

Supplementary Figure 1. Visualization of simultaneously measured SNP and CNV in 100 human genomic DNA

CYP2A6 / 2A7	Ct (CYP2A6 -48T>G) Average of 3-repeats										Ct (CYP2A7) Average of 3-repeats										
Sample Number	11	12	13	14	15	16	17	18	19	20	11	12	13	14	15	16	17	18	19	20	Genomic DNA
IC1: CN2 (Cy5)	29.25	29.29	29.36	29.21	29.46	29.18	29.60	29.21	29.40	29.40	30.09	29.99	29.88	29.83	30.11	29.85	30.08	29.84	30.06	30.14	Threshold : 400
IC2 : CN4 (610)	28.21	28.21	28.22	28.16	28.59	28.10	28.43	28.12	28.28	28.28	29.09	28.98	28.89	28.85	29.17	28.84	29.09	28.80	29.05	29.15	
CYP2A6/2A7 (FAM)	29.37	29.35	29.45	29.25	29.54	28.63	29.64	29.33	29.38	29.50	30.06	29.94	29.82	29.80	30.08	30.65	29.99	29.81	30.04	30.06	
[T] (JOE)	29.25	29.23	30.32	29.21	29.51	28.59	30.56	30.27	30.27	30.56	-	-	-	-	-	-	-	-	-	-	
CN1	30.30	30.37	30.50	30.27	30.34	30.26	30.76	30.30	30.53	30.52	31.09	30.99	30.86	30.82	31.05	30.86	31.07	30.88	31.08	31.13	CN1 constant 100%
CN3	28.64	28.66	28.70	28.60	28.95	28.55	28.92	28.57	28.74	28.75	29.51	29.40	29.30	29.26	29.56	29.26	29.50	29.23	29.47	29.56	CN3 constant 58.5%
CN5	27.87	27.87	27.86	27.82	28.30	27.76	28.06	27.77	27.91	27.92	28.77	28.66	28.57	28.53	28.86	28.52	28.77	28.47	28.72	28.84	CN5 constant 32.2%
CN6	27.59	27.58	27.56	27.54	28.07	27.47	27.75	27.48	27.62	27.63	28.51	28.39	28.31	28.27	28.61	28.25	28.51	28.20	28.45	28.58	CN6 constant 58.6%
																					○ FAM (Total CN) ○ JOE ([T] CN) - CN1 (Calculated) - CN2 (Cy5) - CN3 (Calculated) - CN4 (610) - CN5 (Calculated) - CN6 (Calculated)
[T] Copy Number	2	2	1	2	2	3	1	1	1	1	2A7 Copy Number										
Total Copy Number	2	2	2	2	2	3	2	2	2	2											ND : Not Determined
Diplotype	Tx2	Tx2	Tx1/ Gx1	Tx2	Tx2	Tx3	Tx1/ Gx1	Tx1/ Gx1	Tx1/ Gx1	Tx1/ Gx1	2	2	2	2	2	1	2	2	2	2	

CYP2A6 / 2A7	Ct (CYP2A6 -48T>G) Average of 3-repeats										Ct (CYP2A7) Average of 3-repeats										
Sample Number	21	23	25	26	27	28	30	31	32	33	21	23	25	26	27	28	30	31	32	33	Genomic DNA
IC1: CN2 (Cy5)	29.47	29.22	29.41	29.28	28.77	29.08	29.09	29.33	29.48	29.29	29.86	29.83	30.03	30.03	30.05	29.77	29.96	30.01	30.22	30.11	Threshold : 400
IC2 : CN4 (610)	28.31	28.18	28.31	28.19	27.63	28.07	28.07	28.24	28.35	28.21	28.89	28.84	29.01	29.04	29.08	28.78	28.98	28.98	29.21	29.11	
CYP2A6/2A7 (FAM)	29.50	29.29	29.55	30.33	28.84	29.16	30.19	29.46	29.65	28.68	29.71	29.83	29.89	29.94	31.01	29.80	29.92	29.96	30.12	30.93	
[T] (JOE)	29.29	29.13	30.37	30.06	28.64	29.03	29.98	29.35	30.41	28.62	-	-	-	-	-	-	-	-	-	-	
CN1	30.64	30.27	30.50	30.38	29.90	30.09	30.11	30.42	30.60	30.36	30.84	30.82	31.04	31.01	31.02	30.77	30.94	31.05	31.22	31.12	CN1 constant 100%
CN3	28.79	28.61	28.77	28.64	28.10	28.49	28.49	28.69	28.82	28.66	29.29	29.25	29.43	29.45	29.48	29.19	29.39	29.41	29.63	29.53	CN3 constant 58.5%
CN5	27.94	27.84	27.96	27.84	27.26	27.75	27.74	27.89	27.99	27.87	28.58	28.52	28.69	28.72	28.77	28.46	28.67	28.64	28.89	28.79	CN5 constant 32.2%
CN6	27.63	27.56	27.67	27.55	26.97	27.48	27.47	27.60	27.70	27.59	28.32	28.26	28.42	28.46	28.52	28.20	28.41	28.37	28.62	28.52	CN6 constant 58.6%
32 31 30 29 28 27 26 Ct																					○ FAM (Total CN) ○ JOE ([T] CN) - CN1 (Calculated) - CN2 (Cy5) - CN3 (Calculated) - CN4 (610) - CN5 (Calculated) - CN6 (Calculated)
[T] Copy Number	2	2	1	1	2	2	1	2	1	3	2A7 Copy Number										
Total Copy Number	2	2	2	1	2	2	1	2	2	3											ND : Not Determined
Diplotype	Tx2	Tx2	Tx1/ Gx1	Tx1	Tx2	Tx2	Tx1	Tx2	Tx1/ Gx1	Tx3	2	2	2	2	1	2	2	2	2	1	

