



## Gene Comprehensive Nutrigenomic Report

Accession Number: #####  
Specimen Collected: ##/##/####  
Specimen Received: ##/##/####  
Report Generated: December 4, 2019  
Specimen Type: Buccal Swab  
Provider: #####  
Patient Name: #####  
Patient DOB: ##/##/####  
Patient Gender: Female

Do not make any decisions about your health solely based on the information contained in this report.  
Always consult with a licensed and experienced health practitioner when you receive this report.

##### – 60 – Female

(-/-) No clinical abnormality

(+/-) Heterozygous result

(+/+) Homozygous result

rsID	Gene	Genetic Result	Therapeutics Associated With Positive Result	Highly Recommended Therapeutics / Neurobiologix Formulas	Provider Discretion: As Needed Formula Recommendations	Lifestyle Recommendations	Laboratory Recommendations
<b>Best Diets for Weight Loss</b>							
rs1799883	FABP2 A54T	+/-	Increased Absorption of Fatty Acids from Foods	<b>Low Fat / High Protein Diet Should Work Well for You</b> (2.5 times Expected Weight Loss with Low Fat Diet)			
rs1801282	PPARG	+/-	Polymorphism Causes Increase in Fat Storage and Decrease in Fat Mobilization  Polymorphism Causes Increase Cellular Uptake of Glucose and Can Lead to Hypoglycemia				
rs9939609	FTO	-/-	Lower Calorie Intake		<b>"Zone" Diet May Be Beneficial to Weight Loss</b>  <b>Plant Based Diet May Be Beneficial to Weight Loss</b>		
rs17300539	ADIPOQ	-/-	Need to Limit Saturated Fat and Ingest Low Glycemic Foods				
<b>Fatty Acid Metabolism</b>							
rs5082	APOA2	+/-	Increased Incidence of Obesity			Having Some High Quality Unsaturated Fats in Your Diet Should Not Affect Weight Loss	
rs662799	APOA5	-/-	Increased Risk of Hyperlipidemia				
rs662799	APOA5	-/-					
<b>Satiety Genes</b>							
rs1137101	LEPR	+/-	Decreased Leptin Receptor Response Indicates Decreased Satiety	<b>Calorie Restriction (Small Portion / Lower Calorie) Should Work Well for You</b>		You May Benefit from Leptin Drugs and Appetite Suppressants	

rs696217	GHRL	-/-	Increased Hunger Reponse Increased Reward System for Alcohol and Sweets				
rs1800206	PPARA	-/-	Poor Response to Fasting		<b>"Fasting Mimicking Diet" May Be Beneficial for Weight Loss</b>	<b>Intermittent Fasting Should Benefit Weight Loss</b>	

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rsID	Gene	Genetic Result	Therapeutics Associated With Positive Result	Highly Recommended Therapeutics / Neurobiologix Formulas	Provider Discretion: As Needed Formula Recommendations	Lifestyle Recommendations	Laboratory Recommendations
<b>Intensity of Exercise Needed for Weight Loss</b>							
rs4994	ADRB3	+/-	<b>Positive Result Indicates Lower Than Expected Weight Loss Potential with Exercise</b>	<b>High Intensity Interval Training (More than 30 Mins of Exercise with Heart Rate &gt; 70% of Maximum) Required for Significant Weight Loss</b>			
rs1042714	ADRB2	+/-					
rs17300539	ADIPOQ	-/-					
<b>Insulin Resistance Risk</b>							
rs510432	ATG5	+/-	Curcumin, Lithium Orotate, D-Chiro-Inositol, Catechins, Resveratrol, Caffeine, 12-15 Hour Fasting	<b>Consider DCI 500mg™</b> <b>Consider N.A.S. Enhancer™</b>		12-15 Hour Fasting Should Work Well for You Unless PPARA Polymorphism is Present	Routine Blood Sugar, Insulin and Hb A1c
rs10210302	ATG16L1	+/-					

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rsID	Gene	Genetic Result	Therapeutics Associated With Positive Result	Highly Recommended Therapeutics / Neurobiologix Formulas	Provider Discretion: As Needed Formula Recommendations	Lifestyle Recommendations	Laboratory Recommendations
Lactose Intolerance							
rs4988235	MCM6	+/+	High Incidence of Lactose Intolerance	<b>You Possess a High Risk of Lactose Intolerance</b>		Avoid Lactose (Milk Products)	
Caffeine Metabolism							
rs762551	CYP1A2	-/-	Slow Metabolizer of Caffeine				
Chromium and Low Dose Naltrexone Efficacy							
rs1076560	DRD2	-/-	Polymorphism Indicates Better Response Rate to Chromium Picolinate and Low Dose Naltrexone				
Effect from Green Tea Extract / Green Coffee Bean Extract							
rs4680	COMT V158M	+/-	Improved Response to Green Tea and Green Coffee Bean Extracts	<b>You Should Benefit from Green Tea Extract or Green Coffee Bean Extract for Weight Loss</b>			
Salt Sensitivity							
rs4343	ACE	+/-	Increased Risk of Salt Retention and Hypertension	<b>Be Cautious with High Salt Foods</b>		Recommend Reducing Your Salt Intake	
rs699	AGT	+/+					
Sugar Sensitivity and Mood							
rs1800544	ADRA2A	+/+	Increased Risk of Anti-Psychotic or Anti-Depressant Induced Weight Gain			High Risk of Major Weight Gain with Anti-Psychotic and Anti-Depressant Medications	

