

**CBCS Phase 1 & Phase 2 (Invasive & CIS)
List of SNPs using Illumina or Taqman Assay**

Table of Contents

1. Ancestry Informative Markers (AIMS).....	2
2. DNA Repair	
a) Nucleotide Excision Repair and Transcription-coupled Repair.....	3
b) Double-Strand Break Repair -- Homologous Recombination Repair.....	5
c) Base Excision Repair.....	8
d) Direct Repair.....	10
e) Double-Strand Break Repair -- Non-Homologous End Joining	11
f) Bypass Polymerases	12
3. Damage Recognition, Cell Cycle Control, Cellular Proliferation and Cellular Adhesion.....	13
4. Hormone Metabolism	22
5. Oxidative Metabolism	30
6. Additional Genotypes from NCI Collaboration	32
7. Drug Metabolizing Enzymes (DMEs)	33
8. Hypothalamic-Pituitary-Adrenal (HPA) Axis	34
9. Hypothalamic-Pituitary-Ovarian (HPO) Axis	35
10. Inflammation	36
11. miRNA associated Genes	38
12. Obesity/Insulin Resistance Pathway Genes	39
13. Additional known candidate genes	44
14. Chuck Perou's candidates, including subtype specific genes	46
15. Replication of GWAS results	47
16. Additional genes in the FGFR2 and other pathways identified by GWAS	51
17. Additional SNPs	52

1. Ancestry Informative Markers (AIMS)

144 SNPs, all were done using the Illumina assay (N=3748).

rs12094678	rs2687427	rs9297712	rs2246695
rs11264110	rs9306906	rs7021690	rs710052
rs7161	rs4619931	rs10124991	rs12900552
rs6666101	rs12640848	rs1415723	rs1470608
rs7512316	rs7689609	rs3861709	rs12900262
rs4659762	rs6535244	rs10962612	rs7086
rs12129648	rs385194	rs1885167	rs4923940
rs798443	rs1372894	rs1412521	rs12594483
rs12612040	rs2100438	rs870272	rs567357
rs1508061	rs316598	rs2488465	rs6494466
rs3755446	rs13169284	rs1335826	rs9806307
rs10195705	rs16891982	rs9416972	rs4506877
rs1257010	rs10056388	rs1733731	rs4350528
rs4149436	rs13173738	rs2184033	rs9923864
rs17049450	rs10041728	rs4529792	rs7187359
rs17261772	rs33957	rs9416026	rs12926237
rs1117382	rs1917028	rs1911999	rs11150219
rs1372115	rs1380014	rs1125217	rs7189172
rs12692701	rs13178470	rs7107482	rs1862819
rs1982235	rs6556352	rs11607932	rs4792105
rs7424137	rs2451563	rs11223503	rs12945601
rs12997060	rs10806263	rs2416791	rs1043809
rs10202705	rs6937164	rs1490728	rs2593595
rs3791896	rs4896780	rs10842753	rs4793237
rs11901793	rs10952147	rs7134682	rs228768
rs155409	rs7810554	rs328744	rs11652805
rs1303629	rs7788641	rs3759171	rs4789070
rs13318432	rs17520733	rs2596793	rs897351
rs2660769	rs10254729	rs645510	rs8113143
rs1462309	rs10255169	rs9525462	rs1991818
rs6414248	rs344454	rs9543532	rs1011643
rs13080353	rs4602918	rs4885162	rs2426515
rs6765491	rs4143633	rs9530646	rs6023376
rs9849733	rs1870571	rs6491743	rs4811651
rs833282	rs12676654	rs1477921	rs2075902
rs4859147	rs13261248	rs222674	rs4823460

2. DNA Repair

a. Nucleotide Excision Repair and Transcription-coupled Repair

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>XPD</i> (<i>ERCC2</i>) TAGS	Failed		rs1799793 rs13181
<i>RAD23B</i> (<i>RD23B, hHR23B</i>)	rs1805330		rs1805329
<i>XPG</i> (<i>ERCC5</i>)	rs2227869 rs2296148 rs4150313 rs2020915 rs4987063 rs2296147 rs4150360		rs17655
<i>XPC</i>	rs3731062 rs2228000 rs3731055 rs2279017 rs35629274 rs3731124		rs2228001
<i>XPF</i> (<i>ERCC4</i>)	rs1799802 rs744154 rs3136091 rs254942 rs4986933		rs1800067 rs2020955 (mostly AA)
<i>ERCC6</i> (<i>CKN2, CSB, RAD26</i>)	rs4253072 rs2228528 rs4253132 rs2229761 rs4253230		rs2228527 rs4253211
<i>ERCC1</i> TAGS	rs3212930 rs3212935		rs3212986

