

Medically Actionable Genes List (work in process)

Updated: 8/1/09

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<http://melanieswan.com/documents/MAGL.xls>

Most people do not realize that there are many genes which are medically actionable now.
 Not all data is available for every entry; information is added as it becomes available.
 The information was collected from technical conference talks and academic journal articles.

Category and Condition	Locus ex: 10p14	Chromosome ex: 10	Gene	Variant/SNP RSID	Allele Variant	# cases / # controls	Updated: 7/31/09 Citation/Note
Pharmacogenomics							
Poor metabolizer / Ultrafast metabolizer			CYP2D6	rs1799853 rs1057910 rs9923231			poor drug metabolizers (10% cauc) http://elcamino.dnadirect.com/elcamino/simple/ssri/patient-articles/what-testing-for-cyp2d6-and-cyp2c19-tells-you.html
Zinc transport/ Insulin transport			SLC30A4 SLC30A5				genes involved in zinc transportation; insulin transport from the pancreas 13% of the caucasian population has a zinc transport gene mutation
Heart disease							
MI: myocardial infarction	9p21	9	CDKN2A/B				Genome-wide case-control assn study, 9p21 is the strongest GWA marker so far
Hypertrophic cardiomyopathy			MYH7 MYBPC3 TNNT2 TNNI3 TPMI MYL2 etc				Mutations and missense polymorphisms in these sarcomere protein genes which cause hypertrophic cardiomyopathy (HCM) Genetic Basis for Hypertrophic Cardiomyopathy: From Bench to the Clinics http://www.ncbi.nlm.nih.gov/pubmed/17916152
Hypertensior			STK39				A gene that affects how the kidneys process salt, gene variants have higher blood pressure n=7,125 Wang, Y. Whole-genome association study identifies STK39 as a hypertension susceptibility gene http://www.ncbi.nlm.nih.gov/pubmed/19114657
Diabetes							
Type 2			TCF7L2 IGF2BP2 CDKN2A/B FTO CDKAL1 KCNJ11 HHEX/IDE SLC30A8 PPARG	rs10885409			NHGRI's GWAS study finding of the first nine genetic risk variants for type 2 diabetes Could identify higher-risk individuals early in life
Neurological disease							
Alzheimer's Disease			APOE*e4 Kibra, GAB2				Correlation with mid-life cholesterol levels and AD - study citation in CGS talk http://www.news-medical.net/news/2006/10/20/20650.aspx
Cancer							
Breast	16q12	chr8.12842480		rs3803662 rs13281615 rs2981582 TNRC9 rs3803662			Cited by Navigenics, 23andme and deCODEme Cited by Navigenics Cited by Navigenics Cited by Navigenics

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	2q35	chr2.21761407		rs13387042			Cited by Navigenics and deCODEme
	5q11		MAP3K1	rs889312			Cited by Navigenics and deCODEme
	10q26			rs1219648			Cited by 23andme and deCODEme
			FGFR2	rs2981582			Cited by Navigenics
			CASP8	rs1045485			Cited by Navigenics
				rs3834129			Cited by Navigenics
			LSP1				Cited by Navigenics
	5p12			rs4415084			Cited by deCODEme
	8q24			rs13281615			Cited by deCODEme
	11p15			rs3817198			Cited by deCODEme
			185delAG				Cited by 23andme; BRCA1 mutation
			5382insC				Cited by 23andme; BRCA1 mutation
			6174delT				Cited by 23andme; BRCA2 mutation
				rs11249433			Nature Genetics June/July 2009
				rs999737			Nature Genetics June/July 2009
Testicular Germ Cell Tumors (TGCT)			KITLG				Stronger indication of genetic predisposition established
			SPRY4				High marks for GWAS, Stephen Chanock
			BAK1				http://www.nature.com/ng/journal/v41/n7/full/ng0709-765.html
Colorectal Cancer			p16, vimentin			20/50	p16, vimentin (aberrant methylation) p16 (tumor suppression) was hypermethylated in 40% (only!) of cases tested (20/50)
General phenotypic traits							
Height, BMI							A large-scale genome-wide association study of Asian populations
Pulse rate				rs12731740			uncovers genetic factors influencing eight quantitative traits.
Pulse rate				rs12110693			http://www.ncbi.nlm.nih.gov/pubmed/19396169
Systolic blood				rs17249754			Nat Genet. 2009 May;41(5):527-34. Epub 2009 Apr 26
Bone density				rs7776725			
Waist/hip ratio				rs2074356			