the overall performance of the assay, from amplification to detection, by querying a particular base in a non-polymorphic region of the genome. It help to compare assay performance across different samples. One non-polymorphic control has been designed for each of the four nucleotides (A, T, C, and G) (*High signal intensity for NP (A,T) in Red channel and NP(C,G) in Green channel*).



Sample QC table:

S.No	Sample ID	Call Rate	Gender	Barcode	Position
1	V1	0.979884	Female	2.03464E+11	R01C01
2	V2	0.9870607	Male	2.03464E+11	R02C01
3	V3	0.9886327	Male	2.03464E+11	R03C01
4	V4	0.9890164	Male	2.03464E+11	R04C01
5	V5	0.9853075	Female	2.03464E+11	R05C01
6	V6	0.9886178	Male	2.03464E+11	R06C01
7	V7	0.9886935	Male	2.03464E+11	R07C01
8	V8	0.9849524	Female	2.03464E+11	R08C01
9	V9	0.9850555	Female	2.03464E+11	R09C01
10	V10	0.9846755	Female	2.03464E+11	R10C01
11	V11	0.9882354	Male	2.03464E+11	R11C01
12	V12	0.9840634	Male	2.03464E+11	R12C01
13	V13	0.9642628	Unknown	2.03464E+11	R01C02
14	V14	0.9821885	Female	2.03464E+11	R02C02
15	V15	0.9842608	Female	2.03464E+11	R03C02
16	V16	0.9848953	Female	2.03464E+11	R04C02
17	V17	0.9887295	Male	2.03464E+11	R05C02
18	V18	0.9851387	Female	2.03464E+11	R06C02
19	V19	0.9849263	Female	2.03464E+11	R07C02
20	V20	0.9890896	Male	2.03464E+11	R08C02
21	V21	0.9888636	Male	2.03464E+11	R09C02
22	V23	0.9887258	Male	2.03464E+11	R10C02
23	V24	0.9849499	Female	2.03464E+11	R11C02
24	V25	0.977505	Male	2.03464E+11	R12C02
25	V27	0.9889853	Male	2.03498E+11	R01C01
26	V29	0.988804	Male	2.03498E+11	R02C01
27	V30	0.989189	Male	2.03498E+11	R03C01
28	V32	0.9891579	Male	2.03498E+11	R04C01
29	V33	0.9889468	Male	2.03498E+11	R05C01
30	V36	0.9890064	Male	2.03498E+11	R06C01
31	V37	0.9850518	Female	2.03498E+11	R07C01
32	V38	0.988799	Male	2.03498E+11	R08C01
33	V39	0.9852281	Female	2.03498E+11	R09C01
34	V40	0.98866	Male	2.03498E+11	R10C01
35	V41	0.9887581	Male	2.03498E+11	R11C01
36	V42	0.9844632	Male	2.03498E+11	R12C01
37	V43	0.9847624	Female	2.03498E+11	R01C02
38	V44	0.9889891	Male	2.03498E+11	R02C02
39	V45	0.9894596	Male	2.03498E+11	R03C02
40	V46	0.9854776	Female	2.03498E+11	R04C02
41	V47	0.9896074	Male	2.03498E+11	R05C02

42	V48	0.9896359	Male	2.03498E+11	R06C02
43	V49	0.9896049	Male	2.03498E+11	R07C02
44	V50	0.9859458	Female	2.03498E+11	R08C02
45	V51	0.9897477	Male	2.03498E+11	R09C02
46	V53	0.9892101	Male	2.03498E+11	R10C02
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48	V55	0.9800032	Female	2.03498E+11	R12C02
49	V56	0.9877486	Male	2.03498E+11	R01C01
50	V57	0.9881646	Male	2.03498E+11	R02C01
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52	V59	0.9854094	Female	2.03498E+11	R04C01
53	V60	0.989107	Male	2.03498E+11	R05C01
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62	V69	0.9809965	Female	2.03498E+11	R02C02
63	V70	0.9858129	Female	2.03498E+11	R03C02
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65	V72	0.9858551	Female	2.03498E+11	R05C02
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85	V97	0.9844496	Female	2.03498E+11	R01C02

86	V99	0.9849499	Female	2.03498E+11	R02C02
87	V100	0.985058	Female	2.03498E+11	R03C02
88	V101	0.9851362	Female	2.03498E+11	R04C02
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96	V113	0.9809828	Female	2.03498E+11	R12C02
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99	V116	0.985202	Female	2.03498E+11	R03C01
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118	V156	0.9870036	Male	2.03498E+11	R10C02
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137	V178	0.9897986	Male	2.03498E+11	R05C02
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160	V205	0.9899054	Male	2.03498E+11	R04C02
161	V206	0.9896446	Male	2.03498E+11	R05C02
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173	V220	0.9860178	Female	2.03498E+11	R05C01

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176	V223	0.9862326	Female	2.03498E+11	R08C01
177	V224	0.9898992	Male	2.03498E+11	R09C01
178	V225	0.9863009	Female	2.03498E+11	R10C01
179	V226	0.9895751	Male	2.03498E+11	R11C01
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185	V232	0.9898247	Male	2.03498E+11	R05C02
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191	V238	0.988922	Male	2.03498E+11	R11C02
192	V239	0.9887432	Male	2.03498E+11	R12C02
193	V240	0.9838362	Female	2.03498E+11	R01C01
194	V241	0.9884154	Male	2.03498E+11	R02C01
195	V242	0.9851163	Female	2.03498E+11	R03C01
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198	V245	0.989359	Male	2.03498E+11	R06C01
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201	V249	0.9895589	Male	2.03498E+11	R09C01
202	V250	0.9857024	Female	2.03498E+11	R10C01
203	V251	0.9855385	Female	2.03498E+11	R11C01
204	V252	0.9879969	Male	2.03498E+11	R12C01
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206	V254	0.985392	Female	2.03498E+11	R02C02
207	V255	0.9854292	Female	2.03498E+11	R03C02
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214	V262	0.9892448	Male	2.03498E+11	R10C02
215	V263	0.9857384	Male	2.03498E+11	R11C02
216	V264	0.9789577	Male	2.03498E+11	R12C02
217	V265	0.9221137	Unknown	2.03498E+11	R01C01

218	V266	0.9656969	Unknown	2.03498E+11	R02C01
219	V267	0.9846482	Male	2.03498E+11	R03C01
220	V269	0.9886885	Male	2.03498E+11	R04C01
221	V270	0.985716	Female	2.03498E+11	R05C01
222	V271	0.989251	Male	2.03498E+11	R06C01
223	V272	0.9895031	Male	2.03498E+11	R07C01
224	V273	0.9856999	Female	2.03498E+11	R08C01
225	V275	0.9895192	Male	2.03498E+11	R09C01
226	V278	0.9892684	Male	2.03498E+11	R10C01
227	V279	0.9890263	Male	2.03498E+11	R11C01
228	V282	0.9855335	Male	2.03498E+11	R12C01
229	V285	0.8814484	Unknown	2.03498E+11	R01C02
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232	V289	0.9881795	Male	2.03498E+11	R04C02
233	V290	0.9891753	Male	2.03498E+11	R05C02
234	V291	0.985721	Female	2.03498E+11	R06C02
235	V292	0.9858675	Female	2.03498E+11	R07C02
236	V295	0.9895416	Male	2.03498E+11	R08C02
237	V297	0.9856899	Female	2.03498E+11	R09C02
238	V298	0.9893553	Male	2.03498E+11	R10C02
239	V299	0.9857782	Female	2.03498E+11	R11C02
240	V300	0.9821922	Male	2.03498E+11	R12C02
241	V301	0.9841205	Female	2.03498E+11	R01C01
242	V302	0.9885396	Male	2.03498E+11	R02C01
243	V308	0.9882987	Male	2.03498E+11	R03C01
244	V309	0.9892548	Male	2.03498E+11	R04C01
245	V311	0.9892597	Male	2.03498E+11	R05C01
246	V312	0.9891579	Male	2.03498E+11	R06C01
247	V313	0.9892125	Male	2.03498E+11	R07C01
248	V314	0.9893839	Male	2.03498E+11	R08C01
249	V317	0.9893876	Male	2.03498E+11	R09C01
250	V322	0.9890896	Male	2.03498E+11	R10C01
251	V323	0.9846681	Male	2.03498E+11	R11C01
252	V324	0.9820929	Male	2.03498E+11	R12C01
253	V327	0.9885855	Male	2.03498E+11	R01C02
254	V328	0.9852517	Female	2.03498E+11	R02C02
255	V329	0.9850903	Female	2.03498E+11	R03C02
256	V330	0.9858042	Female	2.03498E+11	R04C02
257	V332	0.9852939	Female	2.03498E+11	R05C02
258	V333	0.9854603	Female	2.03498E+11	R06C02
259	V334	0.9894422	Male	2.03498E+11	R07C02
260	V335	0.9816161	Female	2.03498E+11	R08C02
261	V336	0.9891169	Male	2.03498E+11	R09C02

262	V337	0.9883595	Male	2.03498E+11	R10C02
263	V338	0.9822071	Female	2.03498E+11	R11C02
264	V339	0.9758151	Female	2.03498E+11	R12C02
265	V341	0.9857322	Female	2.03498E+11	R01C01
266	V342	0.9855993	Female	2.03498E+11	R02C01
267	V345	0.9825448	Female	2.03498E+11	R03C01
268	V347	0.9857595	Female	2.03498E+11	R04C01
269	V351	0.9898023	Male	2.03498E+11	R05C01
270	V354	0.9856453	Female	2.03498E+11	R06C01
271	V355	0.9897949	Male	2.03498E+11	R07C01
272	V356	0.985649	Female	2.03498E+11	R08C01
273	V357	0.985536	Female	2.03498E+11	R09C01
274	V358	0.9855149	Female	2.03498E+11	R10C01
275	V359	0.9856341	Female	2.03498E+11	R11C01
276	V360	0.9816149	Female	2.03498E+11	R12C01
277	V363	0.9895093	Male	2.03498E+11	R01C02
278	V364	0.9896061	Male	2.03498E+11	R02C02
279	V365	0.9898607	Male	2.03498E+11	R03C02
280	V366	0.9859085	Female	2.03498E+11	R04C02
281	V367	0.9900122	Male	2.03498E+11	R05C02
282	V368	0.9897712	Male	2.03498E+11	R06C02
283	V369	0.9897054	Male	2.03498E+11	R07C02
284	V370	0.9898955	Male	2.03498E+11	R08C02
285	V371	0.9857707	Female	2.03498E+11	R09C02
286	V372	0.9856527	Female	2.03498E+11	R10C02
287	V373	0.984868	Female	2.03498E+11	R11C02
288	V374	0.9824356	Female	2.03498E+11	R12C02
289	V375	0.9900184	Male	2.03498E+11	R01C01
290	V378	0.9901922	Male	2.03498E+11	R02C01
291	V379	0.9863455	Female	2.03498E+11	R03C01
292	V391	0.9862413	Female	2.03498E+11	R04C01
293	V392	0.9863046	Female	2.03498E+11	R05C01
294	V394	0.9903002	Male	2.03498E+11	R06C01
295	V395	0.989292	Male	2.03498E+11	R07C01
296	V397	0.9862462	Female	2.03498E+11	R08C01
297	V399	0.9903784	Male	2.03498E+11	R09C01
298	V400	0.990222	Male	2.03498E+11	R10C01
299	V401	0.9895242	Male	2.03498E+11	R11C01
300	V402	0.9859644	Male	2.03498E+11	R12C01
301	V403	0.9902158	Male	2.03498E+11	R01C02
302	V404	0.9901165	Male	2.03498E+11	R02C02
303	V405	0.9903772	Male	2.03498E+11	R03C02
304	V406	0.9861432	Female	2.03498E+11	R04C02
305	V407	0.9903312	Male	2.03498E+11	R05C02

306	V408	0.9901785	Male	2.03498E+11	R06C02
307	V409	0.9902294	Male	2.03498E+11	R07C02
308	V410	0.9904778	Male	2.03498E+11	R08C02
309	V411	0.9903424	Male	2.03498E+11	R09C02
310	V412	0.9906318	Male	2.03498E+11	R10C02
311	V413	0.9889928	Male	2.03498E+11	R11C02
312	V414	0.9862102	Male	2.03498E+11	R12C02
313	V415	0.9899302	Male	2.03498E+11	R01C01
314	V416	0.9901376	Male	2.03498E+11	R02C01
315	V417	0.9903722	Male	2.03498E+11	R03C01
316	V418	0.9903896	Male	2.03498E+11	R04C01
317	V419	0.9902754	Male	2.03498E+11	R05C01
318	V420	0.9861655	Female	2.03498E+11	R06C01
319	V421	0.9863269	Female	2.03498E+11	R07C01
320	V422	0.9863555	Female	2.03498E+11	R08C01
321	V423	0.9903822	Male	2.03498E+11	R09C01
322	V424	0.9901934	Male	2.03498E+11	R10C01
323	V425	0.9855509	Female	2.03498E+11	R11C01
324	V426	0.9879783	Male	2.03498E+11	R12C01
325	V427	0.9861022	Female	2.03498E+11	R01C02
326	V428	0.9862164	Female	2.03498E+11	R02C02
327	V429	0.9862065	Female	2.03498E+11	R03C02
328	V430	0.9862549	Female	2.03498E+11	R04C02
329	V431	0.9862673	Female	2.03498E+11	R05C02
330	V432	0.9863369	Female	2.03498E+11	R06C02
331	V433	0.9864387	Female	2.03498E+11	R07C02
332	V434	0.9903002	Male	2.03498E+11	R08C02
333	V435	0.9904566	Male	2.03498E+11	R09C02
334	V436	0.9903995	Male	2.03498E+11	R10C02
335	V437	0.9855646	Female	2.03498E+11	R11C02
336	V438	0.9880007	Male	2.03498E+11	R12C02
337	V439	0.9899389	Male	2.03498E+11	R01C01
338	V440	0.9901301	Male	2.03498E+11	R02C01
339	V441	0.9863741	Female	2.03498E+11	R03C01
340	V442	0.9902816	Male	2.03498E+11	R04C01
341	V443	0.9903312	Male	2.03498E+11	R05C01
342	V444	0.9903337	Male	2.03498E+11	R06C01
343	V445	0.990217	Male	2.03498E+11	R07C01
344	V446	0.9902741	Male	2.03498E+11	R08C01
345	V447	0.9902419	Male	2.03498E+11	R09C01
346	V448	0.9864337	Female	2.03498E+11	R10C01
347	V449	0.9903399	Male	2.03498E+11	R11C01
348	V450	0.9874122	Male	2.03498E+11	R12C01
349	V451	0.9902158	Male	2.03498E+11	R01C02

350	V452	0.9904132	Male	2.03498E+11	R02C02
351	V453	0.9863158	Female	2.03498E+11	R03C02
352	V454	0.9903983	Male	2.03498E+11	R04C02
353	V455	0.9861928	Female	2.03498E+11	R05C02
354	V456	0.9904306	Male	2.03498E+11	R06C02
355	V457	0.98643	Female	2.03498E+11	R07C02
356	V458	0.9904082	Male	2.03498E+11	R08C02
357	V459	0.9903735	Male	2.03498E+11	R09C02
358	V460	0.9904964	Male	2.03498E+11	R10C02
359	V462	0.9903312	Male	2.03498E+11	R11C02
360	V463	0.9877114	Male	2.03498E+11	R12C02
361	V464	0.989184	Male	2.03509E+11	R01C01
362	V466	0.9893926	Male	2.03509E+11	R02C01
363	V467	0.9894261	Male	2.03509E+11	R03C01
364	V468	0.985798	Female	2.03509E+11	R04C01
365	V469	0.9894025	Male	2.03509E+11	R05C01
366	V470	0.9857918	Female	2.03509E+11	R06C01
367	V471	0.9895627	Male	2.03509E+11	R07C01
368	V472	0.9894969	Male	2.03509E+11	R08C01
369	V474	0.9894137	Male	2.03509E+11	R09C01
370	V475	0.9894695	Male	2.03509E+11	R10C01
371	V476	0.9831148	Female	2.03509E+11	R11C01
372	V477	0.9822208	Female	2.03509E+11	R12C01
373	V478	0.9855546	Female	2.03509E+11	R01C02
374	V479	0.9858477	Female	2.03509E+11	R02C02
375	V480	0.9898085	Male	2.03509E+11	R03C02
376	V481	0.9860774	Female	2.03509E+11	R04C02
377	V482	0.9860153	Female	2.03509E+11	R05C02
378	V485	0.9897912	Male	2.03509E+11	R06C02
379	V486	0.9858266	Female	2.03509E+11	R07C02
380	V487	0.9895825	Male	2.03509E+11	R08C02
381	V488	0.9860004	Female	2.03509E+11	R09C02
382	V489	0.9891182	Male	2.03509E+11	R10C02
383	V490	0.9821761	Female	2.03509E+11	R11C02
384	V491	0.9824455	Female	2.03509E+11	R12C02
385	V492	0.9898421	Male	2.03604E+11	R01C01
386	V493	0.9895738	Male	2.03604E+11	R02C01
387	V494	0.9864238	Female	2.03604E+11	R03C01
388	V495	0.9862549	Female	2.03604E+11	R04C01
389	V496	0.9901388	Male	2.03604E+11	R05C01
390	V502	0.9900097	Male	2.03604E+11	R06C01
391	V503	0.9899997	Male	2.03604E+11	R07C01
392	V504	0.9901189	Male	2.03604E+11	R08C01
393	V505	0.9898048	Male	2.03604E+11	R09C01

394	V506	0.9899004	Male	2.03604E+11	R10C01
395	V507	0.9859731	Female	2.03604E+11	R11C01
396	V508	0.9824815	Female	2.03604E+11	R12C01
397	V509	0.9898222	Male	2.03604E+11	R01C02
398	V510	0.9897501	Male	2.03604E+11	R02C02
399	V511	0.9891045	Male	2.03604E+11	R03C02
400	V512	0.9899141	Male	2.03604E+11	R04C02
401	V513	0.99015	Male	2.03604E+11	R05C02
402	V514	0.9865156	Female	2.03604E+11	R06C02
403	V515	0.9902021	Male	2.03604E+11	R07C02
404	V516	0.9863741	Female	2.03604E+11	R08C02
405	V517	0.9901189	Male	2.03604E+11	R09C02
406	V518	0.9902083	Male	2.03604E+11	R10C02
407	V519	0.9888003	Male	2.03604E+11	R11C02
408	V520	0.9874593	Male	2.03604E+11	R12C02

Data Filtering and Preprocessing :

1. Raw Data : Plink Input files (SNP=805379, Samples = 408, 237 males, 166 females, 5 ambiguous)

- .ped files : SNP genotype data for all samples
- .map file : SNP location information (Chromosome, Genomic Position)

2. Quality Control and Data Pre processing: Plink

2.0. SNP Filtering based on X, Y, XY & MT: 42878

2.1 Sample Genotyping rate (per sample): MIND 0.1 : Samples with missing genotypes more 10 % will be removed.

Results: One sample (V285) removed due to low genotyping rate (< 90% SNP calls).

