The American Journal of Human Genetics Volume 87

## **Supplemental Data**

## **Spoiling the Whole Bunch: Quality Control Aimed**

# at Preserving the Integrity of High-Throughput Genotyping

Anna Pluzhnikov, Jennifer E. Below, Anuar Konkashbaev, Anna Tikhomirov, Emily Kistner-Griffin, Cheryl A. Roe, Dan L. Nicolae, and Nancy J. Cox



Figure S1.

Flow chart of recommended quality control measures.



### Figure S2.

Normalized signal intensity plots for the rest of the SNPs that retain a highly significant

plate bias (P-value < 10-7) upon removal of eight problem samples, but before the recall. Panels (A)-(E) correspond to Nsp-specific SNPs, (F)-(K), to Sty-specific, and (L)-(X), to Nsp/Sty SNPs.



### Figure S3.

Log-transformed *normalized* probe intensity and contrast distributions for the samples on plate 4RWG569. As in Figure 3, the top row corresponds to the panel of Nspspecific, and the bottom row, Sty-specific SNPs. Solid black curves representing the eight samples with Sty target preparation failure remain completely distinct. However, in contrast to Figure 3, the sample identified by SQC and CQC as failing on both Sty and Nsp panels appears "normal" in all four plots.



Figure S4.

Restriction fragment length distribution of the full (A,C) and CQC panel (B,D) set of enzyme-specific probes . The top row corresponds to Nsp, and the bottom row, to Sty enzyme.

