

CHARGE ExomeChip QC Controls

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Goals:

The ultimate goal will be to exchange raw genotyping data (R, Theta, etc.) without phenotypes to allow joint calling of all variants across the entire CHARGE+ project. In addition to the standard Illumina GenomeStudio calling algorithms, we can then explore other statistical methods for calling of rare variants.

However, in the event that joint calling proves too complicated or time consuming for preliminary analyses, we have also selected a set of common controls to be included along with each cohort's samples.

These controls will serve two purposes:

1. Provide a set of samples with known 1000 Genome ascertained genotypes at Exome Chip loci to assess the quality of the genotype calls.
2. Serve as cross-study replicates in the event that we aren't able to do joint calling across all of CHARGE+ to address similarities and differences in local calling algorithms.

Methods:

Each cohort will include at least 96 Coriell Samples in their project, ideally with at least one common control sample per 96 well plate. **The 96 samples listed in Appendix B are proposed to be genotyped by all studies.** Further information about those samples are found at Coriell:

- 48 samples from European origin individuals (CEU), [HAPMAPPT01-European](#)
- 48 samples from African origin individuals (YRI), [HAPMAPPT03-Yoruban-African](#)

Each panel contains 50 micrograms of DNA from unrelated individuals within the specified population at a cost of \$8,000 per panel. The DNAs are also available individually at \$85. These 96 samples listed in Appendix B should be genotyped by all studies.

The minimum for each genome center will be to genotype 24 males and 24 females from each population. Genome centers may elect to add the children of these 24 parents if the center wants to genotype trios. These samples, listed in Appendix C, will be used for additional QC if available. A full list of samples is also available as an Excel spreadsheet [Exomechip_qc_controls.xlsx](#).

Features of this plan:

1. Maximizes number of unrelateds to increase the number of alleles available.
2. Uses samples with public genotyping and sequencing data available for comparisons.
3. Includes both Caucasian samples, the population with the largest number of samples in ESP, and Yoruban samples, the population with the larger number of alleles.
4. Availability of samples to all genome centers.
5. Relies on the genome center to obtain the DNAs needed, rather than a central lab.

In addition to these samples, members of the cohort previously sequenced in ESP or CHARGE-S will serve as additional QC samples for these rare variants (Common SNPs on the Exome Chip will also be useful for "fingerprinting" against existing GWAS for detection of sample swaps). For the samples previously sequenced in ESP, this will have the benefit of generating genotypes on those from whom the exome chip contents were derived.

Appendix A: Example plating schemes

- CHS will include 1 internal duplicate per plate (in a fixed position) along with two blank wells for common control samples (1 “moving” to identify plates; 1 fixed to the last plate spot to provide plate orientation) to be plated by the Cedars-Sinai genotyping lab.
- *Cohorts that are unable to intersperse controls across plates may need to resort to a single plate of common controls.*

Appendix B: Required Unrelated Samples

Full list of 96 required samples and 48 optional samples listed below. Also available as an Excel spreadsheet ([Exomechip_qc_controls.xlsx](#))