2.2 SNP Genotyping rate (Per SNP): GENO 0.1: SNPs with missing genotypes > 10% across all samples will be removed.

Results : 8201 SNPs removed due to low genotyping rate.

2.3 Minor Allele frequency (MAF 0.05): Remove SNPs with minor allele frequency < 0.05 due to genotyping error or bias or Monomorphic markers.

Results : 421600 SNPs removed with MAF < 0.05.

2.4 Hardy Weinberg Equilibrium error (HWE 0.001) : Remove SNP showing deviation from HWE with p-value 0.001.

Results: 2231 SNP removed with HWE error.

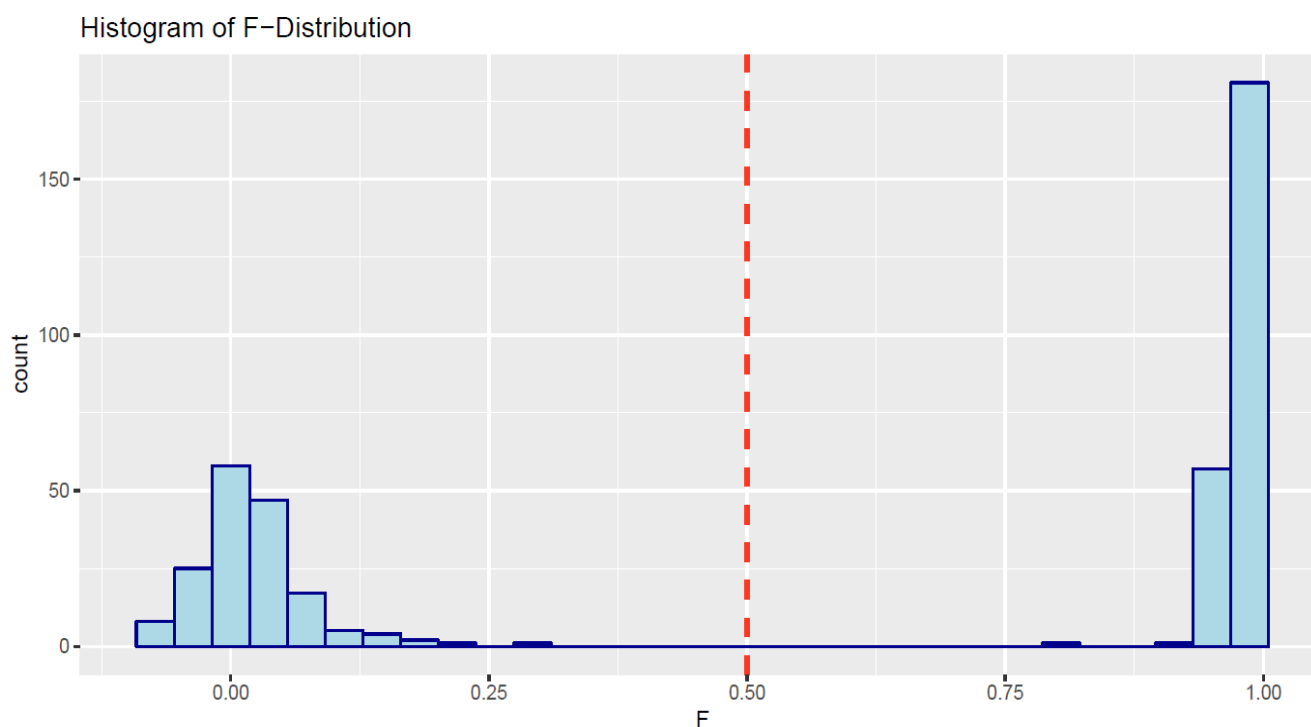
2.5 LD pruning: Remove SNPs in high LD required only for IBS analysis. LD pruning not required before Association Analysis thus performed on complete dataset.

Results: 119286

Gender Prediction & Imputation : Plink Vs Genome Studio

Plink compares sex assignments in the input dataset with those imputed from X chromosome inbreeding coefficients. By default F estimates smaller than 0.2 yield female calls, and values larger than 0.8 yield male calls. Based on distribution of F estimates (gap between a tight male clump at the right side of the distribution and females at left side) cut off point estimation.

**Please note: Previous Gender information share in Sample Table is based on Genome Studio.*



Gender Check & Imputation (Plink Vs Genome Studio)

FID	IID	Genome studio prediction	Plink Prediction (1=Male, 2=Female)	STATUS	F
13	V13	0	1	PROBLEM	0.9318
217	V265	0	2	PROBLEM	0.02528
218	V266	0	1	PROBLEM	0.948
229	V285	0	1	PROBLEM	0.8103
230	V286	0	2	PROBLEM	-0.08407
1	V1	2	2	OK	0.02372
2	V2	1	1	OK	0.9676
3	V3	1	1	OK	0.9712
4	V4	1	1	OK	0.9684
5	V5	2	2	OK	0.03578
6	V6	1	1	OK	0.9716
7	V7	1	1	OK	0.9735
8	V8	2	2	OK	0.05696
9	V9	2	2	OK	0.03771
10	V10	2	2	OK	0.02723
11	V11	1	1	OK	0.9704
12	V12	1	1	OK	0.961
14	V14	2	2	OK	0.2939
15	V15	2	2	OK	0.03374
16	V16	2	2	OK	0.1583
17	V17	1	1	OK	0.9692
18	V18	2	2	OK	0.03761
19	V19	2	2	OK	0.05682
20	V20	1	1	OK	0.9728
21	V21	1	1	OK	0.9751
22	V23	1	1	OK	0.9708
23	V24	2	2	OK	0.01846
24	V25	1	1	OK	0.9576
25	V27	1	1	OK	0.97
26	V29	1	1	OK	0.9644
27	V30	1	1	OK	0.9716
28	V32	1	1	OK	0.9673
29	V33	1	1	OK	0.9724
30	V36	1	1	OK	0.9712
31	V37	2	2	OK	0.06842
32	V38	1	1	OK	0.972
33	V39	2	2	OK	0.03766
34	V40	1	1	OK	0.974
35	V41	1	1	OK	0.9708

36	V42	1	1	OK	0.9684
37	V43	2	2	OK	0.07594
38	V44	1	1	OK	0.97
39	V45	1	1	OK	0.9684
40	V46	2	2	OK	0.06038
41	V47	1	1	OK	0.9728
42	V48	1	1	OK	0.9677
43	V49	1	1	OK	0.972
44	V50	2	2	OK	0.000434
45	V51	1	1	OK	0.9732
46	V53	1	1	OK	0.9713
47	V54	1	1	OK	0.9687
48	V55	2	2	OK	0.08563
49	V56	1	1	OK	0.9727
50	V57	1	1	OK	0.9692
51	V58	1	1	OK	0.9696
52	V59	2	2	OK	0.05543
53	V60	1	1	OK	0.9704
54	V61	2	2	OK	0.05105
55	V62	2	2	OK	0.01931
56	V63	2	2	OK	0.1812
57	V64	2	2	OK	0.02736
58	V65	2	2	OK	0.01884
59	V66	1	1	OK	0.9708
60	V67	1	1	OK	0.9652
61	V68	2	2	OK	0.03777
62	V69	2	2	OK	-0.02831
63	V70	2	2	OK	-0.00523
64	V71	2	2	OK	-0.02639
65	V72	2	2	OK	0.01475
66	V73	2	2	OK	-0.01562
67	V74	1	1	OK	0.9704
68	V75	2	2	OK	-0.01154
69	V76	2	2	OK	0.02949
70	V77	2	2	OK	-0.00928
71	V78	2	2	OK	0.05023
72	V79	2	2	OK	0.04041
73	V80	2	2	OK	0.001889
74	V81	1	1	OK	0.9687
75	V82	2	2	OK	-0.02034
76	V84	2	2	OK	-0.03562
77	V85	2	2	OK	0.001706
78	V87	2	2	OK	-0.05333
79	V88	2	2	OK	0.0852