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>POLE</i>	rs5744934 rs5745068 rs5744991 rs4883543 rs5744776 rs5744760		
<i>POLD2</i>	rs3087370		
<i>POLH</i>	rs35675573 rs9333555 rs6899628		
<i>RAD23A</i>	rs2974752 rs4987202		
<i>RFC3</i>	rs3135525 rs3135530 rs3135638 rs3135640 rs3135635		
<i>XPA</i>	rs1800975 rs3176750 rs3176753		
<i>Ligase 1 (LIG1)</i>	rs3730933 rs20579 rs3731003 rs3730980 rs4987070		
<i>ERCC8 (CKN1, CSA)</i>	rs3117 rs4647153		
<i>BRCA1 (see DSB on page 5)</i>			

b. Double-Strand Break Repair -- Homologous Recombination Repair

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>XRCC3</i> TAGS	rs1799794 rs1799796		rs861539
<i>NBS1</i> (<i>NBN</i>)	rs2308962 rs769420		rs1805794
<i>XRCC2</i>	rs3218385		rs3218536
<i>BRCA1</i>	rs4986850 rs1799950 rs16941 rs16942 rs1799966 rs799917 rs3737559 rs799923	rs4986852	
<i>BRCA2</i>	rs144848 rs4987117 rs766173 rs1801406 rs1799955 rs15869		rs144848 (repeated using Illumina assay) rs206340
<i>ZNF350</i> (<i>ZBRK - BRCA1 interacting protein</i>) TAGS	rs2278420 rs4986771 rs2278415 rs11879758 rs2278417 rs4986770 rs4988334 rs8102072	rs4986773 rs3764538	
<i>BRIP1</i> (<i>BACH1 - BRCA1 interacting protein</i>) TAGS	rs4986764 rs7213430 rs4988350 rs4988346	rs4988351 rs2048718 rs1978111	

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>BARD1</i> TAGS	rs28997576 rs2229571 rs10932568 rs10221582 rs10932573 rs12474696 rs12477063 rs1542173 rs16852761 rs16852798 rs16852799 rs17487827 rs1979028 rs2075622 rs2888294 rs3768704 rs3768707 rs3768708 rs3820727 rs4672729 rs6706777 rs6712055 rs6749828 rs6751923 rs6753417 rs6756902 rs7557557 rs7566806 rs7585356	rs1048108 rs3738888	
<i>PALB2</i> (<i>BRCA2</i> partner associated with Fanconi anemia)	rs3809683 rs249954		
<i>RAD51</i> TAGS	rs1801320 rs7174493		
<i>RPA1</i>	rs9914073		
<i>RAD52</i>	rs4987206		

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>RAD54L</i>	rs10437093 rs10437094 rs4506412		
<i>MRE11A</i>	rs1805360 rs1805362		
<i>WRN</i>	rs2230009 rs1800391 rs2725362 rs1346044		

c. Base Excision Repair

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>XRCC1</i> TAGS	rs25496 rs2682558		rs1799782 rs25489 rs25487
<i>APE1</i> (<i>APEX1</i>)	rs1048945		rs3136820
<i>OGG1</i> (<i>HOGG1</i>)	rs1805373		rs1052133
<i>MYH</i> (<i>MUTYH</i>)	rs3219497 rs3219484		rs3219489
<i>MBD4</i>	rs140696 rs2307289		
<i>MPG</i>	rs710080		
<i>NTHL1</i>	rs3087468		
<i>TDG</i>	rs4135113		
<i>UNG</i> (<i>UNG1</i>)	rs3219275		
<i>POLB</i>	rs3136797		
<i>Ligase 3</i> (<i>LIG3</i>)	rs3136025 rs4796030		
<i>NEIL1</i>	rs5745926		
<i>NEIL2</i>	rs8191613 rs8191664 rs1534862		
<i>SMUG1</i>	rs3136391 rs3087404		

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>POLE2</i>	rs34857719		
<i>PCNA</i>	rs25406 rs17352 rs17349		
<i>RFC1</i>	rs17288820 rs2066791 rs17287851		
<i>FEN1</i>	rs412334		
<i>PARP1</i> (<i>ADPRT1</i>)	rs1136410 rs1805409		
<i>PARP3</i> (<i>ADPRTL2</i>)	rs34224216 rs323870		

d. Direct Repair

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>MGMT</i>	rs2308327 rs2020893		rs12917

e. Double-Strand Break Repair -- Non-Homologous End Joining

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>XRCC4</i>	rs1805377 rs28360136 rs28383138 rs3734091 rs2075685		rs2075685
<i>DNA-PKcs (PRKDC)</i>	rs1231202 rs10109984 rs4278157 rs7830743		
<i>Ku70</i>	rs132793 rs132770 rs7284074		
<i>Ku80</i>	rs3835 rs3834 rs1051677 rs1051685 rs2440		
<i>Ligase 4 (LIG4)</i>	rs2232642 rs1805386 rs1805388		
<i>PNKP (polynucleotide kinase) TAGS</i>	rs3739186 rs2257103		

f. Bypass Polymerases

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>POLG</i>	rs3087374 rs2307450		
<i>POLI</i>	rs3730823 rs3218786 rs8305		
<i>POLL</i>	rs3730477 rs3730475 rs3730463		
<i>POLM</i>	rs28382653 rs28382644 rs28382635		
<i>POLQ</i>	rs487848 rs3218651 rs532411 rs1381057 rs3218634 rs3218637 rs3218649 rs702017		
<i>REV1L</i>	rs3087403 rs3087386 rs3087399		
<i>REV3L</i>	rs458017 rs17539651		