Ancestry	Panel	Type	Catalog ID	Priority	Sex	Family	Relationship
European	HAPMAPPT01	unrelateds	GM12892	1	Female	1463	Maternal Grandmother
European	HAPMAPPT01	unrelateds	GM12891	3	Male	1463	Maternal Grandfather
European	HAPMAPPT01	unrelateds	GM12873	5	Female	1459	Paternal Grandmother
European	HAPMAPPT01	unrelateds	GM12872	7	Male	1459	Paternal Grandfather
European	HAPMAPPT01	unrelateds	GM12875	9	Female	1459	Maternal Grandmother
European	HAPMAPPT01	unrelateds	GM12874	11	Male	1459	Maternal Grandfather
European	HAPMAPPT01	unrelateds	GM12813	13	Female	1454	Paternal Grandmother
European	HAPMAPPT01	unrelateds	GM12815	15	Female	1454	Maternal Grandmother
European	HAPMAPPT01	unrelateds	GM12812	17	Male	1454	Paternal Grandfather
European	HAPMAPPT01	unrelateds	GM12814	19	Male	1454	Maternal Grandfather
European	HAPMAPPT01	unrelateds	GM12761	21	Female	1447	Paternal Grandmother
European	HAPMAPPT01	unrelateds	GM12763	23	Female	1447	Maternal Grandmother
European	HAPMAPPT01	unrelateds	GM12760	25	Male	1447	Paternal Grandfather
European	HAPMAPPT01	unrelateds	GM12762	27	Male	1447	Maternal Grandfather
European	HAPMAPPT01	unrelateds	GM12751	29	Female	1444	Maternal Grandmother
European	HAPMAPPT01	unrelateds	GM12750	31	Male	1444	Maternal Grandfather
European	HAPMAPPT01	unrelateds	GM12004	33	Female	1420	Paternal Grandmother
European	HAPMAPPT01	unrelateds	GM12006	35	Female	1420	Maternal Grandmother
European	HAPMAPPT01	unrelateds	GM12003	37	Male	1420	Paternal Grandfather
European	HAPMAPPT01	unrelateds	GM12005	39	Male	1420	Maternal Grandfather
European	HAPMAPPT01	unrelateds	GM12249	41	Female	1416	Paternal Grandmother
European	HAPMAPPT01	unrelateds	GM12248	43	Male	1416	Paternal Grandfather
European	HAPMAPPT01	unrelateds	GM12236	45	Female	1408	Paternal Grandmother
European	HAPMAPPT01	unrelateds	GM12154	47	Male	1408	Paternal Grandfather
European	HAPMAPPT01	unrelateds	GM12156	49	Female	1408	Maternal Grandmother
European	HAPMAPPT01	unrelateds	GM12155	51	Male	1408	Maternal Grandmother
European	HAPMAPPT01	unrelateds	GM12234	53	Female	1375	Maternal Grandmother
European	HAPMAPPT01	unrelateds	GM12264	55	Male	1375	Maternal Grandfather
European	HAPMAPPT01	unrelateds	GM11995	57	Female	1362	Maternal Grandmother
European	HAPMAPPT01	unrelateds	GM11993	59	Female	1362	Paternal Grandmother
European	HAPMAPPT01	unrelateds	GM11992	61	Male	1362	Paternal Grandfather
European	HAPMAPPT01	unrelateds	GM11994	63	Male	1362	Maternal Grandfather
European	HAPMAPPT01	unrelateds	GM12717	65	Female	1358	Paternal Grandmother
European	HAPMAPPT01	unrelateds	GM12716	67	Male	1358	Paternal Grandfather
European	HAPMAPPT01	unrelateds	GM11830	69	Female	1350	Paternal Grandmother
European	HAPMAPPT01	unrelateds	GM11832	71	Female	1350	Maternal Grandmother
European	HAPMAPPT01	unrelateds	GM11831	73	Male	1350	Maternal Grandfather
European	HAPMAPPT01	unrelateds	GM11829	75	Male	1350	Paternal Grandfather
European	HAPMAPPT01	unrelateds	GM11840	77	Female	1349	Maternal Grandmother
European	HAPMAPPT01	unrelateds	GM11839	79	Male	1349	Maternal Grandfather
European	HAPMAPPT01	unrelateds	GM11882	81	Female	1347	Maternal Grandmother
European	HAPMAPPT01	unrelateds	GM11881	83	Male	1347	Maternal Grandfather
European	HAPMAPPT01	unrelateds	GM12044	85	Female	1346	Paternal Grandmother
European	HAPMAPPT01	unrelateds	GM12043	87	Male	1346	Paternal Grandfather
European	HAPMAPPT01	unrelateds	GM07345	89	Female	1345	Maternal Grandmother
European	HAPMAPPT01	unrelateds	GM07357	91	Male	1345	Maternal Grandfather
European	HAPMAPPT01	unrelateds	GM12057	93	Female	1344	Paternal Grandmother
European	HAPMAPPT01	unrelateds	GM12056	95	Male	1344	Paternal Grandfather