80	V89	2	2	OK	0.04317
81	V90	2	2	OK	0.1234
82	V91	1	1	OK	0.9708
83	V92	2	2	OK	0.1446
84	V93	2	2	OK	-0.02261
85	V97	2	2	OK	0.05482
86	V99	2	2	OK	0.05416
87	V100	2	2	OK	0.06709
88	V101	2	2	OK	-0.00518
89	V102	2	2	OK	-0.01338
90	V103	1	1	OK	0.9672
91	V104	2	2	OK	0.006309
92	V106	2	2	OK	-0.03207
93	V109	2	2	OK	0.03836
94	V111	2	2	OK	0.06458
95	V112	2	2	OK	0.119
96	V113	2	2	OK	0.12
97	V114	2	2	OK	0.0701
98	V115	2	2	OK	0.02115
99	V116	2	2	OK	0.00455
100	V117	2	2	OK	0.08183
101	V123	2	2	OK	0.01741
102	V124	1	1	OK	0.9675
103	V125	1	1	OK	0.9701
104	V126	1	1	OK	0.9689
105	V129	2	2	OK	-0.01513
106	V130	1	1	OK	0.9675
107	V138	2	2	OK	-0.00484
108	V139	2	2	OK	0.05858
109	V140	2	2	OK	0.1228
110	V141	2	2	OK	0.05095
111	V142	1	1	OK	0.9712
112	V143	1	1	OK	0.9708
113	V144	2	2	OK	0.1509
114	V145	2	2	OK	0.03669
115	V149	1	1	OK	0.9695
116	V150	1	1	OK	0.9689
117	V155	2	2	OK	-0.02122
118	V156	1	1	OK	0.9658
119	V157	1	1	OK	0.9696
120	V159	1	1	OK	0.9658
121	V161	2	2	OK	-0.01988
122	V162	1	1	OK	0.972
123	V164	2	2	OK	-0.01388

124	V165	2	2	OK	-0.05904
125	V166	2	2	OK	-0.02089
126	V167	1	1	OK	0.9716
127	V168	1	1	OK	0.9716
128	V169	1	1	OK	0.9685
129	V170	1	1	OK	0.9704
130	V171	2	2	OK	0.00335
131	V172	1	1	OK	0.9645
132	V173	2	2	OK	0.01049
133	V174	1	1	OK	0.9704
134	V175	2	2	OK	-0.03232
135	V176	1	1	OK	0.9717
136	V177	1	1	OK	0.9697
137	V178	1	1	OK	0.9665
138	V180	1	1	OK	0.9685
139	V181	2	2	OK	0.03502
140	V182	1	1	OK	0.9712
141	V183	1	1	OK	0.9713
142	V184	1	1	OK	0.9744
143	V185	1	1	OK	0.9708
144	V188	1	1	OK	0.9757
145	V189	1	1	OK	0.9724
146	V190	2	2	OK	-0.03415
147	V191	1	1	OK	0.9728
148	V192	1	1	OK	0.9685
149	V193	1	1	OK	0.9681
150	V194	2	2	OK	0.004839
151	V195	1	1	OK	0.9728
152	V196	1	1	OK	0.9673
153	V197	1	1	OK	0.9689
154	V199	2	2	OK	-0.07144
155	V200	1	1	OK	0.9693
156	V201	1	1	OK	0.9699
157	V202	1	1	OK	0.9748
158	V203	1	1	OK	0.9708
159	V204	1	1	OK	0.9712
160	V205	1	1	OK	0.9701
161	V206	1	1	OK	0.9716
162	V207	2	2	OK	-0.07639
163	V208	1	1	OK	0.9685
164	V209	1	1	OK	0.9716
165	V210	1	1	OK	0.9685
166	V211	1	1	OK	0.972
167	V212	1	1	OK	0.9671

168	V213	1	1	OK	0.9683
169	V214	1	1	OK	0.9692
170	V215	1	1	OK	0.9736
171	V216	2	2	OK	-0.0592
172	V218	1	1	OK	0.9728
173	V220	2	2	OK	-0.02526
174	V221	1	1	OK	0.9736
175	V222	1	1	OK	0.9725
176	V223	2	2	OK	0.01573
177	V224	1	1	OK	0.9725
178	V225	2	2	OK	-0.02377
179	V226	1	1	OK	0.9716
180	V227	1	1	OK	0.9706
181	V228	1	1	OK	0.9748
182	V229	1	1	OK	0.972
183	V230	1	1	OK	0.9728
184	V231	1	1	OK	0.9736
185	V232	1	1	OK	0.9697
186	V233	1	1	OK	0.9712
187	V234	1	1	OK	0.9704
188	V235	1	1	OK	0.9701
189	V236	2	2	OK	-0.07711
190	V237	1	1	OK	0.972
191	V238	1	1	OK	0.9688
192	V239	1	1	OK	0.9687
193	V240	2	2	OK	-0.01607
194	V241	1	1	OK	0.9656
195	V242	2	2	OK	-0.00039
196	V243	2	2	OK	-0.00125
197	V244	2	2	OK	-0.00652
198	V245	1	1	OK	0.9716
199	V247	1	1	OK	0.9685
200	V248	1	1	OK	0.974
201	V249	1	1	OK	0.9685
202	V250	2	2	OK	-0.02648
203	V251	2	2	OK	0.03051
204	V252	1	1	OK	0.9687
205	V253	1	1	OK	0.9676
206	V254	2	2	OK	0.1409
207	V255	2	2	OK	0.03893
208	V256	1	1	OK	0.9677
209	V257	2	2	OK	0.03322
210	V258	1	1	OK	0.9685
211	V259	1	1	OK	0.9693

212	V260	2	2	OK	2.39E-05
213	V261	2	2	OK	-0.03745
214	V262	1	1	OK	0.9716
215	V263	1	1	OK	0.9653
216	V264	1	1	OK	0.9645
219	V267	1	1	OK	0.9659
220	V269	1	1	OK	0.9708
221	V270	2	2	OK	-0.03893
222	V271	1	1	OK	0.9673
223	V272	1	1	OK	0.9704
224	V273	2	2	OK	0.008628
225	V275	1	1	OK	0.9705
226	V278	1	1	OK	0.9692
227	V279	1	1	OK	0.9728
228	V282	1	1	OK	0.9719
231	V288	1	1	OK	0.9617
232	V289	1	1	OK	0.9668
233	V290	1	1	OK	0.9701
234	V291	2	2	OK	-0.03615
235	V292	2	2	OK	0.00219
236	V295	1	1	OK	0.9704
237	V297	2	2	OK	-0.01254
238	V298	1	1	OK	0.9684
239	V299	2	2	OK	-0.03869
240	V300	1	1	OK	0.9677
241	V301	2	2	OK	-0.01004
242	V302	1	1	OK	0.9699
243	V308	1	1	OK	0.9714
244	V309	1	1	OK	0.9719
245	V311	1	1	OK	0.9704
246	V312	1	1	OK	0.9665
247	V313	1	1	OK	0.9696
248	V314	1	1	OK	0.972
249	V317	1	1	OK	0.9712
250	V322	1	1	OK	0.9724
251	V323	1	1	OK	0.9645
252	V324	1	1	OK	0.9624
253	V327	1	1	OK	0.9743
254	V328	2	2	OK	-0.00714
255	V329	2	2	OK	-0.06434
256	V330	2	2	OK	0.01156
257	V332	2	2	OK	-0.0324
258	V333	2	2	OK	0.007904
259	V334	1	1	OK	0.9728

260	V335	2	2	OK	0.01757
261	V336	1	1	OK	0.9696
262	V337	1	1	OK	0.9727
263	V338	2	2	OK	0.02528
264	V339	2	2	OK	0.08531
265	V341	2	2	OK	0.02425
266	V342	2	2	OK	-0.01239
267	V345	2	2	OK	0.02926
268	V347	2	2	OK	0.01576
269	V351	1	1	OK	0.9677
270	V354	2	2	OK	0.006285
271	V355	1	1	OK	0.9728
272	V356	2	2	OK	-0.00446
273	V357	2	2	OK	0.09138
274	V358	2	2	OK	-0.01432
275	V359	2	2	OK	0.03482
276	V360	2	2	OK	-0.00168
277	V363	1	1	OK	0.9701
278	V364	1	1	OK	0.9677
279	V365	1	1	OK	0.9693
280	V366	2	2	OK	-0.06032
281	V367	1	1	OK	0.9729
282	V368	1	1	OK	0.9669
283	V369	1	1	OK	0.9681
284	V370	1	1	OK	0.9713
285	V371	2	2	OK	0.006057
286	V372	2	2	OK	-0.01506
287	V373	2	2	OK	-0.03472
288	V374	2	2	OK	0.01378
289	V375	1	1	OK	0.9673
290	V378	1	1	OK	0.9721
291	V379	2	2	OK	0.02116
292	V391	2	2	OK	0.007487
293	V392	2	2	OK	-0.00472
294	V394	1	1	OK	0.9709
295	V395	1	1	OK	0.9682
296	V397	2	2	OK	-0.00837
297	V399	1	1	OK	0.9709
298	V400	1	1	OK	0.9709
299	V401	1	1	OK	0.9728
300	V402	1	1	OK	0.9741
301	V403	1	1	OK	0.9725
302	V404	1	1	OK	0.9744
303	V405	1	1	OK	0.9678

304	V406	2	2	OK	0.01508
305	V407	1	1	OK	0.9701
306	V408	1	1	OK	0.9693
307	V409	1	1	OK	0.9709
308	V410	1	1	OK	0.9709
309	V411	1	1	OK	0.9709
310	V412	1	1	OK	0.9701
311	V413	1	1	OK	0.968
312	V414	1	1	OK	0.9656
313	V415	1	1	OK	0.9721
314	V416	1	1	OK	0.9697
315	V417	1	1	OK	0.9681
316	V418	1	1	OK	0.9732
317	V419	1	1	OK	0.9701
318	V420	2	2	OK	0.01004
319	V421	2	2	OK	0.0235
320	V422	2	2	OK	-0.02676
321	V423	1	1	OK	0.967
322	V424	1	1	OK	0.9673
323	V425	2	2	OK	0.1771
324	V426	1	1	OK	0.9729
325	V427	2	2	OK	0.01606
326	V428	2	2	OK	0.0326
327	V429	2	2	OK	0.2183
328	V430	2	2	OK	0.0219
329	V431	2	2	OK	0.02911
330	V432	2	2	OK	-0.0033
331	V433	2	2	OK	0.01092
332	V434	1	1	OK	0.9701
333	V435	1	1	OK	0.9725
334	V436	1	1	OK	0.9729
335	V437	2	2	OK	0.02079
336	V438	1	1	OK	0.9746
337	V439	1	1	OK	0.9669
338	V440	1	1	OK	0.9665
339	V441	2	2	OK	0.000383
340	V442	1	1	OK	0.9697
341	V443	1	1	OK	0.9693
342	V444	1	1	OK	0.9709
343	V445	1	1	OK	0.9697
344	V446	1	1	OK	0.974
345	V447	1	1	OK	0.9662
346	V448	2	2	OK	-0.00771
347	V449	1	1	OK	0.9685