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rsID	Gene	Genetic Result	Therapeutics Associated With Positive Result	Highly Recommended Therapeutics / Neurobiologix Formulas	Provider Discretion: As Needed Formula Recommendations	Lifestyle Recommendations	Laboratory Recommendations
Inflammatory Environmental							
rs10156191	AOC1	-/-	Risk of Histamine Food Reaction				
rs11558538	HNMT	-/-					
rs12995000	HNMT	+/-					
rs492602	FUT2	+/-	Prebiotics and Probiotics Needed		Consider Biotic Blend Pro™ Every 2-3 Days if GI Inflammation is Present		
rs2187668	HLA DQA1	-/-	High Risk of Gluten and Casein Sensitivity				
rs2858331	HLA DQA2	-/-	Broad Spectrum Enzyme				

# Summary for Diet / Wellness

## Highly Recommended Therapeutics / Neurobiologix Formulas

- Low Fat / High Protein Diet Should Work Well for You (2.5 times Expected Weight Loss with Low Fat Diet)
- Calorie Restriction (Small Portion / Lower Calorie) Should Work Well for You
- High Intensity Interval Training (More than 30 Mins of Exercise with Heart Rate > 70% of Maximum) Required for Significant Weight Loss
- Consider DCI 500mg™
- Consider N.A.S. Enhancer™
- You Possess a High Risk of Lactose Intolerance
- You Should Benefit from Green Tea Extract or Green Coffee Bean Extract for Weight Loss
- Be Cautious with High Salt Foods

## Provider Discretion

- "Zone" Diet May Be Beneficial to Weight Loss
- Plant Based Diet May Be Beneficial to Weight Loss
- "Fasting Mimicking Diet" May Be Beneficial for Weight Loss
- Consider Biotic Blend Pro™ Every 2-3 Days if GI Inflammation is Present

## Lifestyle Recommendations

- Having Some High Quality Unsaturated Fats in Your Diet Should Not Affect Weight Loss
- You May Benefit from Leptin Drugs and Appetite Suppressants
- Intermittent Fasting Should Benefit Weight Loss
- 12-15 Hour Fasting Should Work Well for You Unless PPARA Polymorphism is Present
- Avoid Lactose (Milk Products)
- Recommend Reducing Your Salt Intake
- High Risk of Major Weight Gain with Anti-Psychotic and Anti-Depressant Medications

## Laboratory Recommendations

- Routine Blood Sugar
- Insulin and Hb A1c

# LOW FAT/HIGH PROTEIN DIET

## FOOD SOURCES



Egg whites



Tofu



Lean cuts of beef, pork (loin, etc.)



Turkey/chicken



Fish (cod, tilapia, shrimp, etc.)



Low or non-fat yogurt



Low-fat cottage cheese



Fat free milk



Vegetables (spinach, lettuce, mushrooms, broccoli)



Fruits (oranges, apples, melons, bananas)



Grains (quinoa)

**Low Fat / High Protein Diet Definition:** This diet plan limits total fat and carbohydrate intake while increasing protein intake to improve satiety and promote healthy weight loss.



## BENEFITS:



Improved blood lipids



Reductions in body weight & triglycerides



Limits risk of developing cardiovascular disease

## AVOID

- High Fat Meats
- Whole Milk
- Nuts/Oils
- Limit Grains



# ZONE DIET

## FOOD SOURCES



Proteins (lean beef, pork, lamb, chicken, turkey breast, fish, tofu, egg whites)



Oils/Fats (avacados, peanuts, almonds, peanut butter, oils)



Dairy (low fat cheese, milk & yogurt)



Carbohydrates (fruits, vegetables, grains/oatmeal)

**Zone Diet Definition:** The Zone Diet is a weight loss plan based upon the idea that the right ratio of carbohydrates to proteins and fats can control levels of insulin in the bloodstream.



## BENEFITS:



Increased energy



Increased physical performance



Heightened mental focus & productivity



Metabolic state of hormonal efficiency

## AVOID

- High sugar fruits
- High sugar or starchy vegetables
- Refined & processed carbs
- Foods with added sugar
- Soft drinks
- Coffee & tea

# HIIT EXERCISE

## TYPES



Sit-Ups



Sprints



Jumping Exercises



Push-Ups



Jumping Jacks



Pull-Ups

**HIIT (High-Intensity Interval Training) Definition:** A training technique in which you give all-out, one hundred percent effort through quick, intense bursts of exercise, followed by short, sometimes active, recovery periods. This type of training gets and keeps your heart rate up and burns more fat in less time.