Ancestry	Panel	Type	Catalog ID	Priority	Sex	Family	Relationship
African	HAPMAPPT03	unrelateds	GM19238	2	Female	Y117	Mother
African	HAPMAPPT03	unrelateds	GM19239	4	Male	Y117	Father
African	HAPMAPPT03	unrelateds	GM19193	6	Female	Y112	Mother
African	HAPMAPPT03	unrelateds	GM19192	8	Male	Y112	Father
African	HAPMAPPT03	unrelateds	GM19099	10	Female	Y105	Mother
African	HAPMAPPT03	unrelateds	GM19098	12	Male	Y105	Father
African	HAPMAPPT03	unrelateds	GM19131	14	Female	Y101	Mother
African	HAPMAPPT03	unrelateds	GM19130	16	Male	Y101	Father
African	HAPMAPPT03	unrelateds	GM19127	18	Female	Y077	Mother
African	HAPMAPPT03	unrelateds	GM19128	20	Male	Y077	Father
African	HAPMAPPT03	unrelateds	GM19143	22	Female	Y074	Mother
African	HAPMAPPT03	unrelateds	GM19144	24	Male	Y074	Father
African	HAPMAPPT03	unrelateds	GM19152	26	Female	Y072	Mother
African	HAPMAPPT03	unrelateds	GM19153	28	Male	Y072	Father
African	HAPMAPPT03	unrelateds	GM19140	30	Female	Y071	Mother
African	HAPMAPPT03	unrelateds	GM19141	32	Male	Y071	Father
African	HAPMAPPT03	unrelateds	GM19116	34	Female	Y060	Mother
African	HAPMAPPT03	unrelateds	GM19119	36	Male	Y060	Father
African	HAPMAPPT03	unrelateds	GM19122	38	Female	Y058	Mother
African	HAPMAPPT03	unrelateds	GM19223	40	Male	Y058	Father
African	HAPMAPPT03	unrelateds	GM19159	42	Female	Y056	Mother
African	HAPMAPPT03	unrelateds	GM19160	44	Male	Y056	Father
African	HAPMAPPT03	unrelateds	GM19206	46	Female	Y051	Mother
African	HAPMAPPT03	unrelateds	GM19206	48	Male	Y051	Father
African	HAPMAPPT03	unrelateds	GM19209	50	Female	Y050	Mother
African	HAPMAPPT03	unrelateds	GM19210	52	Male	Y050	Father
African	HAPMAPPT03	unrelateds	GM19204	54	Female	Y048	Mother
African	HAPMAPPT03	unrelateds	GM19203	56	Male	Y048	Father
African	HAPMAPPT03	unrelateds	GM19172	58	Female	Y047	Mother
African	HAPMAPPT03	unrelateds	GM19171	60	Male	Y047	Father
African	HAPMAPPT03	unrelateds	GM19201	62	Female	Y045	Mother
African	HAPMAPPT03	unrelateds	GM19200	64	Male	Y045	Father
African	HAPMAPPT03	unrelateds	GM19137	66	Female	Y043	Mother
African	HAPMAPPT03	unrelateds	GM19138	68	Male	Y043	Father
African	HAPMAPPT03	unrelateds	GM19102	70	Female	Y042	Mother
African	HAPMAPPT03	unrelateds	GM19101	72	Male	Y042	Father
African	HAPMAPPT03	unrelateds	GM19093	74	Female	Y040	Mother
African	HAPMAPPT03	unrelateds	GM19092	76	Male	Y040	Father
African	HAPMAPPT03	unrelateds	GM18912	78	Female	Y028	Mother
African	HAPMAPPT03	unrelateds	GM18913	80	Male	Y028	Father
African	HAPMAPPT03	unrelateds	GM18861	82	Female	Y024	Mother
African	HAPMAPPT03	unrelateds	GM18862	84	Male	Y024	Father
African	HAPMAPPT03	unrelateds	GM18855	86	Female	Y023	Mother
African	HAPMAPPT03	unrelateds	GM18856	88	Male	Y023	Father
African	HAPMAPPT03	unrelateds	GM18852	90	Female	Y018	Mother
African	HAPMAPPT03	unrelateds	GM18853	92	Male	Y018	Father
African	HAPMAPPT03	unrelateds	GM18870	94	Female	Y017	Mother
African	HAPMAPPT03	unrelateds	GM18871	96	Male	Y017	Father