348	V450	1	1	OK	0.9698
349	V451	1	1	OK	0.9724
350	V452	1	1	OK	0.9721
351	V453	2	2	OK	0.04898
352	V454	1	1	OK	0.9705
353	V455	2	2	OK	0.07683
354	V456	1	1	OK	0.9689
355	V457	2	2	OK	-0.00859
356	V458	1	1	OK	0.9713
357	V459	1	1	OK	0.9717
358	V460	1	1	OK	0.9705
359	V462	1	1	OK	0.9681
360	V463	1	1	OK	0.9738
361	V464	1	1	OK	0.9693
362	V466	1	1	OK	0.9696
363	V467	1	1	OK	0.9724
364	V468	2	2	OK	0.0357
365	V469	1	1	OK	0.9677
366	V470	2	2	OK	0.04411
367	V471	1	1	OK	0.972
368	V472	1	1	OK	0.9732
369	V474	1	1	OK	0.9716
370	V475	1	1	OK	0.97
371	V476	2	2	OK	0.06866
372	V477	2	2	OK	0.01454
373	V478	2	2	OK	0.0428
374	V479	2	2	OK	0.000348
375	V480	1	1	OK	0.9665
376	V481	2	2	OK	-0.04053
377	V482	2	2	OK	0.04596
378	V485	1	1	OK	0.9732
379	V486	2	2	OK	-0.01284
380	V487	1	1	OK	0.9685
381	V488	2	2	OK	0.02667
382	V489	1	1	OK	0.9708
383	V490	2	2	OK	0.008474
384	V491	2	2	OK	0.02182
385	V492	1	1	OK	0.97
386	V493	1	1	OK	0.9693
387	V494	2	2	OK	-0.02561
388	V495	2	2	OK	0.01557
389	V496	1	1	OK	0.9689
390	V502	1	1	OK	0.9728
391	V503	1	1	OK	0.9701

392	V504	1	1	OK	0.9724
393	V505	1	1	OK	0.9712
394	V506	1	1	OK	0.9673
395	V507	2	2	OK	0.01922
396	V508	2	2	OK	0.04554
397	V509	1	1	OK	0.9653
398	V510	1	1	OK	0.9717
399	V511	1	1	OK	0.9728
400	V512	1	1	OK	0.9716
401	V513	1	1	OK	0.9677
402	V514	2	2	OK	-0.04071
403	V515	1	1	OK	0.9705
404	V516	2	2	OK	0.03865
405	V517	1	1	OK	0.972
406	V518	1	1	OK	0.9705
407	V519	1	1	OK	0.9708
408	V520	1	1	OK	0.9725

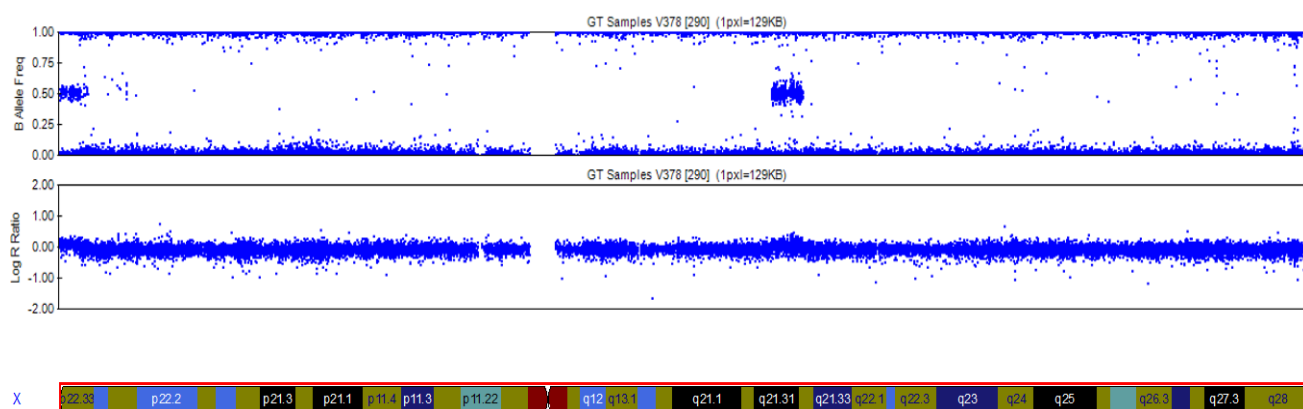
Gender Conflict samples (Ambiguous prediction): Gender re-checked based on logR ratio and B allele Frequency Plot (X & Y Chromosome) and found to be concordant with Plink Gender estimation.

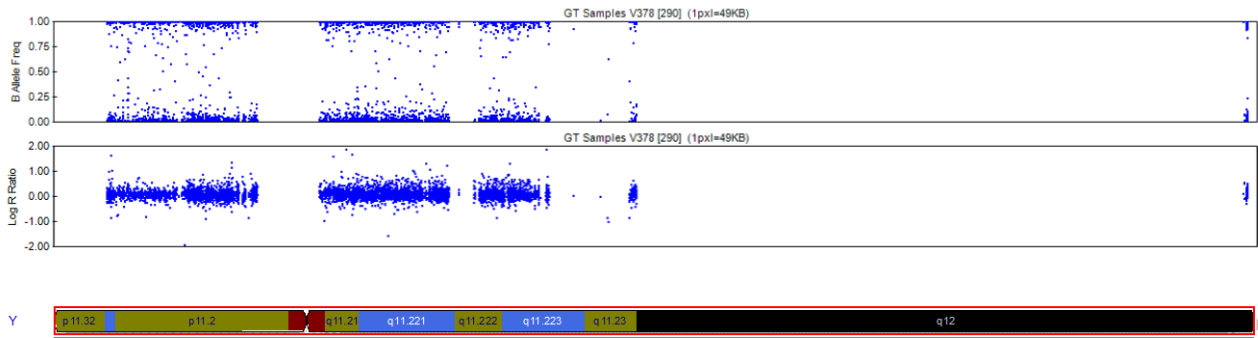
- **Male :** Loss of Heterozygote calls (missing AB cluster at 0.5 scale) in BAF plot at Chromosome X and A/- & B/- calls at chromosome Y.

- **Female:** No loss of heterozygote calls (AB cluster at 0.5 scale) in BAF plot at Chromosome X and Noise can be observed at chromosome Y.

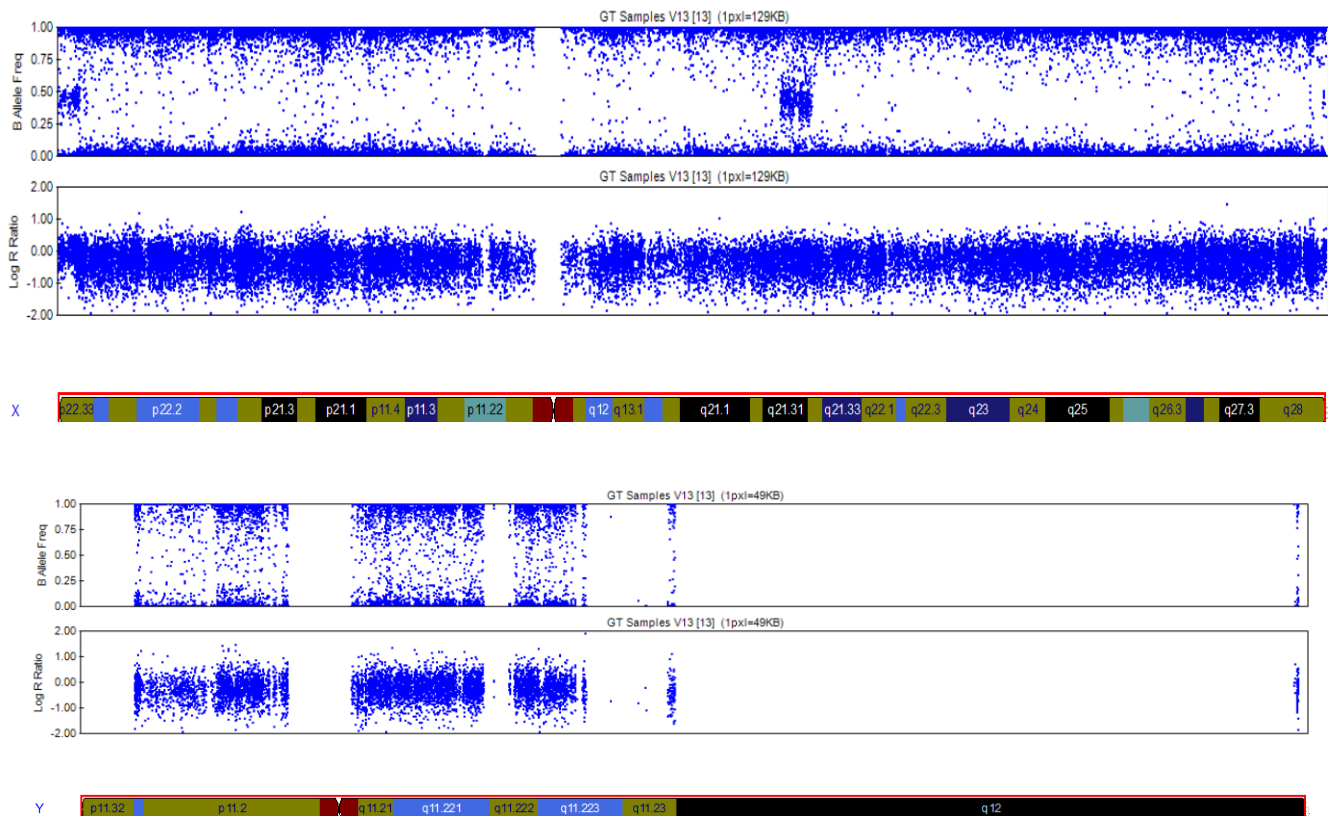
**Note: Due to low data noise and low call rate (< 0.99) Genome studio could not able to predict the Gender.*

