

Estrogen Metabolism SNPs

Gene & Variation	rsID	Risk Allele	Function	Health Risks	Actions to Take to Protect																					Lab Testing																				
					Avoid/Reduce These													Increase These																												
					Xeno-Estrogens *	Caffeine	Alcohol	Exposure to cigarette smoke	Exposure to gas fumes	Acid-blocking Rx	Tylenol & other Rx	Char-broiled meat	Processed meats	Sugar	Processed Foods	Cosmetics and cleaners	Pesticides	Heated & oxidized fats	Exercise and Sweat	Hydration	Phase1 enhancing foods**	Omega 3 fats	Antioxidant intake	Foods that enhance Phase 1	Green Leafy Veggies	Sulfur-rich foods/MSM	Cruciferous vegetables	Mindfulness practices	Glutathione precursors	Glucoraphanin/Sulforaphane	Milk Thistle/Liver Support	Iron if deficient	Selenium	B vitamin intake	Folate (Vitamin B9)	Vitamin B12	Vitamin A	Vitamin C	Vitamin D	Vitamin E	CBC	Vitamin D	Homocysteine	Hemoglobin A1c	Blood Spot Fatty Acid	DUTCH Test
CYP1A1 *2CA4889G C2453A	rs1048943 rs1799814	C T	• Detoxifies polycyclic aromatic hydrocarbons • Metabolizes excess estrogen • Responsible for 2-OH estrogen.	• uterine fibroids • fibrocystic breasts • cancer	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓				✓				✓				✓	✓	✓	✓	✓	✓	✓	✓		
CYP1A2 164A>C	rs762551	C	• Caffeine metabolism • Estrogen metabolism – upregulation of 4-OH, • Detoxifies acetaminophen • Detoxifies aflatoxin	• uterine fibroids • fibrocystic breasts • breast cancer • ovarian cancer • prostate cancer	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓				✓					✓				✓	✓	✓	✓	✓	✓	✓	✓	
CYP1B1 L432V N453S R48G	rs1056836 rs1800440 rs10012	C C C	• Helps detoxify estrogen • upregulation of the 4-OH estrogen	• uterine fibroids • fibrocystic breasts • breast cancer	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓				✓					✓				✓	✓	✓	✓	✓	✓	✓	✓	
CYP3A4 *1B	Rs2740579	C	• Breakdown of over 60% of drugs, including steroids • breakdown of organophosphate insecticides (e.g. parathion). • 16-OH estrogen.	• Prostate cancer • uterine fibroids • fibrocystic • breast cancer	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓				✓						✓				✓	✓	✓	✓	✓	✓	✓	✓
HSD3	rs3765945	C	• Aids with the production of all classes of steroid hormones	• Essential hypertension • aldosterone level	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓				✓						✓				✓	✓	✓	✓	✓	✓	✓	✓



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					Xeno-Estrogens *	Caffeine	Alcohol	Exposure to cigarette smoke	Exposure to gas fumes	Acid-blocking Rx	Tylenol & other Rx	Char-broiled meat	Processed meats	Sugar	Processed Foods	Cosmetics and cleaners	Pesticides	Heated & oxidized fats		Exercise and Sweat	Hydration	Phase1 enhancing foods**	Omega 3 fats	Antioxidant intake	Foods that enhance Phase 1	Green Leafy Veggies	Sulfur-rich foods/MSM	Cruciferous vegetables	Mindfulness practices	Glutathione precursors	Glucoraphanin/Sulforaphane	Milk Thistle/Liver Support	Iron if deficient	Selenium	B vitamin intake	Folate (Vitamin B9)	Vitamin B12	Vitamin A	Vitamin C	Vitamin D	Vitamin E	CBC	Vitamin D	Homocysteine	Hemoglobin A1c	Blood Spot Fatty Acid	DUTCH Test
ESR1	rs2881766 rs9383938 rs2881766 rs1999805	G T T C	• Binds estrogen • Mediates functions, turning genes on and off. • Bound estrogen influences: ➤ Development of the reproductive system ➤ Bone growth ➤ Glucose and lipid metabolism	• (G) Endometrial Cancer • (T) Increased risk of pregnancy induced hypertension • (T) Breast Cancer • (C) Bone density	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓			✓							✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓
SRD5A1	rs3822430	G	• Steroid 5-alpha reductase 1 • Converts testosterone into 5-alpha-dihydrotestosterone and progesterone or corticosterone 5-alpha-3-oxosteroids. • Sexual differentiation & androgen physiology. • Associated with dihydro-testosterone (DHT)	• Prostate cancer progression in men • PCOS and hirsutism in women.	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓			✓							✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	
SRD5A2	rs559555	T	• Steroid 5-alpha reductase 2 • Enzyme is responsible for a converting testosterone to DHT	• Prostate cancer • (T) associated with a reduced recurrence rate after radical prostatectomy	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓			✓							✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	