Appendix C: Additional Samples for Trios

Some of the laboratories would like to genotype trios. This is the list of those individuals that complete trios for the parents in the previous tables

Ancestry	Panel	Type	Catalog ID	Priority	Sex	Family	Relationship
European	HAPMAPPT01	Children	GM12878	1	Female	1463	Mother
European	HAPMAPPT01	Children	GM12865	3	Female	1459	Mother
European	HAPMAPPT01	Children	GM12864	5	Male	1459	Father
European	HAPMAPPT01	Children	GM12802	7	Female	1454	Mother
European	HAPMAPPT01	Children	GM12801	9	Male	1454	Father
European	HAPMAPPT01	Children	GM12753	11	Female	1447	Mother
European	HAPMAPPT01	Children	GM12752	13	Male	1447	Father
European	HAPMAPPT01	Children	GM12740	15	Female	1444	Mother
European	HAPMAPPT01	Children	GM10839	17	Female	1420	Mother
European	HAPMAPPT01	Children	GM10838	19	Male	1420	Father
European	HAPMAPPT01	Children	GM10835	21	Male	1416	Father
European	HAPMAPPT01	Children	GM10831	23	Female	1408	Mother
European	HAPMAPPT01	Children	GM10830	25	Male	1408	Father
European	HAPMAPPT01	Children	GM10863	27	Female	1375	Mother
European	HAPMAPPT01	Children	GM10861	29	Female	1362	Mother
European	HAPMAPPT01	Children	GM10860	31	Male	1362	Father
European	HAPMAPPT01	Children	GM12707	33	Male	1358	Father
European	HAPMAPPT01	Children	GM10855	35	Female	1350	Mother
European	HAPMAPPT01	Children	GM10856	37	Male	1350	Father
European	HAPMAPPT01	Children	GM10854	39	Female	1349	Mother
European	HAPMAPPT01	Children	GM10858	41	Female	1347	Mother
European	HAPMAPPT01	Children	GM10857	43	Female	1346	Mother
European	HAPMAPPT01	Children	GM07348	45	Female	1345	Mother
European	HAPMAPPT01	Children	GM10851	47	Male	1344	Father
African	HAPMAPPT03	Children	GM19240	2	Female	Y117	Child
African	HAPMAPPT03	Children	GM19194	4	Male	Y112	Child
African	HAPMAPPT03	Children	GM19100	6	Female	Y105	Child
African	HAPMAPPT03	Children	GM19132	8	Female	Y101	Child
African	HAPMAPPT03	Children	GM19129	10	Female	Y077	Child
African	HAPMAPPT03	Children	GM19145	12	Male	Y074	Child
African	HAPMAPPT03	Children	GM19154	14	Male	Y072	Child
African	HAPMAPPT03	Children	GM19142	16	Male	Y071	Child
African	HAPMAPPT03	Children	GM19120	18	Male	Y060	Child
African	HAPMAPPT03	Children	GM19221	20	Female	Y058	Child
African	HAPMAPPT03	Children	GM19161	22	Male	Y056	Child
African	HAPMAPPT03	Children	GM19208	24	Male	Y051	Child
African	HAPMAPPT03	Children	GM19211	26	Male	Y050	Child
African	HAPMAPPT03	Children	GM19205	28	Male	Y048	Child
African	HAPMAPPT03	Children	GM19173	30	Male	Y047	Child
African	HAPMAPPT03	Children	GM19202	32	Female	Y045	Child
African	HAPMAPPT03	Children	GM19139	34	Male	Y043	Child
African	HAPMAPPT03	Children	GM19103	36	Male	Y042	Child
African	HAPMAPPT03	Children	GM19094	38	Female	Y040	Child
African	HAPMAPPT03	Children	GM18914	40	Male	Y028	Child
African	HAPMAPPT03	Children	GM18863	42	Male	Y024	Child
African	HAPMAPPT03	Children	GM18857	44	Male	Y023	Child
African	HAPMAPPT03	Children	GM18854	46	Male	Y018	Child
African	HAPMAPPT03	Children	GM18872	48	Male	Y017	Child