1. V378 (Male)

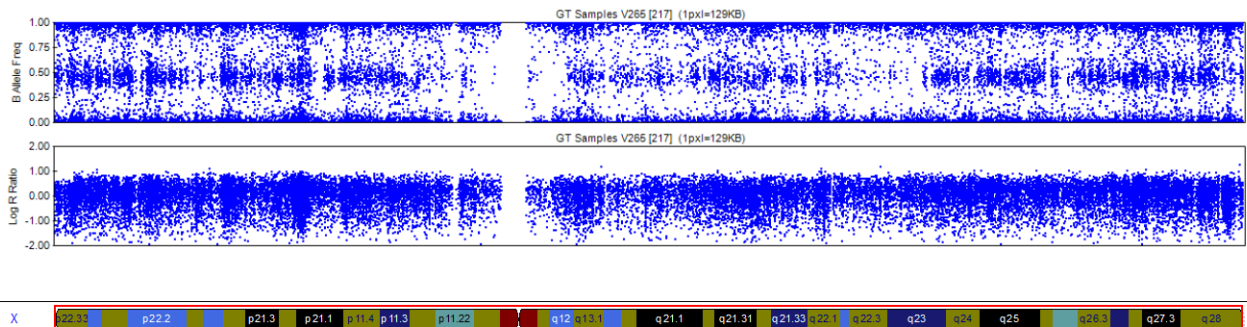


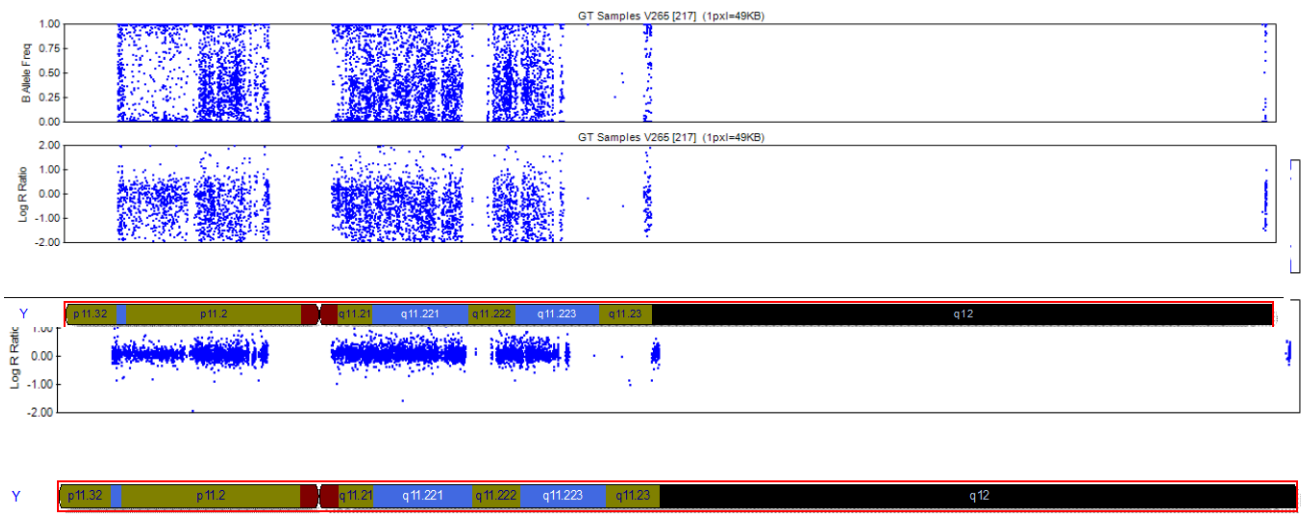


2. V13 (Male)

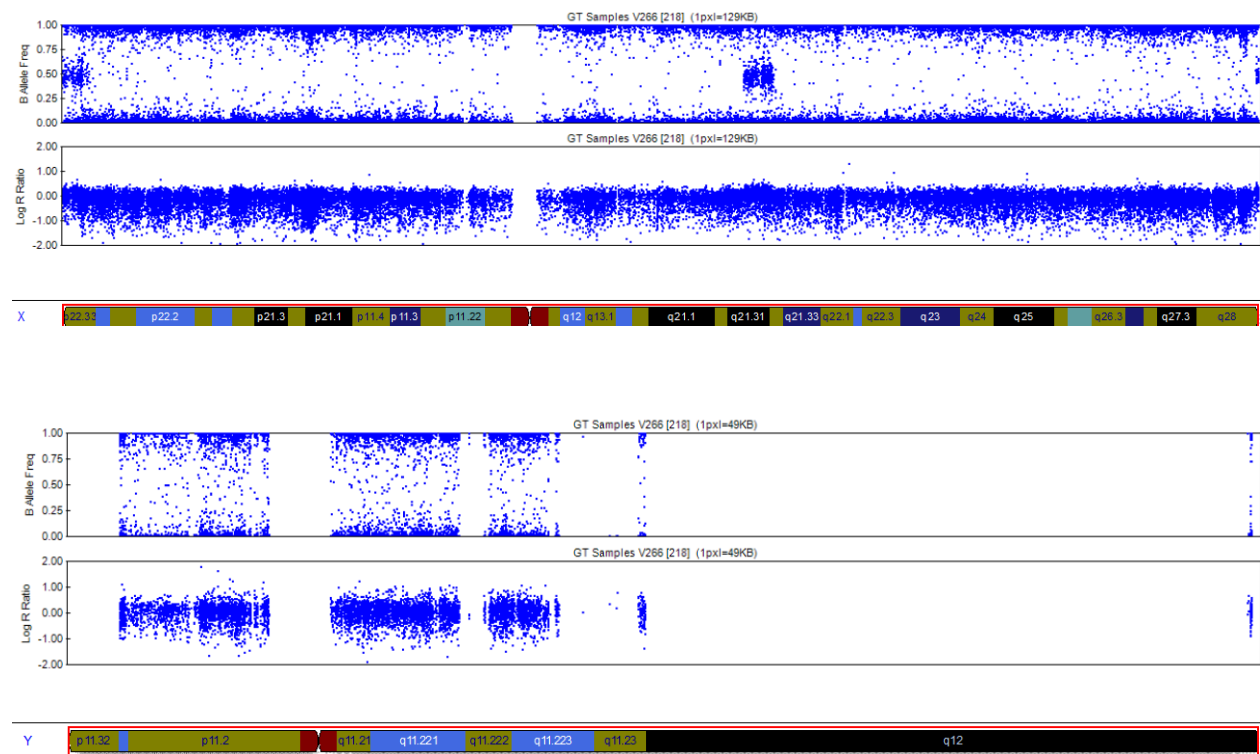


3. V265 (Female)

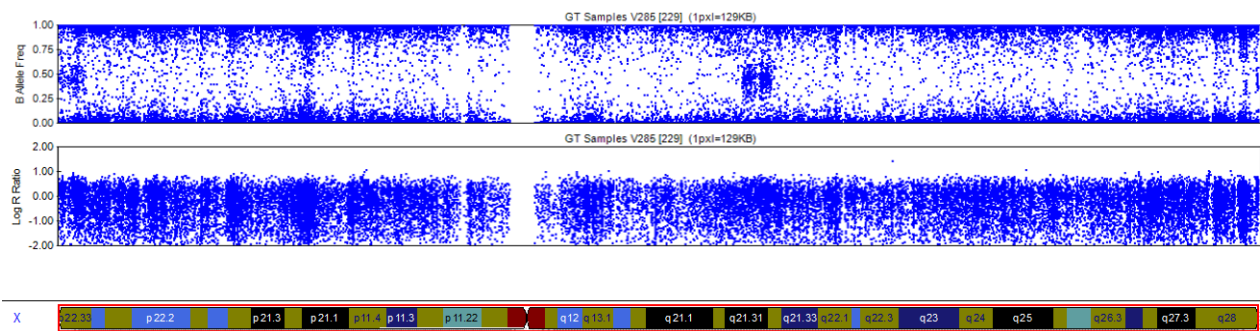