3. Damage Recognition, Cell Cycle Control, Cellular Proliferation and Cellular Adhesion

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>TP53</i> <i>See section 6</i>			
<i>PRKAG2</i> <i>(TP53 responsive genes)</i>	rs2241053 rs41317142 rs8961	rs1860746	
<i>NSMCE2</i> <i>(TP53 responsive genes)</i>	rs10091027 rs10093674 rs10101690 rs10108110 rs10110845 rs10112218 rs10112737 rs10283134 rs10956238 rs11779422 rs11994861 rs11996045 rs12679602 rs12681602 rs13260832 rs16900402 rs16900403 rs16900479 rs16900487 rs17403327 rs17403341 rs2293890 rs2384890 rs3955404 rs4242367 rs4350000 rs4870935 rs4870938 rs4871570 rs4871584 rs6651237 rs6984563 (Continued on next page)	rs13249982	

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>NSMCE2</i> (continued)	rs6988871 rs6989407 rs6992545 rs6995096 rs6997862 rs7002315 rs7003102 rs7004396 rs7008046 rs7008482 rs7010385 rs7010992 rs7015414 rs7357375 rs7815471 rs7824635 rs7825799 rs7831918 rs7839468 rs9297712 rs9987342		
<i>TP73</i> TAGS	rs1885867 rs12731705 rs17379833 rs2181484 rs3765695 rs3765701 rs3765703 rs3765731		
<i>MDM2</i>	rs1144945 rs1695144 rs1695147 rs1846402 rs3730536 rs3730646 rs1625525 rs2291857 rs3730504 rs3730529 rs3730591 rs3730648 rs769412		rs2279744

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>RB1</i>	rs2854344 rs198580		
<i>AURKA</i> (<i>STK15</i> , <i>STK 6</i>)	rs1047972 rs34987347 rs1468056 rs16979826 rs16979829 rs16979865 rs2064863 rs2180691 rs2236207 rs2273535 rs2298016 rs6014711 rs6014712 rs6024840 rs6092309 rs6099120 rs6099122 rs6099126 rs6099127 rs6099128 rs1468055 rs6024836 rs911162	rs33923703 rs6099119 rs911162	
<i>MYBL2</i>	rs619289 rs285205 rs387769 rs6017147 rs419842 rs285164 rs442143 rs7347231		rs2070235

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>ATM</i>	rs3092992 rs664143 rs170548 rs3092993 rs1800054 rs4986761 rs1800056 rs1800057 rs1800058 rs1801516		
<i>CHEK1</i>	rs506504 rs521102		
<i>CHEK2</i>	rs1033667 rs17507066 rs2017309 rs2073327 rs2236142 rs5762746 rs9608698 rs5752777 rs5762760 rs5762764 rs6005843 rs6519761 rs9620817		
<i>HUS1</i>	rs2037483 rs2307254		
<i>RAD1</i> (<i>REC1</i>)	rs1805327		
<i>CCND1</i> (<i>PRAD-1</i>)	rs603965 rs3918298 rs649392 rs678653 rs3212860 rs7178		
<i>CDK7</i>	rs2972388 rs34584424		

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>CCNH</i> (<i>Cyclin H</i>)	rs2266690 rs2266691 rs2266692		
<i>CDKN2A</i>	rs3731249 rs3731257 rs10811661 rs518394 rs564398	rs1011970 rs10757278	
<i>XRCC9</i> (<i>FANCG</i>)	rs587118 rs4986940 rs2237857		
<i>CDH1</i> (<i>E-cadherin</i>)	rs10431923 rs10500545 rs11640099 rs11863122 rs12185157 rs12599393 rs12919719 rs12930371 rs16260 rs16958356 rs1862748 rs2010724 rs2276329 rs2902185 rs4076177 rs4783676 rs6499200 rs7188750 rs7203904 rs8049967 rs8057342 rs8059139 rs9941051 rs1125557 rs12597188 rs8061932 rs1078621 rs12931189 rs17690554 rs17715799 rs2276330		

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>APC</i> (Adenomatous polyposis coli)	rs459552 rs2439591 rs11948681 rs11953943 rs17164132 rs1734242 rs1734245 rs2020383 rs2545169 rs2707761 rs41115 rs464708 rs7731851 rs9686237 rs2439595 rs414098 rs11954856 rs2909962		
<i>Beta-catenin</i> <i>CTNNB1</i>	rs2953 rs4135385 rs1880481 rs11564473 rs4016435 rs11564450 rs1722845 rs6792748 rs1798802		
<i>EGF</i> (Epidermal growth factor)	rs4444903 rs11568911 rs11568943 rs11568992 rs2237051 rs11569017 rs11569046 rs4698803 rs11568846		

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>EGFR</i> (HER1; <i>Epidermal growth factor receptor</i>)	rs1015793 rs10225877 rs10228436 rs10229932 rs10244987 rs10245472 rs10251695 rs10269512 rs10488140 rs10488141 rs11487218 rs11976696 rs11977388 rs11977660 rs12531163 rs12535226 rs12538489 rs12669749 rs12718939 rs13244925 rs1404908 rs1554718 rs1558544 rs17151957 rs17172428 rs17172432 rs17172451 rs2075109 rs2075112 rs2241055 rs2293348 rs2472520 rs3778866 rs3823585 rs4140770 rs4947974 rs4947979 rs6593205 rs6593206 rs6944906 rs6957176 (Continued on next page)		