CYP2A6 / 2A7	Ct (CYP2A6 -48T>G) Average of 3-repeats										Ct (CYP2A7) Average of 3-repeats																																																																																				
Sample Number	34	35	36	38	39	40	41	42	43	44	34	35	36	38	39	40	41	42	43	44	Genomic DNA																																																																										
IC1: CN2 (Cy5)	29.09	29.30	29.34	29.24	29.23	29.35	29.31	29.33	29.27	29.38	29.82	29.86	29.89	29.90	29.80	29.89	29.96	29.83	29.96	30.05	Threshold : 400																																																																										
IC2 : CN4 (610)	28.04	28.22	28.23	28.19	28.15	28.26	28.19	28.24	28.18	28.28	28.94	28.97	29.06	29.03	28.95	29.01	29.03	28.91	29.08	29.17																																																																											
CYP2A6/2A7 (FAM)	28.52	30.31	29.47	29.39	29.39	30.47	28.33	29.40	29.29	30.39	30.79	29.98	29.46	30.05	29.96	30.04	34.30	30.02	30.08	30.13																																																																											
[T] (JOE)	29.08	40.86	29.34	30.31	38.70	30.25	28.71	30.27	29.18	30.19	-	-	-	-	-	-	-	-	-	-																																																																											
CN1	30.14	30.38	30.45	30.30	30.30	30.44	30.43	30.43	30.36	30.48	30.71	30.75	30.72	30.78	30.65	30.77	30.89	30.74	30.84	30.93	CN1 constant 100%																																																																										
CN3	28.48	28.67	28.69	28.63	28.60	28.72	28.66	28.69	28.64	28.74	29.31	29.34	29.40	29.39	29.30	29.38	29.42	29.29	29.45	29.53	CN3 constant 58.5%																																																																										
CN5	27.71	27.87	27.88	27.85	27.80	27.91	27.83	27.88	27.83	27.93	28.66	28.69	28.79	28.74	28.67	28.73	28.73	28.61	28.80	28.88	CN5 constant 32.2%																																																																										
CN6	27.43	27.59	27.58	27.57	27.52	27.63	27.54	27.60	27.55	27.64	28.42	28.45	28.57	28.51	28.45	28.50	28.49	28.37	28.57	28.65	CN6 constant 58.6%																																																																										
Ct 32 31 30 29 28 27 26																																																																																															

