

Supplementary Figure 5: Variant calling and phenotype prediction of human HCCOs

	Sample		Position in GRCh37	dbSNP	Amino Acid Change	Variant Allele	Function	Total Read Depth	Read Depth (specific position)	Variant Allele Frequency (FreeBayes)	Variant Allele Frequency (BCFtools)	Detected by BCFtools	Likely Allele	Likely Diploptype	Likely Phenotype		
1	SN3	HCCO-C798	42524947C>T	rs3892097	Splicing Defect	CYP2D6*4	Loss of Function	2588 (2583)	84	68%	69%	yes	Allele 1 (*4)	*4/*1	Intermediate metabolizer		
			42526694G>A	rs1065852	P34S	present in several alleles and suballeles (including *4)	Loss of Function		42	79%	77%	yes					
			42522613G>C	rs1135840	S486T	present in several alleles and suballeles (including *4)	Normal		77	35%	34%	yes					
			42523528C>T	rs1058172	R365H	present in several alleles and suballeles (including *4)	Normal		150	35%	27%	yes					
2	SN5	HCCO-C948	42524947C>T	rs3892097	Splicing Defect	CYP2D6*4	Loss of Function	3987 (3978)	122	39%	37%	yes	Allele 1 (*4)	*4/*20 or *150	20 gives no function, 150 gives normal function => (intermediate or poor metabolizer)		
			42522613G>C	rs1135840	S486T	present in several alleles and suballeles (including *4)	Normal		103	51%	51%	yes					
			42526694G>A	rs1065852	P34S	present in several alleles and suballeles (including *44)	Loss of Function		73	26%	30%	yes					
			42523528C>T	rs1058172	R365H	present in several alleles and suballeles (including *4)	Normal		230	20%	20%	yes					
			42525811T>C	rs28371704	H94R	present in several alleles and suballeles (including *4)	Normal		60	43%		no	Allele 2 (*20 or *150)				
			42524814A>G	rs199535154	L213P	Present in several alleles	Normal		60		22%	yes, but not by FreeBayes					
			42524815G>A	rs150163869		Present in several alleles and suballeles	Normal		60		20%	yes, but not by FreeBayes					
3	SN7	HCCO-C949	42524947C>T	rs3892097	Splicing Defect	CYP2D6*4	Loss of Function	3754 (3746)	109	44%	45%	yes	Allele 1 (*4)	*4/*1	Intermediate metabolizer		
			42522613G>C	rs1135840	S486T	present in several alleles and suballeles (including *4)	Normal		71	54%	55%	yes					
			42526694G>A	rs1065852	P34S	present in several alleles and suballeles (including *4)	Loss of Function		55	62%	60%	yes					
			42525811T>C	rs28371704	H94R	present in several alleles and suballeles (including *4)	Normal		71	32%	32%	no					
			42523528C>T	rs1058172	R365H	present in several alleles and suballeles (including *4)	Normal		209	23%	20%	yes					
			42525821G>T	rs28371703	L91M	present in several alleles and suballeles (including *4)	Normal		63	33%	32%	yes					
4	SN9	HCCO-C975						1561 (1553)							Normal metabolizer		
5	SN10	HCCO-D045						2596 (2591)							Normal metabolizer		
6	SN11	HCCO-D045						989 (986)							Normal metabolizer		
7	SN15	HCCO-D324						5481 (5462)							Normal metabolizer		
8	SN16	HCCO-D359	42524947C>T	rs3892097	Splicing Defect	CYP2D6*4	Loss of Function	18271 (18171)	305	21%	21%	yes	Allele 1 (*4)	*4/*1	Intermediate metabolizer		
			42526694G>A	rs1065852	P34S	present in several alleles and suballeles (including *4)	Loss of Function		807	31%	32%	yes					
			42523528C>T	rs1058172	R365H	present in several alleles and suballeles (including *4)	Normal		1113	22%		no					
			42523943A>G	rs16947	R296C	present in several alleles and suballeles (including *4)	Normal		853	19%		no					
9	SN17	HCCO-D386	42523943A>G	rs16947	R296C	present in several alleles (including *33) and suballeles	Normal	10737 (10672)	462	99%		no			Normal metabolizer		
10	SN18	HCCO-D415	42522613G>C	rs1135840	S486T	present in several alleles and suballeles (including *4)	Normal	10790 (10734)	214	37%	37%	yes	Allele 1(*1)	*1/*1	Normal metabolizer		
			42523943A>G	rs16947	R296C	present in several alleles and suballeles (including *4)	Normal		496	48%	46%	yes					
11	SN19	HCCO-D455	42523943A>G	rs16947	R296C	present in several alleles and suballeles (including *4)	Normal	9610 (9548)	443	100%		no			Normal metabolizer		