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>EGFR</i> (continued)	rs6957999 rs6958497 rs6964647 rs6964705 rs6970262 rs712831 rs723527 rs729969 rs759158 rs759160 rs759162 rs759167 rs759170 rs763317 rs7781264 rs7784148 rs7785854 rs7803520 rs7803660 rs845550 rs845552 rs845555 rs845559 rs917880 rs9649847 rs9692301 rs10244108 rs11770506 rs11971051 rs12535578 rs12666347 rs1344307 rs17172446 rs17586344 rs2293347 rs2330951 rs2740762 rs4947490 rs4947972 rs4947986 rs6593202 rs6947594 (Continued on next page)		

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>EGFR</i> (continued)	rs6954351 rs7780270 rs7796139 rs7809394 rs845554 rs845562 rs980653 rs11536635 rs13222385 rs884904 rs6593210 rs1861003		
<i>AIB1</i> (interacts with <i>EGFR</i>)	rs6094752 rs2230782 rs2230781 rs12273		
<i>HER2</i>	rs1801200 rs2952156 rs2517955		Codon 655 Ile/Val
<i>MAPK8</i> TAGS	rs11598320		

4. Hormone metabolism

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>AR</i> (<i>Androgen receptor</i>)	rs1337076 rs6154 rs2362520 rs962458 rs1204038 rs2361634 rs1337082		
<i>CYP19A1</i> (<i>aka CYP19</i>)	rs700518 rs28757081 rs2446405 rs2445765 rs2470144 rs2445762 rs1004984 rs1902584 rs3751591 rs28566535 rs2445759 rs936306 rs1902586 rs749292 rs6493494 rs1008805 rs727479 rs2414096 rs28757184 rs700519 rs17601241 rs10046 rs4646 rs2236722		
<i>CYP17</i> (<i>CYP17A1</i>)	rs17115144 rs2486758 rs1004467 rs10786712 rs17115104 rs3740397 rs3781287		

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>17B-HSD</i> (<i>HSD17B1</i>)	rs676387 rs598126 rs2830 rs597255		
<i>HSD17B2</i>	rs4445895 rs8191136 rs11648233 rs1364285 rs1364286 rs1424151 rs16956274 rs16956326 rs2911418 rs2911420 rs2955153 rs2955159 rs2955162 rs2955163 rs2966244 rs2966245 rs2966246 rs2966248 rs2966250 rs3111351 rs4243229 rs6564962 rs7196087 rs7196807 rs7200696 rs723013 rs8045494 rs8049423 rs8050327 rs8052451 rs8059915 rs8191072 rs8191102 rs8191232 rs9319572 rs9889094 rs1364287 rs2042429 rs6564964 rs8191167		

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>HSD3B1</i>	rs6428830 rs10754400 rs6203 rs6671149 rs6205 rs932603 rs3765945		
<i>CYP1A1</i> (<i>AHH</i> , <i>aryl hydrocarbon</i> <i>Hydrolase</i>)	rs4986882 rs1799814 rs4986883		M1, M2, M3, M4 Done on part of CBCS Phase 1
<i>CYP1A2</i>	rs762551 rs28399424 rs17861157		
<i>CYP1B1</i> TAGS	rs1056836 rs1800440		rs1056836 (repeated using Illumina assay)
<i>CYP3A4</i>	rs2740574 rs4986910 rs4986909 rs12721636 rs28988603 rs12721626 rs10267228 rs12333983 rs17161886 rs2242480 rs2687117 rs4646437 rs4646440 rs7801671		rs2740574 (repeated using Illumina assay)

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>CYP3A5</i>	rs28365083 rs776746 rs15524 rs10224569 rs10256106 rs10259288 rs4646456 rs4646458 rs6976017 rs6977165 rs4646450		
<i>CYP11B2</i> TAGS (also listed in Section 8)	rs4545 rs4537 rs3802230 rs6440		
<i>CYP11A</i> (<i>CYP11A1</i>)	rs3825944 rs3803463 rs1484215 rs7173655 rs2277604 rs2277602 rs1843091		
<i>COMT</i>	rs5031015 rs9332380		rs4680 (Phase I only)
<i>SULT1A2</i> TAGS	rs1059491		
<i>AHR</i> (aryl hydrocarbon receptor)	rs2066853		
<i>ER alpha</i> <i>ESR1</i>	rs3020314 rs851984 rs2881766 rs2077647 rs1801132 rs6905370 rs3798577 rs9341070 (Continued on next page)	rs543650	

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>ER alpha</i> <i>ESR1</i> (continued)	rs851982 rs2234693 rs12212176 rs1514348 rs1884053 rs2813543 rs2982712 rs3003925 rs3020383 rs3020404 rs3020407 rs3020410 rs4583998 rs6557170 rs6914211 rs7761133 rs9322336 rs11155813 rs11155818 rs11964865 rs12154178 rs13192976 rs17081703 rs17081740 rs17082028 rs1709183 rs1884049 rs2144025 rs2207232 rs2207396 rs2982684 rs3003917 rs3020318 rs3020364 rs3020371 rs3020375 rs3020381 rs3020401 rs3020434 rs3778082 rs3778099 rs3798569 rs3798577 (Continued on next page)		