CYP2A6 / 2A7	Ct (CYP2A6 -48T>G) Average of 3-repeats										Ct (CYP2A7) Average of 3-repeats										
Sample Number	56	57	58	59	61	62	63	64	66	67	56	57	58	59	61	62	63	64	66	67	Genomic DNA
IC1: CN2 (Cy5)	29.89	29.37	29.44	29.64	29.62	29.23	29.38	29.30	29.86	29.35	30.12	29.93	29.98	30.15	29.92	29.80	30.07	29.82	30.24	30.21	Threshold : 400
IC2 : CN4 (610)	28.75	28.33	28.39	28.52	28.49	28.25	28.36	28.24	28.76	28.36	29.10	28.95	28.97	29.09	28.94	28.86	29.08	28.86	29.18	29.15	
CYP2A6/2A7 (FAM)	29.90	29.37	29.36	29.86	29.58	30.29	29.46	29.41	29.15	30.39	30.13	30.00	30.02	30.17	29.98	29.93	30.13	29.93	31.14	30.72	
[T] (JOE)	30.91	30.34	29.37	30.76	29.55	30.18	29.50	30.51	29.75	30.39	-	-	-	-	-	-	-	-	-	-	
CN1	31.03	30.41	30.50	30.75	30.74	30.21	30.39	30.36	30.95	30.34	31.14	30.91	30.99	31.20	30.90	30.73	31.06	30.78	31.30	31.27	CN1 constant 100%
CN3	29.23	28.76	28.83	28.99	28.96	28.66	28.78	28.68	29.22	28.77	29.53	29.36	29.39	29.53	29.35	29.25	29.49	29.26	29.62	29.59	CN3 constant 58.5%
CN5	28.39	28.00	28.05	28.16	28.13	27.94	28.03	27.90	28.41	28.04	28.78	28.63	28.64	28.75	28.63	28.56	28.76	28.56	28.84	28.81	CN5 constant 32.2%
CN6	28.09	27.73	27.77	27.87	27.84	27.68	27.77	27.62	28.12	27.78	28.51	28.38	28.38	28.48	28.37	28.32	28.50	28.30	28.56	28.53	CN6 constant 58.6%
32 31 30 29 28 27 26																					○ FAM (Total CN) ○ JOE ([T] CN) - CN1 (Calculated) - CN2 (Cy5) - CN3 (Calculated) - CN4 (610) - CN5 (Calculated) - CN6 (Calculated)
[T] Copy Number	1	1	2	1	2	1	2	1	2	1	2A7 Copy Number										
Total Copy Number	2	2	2	2	2	1	2	2	3	1									1	1	ND : Not Determined
Diplotype	Tx1/ Gx1	Tx1/ Gx1	Tx2	Tx1/ Gx1	Tx2	Tx1	Tx2	Tx1/ Gx1	Tx2/ Gx1	Tx1	2	2	2	2	2	2	2	2	1	1	

CYP2A6 / 2A7	Ct (CYP2A6 -48T>G) Average of 3-repeats										Ct (CYP2A7) Average of 3-repeats											
Sample Number	68	69	70	71	73	75	76	77	78	79	68	69	70	71	73	75	76	77	78	79	Genomic DNA	
IC1: CN2 (Cy5)	29.15	29.36	29.52	29.16	29.22	29.38	29.69	29.51	29.54	29.58	29.77	29.82	29.91	29.58	29.76	29.96	30.03	30.01	30.08	30.05	Threshold : 400	
IC2 : CN4 (610)	28.14	28.31	28.43	28.13	28.17	28.29	28.58	28.43	28.47	28.48	28.77	28.80	28.92	28.60	28.77	28.91	29.04	28.99	29.08	29.06		
CYP2A6/2A7 (FAM)	29.24	29.36	29.57	28.54	29.28	28.38	29.69	29.40	31.21	30.43	29.82	29.80	29.85	30.50	29.75	33.84	30.05	30.03	30.03	30.07		
[T] (JOE)	30.28	29.38	30.58	28.60	30.41	28.51	30.75	29.45	37.60	30.39	-	-	-	-	-	-	-	-	-	-		
CN1	30.16	30.41	30.61	30.18	30.28	30.47	30.79	30.59	30.60	30.67	30.76	30.83	30.90	30.57	30.76	31.01	31.03	31.03	31.07	31.04	CN1 constant 100%	
CN3	28.56	28.74	28.88	28.56	28.61	28.75	29.04	28.88	28.91	28.94	29.18	29.22	29.33	29.01	29.18	29.34	29.45	29.42	29.49	29.47	CN3 constant 58.5%	
CN5	27.82	27.97	28.08	27.80	27.83	27.94	28.23	28.08	28.13	28.13	28.45	28.48	28.60	28.28	28.45	28.57	28.72	28.67	28.76	28.74	CN5 constant 32.2%	
CN6	27.55	27.69	27.79	27.53	27.55	27.66	27.94	27.79	27.85	27.84	28.19	28.21	28.34	28.02	28.18	28.29	28.45	28.40	28.50	28.48	CN6 constant 58.6%	
32 31 30 29 28 27 26 Ct																						○ FAM (Total CN) ○ JOE ([T] CN) - CN1 (Calculated) - CN2 (Cy5) - CN3 (Calculated) - CN4 (610) - CN5 (Calculated) - CN6 (Calculated)
[T] Copy Number	1	2	1	3	1	4	1	2	0	1	2A7 Copy Number											
Total Copy Number	2	2	2	3	2	4	2	2	1	1											ND : Not Determined	
Diplotype	Tx1/ Gx1	Tx2	Tx1/ Gx1	Tx3	Tx1/ Gx1	Tx4	Tx1/ Gx1	Tx2	Gx1	Tx1	2	2	2	1	2	ND	2	2	2	2		

CYP2A6 / 2A7	Ct (CYP2A6 -48T>G) Average of 3-repeats										Ct (CYP2A7) Average of 3-repeats										
Sample Number	80	81	83	85	86	87	88	89	90	91	80	81	83	85	86	87	88	89	90	91	Genomic DNA
IC1: CN2 (Cy5)	29.67	29.43	29.23	29.41	29.44	29.09	29.16	29.34	29.48	29.30	29.88	29.92	29.90	30.09	29.84	29.74	29.89	29.95	30.08	30.30	Threshold : 400
IC2 : CN4 (610)	28.56	28.41	28.21	28.34	28.38	28.14	28.14	28.29	28.42	28.31	28.88	28.92	28.90	29.10	28.85	28.72	28.88	28.94	29.07	29.34	
CYP2A6/2A7 (FAM)	30.67	29.42	29.24	29.41	30.48	29.07	29.17	29.31	29.43	30.34	29.83	29.87	29.80	30.07	29.74	30.51	29.79	29.86	30.02	30.21	
[T] (JOE)	30.52	30.39	29.24	29.41	30.41	29.07	30.16	29.33	29.43	38.60	-	-	-	-	-	-	-	-	-	-	
CN1	30.79	30.45	30.25	30.48	30.49	30.05	30.17	30.40	30.55	30.29	30.88	30.92	30.89	31.08	30.82	30.75	30.90	30.96	31.09	31.26	CN1 constant 100%
CN3	29.02	28.84	28.64	28.79	28.82	28.53	28.56	28.73	28.86	28.72	29.29	29.34	29.32	29.51	29.26	29.14	29.30	29.36	29.49	29.74	CN3 constant 58.5%
CN5	28.20	28.09	27.88	28.00	28.04	27.83	27.82	27.95	28.08	27.99	28.55	28.60	28.58	28.79	28.54	28.40	28.56	28.62	28.74	29.03	CN5 constant 32.2%
CN6	27.91	27.82	27.62	27.72	27.77	27.58	27.55	27.67	27.80	27.73	28.29	28.34	28.32	28.53	28.28	28.13	28.29	28.35	28.48	28.78	CN6 constant 58.6%
32 31 30 29 28 27 26																					○ FAM (Total CN) ○ JOE ([T] CN) - CN1 (Calculated) - CN2 (Cy5) - CN3 (Calculated) - CN4 (610) - CN5 (Calculated) - CN6 (Calculated)
[T] Copy Number	1	1	2	2	1	2	1	2	2	0	2A7 Copy Number										
Total Copy Number	1	2	2	2	1	2	2	2	2	1											ND : Not Determined
Diplotype	Tx1	Tx1/ Gx1	Tx2	Tx2	Tx1	Tx2	Tx1/ Gx1	Tx2	Tx2	Gx1	2	2	2	2	2	1	2	2	2	2	