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CYP19A1 (Aromatase)	rs1008805 rs700518	G C	• Encodes for a cytochrome P450 enzyme aromatase • catalyzes drug metabolism reactions • Synthesis of cholesterol, steroids, and other lipids. • formation of aromatic C18 estrogens from C19 androgens. • Catalyzes the last steps of estrogen biosynthesis important for sexual development	• Low estrogen • increased androgens • impaired sexual development in females • Breast cancer • unusual bone growth • insulin resistance in males. • Hypertension and Preeclampsia • Bone loss • endometriosis-related infertility.	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓			✓				✓				✓	✓	✓	✓	✓	✓	✓	✓		
SULT2A1	rs2547231	A	• Sulfonation of steroids and bile acids in the liver and adrenal glands	• Gallstone disease	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓			✓					✓			✓	✓	✓	✓	✓	✓	✓	✓		
SULT1E1	rs9282861 rs1042157	C A	• Catalyzes transformation of estrone into estrone sulfate • Catalyzes the sulfate conjugation of many hormones, neurotransmitters, drugs, and xenobiotic compounds. • May control levels of estrogen receptors	• Breast Cancer • associated with a poor response to tamoxifen, a chemotherapeutic drug used to treat breast cancer	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓			✓					✓			✓	✓	✓	✓	✓	✓	✓	✓		



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STS	rs1473666	?	Conversion of sulfated steroid precursors to estrogens during pregnancy.	- Fatty liver - Inflammation, - Insulin resistance - Weight gain - Obesity - Rheumatoid arthritis	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓				✓				✓			✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	
AKR1C4	rs17134585 rs1832007	T G, A	- Catalyzes DHT into 5-alpha-androstan-3-alpha,17-beta-diol 20-alpha-hydroxysteroid dehydrogenase activity.	- Blood metabolite measurement - Triglyceride measurement	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓				✓				✓			✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	
CYP11B1			Encodes for enzyme 11-beta-hydroxylase	- Congenital adrenal hyperplasia - Hypertension.	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓				✓						✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	
CYP11B2	rs1799998 rs1799998	A T	- Codes for aldosterone - Breaks down steroid-based drugs - Maintains intracellular salt and fluid levels. - Estrogen regulation of adrenal gland	- Recurrent cerebral ischemia - Renal sodium excretion and potassium retention resulting in hyponatremia, hyperkalemia, metabolic acidosis	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓				✓						✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓



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HSD11B1	rs3753519 rs12086634	T G, A	- Codes for hydroxysteroid 11-beta dehydrogenase. - Active cortisol production and amplification of glucocorticoids in the liver and adipose tissue. - Catalyzes conversion of cortisol to cortisone, and cortisone to cortisol.	- Pediatric-onset central obesity and insulin resistance. - Hypertension - hyperglycemia - High HDL Cognitive decline with aging	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓				✓					✓			✓	✓	✓	✓	✓	✓	✓	✓	✓
BRCA1 A1708E R1699W	rs28897696 rs55770810	T T	- Tumor suppressor gene. - Repairing DNA damage - Stopping the cell cycle to prevent passing damaged DNA to new cells. Regulating the production and activity of proteins implicated in cancer	Significantly increases risk of developing breast and ovarian cancer.	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓			✓					✓			✓	✓	✓	✓	✓	✓	✓	✓	✓
BRCA2	rs11571833	T	- tumor suppressor. - Repair of damaged DNA - regulate cytokinesis (part of cell division)	- Early age breast/ ovarian cancer – risk increase is 60 to 85% - Ovarian cancer - Estrogen receptor negative ER(-) breast, lung, ovarian, prostate, colorectal cancer	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓			✓					✓			✓	✓	✓	✓	✓	✓	✓	✓	✓



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HSD17B1-8	rs598126 rs2010750	T, CC C, GG	- Estrogen activation and androgen inactivation - Establishing E2 concentration gradient between serum and peripheral tissues. - Expressed in the placenta, ovarian granulosa cells, endometrium, adipose tissue, and prostate.	- Increase in Alzheimer’s - Increase in cholinergic. - Dramatic declines in estrogen following menopause - Endometriosis - Estrogen receptor-negative tumors in colorectal cancer	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓			✓					✓			✓	✓	✓	✓	✓	✓	✓	✓	✓	✓
COMT	rs4680 rs4633	A G	- Converts hydroxy to methoxy estrone. - Reduces estrogen dominance symptoms: PMS, heavy bleeding, fibroids, endometriosis, etc. - Metabolism of dopamine, serotonin and norepinephrine	4680 (A) Worrier 4680 (G) Warrior, Schizophrenia 4633 (G) Breast and endometrial cancer	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓			✓					✓			✓	✓	✓	✓	✓	✓	✓	✓	✓	✓
SULT1A1	rs1042157	A	Sulfate conjugation of: hormones, neurotransmitters, drugs, and xenobiotic compounds	- Breast Cancer, - Male Breast Cancer - Colon Cancer - Lung Cancer - Sensitivities to food colorings, phenol	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓			✓					✓			✓	✓	✓	✓	✓	✓	✓	✓	✓	✓



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SHBG	rs179941	A	• Codes for Sex Hormone Binding Globulin Protein binds to and makes inactive sex hormones testosterone and estrogen.	• Decreased availability of free testosterone and estrogen	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓			✓					✓			✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	
BARD-1	rs6435862 rs3768716	G C	• Encodes a protein that interacts with the N-terminal of the BRCA1 gene (involved in tumor suppression	• Breast Cancer • Ovarian Cancer • Neuroblastoma	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓			✓					✓			✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	
GSTM1	rs1056806	T	• Codes for Glutathione transferease • Involved in degradation of xenoestrogens, toxins and carcinogens.	• Drug induced liver injury • Lung cancer • Liver cancer • Chromosome aberrations • Asthma • Squamous cell carcinoma	✓	✓	✓	✓	✓		✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓			✓					✓			✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓