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>ER alpha</i> <i>ESR1</i> (continued)	rs3798758 rs3866461 rs4870056 rs6557171 rs6557177 rs6901451 rs6903763 rs6911230 rs712221 rs7739274 rs7754762 rs7755185 rs7759411 rs7775047 rs827421 rs926777 rs9322332 rs9322335 rs9322337 rs9322338 rs9340817 rs9340835 rs9340888 rs9341008 rs9341062 rs9371236 rs9383599 rs9397459 rs9397462 rs9397463 rs9397472 rs985191 rs985695 rs9341052 rs3020403 rs3020368 rs9340944 rs532010 rs9383951 rs2459107 rs2474148 rs2982699 (Continued on next page)		

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>ER alpha</i> <i>ESR1</i> (continued)	rs9340971 rs12523805 rs12055837 rs11155833 rs13192678		
<i>Progesterone receptor</i> <i>PGR</i>	rs10895068 rs11224566 rs11224575 rs11224579 rs1824128 rs2124761 rs495997 rs501732 rs503602 rs538915 rs543936 rs546763 rs548668 rs555653 rs578029 rs596223 rs653752 rs660149 rs679275 rs693765 rs11224570 rs11224565 rs492827 rs11224590 rs11571247 rs11224591		
<i>SHBG</i>	rs1799941		

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>POR</i> <i>Cytochrome P450</i> <i>Oxidoreductase</i>	rs10262966 rs2228104 rs10281803 rs10954724 rs10954732 rs17685 rs239960 rs2868178 rs2868180 rs4732513 rs6949454 rs6964407 rs6964550 rs7796654 rs7804806 rs8509 rs917720		

University of Calgary (Alastair Cribb) collaboration: CYP2C9 and CYP2C19

<i>CYP2C9</i> TAGS	rs28371685 rs2256871 rs1934967 rs2475376 rs4086116 rs9332242 rs1934969 rs2253635 rs9332121 rs9332223 rs9332232		
<i>CYP2C19</i> TAGS	rs3758581 rs6413438 rs17878459 rs10509676 rs11597626 rs4388808 rs4917623 rs12247175 rs12767583 rs4451645 rs6583954 rs7088784		

5. Oxidative Metabolism

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>MnSOD (SOD2)</i> TAGS	rs4987023		rs4880
<i>PTGS2 (COX2)</i> (also listed in Section 10)	rs11567822 rs5271 rs2206593 rs5275 rs20424 rs20417 rs5277 rs4648298		rs5273 (AA only)
<i>PPARG</i> TAGS	Failed		rs1801282
<i>PPARD</i>	rs2016520		
<i>PPARA</i>	rs1800206		
<i>INOS</i>	rs2297518		
<i>ENOS</i>	rs1799983		
<i>GSTM1</i>	Failed		Null deletion Part of CBCS Phase 1
<i>GSTP1</i>	rs1138272 rs4986949 rs1695 rs4147581		rs1695 (Done on part of CBCS Phase; repeated using Illumina assay for CBCS 1 & 2)
<i>GSTT1</i>	Failed		Null deletion Part of CBCS Phase 1
<i>MPO</i>	rs7208693 rs2759 rs2071409 rs35433825		rs2333227

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>HFE</i>	Not done		rs1799945 rs1800562
<i>CYP2E1</i>	rs6413420 rs28969387 rs6413423 rs6413424 rs9919378		
<i>EPHX1</i>	rs2234697 rs2854461		
<i>NQO1</i> (also listed in Section 6)	rs10517 rs1800566 rs34755915 rs689453 rs689452		rs34906225 rs10517 rs1800566 rs34755915 rs689453 rs689452 Done for 1494 Phase 1 samples. Some of the SNPs are repeated using Illumina assay for CBCS 1 & 2.

6. Additional genotypes from NCI collaboration

Repeat those that were previously done on the N = 1494 Phase 1 CBCS samples
Chanock NCI (collaboration)

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Previously done on N=1494 Phase 1 CBCS samples
<i>GATA3</i> TAGS	rs1149901 rs3802604 rs570613 rs422628		rs1149901 rs3802604 rs3781092 rs570613 rs422628
<i>TP53</i>	rs9894946 rs1614984 rs4968187 rs12951053 rs17880604 rs1800372 rs2909430 rs8079544	rs1042522	rs17887200 rs9894946 rs1614984 rs4968187 rs12951053 rs12947788 rs17880604 rs1625895 rs35163653 rs1800372 rs2909430 rs9895829 rs1042522 rs1642785 rs8079544
<i>FOXA1</i>	rs2145146 rs1057536		rs33984772
<i>NQO1</i> (also listed in Section 5)	rs10517 rs1800566 rs34755915 rs689453 rs689452		rs34906225 rs10517 rs1800566 rs34755915 rs689453 rs689452
<i>XBP1</i>	rs2267131 rs2097461 rs35771921		rs2267131 rs2097461 rs35873774 rs35771921

7. Drug Metabolizing Enzymes

NOTE: CYP19, CYP3A4, CYP3A5, CYP1B1 and CYP17 are listed elsewhere (Section 4).