**HIIT  
TYPICAL  
EXERCISE**

## BENEFITS:



Boosts energy



Boosts metabolism



Reduces heart rate, blood sugar, and blood pressure



Helps with muscle gain



Can improve oxygen consumption



Burns calories in a short amount of time and helps you lose fat

## Gene Information Key

rsID	Gene	"-" variant	"+" variant
rs4343	ACE	A	G
rs17300539	ADIPOQ	G	A
rs1800544	ADRA2A	G	C
rs1042714	ADRB2	C	G
rs4994	ADRB3	A	G
rs699	AGT	A	G
rs10156191	AOC1	C	T
rs5082	APOA2	A	G
rs662799	APOA5	A	G
rs10210302	ATG16L1	C	T
rs510432	ATG5	C	T
rs4680	COMT V158M	G	A
rs762551	CYP1A2	A	C
rs1076560	DRD2	C	A
rs1799883	FABP2 A54T	C	T
rs9939609	FTO	T	A
rs492602	FUT2	A	G
rs696217	GHRL	G	T
rs2187668	HLA-DQA1	C	T
rs2858331	HLA-DQA2	A	G
rs11558538	HNMT	C	T
rs12995000	HNMT	C	T
rs1137101	LEPR	A	G
rs4988235	MCM6	G	A
rs1800206	PPARA	C	G
rs1801282	PPARG	C	G

## Definitions

GASTROINTESTINAL	
MCM6	A mutation in a DNA control region located in the MCM6 gene is associated with expression of the lactase gene. Individuals homozygous for this polymorphism are more likely to have hypolactasia, or lactose intolerance.
General	
ADIPOQ	This gene is expressed in adipose tissue exclusively and encodes for the protein adiponectin. Adiponectin is involved with metabolic and hormonal processes. Mutations in this gene are associated with adiponectin deficiency.
ADRB2	The protein encoded by this gene belongs to the family of beta adrenergic receptors that mediate catecholamine sensitivity. This receptor is located mainly in the adipose tissue and is involved in the regulation of lipolysis and thermogenesis.
ADRB3	The protein encoded by this gene belongs to the family of beta adrenergic receptors that mediate catecholamine sensitivity. This receptor is located mainly in the adipose tissue and is involved in the regulation of lipolysis and thermogenesis.
APOA2	This gene encodes apolipoprotein (apo-) A-II, which is the second most abundant protein of the high density lipoprotein particles. Defects in this gene may result in apolipoprotein A-II deficiency or hypercholesterolemia.
APOA5	The protein encoded by this gene is an apolipoprotein that plays an important role in regulating the plasma triglyceride levels, a major risk factor for coronary artery disease. It is a component of high density lipoprotein. Mutations in this gene have been associated with hypertriglyceridemia and hyperlipoproteinemia type 5.
CYP1A2	This gene encodes a member of the cytochrome P450 superfamily of enzymes typically found in the liver. These enzymes catalyze many reactions involved in drug metabolism and synthesis of cholesterol, steroids, lipids, and caffeine.
FABP2 A54T	The protein encoded by this gene is an intracellular fatty acid-binding protein that participates in the uptake, intracellular metabolism, and transport of long-chain fatty acids. The encoded protein is also involved in the modulation of cell growth and proliferation. This protein binds saturated long-chain fatty acids with high affinity, and acts as a lipid sensor to maintain energy homeostasis.
FTO	This gene codes for a nuclear protein non-haem iron and 2-oxoglutarate-dependent oxygenase superfamily. This enzyme functions to reverse alkylated DNA and RNA damage by oxidative demethylation. Studies indicate a strong association with body mass index, obesity risk, and type 2 diabetes.
GHRL	This gene encodes the ghrelin-obestatin preproprotein that is cleaved to yield two peptides, ghrelin and obestatin. Ghrelin is a powerful appetite stimulant and plays an important role in energy homeostasis. Its secretion is initiated when the stomach is empty, whereupon it binds to the growth hormone secretagogue receptor in the hypothalamus which results in the secretion of growth hormone (somatotropin). Ghrelin is thought to regulate multiple activities, including hunger, reward perception, gastric acid secretion, gastrointestinal motility, and pancreatic glucose-stimulated insulin secretion.
LEPR	This gene codes for the Leptin Receptor which is associated with the cytosolic STAT proteins. This receptor for leptin (an adipocyte-specific hormone that regulates body weight) is involved in the regulation of fat metabolism with mutations in this gene have been associated with obesity.
PPARA	The peroxisome proliferators induce the production of intracellular peroxisomes that contain enzymes for respiration and for cholesterol and lipid metabolism. The action of peroxisome proliferators is mediated via specific receptors, called PPARs, which belong to the steroid hormone receptor superfamily. PPARs affect the expression of target genes involved in cell proliferation, cell differentiation and in immune and inflammation responses. This gene encodes the subtype PPAR-alpha, which is a nuclear transcription factor.
PPARG	This gene encodes a nuclear factor called peroxisome proliferator-activated receptor (PPAR). PPARs form heterodimers with retinoid X receptors (RXRs) and these heterodimers regulate transcription of various genes. The protein encoded by this gene is PPAR-gamma and is a regulator of adipocyte differentiation.
HYPERTENSION	The polymorphisms in this category will increase the risk of developing hypertension.
ACE	Angiotensin-converting enzyme (ACE) is an important target for therapeutic drugs treating hypertension and heart failure. The best studied single nucleotide polymorphism in the ACE gene (rs4343) has been linked to a wide variety of human phenotypes: nephropathy and renal disease, cancer, and even sports performance. Interestingly, rs4343 is a member of a large family of human mutations called Alu elements.
AGT	The AGT gene codes for the angiotensinogen protein, a key regulator of blood pressure and body fluid homeostasis. Individuals carrying two copies of the rs699 C allele are at increased risk of hypertension-related disorders such as pre-eclampsia.
INFLAMMATORY	This Enzyme category has significant effects on the inflammatory state of a person's body. Polymorphisms in these specific enzymes will significantly increase the levels of inflammation in the body. By supplementing these enzyme deficiencies, the patient will effectively reduce inflammatory damage to the body.