		SQC	panel	SQC all		CQC panel				CQC all			
GoKinD ID	Plate ID	score Nsp	score Sty	score Nsp	score Sty	score	score random	score Nsp	score Sty	score	score random	score Nsp	score Sty
G325	4RWG569	-0.19	0.20	-0.14	0.18	0.00	1.72	3.99	-0.07	0.00	1.39	3.56	-0.02
G306	4RWG569	-0.19	0.14	-0.15	0.13	0.00	1.59	3.94	0.00	0.00	1.26	3.30	-0.01
G537	4RWG569	-0.14	0.17	-0.08	0.17	0.00	1.43	3.62	-0.10	0.00	1.15	2.93	-0.02
G544	4RWG569	-0.12	0.18	-0.08	0.17	0.00	1.30	4.39	-0.03	0.00	1.07	2.87	-0.02
G1987	4RWG569	-0.21	0.17	-0.14	0.16	0.00	1.40	4.39	-0.10	0.00	1.31	3.42	-0.04
G540	4RWG569	-0.23	0.15	-0.17	0.15	0.00	1.27	4.19	-0.03	0.00	1.46	3.68	-0.01
G585	4RWG569	-0.16	0.16	-0.15	0.17	0.00	2.03	3.99	0.00	0.00	1.49	3.70	-0.01
G602	4RWG569	-0.13	0.15	-0.10	0.16	0.00	1.43	4.19	-0.03	0.00	1.26	3.29	-0.04
G139	4BWG569	0.12	0.11	0.16	0.19	-0.10	-0.10	1.22	1.76	-0.13	-0.13	-0.29	-0.11
G895	3KYJ479	-0.14	0.15	-0.14	0.13	0.00	1.75	3.82	0.00	0.00	1.43	3.46	-0.04
G925	3KYJ479	-0.14	0.12	-0.14	0.13	0.00	0.92	3.48	0.00	0.00	1.25	3.39	-0.01
G5911	1CWO500	0.11	-0.07	0.15	0.07	0.35	0.35	0.88	2.41	0.79	0.79	0.26	0.34
G1062*	5GBM583	0.07	-0.04	0.13	-0.01	0.00	0.00	-0.51	2.54	-0.15	-0.15	-0.20	1.44
G4663	1UBP242	0.15	-0.02	0.19	0.14	0.10	0.10	0.62	2.34	0.56	0.56	0.12	0.09
G816	9BHI 593	-0.17	-0.14	0.09	0.11	0.06	0.06	4.30	3.79	0.33	0.33	0.23	-0.16
G1370	1SHK635	-0.04	-0.11	-0.06	-0.03	0.00	2.86	2 41	4 54	3.08	3.08	3.09	2.38
G5610	90YW461	0.06	-0.05	-0.06	-0.06	0.00	2.86	-1 64	2.68	2.28	2.28	2.28	2.03
G4450	9RHI 593	-0.02	-0.00	0.00	0.00	-0.38	-0.38	1 30	1 /0	0.56	0.56	0.50	-0.02
G935	9RHI 593	-0.02	-0.07	-0.06	-0.06	0.13	-0.00	1.05	0.91	0.33	0.33	0.50	0.02
G5065		-0.08	-0.07	-0.00	-0.00	0.13	0.13	1.00	1.42	0.33	0.33	0.40	0.00
G3005		-0.07	-0.08	-0.00	-0.05	0.35	0.35	1.01	0.64	0.54	0.94	0.92	0.04
G4005	SHILDSS	-0.07	-0.07	0.00	-0.00	0.35	1 10	1.00	4.57	1.05	1.05	0.40	0.13
63729	0GXO476	0.00	-0.07	0.15	0.10	0.00	1.10	0.00	4.57	1.25	1.25	0.00	0.91
03579	90X0476	0.04	-0.07	0.05	0.04	0.00	0.99	0.99	0.70	0.50	1.04	0.62	0.23
G3563	9670476	0.07	0.05	0.08	0.11	0.57	0.57	0.71	0.78	0.52	0.52	0.59	0.00
NA12144	8WWR250	0.04	-0.03	-0.06	-0.03	0.00	2.73	0.96	3.01	2.78	2.78	2.76	1.89
G1099	7FDZ321	0.10	0.05	0.09	0.10	0.79	0.79	0.93	1.93	0.62	0.62	0.50	-0.01
G2279	7FDZ321	0.08	0.01	0.06	0.07	0.10	0.10	1.39	1.49	0.63	0.63	0.55	0.14
G4097	9RHL593	-0.16	-0.12	0.07	0.09	0.19	0.19	4.33	3.46	0.40	0.40	0.38	0.00
G3433	9RHL593	-0.19	-0.16	0.07	0.09	0.16	0.16	4.70	3.56	0.72	0.72	0.74	0.00
G2450	9RHL593	-0.20	-0.14	0.06	0.08	0.32	0.32	3.99	3.22	0.65	0.65	0.74	0.00
G718	9RHL593	-0.16	-0.10	0.07	0.08	0.16	0.16	4.45	3.29	0.30	0.30	0.26	-0.21
G3625	9RHL593	-0.16	-0.11	0.08	0.09	0.25	0.25	4.08	3.29	0.30	0.30	0.14	-0.10
G783	9RHL593	-0.16	-0.13	0.06	0.07	0.22	0.22	4.39	3.18	0.23	0.23	0.14	0.00
G4096	9RHL593	-0.18	-0.16	0.02	0.04	0.25	0.25	3.99	2.91	0.63	0.63	0.62	-0.01
G853	9RHL593	-0.20	-0.17	0.07	0.09	0.38	0.38	3.88	3.22	0.44	0.44	0.38	-0.16
G4141	5ZCV995	-0.02	-0.14	0.07	-0.07	0.00	0.79	1.27	4.00	0.00	0.55	-0.01	2.97
G4231	5ZCV995	0.07	-0.10	-0.04	-0.05	0.00	2.89	0.00	3.46	2.77	2.77	2.34	2.76
G4220**	5ZCV995	-0.01	-0.08	0.00	-0.01	0.00	1.14	0.68	4.13	0.00	1.33	0.32	2.62
G5199	5ZCV995	0.07	-0.05	0.09	0.04	-0.06	-0.06	0.85	2.51	0.50	0.50	0.00	0.18
G5336	5ZCV995	-0.06	-0.10	-0.04	-0.06	0.00	0.60	0.65	3.29	1.02	1.02	0.00	1.99
NA12750	5ZCV995	-0.04	-0.09	-0.04	-0.07	0.00	0.19	-0.20	3.46	0.00	0.44	-0.47	2.41
G2977	6KAE204	-0.02	-0.05	-0.03	0.01	0.00	3.18	4.39	2.34	2.84	2.84	2.91	2.19
G1882	1DXJ626	0.02	-0.07	-0.07	-0.02	0.00	2.80	1.42	3.49	2.90	2.90	3.06	2.34
G2504	1DXJ626	0.05	-0.05	-0.03	-0.01	0.00	1.11	-0.03	2.07	1.14	1.14	1.25	0.53
G503	6PKP514	0.11	0.07	0.08	0.12	0.67	0.67	-0.06	-0.41	0.61	0.61	0.61	-0.09
G5728	6PKP514	0.16	-0.01	0.16	0.10	1.18	1.18	0.11	1.76	0.60	0.60	0.04	0.30
G257	6PKP514	0.05	0.06	0.02	0.04	0.00	1.18	0.00	2.03	1.46	1.46	1.37	0.78
G4695	3FQM386	0.01	0.02	0.09	0.10	1.18	1.18	2.58	1.96	0.72	0.72	0.67	0.00
G4366	0VXC556	-0.12	-0.11	-0.09	-0.06	0.00	1.37	3.54	1.49	1.50	1.50	1.70	0.56
G150	0VXC556	-0.18	-0.07	-0.05	-0.01	0.00	2.64	4.64	2.34	2.66	2.66	2.83	1.93
G6019	3ECO791	0.09	-0.07	0.16	0.11	0.83	0.83	1.47	3.12	0.96	0.96	0.39	0.38
G4651	cherry_pick_redo1	0.09	-0.02	0.14	0.11	0.00	1.72	1.67	3.93	1.16	1.16	0.71	0.64
*This samp	le was removed from	m the analys	ses due to l	ow call rate									

\*\*This sample was removed from the analyses due to sex misspecification

### Table S1.

Problem GoKinD samples identified by SQC and CQC tests. The problem samples discussed above appear in the top eight rows. Shaded rows next to them correspond to additional failed samples identified by all four variants of the tests. Figures in boldface correspond to score values meeting the threshold for sample removal for a specific QC test.