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)
<i>ABCG2 / BDRP</i>	rs2231137 rs2231142 rs1061018	
<i>ABCB1 / MDR1</i>	rs1128503 rs2235013 rs10276036 rs2235015 rs2235040	

8. Hypothalamic-Pituitary-Adrenal (HPA) Axis – Dorgan Fox Chase collaboration

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)
<i>NR3C1 / GR</i>	rs6190	
<i>CYP11B1</i>	rs6410 rs6387 rs4541 rs5292	
<i>CYP11B2</i> TAGS	rs4545 rs4537 rs3802230 rs6440	

9. Hypothalamic-Pituitary-Ovarian (HPO) Axis -- Dorgan Fox Chase Collaboration

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)
<i>GnRH1</i>	rs6185 rs17790824	
<i>GnRHR</i>	rs2627260 rs2062301	
<i>FSHB</i>	rs5030777 rs506306	
<i>FSHR</i>	rs6165 rs6166	
<i>LHCGR</i>	rs12470652 rs2293275	

10. Inflammation

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>IL12 / IL12B</i>	rs3212227		
<i>CRP</i>	rs3093058 rs1417938 rs1800947 rs1205 rs2808630		
<i>IL1A</i>	rs1800587 rs17561		
<i>IL1B</i>	rs1143634 rs1143630 rs1143643		
<i>TNFA</i>	rs1800630 rs1799964		
<i>IL1RN</i>	rs419598 rs315951 rs1794066 rs380092 rs3087266 rs3181052		
<i>IL5</i>	rs2069812		
<i>IL6</i> (also listed in Section 12)	rs1800795 rs1548216 rs1800796 rs2069824 rs2069835 rs2069840 rs2069842 rs2069843 rs2069845 rs2069827 rs2069832		

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>IL10</i>	rs1800896		
<i>IL16</i>	rs4778889		
<i>TGFB1</i>	rs2241715 rs4803455		rs1800470
<i>VEGF</i>	rs699947 rs1005230 rs2010963 rs25648 rs3025039		
<i>MMP2</i>	rs243849 rs1053605		
<i>MMP3</i>	rs639752 rs650108 rs679620		
<i>MMP9</i>	rs17576 rs3918261 rs3918260 rs3918249		
<i>MMP13</i>	rs2252070		
<i>RECK</i>	rs10814325		
<i>TIMP3</i>	rs9619311		
<i>PTGS2</i> (also listed in Section 5)	rs11567822 rs5271 rs2206593 rs5275 rs20424 rs20417 rs5277 rs4648298		rs5273 (AA only)

11. miRNA associated Genes

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)
<i>miR-9-1</i>	rs12239077 rs16837376	
<i>miR-9-2</i>	rs2304608 rs1501672	
<i>miR-16-1</i>	rs9535416	
<i>miR-34a</i>	rs12128240	
<i>miR-34c</i>	rs4938723	
<i>miR-125a</i>	rs12975333	
<i>miR-185</i>	rs2008591 rs887205 rs2078749	
<i>miR-206</i>	rs6920648 rs16882131	

12. Obesity/Insulin Resistance Pathway Genes

Includes Kadlubar collaboration (Univ Arkansas)

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)
<i>FTO</i>	rs9939609	
<i>IGF1</i> TAGS	rs7965399 rs35767 rs5742612 rs17884626 rs10735380 rs1019731 rs10745940 rs11831436 rs12821878 rs1520220 rs17727841 rs2195240 rs2373721 rs2946834 rs5742614 rs5742626 rs5742632 rs5742657 rs6214 rs7136446 rs7956547 rs9308315 rs972936	
<i>SHC1</i> (<i>IGF1</i> pathway) TAGS	rs3766921	
<i>IRS1</i>	rs1801278	
<i>IRS2</i>	rs11618950	

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)
<i>Adiponectin</i> <i>ADIPOQ</i>	rs12495941 rs1403697 rs1501299 rs16861194 rs16861205 rs16861210 rs6773957 rs822396 rs9842733 rs9877202 rs1063537 rs3821799 rs7649121 rs822391 rs2241766 rs6444174	
<i>ADRB3</i>	rs4994	
<i>IGFBP3</i>	rs3110697 rs2854747 rs2132570	
<i>Leptin</i> <i>(LEP)</i>	rs10244329 rs10954174 rs11760956 rs12706832 rs17151919 rs17151922 rs3793162 rs3828942 rs4236625 rs6976701 rs7795794 rs10954173 rs11761556 rs11763517	

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)
<i>Leptin receptor</i> (LEPR)	rs1137100 rs1137101 rs8179183 rs10157610 rs10158579 rs10749754 rs10889556 rs10889563 rs11208654 rs11208662 rs1171267 rs1171275 rs11801408 rs11808888 rs12042877 rs12405556 rs12564626 rs1327118 rs1409802 rs1475397 rs17097182 rs17127608 rs17127618 rs17127655 rs17127677 rs17127686 rs17127807 rs17127826 rs17127828 rs1751492 rs1782763 rs1887285 rs1938489 rs2025804 rs3790426 rs3790429 rs3790431 rs4655555 rs6413506 rs6588147 rs6693573 (Continued on next page)	