AOC1	The SNP rs10156191 encodes a weaker form of the histamine degradation enzyme Amine Oxidase, Copper Containing 1 (AOC1). This mutation, Thr16Met, is predicted to produce an enzyme with less catalytic activity and associated higher levels of pro-inflammatory amines like histamine and putrescine.
ATG16L1	The ATG16L1 gene encodes a protein that is a vital component of a protein complex necessary for the cellular phenomena known as autophagy. Autophagy is the process of degrading and cleaning of inert debris of the cell. Weakness in autophagy leads to abnormal accumulation of cellular "garbage" that will eventually affect the cellular function and lead to autophagy related disease states in including many neurological and immunological diseases, DM Type 2 and fatty liver disease.
ATG5	Autophagy-related 5 protein (ATG5) is an important intracellular mediator of the autophagy response. ATG5 is involved in a wide range of "quality control" features inside the cell: autophagy vesicle formation, innate immune system signaling, consumption of damaged mitochondria, and apoptosis. Mutations in the ATG5 gene are associated with numerous neurological, immunological and endocrine syndromes.
DRD2	Dopamine receptor D2 is an important component of the neuroinflammation process. Activation of DRD2 signaling is thought to decrease TNFalpha release from inflammatory mast cells. Polymorphisms associated with decreased DRD2 signaling activity are predicted to lead to pro-inflammatory phenotypes.
FUT2	Fucosyltransferase 2 (FUT2) is responsible for producing specific sugar groups that are secreted by the intestinal cells into the bowel to attract "good bacteria" . Polymorphisms in this gene produce "poor secreter" status. Lack of these sugars allows for gut dysbiosis and a higher risk of inflammatory bowel disease.
HLA-DQA1	Major histocompatibility complex, DQ alpha 1 (HLA-DQA1) is a human gene responsible for a cell surface receptor essential to the function of the immune system. Patients with a polymorphism in this gene are at higher risk for auto-immune based inflammatory disease including Celiac disease, Crohn's, Ulcerative Colitis, and gluten sensitivity.
HLA-DQA2	Major histocompatibility complex, DQ alpha 2 (HLA-DQA2) is a human gene responsible for a cell surface receptor essential to the function of the immune system. Patients with a polymorphism in this gene are at higher risk for auto-immune based inflammatory disease including Celiac disease, Crohn's, Ulcerative Colitis, and gluten sensitivity.
HNMT	The HNMT gene encodes the histamine degradative enzyme, histamine N-methyltransferase. HNMT, in contrast to AOC1, requires the methyl donor S-adenosylmethionine and a complete methylation pathway for normal function. Polymorphisms in HNMT gene expression or protein coding are predicted to prolong the pro-inflammatory effects of histamine signaling.
HNMT:Thr105Ile	The HNMT gene encodes the histamine degradative enzyme, histamine N-methyltransferase. HNMT, in contrast to AOC1, requires the methyl donor S-adenosylmethionine and a complete methylation pathway for normal function. Polymorphisms in HNMT gene expression or protein coding are predicted to prolong the pro-inflammatory effects of histamine signaling.
NEUROTRANSMITTER	Neurotransmitters are chemicals that are used to produce specific effects in the nervous system. These specific neurotransmitter genomics assess a person's risk for anxiety, depression and dysphoria.
ADRA2A	ADRA2A (Adrenergic Receptor Alpha 2A) gene that determines sensitivity of the adrenergic nervous system response. Individuals with the G allele at this location predicted to be at higher risk of sugar-induced hyperactivity, and better response to ADHD treatment with typical pharmacological interventions.
COMT V158M	Catechol-O-methyltransferase (COMT) is one of several enzymes that degrade catecholamine neurotransmitters such as dopamine, epinephrine, and norepinephrine. COMT's main function is to inactivate neurotransmitters (dopamine, epinephrine, and norepinephrine) by the addition of a methyl group to the catecholamine. Normal COMT function allows people to rapidly reverse feelings of anxiety or depression. COMT (+/-) patients have sluggish ability to alter anxiety or depression episodes. COMT (+/+) patients are more prone to prolonged episodes of anxiety, depression and OCD.

## Disclaimers

### TESTING:

Testing Performed By: MB

### METHODOLOGY AND LIMITATIONS:

Testing for genetic variation/mutation on listed genes was performed using ProFlex PCR and Real-Time PCR with TaqMan® allele-specific probes on the QuantStudio 12K Flex. All genetic testing is performed by GX Sciences, 4150 Freidrich Lane, Ste H, Austin, TX. 78744. This test will not detect all the known alleles that result in altered or inactive tested genes. This test does not account for all individual variations in the individual tested. Test results do not rule out the possibility that this individual could be a carrier of other mutations/variations not detected by this gene mutation/variation panel. Rare mutations surrounding these alleles may also affect our detection of genetic variations. Thus, the interpretation is given as a probability. Therefore, this genetic information shall be interpreted in conjunction with other clinical findings and familial history for the administration of specific nutrients. Patients should receive appropriate genetic counseling to explain the implications of these test results. Details of assay performance and algorithms leading to clinical recommendations are available upon request. The analytical and performance characteristics of this laboratory developed test (LDT) were determined by GX Sciences' laboratory pursuant to Clinical Laboratory Improvement Amendments (CLIA) requirements.

CLIA #: 45D2144988 Laboratory Director: James Jacobson, PhD

### DISCLAIMER:

This test was developed and its performance characteristics determined by GX Sciences. It has not been cleared or approved by the FDA. The laboratory is regulated under CLIA and qualified to perform high-complexity testing. This test is used for clinical purposes. It should not be regarded as investigational or for research. rsIDs for the alleles being tested were obtained from the dbSNP database (Build 142).

### DISCLAIMER:

UND Result: If you have received the result Variant undetermined (UND) this indicates that we were not able to determine your carrier status based on your raw data. Please refer to the GX Sciences genetic knowledge database for more information: [https://www.gxsciences.com/kb\\_results.asp](https://www.gxsciences.com/kb_results.asp)

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# GX Sciences SNP References

## GASTROINTESTINAL SNP References

### MCM6

• Enattah, N. S. et al. Identification of a variant associated with adult-type hypolactasia. *Nat. Genet.* (2002). doi:10.1038/ng826 • Bersaglieri, T. et al. Genetic Signatures of Strong Recent Positive Selection at the Lactase Gene. *Am. J. Hum. Genet.* (2004). doi:10.1086/421051

## HEALTH PRECAUTIONS SNP References

### ACE

• Angiotensin-converting enzyme inhibition by perindopril in the treatment of cardiovascular disease. (PMID: 19379059) Brugs J.J. ... Simoons M.L.(Expert Rev Cardiovasc Ther 2009) • Triple pharmacological blockade of the renin-angiotensin-aldosterone system in nondiabetic CKD: an open-label crossover randomized controlled trial. (PMID: 18423812) Tylicki L. ... Rutkowski B.(Am. J. Kidney Dis. 2008) • Structural details on the binding of antihypertensive drugs captopril and enalaprilat to human testicular angiotensin I-converting enzyme. (PMID: 15236580) Natesh R. ... Acharya K.R.(Biochemistry 2004) • The ACE gene and human performance: 12 years on. Puthucherry Z1, Skipworth JR, Rawal J, Loosemore M, Van Someren K, Montgomery HE. *Sports Med.* 2011 Jun 1;41(6):433-48. doi: 10.2165/11588720-0. • Meta-analysis of genetic studies in ischemic stroke: thirty-two genes involving approximately 18,000 cases and 58,000 controls. (PMID: 15534175) Casas J.P. ... Sharma P.(Arch. Neurol. 2004)

## HYPERTENSION SNP References

### AGT

• Ahluwalia, Tarunveer Singh, Monica Ahuja, Taranjit Singh Rai, Harbir Singh Kohli, Anil Bhansali, Kamal Sud, and Madhu Khullar. 2009. "ACE Variants Interact with the RAS Pathway to Confer Risk and Protection against Type 2 Diabetic Nephropathy." *DNA and Cell Biology* 28 (3): 141–50. <https://doi.org/10.1089/dna.2008.0810>. • Anton, Raymond F, Gabor Orozci, Stephanie O'Malley, David Couper, Robert Swift, Helen M Pettinati, David Goldman, et al. 2008. "Pharmacogenomics." Edited by Yoshiaki Tsuji. *Nature Genetics* 16 (1). Public Library of Science: 268–78. <https://doi.org/10.1016/j.ejca.2015.06.122>.

## INFLAMMATORY SNP References

### AOC1

• McGrath, A. P. et al. Structure and inhibition of human diamine oxidase. *Biochemistry* 48, 9810–9822 (2009). • McGrath, A. P. et al. Structure and Inhibition of Human Diamine Oxidase - *Biochemistry* (ACS Publications). *Biochemistry* 48, 9810–22 (2009). • Schwelberger, H. G. The origin of mammalian plasma amine oxidases. in *Journal of Neural Transmission* 114, 757–762 (2007). • Solismaa, A. et al. Histaminergic gene polymorphisms associated with sedation in clozapine-treated patients. *Eur. Neuropsychopharmacol.* 27, 442–449 (2017).

### ATG16L1

• Stappenbeck, T. S. et al. Crohn disease: A current perspective on genetics, autophagy and immunity. *Autophagy* (2011). doi:10.4161/auto.7.4.13074 • Salem, M., Ammitzboell, M., Nys, K., Seidelin, J. B. & Nielsen, O. H. ATG16L1: A multifunctional susceptibility factor in crohn disease. *Autophagy* (2015). doi:10.1080/15548627.2015.1017187 • Giubb, D. M. et al. NOD2 and ATG16L1 polymorphisms affect monocyte responses in crohn's disease. *World J. Gastroenterol.* (2011). doi:10.3748/wjg.v17.i23.2829 • Usategui-Martin, R. et al. Polymorphisms in autophagy genes are associated with paget disease of bone. *PLoS One* (2015). doi:10.1371/journal.pone.0128984 • Csöngéi, V. et al. Interaction of the major inflammatory bowel disease susceptibility alleles in Crohn's disease patients. *World J. Gastroenterol.* (2010). doi:10.3748/wjg.v16.i2.176 • Messer, J. S. et al. The Crohn's disease: Associated ATG16L1 variant and Salmonella invasion. *BMJ Open* (2013). doi:10.1136/bmjopen-2013-002790 • Raju, D., Hussey, S. & Jones, N. L. Crohn disease ATG16L1 polymorphism increases susceptibility to infection with *Helicobacter pylori* in humans. *Autophagy* (2012). doi:10.4161/auto.21007 • Rosentul, D. C. et al. Role of autophagy genetic variants for the risk of *Candida* infections. *Med. Mycol.* (2014). doi:10.1093/mmy/my035 • Kuballa, P., Huett, A., Rioux, J. D., Daly, M. J. & Xavier, R. J. Impaired autophagy of an intracellular pathogen induced by a Crohn's disease associated ATG16L1 variant. *PLoS One* (2008). doi:10.1371/journal.pone.0003391 • Gazouli, M. et al. NOD2/CARD15, ATG16L1 and IL23R gene polymorphisms and childhood-onset of Crohn's disease. *World J. Gastroenterol.* (2010). doi:10.3748/wjg.v16.i14.1753 • Salem, M., Nielsen, O. H., Nys, K., Yazdanyar, S. & Seidelin, J. B. Impact of T300A Variant of ATG16L1 on antibacterial response, risk of culture positive infections, and clinical course of Crohn's disease. *Clin. Transl. Gastroenterol.* (2015). doi:10.1038/ctg.2015.47 • Begun, J. et al. Integrated Genomics of Crohn's Disease Risk Variant Identifies a Role for CLEC12A in Antibacterial Autophagy. *Cell Rep.* (2015). doi:10.1016/j.celrep.2015.05.045 • Cheng, J. F., Ning, Y. J., Zhang, W., Lu, Z. H. & Lin, L. T300A polymorphism of ATG16L1 and susceptibility to inflammatory bowel diseases: A meta-analysis. *World J. Gastroenterol.* (2010). doi:10.3748/wjg.v16.i10.1258 • Kabat, A. M. et al. The autophagy gene Atg16l1 differentially regulates Treg and TH2 cells to control intestinal inflammation. *Elife* (2016). doi:10.7554/eLife.12444 • Lassen, K. G. et al. Atg16L1 T300A variant decreases selective autophagy resulting in altered cytokine signaling and decreased antibacterial defense. *Proc. Natl. Acad. Sci.* (2014). doi:10.1073/pnas.1407001111 • Boada-Romero, E. et al. The T300A Crohn's disease risk polymorphism impairs function of the WD40 domain of ATG16L1. *Nat. Commun.* (2016). doi:10.1038/ncomms11821

### ATG5

• Anton, R. F. et al. Pharmacogenomics. *Nat. Genet.* 16, 268–278 (2008). • Martin, L. J. et al. Functional Variant in the Autophagy-Related 5 Gene Promotor is Associated with Childhood Asthma. *PLoS One* 7, e33454 (2012). • Yuan, J. et al. Polymorphisms in autophagy related genes and the coal workers' pneumoconiosis in a Chinese population. *Gene* 632, 36–42 (2017). • White, K. A. M. et al. Variants in autophagy-related genes and clinical characteristics in melanoma: a population-based study. *Cancer Med.* 5, 3336–3345 (2016).

### DRD2

• Sasabe, T., Furukawa, A., Matsusita, S., Higuchi, S. & Ishiura, S. Association analysis of the dopamine receptor D2 (DRD2) SNP rs1076560 in alcoholic patients. *Neurosci. Lett.* (2007). doi:10.1016/j.neulet.2006.10.064 • Anton, R. F. et al. Pharmacogenomics. *Nat. Genet.* (2008). doi:10.1016/j.ejca.2015.06.122 • Clarke, T. K. et al. The dopamine receptor D2 (DRD2) SNP rs1076560 is associated with opioid addiction. *Ann. Hum. Genet.* (2014). doi:10.1111/ahg.12046

### FUT2

• Kimura, K. et al. Diversification of transcriptional modulation: Large-scale identification and characterization of putative alternative promoters of human genes. *Genome Res.* (2006). doi:10.1101/gr.4039406 • Strausberg, R. L. et al. Generation and initial analysis of more than 15,000 full-length human and mouse cDNA sequences. *Proc. Natl. Acad. Sci. U. S. A.* (2002). doi:10.1073/pnas.242603899 • Koda, Y., Soejima, M., Wang, B. & Kimura, H. Structure and expression of the gene encoding secretor-type galactoside 2-alpha-L-fucosyltransferase (FUT2). *Eur. J. Biochem.* (1997). doi:10.1111/j.1432-1033.1997.t01-1-00750.x • Reguigne-Arnould, I. et al. Relative positions of two clusters of human ?-L-fucosyltransferases in 19q (FUT1–FUT2) and 19p (FUT6–FUT3–FUT5) within the microsatellite genetic map of chromosome 19. *Cytogenet. Genome Res.* (1995). doi:10.1159/000134098 • BALL, S. P. et al. The human chromosome 19 linkage group FUT1 (H), FUT2 (SE), LE, LU, PEPD, C3, APOC2, D19S7 and D19S9. *Ann. Hum. Genet.* (1991). doi:10.1111/j.1469-1809.1991.tb00417.x • FUT2 fucosyltransferase 2 (secretor status included) [Bos taurus (cattle)] - Gene - NCBI. *Current neurology and neuroscience reports.* Available at: <https://www.ncbi.nlm.nih.gov/gene/281175>.

## HLA-DQA1

• Kao, H. T. et al. Molecular analysis of the HLA class II genes in two DRw6-related haplotypes, DRw13 DQw1 and DRw14 DQw3. *J. Immunol.* (1989). • Schmidt, H., Williamson, D. & Ashley-Koch, A. HLA-DR15 haplotype and multiple sclerosis: A HuGE review. *American Journal of Epidemiology* (2007). doi:10.1093/aje/kwk118 • Todd, J. a, Fukui, Y., Kitagawa, T. & Sasazuki, T. The A3 allele of the HLA-DQA1 locus is associated with susceptibility to type 1 diabetes in Japanese. *Proc. Natl. Acad. Sci. U. S. A.* (1990). • Marsh, S. G. & Bodmer, J. G. HLA class II nucleotide sequences, 1992. *Immunogenetics* (1993). • Liu, C. P., Bach, F. H. & Wu, S. K. Molecular studies of a rare DR2/LD-5a/DQw3 HLA class II haplotype. Multiple genetic mechanisms in the generation of polymorphic HLA class II genes. *J Immunol* (1988). • Horn, G. T., Bugawan, T. L., Long, C. M., Manos, M. M. & Erlich, H. A. Sequence analysis of HLA class II genes from insulin-dependent diabetic individuals. *Hum. Immunol.* (1988). doi:10.1016/0198-8859(88)90034-1 • Schiffenbauer, J. et al. Complete sequence of the HLA DQ alpha and DQ beta cDNA from a DR5/DQw3 cell line. *J. Immunol.* (1987). • Jonsson, A.-K. et al. Class II genes of the human major histocompatibility complex. Comparisons of the DQ and DX ? and ? genes. *J. Biol. Chem.* (1987).

## HLA-DQA2

• HLA-DQA2 major histocompatibility complex, class II, DQ alpha 2 [Homo sapiens (human)] - Gene - NCBI. Current neurology and neuroscience reports. Available at: <https://www.ncbi.nlm.nih.gov/gene/3118>. • Khalil, I. et al. A combination of HLA-DQ beta Asp57-negative and HLA DQ alpha Arg52 confers susceptibility to insulin-dependent diabetes mellitus. *J. Clin. Invest.* (1990). doi:10.1172/JCI114569 • Khalil, I. et al. Trans-encoded DQ alpha beta heterodimers confer susceptibility to myasthenia gravis disease. *C.R.Acad.Sci.III* (1993). • Hall, M. A., Lanchbury, J. S. S., Bolsover, W. J., Welsh, K. I. & Ciclitira, P. J. Celiac disease is associated with an extended HLA-DR3 haplotype which includes HLA-DPw1. *Hum. Immunol.* (1990). doi:10.1016/0198-8859(90)90052-Q • Kwok, W. W. et al. Polymorphic DQ alpha and DQ beta interactions dictate HLA class II determinants of allo-recognition. *Journal Of Experimental Medicine* (1990). doi:10.1084/jem.171.1.85 • Sollid, L. M. Evidence for a primary association of celiac disease to a particular HLA-DQ alpha/beta heterodimer. *J. Exp. Med.* (1989). doi:10.1084/jem.169.1.345 • Wang, H. & He, R. HLA-DQA and DQB alleles contribute to susceptibility to insulin-dependent diabetes mellitus. *Chinese Med. Sci. J. = Chung-kuo i hsueh k'o hsueh tsa chih* (1993). • Bolsover, W. J., Hall, M. A., Vaughan, R. W., Welsh, K. I. & J. Ciclitira, P. A family study confirms that the HLA-DP associations with celiac disease are the result of an extended HLA-DR3 haplotype. *Hum. Immunol.* (1991). doi:10.1016/0198-8859(91)90012-X • Yu, L. & Sheehy, M. The cryptic HLA-DQA2 (DX alpha) gene is expressed in human B cell lines. *J Immunol.* (1991). • Olerup, O. & Hillert, J. HLA class II-associated genetic susceptibility in multiple sclerosis: a critical evaluation. *Tissue Antigens* (1991). doi:10.1111/j.1399-0039.1991.tb02029.x

## HNMT

• Reference SNP (refSNP) Cluster Report: rs12995000. National Center for Biotechnology Information Available at: [https://www.ncbi.nlm.nih.gov/SNP/snp\\_ref.cgi?rs=rs12995000](https://www.ncbi.nlm.nih.gov/SNP/snp_ref.cgi?rs=rs12995000). • Anton, R. F. et al. Pharmacogenomics. *Nat. Genet.* (2008). doi:10.1016/j.ejca.2015.06.122 • Keeling, B. H. et al. Histamine N-methyltransferase Thr105Ile is not associated with Parkinson's disease or essential tremor. *Parkinsonism Relat. Disord.* (2010). doi:10.1016/j.parkrelidis.2009.08.011

## NEUROTRANSMITTER SNP References

### ADRA2A

• Lochman, J., Balcar, V. J., Š?astný, F. & Šerý, O. Preliminary evidence for association between schizophrenia and polymorphisms in the regulatory Regions of the ADRA2A, DRD3 and SNAP-25 Genes. *Psychiatry Res.* 205, 7–12 (2013). • Comings, D. E. et al. Additive effect of three noradrenergic genes (ADRA2a, ADRA2C, DBH) on attention-deficit hyperactivity disorder and learning disabilities in Tourette syndrome subjects. *Clin. Genet.* 55, 160–172 (1999). • Kumar, S. et al. Significant role of ADRB3 rs4994 towards the development of coronary artery disease. *Coron. Artery Dis.* 25, 29–34 (2014).

### COMT

• Bonifácio, M. J., Palma, P. N., Almeida, L. & Soares-Da-Silva, P. Catechol-O-methyltransferase and its inhibitors in Parkinson's disease. *CNS Drug Reviews* (2007). doi:10.1111/j.1527-3458.2007.00020.x • Golan, D. E., Armstrong, E. J. & Armstrong, A. W. Principles of pharmacology: the pathophysiologic basis of drug therapy. (Wolters Kluwer Health, 2017). • Grossman, M. H., Emanuel, B. S. & Budarf, M. L. Chromosomal mapping of the human catechol-O-methyltransferase gene to 22q11.1?q11.2. *Genomics* (1992). doi:10.1016/0888-7543(92)90316-K • Diamond, A., Briand, L., Fossella, J. & Gehlbach, L. Genetic and Neurochemical Modulation of Prefrontal Cognitive Functions in Children. *Am. J. Psychiatry* (2004). doi:10.1176/appi.ajp.161.1.125 • Robinson, S., Goddard, L., Dritschel, B., Wisley, M. & Howlin, P. Executive functions in children with Autism Spectrum Disorders. *Brain Cogn.* (2009). doi:10.1016/j.bandc.2009.06.007 • Bruder, G. E. et al. Catechol-O-methyltransferase (COMT) genotypes and working memory: Associations with differing cognitive operations. *Biol. Psychiatry* (2005). doi:10.1016/j.biopsych.2005.05.010 • Chen, J. et al. Functional analysis of genetic variation in catechol-O-methyltransferase (COMT): Effects on mRNA, protein, and enzyme activity in postmortem human brain. *Am. J. Hum. Genet.* (2004). doi:10.1086/425589 • Lotta, T. et al. Kinetics of Human Soluble and Membrane-Bound Catechol O-Methyltransferase: A Revised Mechanism and Description of the Thermolabile Variant of the Enzyme. *Biochemistry* (1995). doi:10.1021/bi00013a008 • Stein, M. B., Fallin, M. D., Schork, N. J. & Gelernter, J. COMT polymorphisms and anxiety-related personality traits. *Neuropsychopharmacology* (2005). doi:10.1038/sj.npp.1300787 • Ulmanen, I. et al. Expression and intracellular localization of catechol O-methyltransferase in transfected mammalian cells. *Eur. J. Biochem.* (1997). doi:10.1111/j.1432-1033.1997.0452a.x • Axelrod, J. O-methylation of epinephrine and other catechols in vitro and in vivo. *Science* (80-. ). (1957). doi:10.1126/science.126.3270.400 • Tai, C. H. & Wu, R. M. Catechol-O-methyltransferase and Parkinson's disease. *Acta Medica Okayama* (2002). • Wichers, M. et al. The catechol-O-methyl transferase Val158Met polymorphism and experience of reward in the flow of daily life. *Neuropsychopharmacology* (2008). doi:10.1038/sj.npp.1301520