CYP2A6 / 2A7	Ct (CYP2A6 -48T>G) Average of 3-repeats										Ct (CYP2A7) Average of 3-repeats										
Sample Number	92	93	94	96	97	98	99	100	101	102	92	93	94	96	97	98	99	100	101	102	Genomic DNA
IC1: CN2 (Cy5)	29.28	29.39	29.18	29.28	29.24	29.53	29.50	29.20	29.47	29.52	29.86	29.96	29.59	29.84	29.71	30.04	29.90	29.73	30.18	30.10	Threshold : 400
IC2 : CN4 (610)	28.27	28.34	28.16	28.27	28.21	28.50	28.40	28.14	28.39	28.43	28.92	29.01	28.65	28.86	28.77	29.06	28.95	28.72	29.21	29.12	
CYP2A6/2A7 (FAM)	30.30	28.80	29.33	28.69	29.28	29.49	29.55	29.20	29.47	28.81	29.88	30.02	29.66	30.64	29.80	30.09	29.91	29.75	30.19	30.94	
[T] (JOE)	38.03	28.87	29.33	28.79	30.33	29.58	30.63	29.27	30.57	29.39	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	
CN1	30.29	30.44	30.21	30.29	30.27	30.56	30.59	30.25	30.55	30.61	30.80	30.91	30.53	30.83	30.65	31.01	30.85	30.74	31.16	31.08	CN1 constant 100%
CN3	28.69	28.78	28.58	28.69	28.64	28.92	28.86	28.58	28.83	28.88	29.31	29.40	29.04	29.27	29.16	29.47	29.35	29.14	29.61	29.52	CN3 constant 58.5%
CN5	27.95	28.01	27.83	27.94	27.88	28.17	28.05	27.80	28.04	28.08	28.61	28.70	28.35	28.54	28.47	28.75	28.65	28.39	28.90	28.80	CN5 constant 32.2%
CN6	27.68	27.73	27.56	27.68	27.61	27.89	27.76	27.52	27.75	27.79	28.37	28.45	28.10	28.28	28.23	28.49	28.40	28.13	28.64	28.54	CN6 constant 58.6%
32 31 30 29 28 27 26																					○ FAM (Total CN) ○ JOE ([T] CN) - CN1 (Calculated) - CN2 (Cy5) - CN3 (Calculated) - CN4 (610) - CN5 (Calculated) - CN6 (Calculated)
[T] Copy Number	0	3	2	3	1	2	1	2	1	2	2A7 Copy Number										
Total Copy Number	1	3	2	3	2	2	2	2	2	3											
Diplotype	Gx1	Tx3	Tx2	Tx3	Tx1/ Gx1	Tx2	Tx1/ Gx1	Tx2	Tx1/ Gx1	Tx2/ Gx1	2	2	2	1	2	2	2	2	2	1	ND : Not Determined

CYP2A6 / 2A7	Ct (CYP2A6 -48T>G) Average of 3-repeats										Ct (CYP2A7) Average of 3-repeats																																																																																				
Sample Number	103	107	108	109	110	111	112	113	115	116	103	107	108	109	110	111	112	113	115	116	Genomic DNA																																																																										
IC1: CN2 (Cy5)	29.58	29.27	29.52	29.26	29.52	29.34	29.54	29.36	29.44	29.48	29.69	29.79	29.89	29.90	29.72	29.65	29.89	30.01	29.83	30.13	Threshold : 400																																																																										
IC2 : CN4 (610)	28.44	28.24	28.44	28.22	28.46	28.33	28.45	28.36	28.40	28.40	28.89	28.93	28.99	29.00	28.93	28.81	29.01	29.08	28.98	29.21																																																																											
CYP2A6/2A7 (FAM)	29.62	29.26	29.39	29.31	29.96	30.26	29.63	30.47	0.00	29.49	29.79	29.93	30.79	29.89	29.84	29.78	29.94	30.47	29.86	30.13																																																																											
[T] (JOE)	29.60	29.27	29.35	30.40	37.63	30.26	30.66	30.41	42.17	29.48	-	-	-	-	-	-	-	-	-	-																																																																											
CN1	30.71	30.30	30.60	30.30	30.58	30.35	30.63	30.36	30.49	30.55	30.50	30.65	30.79	30.79	30.52	30.48	30.77	30.93	30.68	31.05	CN1 constant 100%																																																																										
CN3	28.91	28.67	28.89	28.65	28.90	28.75	28.90	28.78	28.83	28.85	29.22	29.29	29.36	29.37	29.26	29.16	29.37	29.46	29.33	29.59	CN3 constant 58.5%																																																																										
CN5	28.08	27.91	28.09	27.89	28.11	28.01	28.10	28.04	28.06	28.06	28.63	28.66	28.70	28.71	28.67	28.55	28.72	28.78	28.70	28.92	CN5 constant 32.2%																																																																										
CN6	27.78	27.64	27.80	27.61	27.83	27.74	27.81	27.78	27.79	27.78	28.42	28.43	28.46	28.48	28.47	28.33	28.49	28.54	28.48	28.68	CN6 constant 58.6%																																																																										
Ct 32 31 30 29 28 27 26																																																																																															