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)
<i>Leptin receptor</i> (<i>LEPR</i>) (continued)	rs6694528 rs6697315 rs6700896 rs7529650 rs7531110 rs7534511 rs7555955 rs9436299 rs9436301 rs9436738 rs9436740 rs9436746 rs9436748 rs970467 rs10493380 rs10889569 rs11585329 rs1171279 rs12145690 rs17412175 rs3762274 rs3790433 rs3806318 rs3828034 rs6657868 rs6704167 rs7518849 rs1627238 rs1343982 rs4655537 rs7526141 rs6678033 rs9436297	
<i>SULT1C2</i>	rs1402467 rs17036092 rs17036104 rs2305484	
<i>CYP39A1</i> TAGS	rs2277119 rs7761731	

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)
<i>GSTM5</i> TAGS	rs4970773	
<i>IL6</i> (also listed in Section 10)	rs1800795 rs1548216 rs1800796 rs2069824 rs2069835 rs2069840 rs2069842 rs2069843 rs2069845 rs2069827 rs2069832	
<i>ANGPTL4</i> Lipid metabolism	rs1044250	
<i>ACC-alpha</i> (Acetyl- carboxylase alpha) Fatty acid synthesis	rs829163 rs11655013	
<i>TADA2L</i> (shares bidirectional promoter with ACC- α)	rs1714987 rs2158235 rs712039	
<i>GH1</i> (growth hormone 1) TAGS	rs2005172	

13. Additional known candidate genes

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)
<i>NFKB</i> (<i>NKIRAS2</i>)	rs34694587 rs35364057	
<i>WISP2</i> (<i>CCN5</i>)	rs2281862	
<i>WISP3</i> (<i>CCN6</i>)	rs3806964	
<i>PDEF</i> (<i>SPDEF</i>)	rs2233639	
<i>PRLR</i> Prolactin receptor	rs16871473 rs401694 rs112461	
<i>RASSF1A</i>	rs2073498	
<i>CYP2J2</i> (promotes metastasis)	rs2280273 rs2280274 rs4388726 rs11572243	
<i>CHFR</i> (checkpoints)	rs3741489 rs2306536	
<i>VCAM1</i> (cell adhesion)	rs34228330 rs3783611 rs3783612 rs3783613 rs3176878	
<i>ATRIP</i> (<i>TREX1</i> , ATR interacting protein)	rs11925638 rs35240314	

Collaboration with Kent Hunter (NCI) Metastasis genes		
<i>LUC7L</i>	rs1203981	
<i>SIPA1</i>	rs931127 rs1466462	
<i>BRD4</i>	rs4808272	
<i>PI16</i>	rs707542	
<i>MTCH1</i>	rs708006	
<i>TTC9C</i>	rs6591720	
<i>RRP1B</i>	rs2155722 rs762400	
<i>VDR</i> (Vitamin D receptor)	rs1544410	

Cooked meat metabolism:			
Gene	Illumina assay (N=3748)	Taqman assay (N=3851)	Completed before 2008 (N varies)
<i>NAT1</i>	rs4986783		Part of Phase 1 data; rs number unknown.
<i>NAT2</i>	rs1801279 rs1801280 rs1208 rs2552 rs4646246		Part of Phase 1 data; rs number unknown.
[Note: SULT1A2, GSTs, CYP1A1: are covered in other sections]			

14. Chuck Perou's candidates (NCI collaboration GWA positives), including subtype specific genes

PGR, TNF alpha, ESR1, ERCC4, GATA3 included in other sections.

Gene	Illumina assay (N=3748)	Taqman assay (N=3851)
<i>CASP10</i> TAGS	rs13010627	
<i>RAP80</i> (<i>RXRIP110</i>)	rs10475633 rs13360277 rs3733876	
<i>Syndecan 1</i>	rs10205485	
<i>Kit oncogene</i>	rs2213181 rs8022	
<i>Cadherin 3</i>	rs1126933	
<i>Nuclear Factor I/B</i>	rs484555	
<i>Aristaless</i> <i>homeobox 4</i>	rs897004 rs4755797	
<i>Bmp7</i>	rs12438 rs6014947 rs10375	
<i>KRAS2</i>	rs12587 rs712 rs9266	

15. Replication of GWA results
(Positive results in Ponder/Easton papers; Hunter; etc.)

Gene name or location	Illumina assay (N=3748)	Taqman assay (N=3851)
<i>CASP8</i>	rs1045485 rs17468277	
<i>FGFR2 / CEK3 Fibroblast growth factor receptor 2: Receptor tyrosine kinase amplified / overexpressed in breast cancer, signal transduction (Ponder; Hunter)</i>	rs2981582 rs1219648 rs2420946 rs11200014 rs2981579 rs2162540 rs3135718 rs2936870 rs2912774 rs1896395	rs2981578 rs1078806 rs10736303 rs3750817
<i>MAP3K1 / MEKK</i>	rs889312	
<i>TNRC9 / TOX3</i>	rs12443621 rs8051542 rs9940048	
<i>LOC643714 Upstream of TNRC9 Non-histone chromatin protein, Maybe transcription factor (Ponder; Stacey ER+, Also in AA opposite direction)</i>	rs3803662 rs16951186 rs8049149	
<i>Chr 16q TOX3 region</i>		rs4784227
<i>Chr 16q TOX3/LOC643714</i>		rs3104746 rs3112562

Gene name or location	Illumina assay (N=3748)	Taqman assay (N=3851)
<i>LSP1</i> / <i>WP43</i>	rs3817198	
<i>Msc 5A1</i>	rs6476643	
<i>H19</i> (involved with <i>IGF2</i>)	rs2107425	
<i>RELN</i>	rs17157903	
<i>TLR1</i> / <i>TLR6</i>	rs7696175	
<i>Others on Ponder list</i> (anonymous) <i>Chr 8q</i> <i>Chr 5p</i> <i>Chr 5q</i> <i>Chr 2p</i>	rs13281615 rs981782 rs30099 rs4666451	
<i>Others on Hunter list</i> (anonymous) <i>Chr 10q</i>	rs10510126	
<i>Others on Hunter list</i> (anonymous) <i>Chr 4p</i>		rs12505080
<i>Others on Stacey ER+ list</i> (anonymous) <i>Chr 2q35</i>	rs13387042	
<i>Chr 1p11.2</i> <i>EMBP1</i>		rs11249433
<i>Chr 3p24</i> <i>SLC4A7</i>		rs4973768

Gene name or location	Illumina assay (N=3748)	Taqman assay (N=3851)
<i>Chr 5q31</i> <i>ARHGAP26</i>		rs153170 rs23282 rs27779 rs13172733 rs7705560
<i>Chr 5</i> <i>ARHGAP26</i>		rs3822397
<i>Chr 2</i>		rs7576160
<i>Chr 8</i>		rs4460408
<i>Chr 3</i> <i>GRM7</i>		rs7610073
<i>Chr 5p12</i> <i>MRPS30</i> <i>Close to centromere</i> <i>Coord</i> <i>44554844</i>		rs4415084 rs10941679
<i>Chr 6q25</i> <i>ESR1</i>		rs6557161
<i>Chr 6q25.1</i> <i>Unknown locus</i> <i>upstream of ESR1</i>		rs851974 rs2046210
<i>Chr 6q22.33</i> <i>ECHDC1</i>		rs2180341
<i>Chr 2</i> <i>DIRC3</i>		rs13388294
<i>Chr 6</i> <i>ESR1</i> <i>(also listed in Section 4)</i>		rs543650

Gene name or location	Illumina assay (N=3748)	Taqman assay (N=3851)
Chr 17q23.2 <i>STXBP4</i>		rs6504950
<i>Chr 17q22</i> <i>COX11, STXBP4</i>		rs7222197
<i>Chr 8q24</i>		rs1562430
<i>LSP1</i>		rs909116
<i>Chr 11q13</i>		rs614367
<i>Chr 9p21</i> <i>CDKN2A/B</i> <i>(also listed in Section 3)</i>		rs1011970 rs10757278
<i>Chr 10</i> <i>different regions</i>		rs2380205 rs10995190 rs704010

16. Additional genes in the FGFR2 and other pathways identified by GWA.
NOT DONE PREVIOUSLY.

Gene or locus	Illumina assay (N=3748)	Taqman assay (N=3851)
<i>N -RAS</i>	rs6681671 rs8453 rs1065634	
<i>MAPKKK (BRAF)</i>	rs34776339	
<i>MAP2K1 (ERK1/2)</i>	rs14303	
<i>MKP3</i>	rs770087	
<i>ER81</i>	rs9639168	
<i>FGFR1</i>	rs2467531	
<i>FGFR3</i>	rs3135868 rs3135901	
<i>FGFR4</i>	rs376618 rs42409	

17. Additional SNPs

Gene name or location	Illumina assay (N=3748)	Taqman assay (N=3851)
<i>FOXC1</i>	rs984253	
<i>TLR4</i>	rs1927911 rs5030717 rs7869402 rs2149356 rs2770146	
<i>SCGB1D2</i>	rs2232950	
<i>Chr 17</i> <i>MYST2 gene,</i> <i>miRNA</i>		rs7211603
TERC Telomere length Codd, Nat Genetics 2010 (Note: May interact with WRN gene)		rs12696304
<i>FANCA</i>		rs1061646
<i>FLT1 / VEGFR</i> (<i>VEGF receptor</i>) -677 Promoter P53 response element T/C		No rs number; variable name is FLT1_T.
<i>OPRM1</i> (<i>Opioid receptor mu 1</i>)		rs563649 rs2075572 rs1799971 rs533586 rs495491 rs609148

Gene name or location	Illumina assay (N=3748)	Taqman assay (N=3851)
CYB5A		rs or ss number
-206 G>-		rs36082929
13T>G		ss159697782 (rs75160992)
65A>G		ss159697395 (rs74339771)
12-992 C>T		rs1790894
155 G>A		rs1803366
178A>G		ss159697398 (rs78009726)
390C>A		rs1803364
*246T>C		ss159816071 (rs76241580)

Gene name or location	Illumina assay (N=3748)	Taqman assay (N=3851)
CYB5R3		rs or ss number
-251G>T		ss159816069 (rs7388347)
-231C>A		ss159816070 (rs75133903)
11M+6C>T		rs8190370
11M+6072C>T		rs8190414
176G>A		ss159830792 (rs111154229)
18-1676C>A		rs751153
890G>A		ss159830793
*138G>A		ss159816065
*863T>C		ss159830807
*392G>C		rs7284807