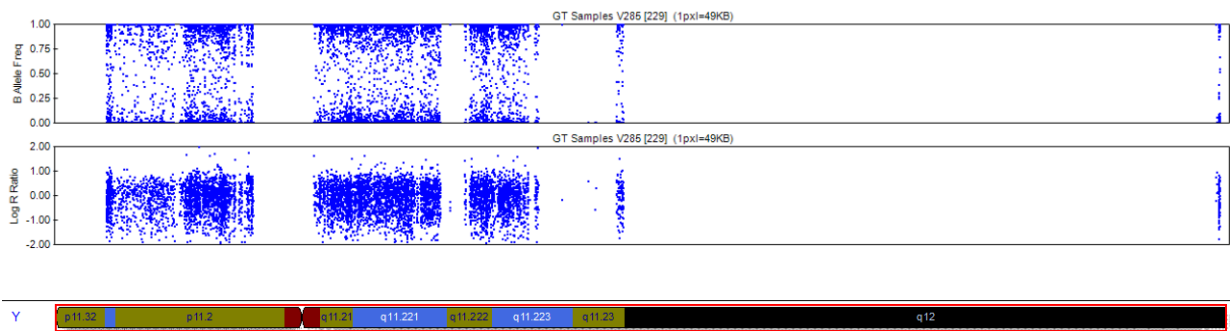


4. V266 (Male)

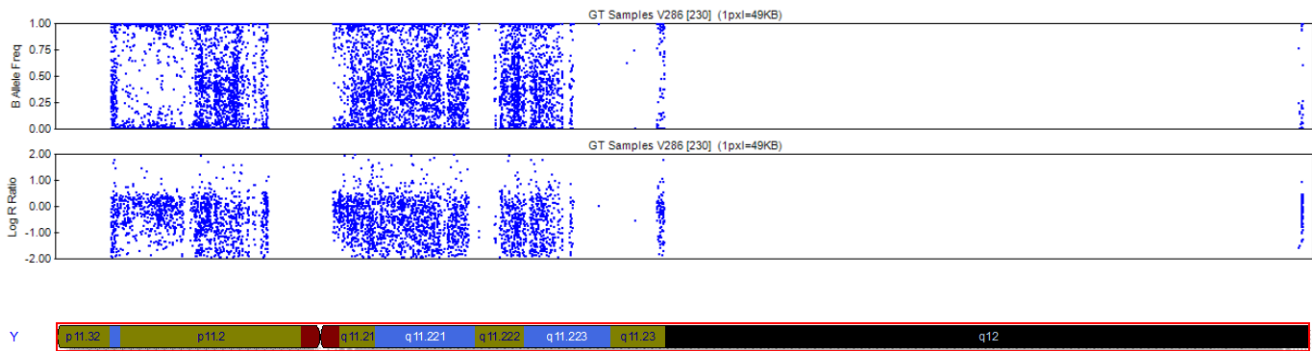
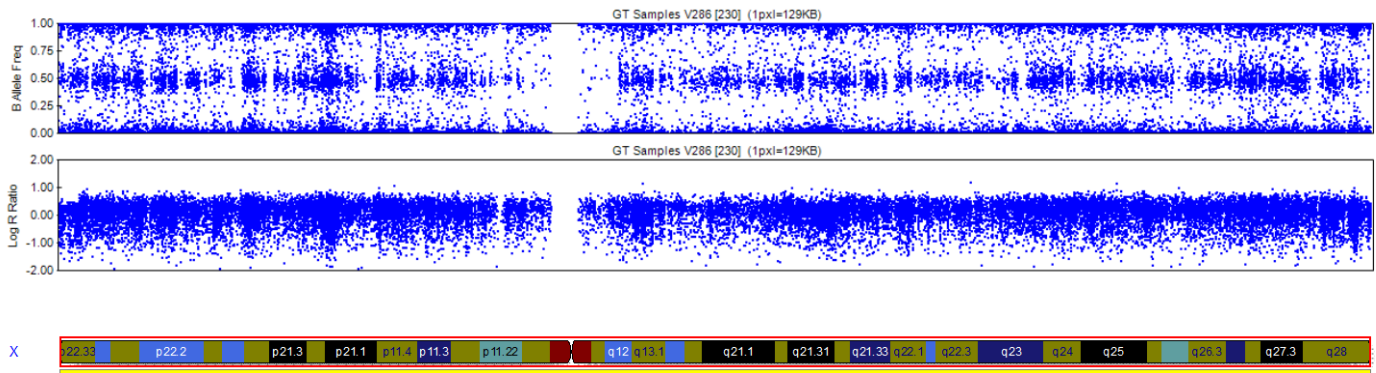


5. V285 (Male)



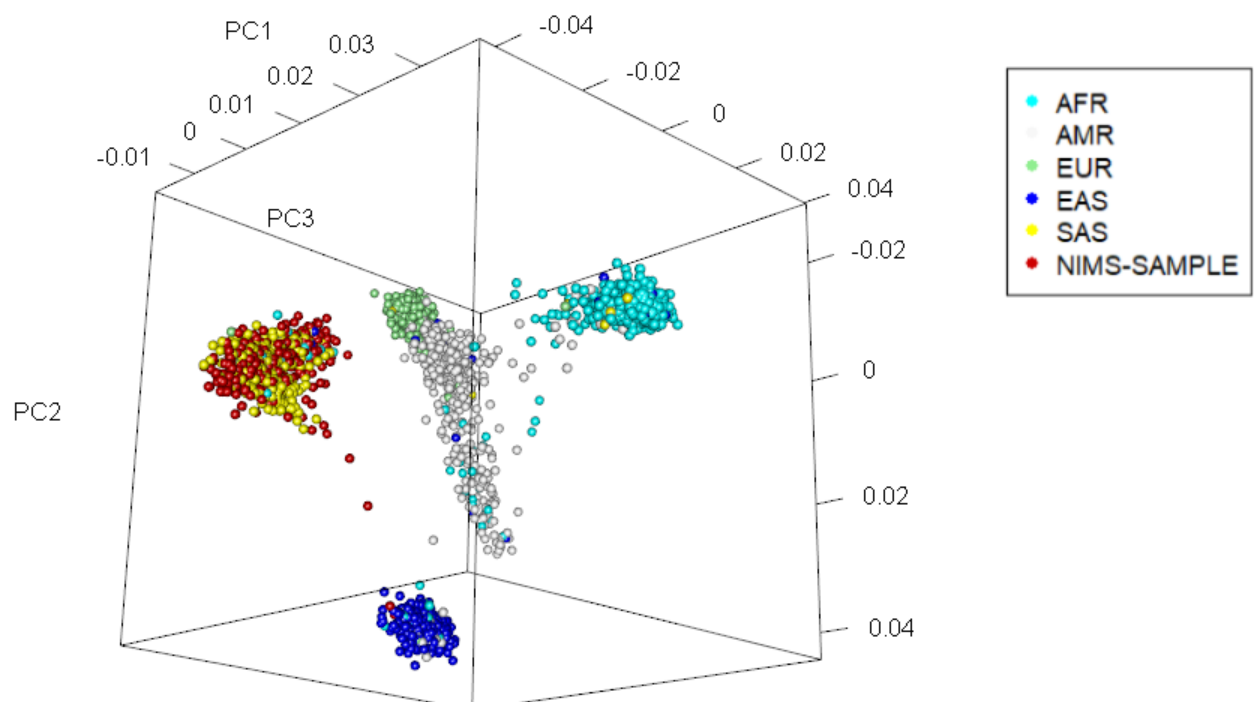
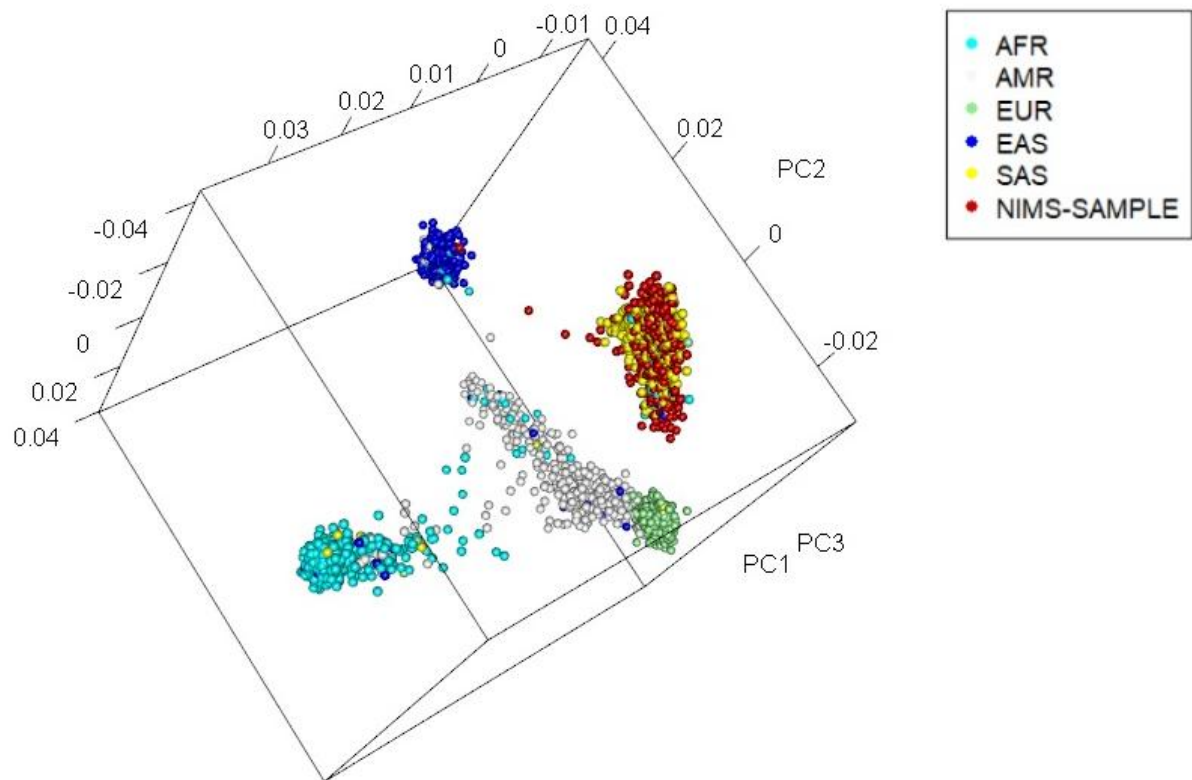


6. V286 (Female)



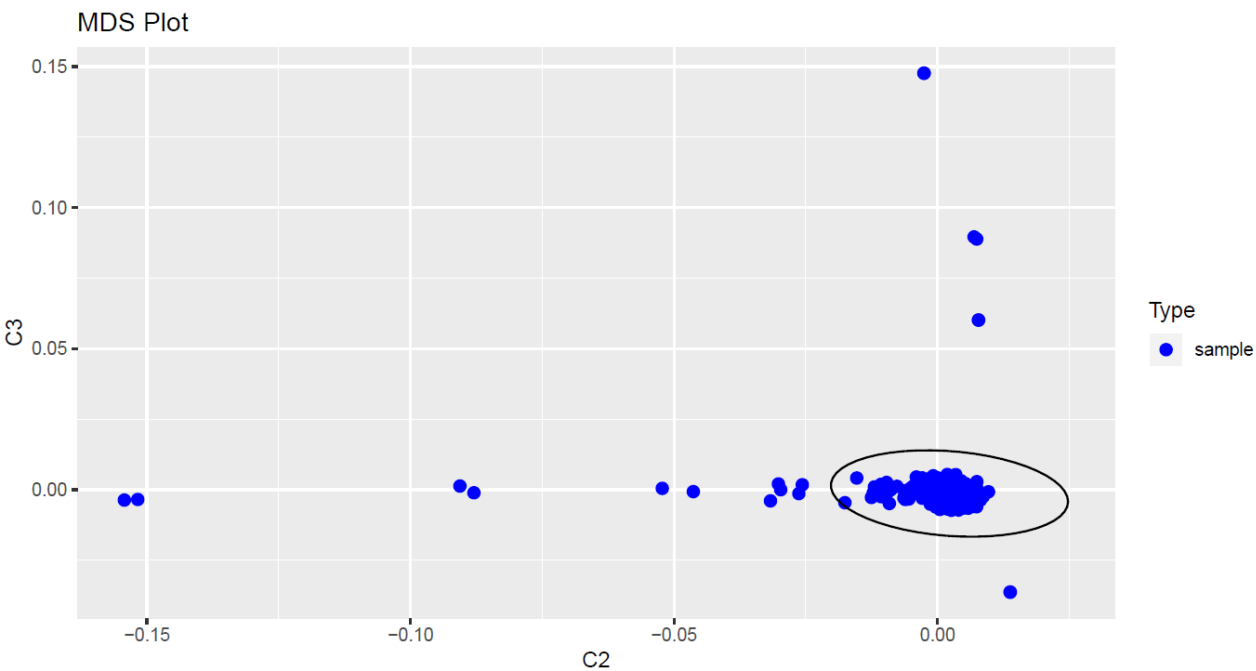
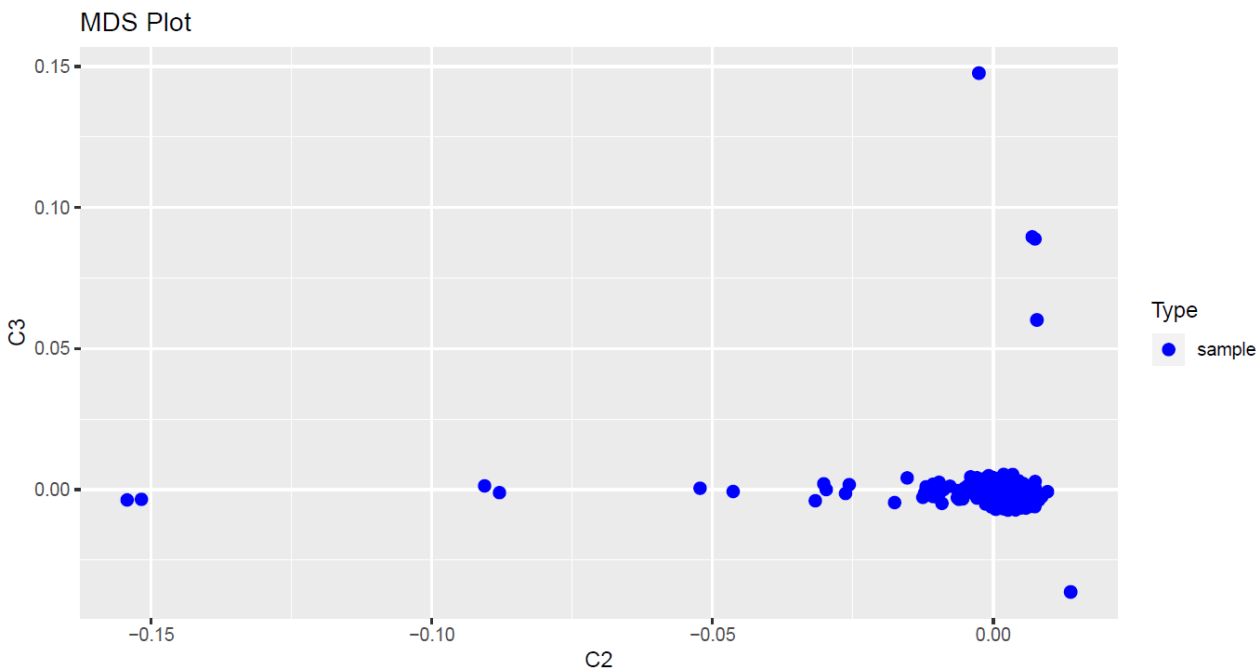
Ancestry Prediction (AIM markers): Nimhans data (N=407) + 1000 Genome (=2504)

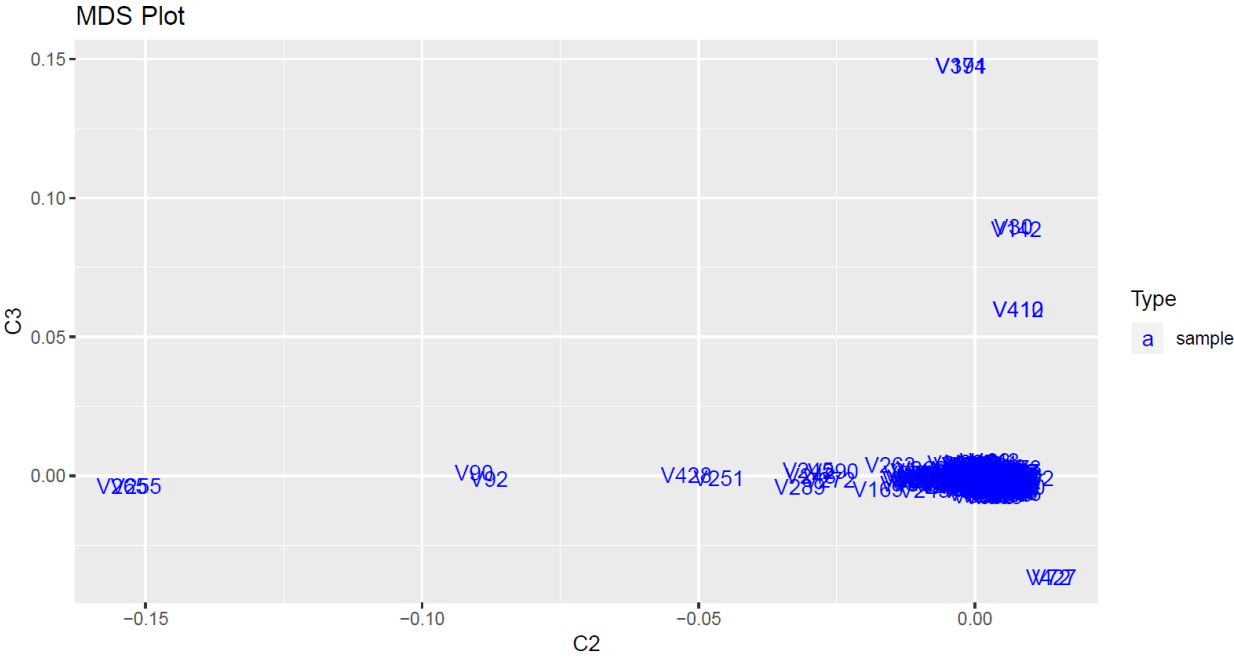
Principle Component Analysis



Population substructure Analysis: NIMHANS samples

Multidimensional Scaling





Principal Component Analysis:

