GX SCIENCES
Innovative Genomic Solutions

# Gene Comprehensive Nutrigenomic Report

Accession Number: ##########
Report Generated: July 25, 2018
Specimen Received: ##/##/####
Created For: ###### ######
DOB: ##/##/####
Male

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.

GX Sciences | 844-258-5564 | www.GXSciences.com
Lab | 4150 Freidrich Lane, Ste H | Austin, TX 78744
Laboratory Director: James W. Jacobson, Ph.D

# ###### ###### – 10 – Male

(-/-) 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 |
|---|---|---|---|---|---|---|---|
| Immune \| Auto Immune \| Inflammatory | | | | | | | |
| Inflammation Cellular | | | | | | | |
| rs10402876 | C3 | +/- | Anti-Inflammatory Therapy: Curcumin, Omega 3s, Resveratrol, Quercetin, Low Dose Naltrexone (LDN), CBD Oil | **CBD Oil, PEA Soothe Support™ , Prescription Low Dose Naltrexone (LDN)** | | Consider Low Inflammatory Diet | Consider Pregnenolone, Cortisol, Progesterone, Testosterone, T cell profile, Sed Rate, ANA, C Reactive Protein, Routine Thyroid Panel, Candida Titer, EBV Titer, Food Allergy Panel, Environmental Allergy Testing |
| rs2569191 | CD14 | +/- | | | | | |
| rs2069812 | IL5 | +/- | | | | | |
| rs1800795 | IL6 | +/+ | | | | | |
| rs1800925 | IL13 | -/- | | | | | |
| rs10181656 | STAT4 | +/+ | | | | | |
| rs361525 | TNF | -/- | | | | | |
| rs1800629 | TNF | -/- | | | | | |
| rs231775 | CTLA4 | +/- | | | | | |
| rs3761847 | TRAF 1 | +/- | | | | | |
| rs243324 | SOCS 1 | +/+ | | | | | |
| rs11209026 | IL23R | -/- | | | | | |
| rs12722489 | IL2RA | -/- | | | | | |
| rs1076560 | DRD2 | -/- | Increased Efficacy of Naltrexone | | | | |

GX Sciences | 844-258-5564 | www.GXSciences.com
Lab | 4150 Freidrich Lane, Ste H | Austin, TX 78744
Laboratory Director: James W. Jacobson, Ph.D

# ###### ###### – 10 – Male

(-/-) 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 |
|---|---|---|---|---|---|---|---|
| Immune \| Auto Immune \| Inflammatory | | | | | | | |
| Autophagy Consideration | | | | | | | |
| rs510432 | ATG5 | +/- | Curcumin, Lithium Orotate, D-Chiro-Inositol, Catechins, Resveratrol, Caffeine, 12 Hour Fasting | **N.A.S. Enhancer™ (NRF2\|Autophagy\|SOD Support), Metabolic Stimulator™** | | Calorie Restriction, 12-15 Hour Fasting | Routine Blood Sugar, Insulin and Hb A1c |
| rs10210302 | ATG16L1 | +/- | | | | | |
| rs2066845 | NOD2 CARD15 | -/- | Increase susceptibility to bacterial GI infections and Crohn's | | | | |
| rs2241880 | ATG16L1 | +/- | | | | | |
| Detoxification | | | | | | | |
| rs819147 | AHCY | -/- | N-Acetyl Cysteine (NAC), Glutathione | | | | |
| rs1021737 | CTH | -/- | | | | | |
| rs1695 | GSTP1 I105V | -/- | Glutathione | | | | |

GX Sciences | 844-258-5564 | www.GXSciences.com
Lab | 4150 Freidrich Lane, Ste H | Austin, TX 78744
Laboratory Director: James W. Jacobson, Ph.D

## ###### ###### – 10 – Male

(-/-) 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 |
|---|---|---|---|---|---|---|---|
| Immune \| Auto Immune \| Inflammatory | | | | | | | |
| Inflammation Environmental | | | | | | | |
| rs10156191 | AOC1 | +/- | Poor Ability To Break Down External Histamine | | | | |
| rs11558538 | HNMT | -/- | | | | | |
| rs12995000 | HNMT | -/- | | | | | |
| rs492602 | FUT2 | +/+ | Probiotics Needed | **Biotic Blend Pro™, Biotic Multi Blend Pro™, Probiotic Boost Daily™, Biotic Boost Chews For Kids™** | | | Consider Microbiome Testing If GI Inflammation Present |
| rs2248814 | NOS2 | +/- | Anti-Infectives, Beta Glucans | | | | |
| rs2187668 | HLA DQA1 | -/- | High Risk of Gluten Based Issues | | | | |
| rs2858331 | HLA DQA2 | +/- | | | | | |
| rs9275224 | HLA DRB2 | +/- | High Reactivity To Mold / Fungi, CBD Oil, Low Dose Naltrexone (LDN) | | | | |
| rs660895 | HLA DRB1 | -/- | | | | | |

# Summary for Inflammatory / Auto-Immune

## Highly Recommended Therapeutics/Neurobiologix Formulas

- **CBD Oil, PEA Soothe Support™, Prescription Low Dose Naltrexone (LDN)**
- **N.A.S. Enhancer™ (NRF2|Autophagy|SOD Support), Metabolic Stimulator™**
- **Biotic Blend Pro™, Biotic Multi Blend Pro™, Probiotic Boost Daily™, Biotic Boost Chews For Kids™**

## Lifestyle Recommendations

- Consider Low Inflammatory Diet
- Calorie Restriction, 12-15 Hour Fasting

## Laboratory Recommendations

- Consider Pregnenolone, Cortisol, Progesterone, Testosterone, T cell profile, Sed Rate, ANA, C Reactive Protein, Routine Thyroid Panel, Candida Titer, EBV Titer, Food Allergy Panel, Environmental Allergy Testing
- Routine Blood Sugar, Insulin and Hb A1c
- Consider Microbiome Testing If GI Inflammation Present

# Gene Information Key

| rsID | Gene | "-" variant | "+" variant |
|---|---|---|---|
| rs10156191 | AOC1 | C | T |
| rs10181656 | STAT4 | C | G |
| rs10210302 | ATG16L1 | C | T |
| rs1021737 | CTH | G | T |
| rs10402876 | C3 | G | C |
| rs1076560 | DRD2 | C | A |
| rs11209026 | IL23R | G | A |
| rs1138272 | GSTP:A114V | C | T |
| rs11558538 | HNMT | C | T |
| rs12722489 | IL2RA | C | T |
| rs12995000 | HNMT | C | T |
| rs1695 | GSTP1:I105V | A | G |
| rs1800629 | TNF | G | A |
| rs1800795 | IL6 | G | C |
| rs1800925 | IL13 | C | T |
| rs2066845 | NOD2/CARD15 | G | C |
| rs2069812 | IL5 | A | G |

| rsID | Gene | "-" variant | "+" variant |
|---|---|---|---|
| rs2187668 | HLA-DQA1 | C | T |
| rs2241880 | ATG16L1 | A | G |
| rs2241880 | ATG16L1 | A | G |
| rs2248814 | NOS2 | G | A |
| rs231775 | CTLA4 | A | G |
| rs243324 | SOCS-1 | G | A |
| rs2569191 | CD14 | T | C |
| rs2858331 | HLA-DQA2 | A | G |
| rs361525 | TNF | G | A |
| rs3761847 | TRAF-1 | A | G |
| rs492602 | FUT2 | A | G |
| rs510432 | ATG5 | C | T |
| rs660895 | HLA-DRB1 | A | G |
| rs819147 | AHCY | T | C |
| rs9275224 | HLA-DRB2 | G | A |

# Definitions

| | |
|---|---|
| DETOXIFICATION | Detoxification enzymes are responsible for clearing environmental chemicals and metabolites from our body. Accumulation of these chemicals and by-products can damage intracellular biochemical functions. Alterations in these systems can have a significant negative effect on the nervous system and immune systems functions. These polymorphisms can result in decreased "quality of life" and even decreased "life-span". |
| AHCY | Adenosylhomocysteinase (AHCY) is an enzyme that breaks down S-adenosylhomocysteine (SAH) to homocysteine and adenosine. Polymorphisms in this gene will lead to lower levels of homocysteine and glutathione. |
| CTH | Glutathione production is dependent on the function of the enzyme cystathionine gamma-lyase (CTH). CTH converts cystathionine to cysteine. Individuals with mutations in the CTH gene are predicted to have decreased glutathione-mediated detoxification. |
| GSTP1 | Glutathione S-transferases (GSTs) are a family of enzymes that play an important role in detoxification. The glutathione S-transferase pi gene (GSTP1) functions in chemical clearance and anti-inflammatory properities. High concentration of GST-p are found in the skin, lungs, sinuses, bladder and the intestinal tract. Polymorphisms of this enzyme allow for increased inflammatory activity in these areas that include eczema, asthma, chronic sinusitis, IBS, "leaky" gut and interstitial cystitis. |
| 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. |
| C3 | Essential for the immune response, C3 is a protein involved in initiation of the complement system. C3 polymorphisms are associated with susceptibility to asthma and other inflammatory disorders. |
| CD14 | The CD14 protein is a macrophage cell surface receptor that binds bacterial cell wall components. As one of the initiators of the innate immune response, fully functional CD14 is necessary for normal response to potential pathogens. Mutations in the CD14 gene are associated with susceptibility to asthma and other allergen-mediated inflammatory processes. |
| CTLA4 | Cytotoxic T-lymphocyte Associated protein 4 (CTLA4) is an important inhibitor of T-cell activity: CTLA4 is part of the signaling cascade that turns off overactive T cells. Mutations in the gene that encodes CTLA4 are associated with a host of diseases characterized by a heightened immune state. |
| 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. |
| HLA-DRB1 | Human leukocyte antigen DRB1 (HLA-DRB1) is an important mediator of the adaptive immune system. HLA-DRB1 protein "presents" antigens from invading pathogens to other cells in the immune system. Mutations in this gene are associated with higher risk of auto-immunity and other chronic inflammatory diseases. |

| | |
|---|---|
| HLA-DRB2 | Human leukocyte antigen DRB2 (HLA-DRB2) is a cell surface receptor involved in mediating the adaptive immune response. Mutations in HLA-DRB2 are associated with susceptibility to chronic inflammation and decreased ability to recover from toxic mold exposure. |
| 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. |
| IL13 | IL13 (Interleukin 13) is a member of the interleukin family of chemical messengers of the immune system. Polymorphisms in this gene are associated with changes in IL13 gene expression and increase the risk of more severe inflammatory responses to allergens. |
| IL23R | A/A and A/G genotypes at rs11209026, a polymorphism in the Interleukin 23 Receptor gene (IL23R), provide a protective effects against Crohn's disease. |
| IL2RA | Polymorphisms in a non-protein coding region of the Interleukin 2 Receptor subtype A (IL2RA) are associated with increased risk of multiple sclerosis in some populations. |
| IL5 | The protein product of the Interleukin 5 gene (IL5) is important for normal development of B lymphocytes and eosinophils (a pro-inflammatory white blood cell). Inactivating mutations in the IL5 gene are associated with susceptibility to certain viral infections and increased aggression of inflammatory response. These polymorphisms are also associated with increased aggression of allergies, asthma and eosinophilia. |
| IL6 | Interleukin 6, IL6, is an important pro-inflammatory cytokine. Polymorphisms in this gene leads to a more aggressive inflammatory response. Patients with IL-6 mutations require assistance with inflammatory control. |
| NOD2/CARD15 | NOD2 is a protein that integrates extracellular sensing of bacterial cell wall components with intracellular pro-inflammatory signaling to the nucleus. Mutations in the NOD2 gene are associated with poor ability to recognize abnormal bacterial pathogens in the intestinal tract and increased risk of intestinal dysbiosis. |
| NOS2 | Nitric Oxide Synthanse 2 (NOS2) is responsible for producing nitric oxide, a biologic mediator used by the nervous system, immune system and in blood vessel function. Polymorphisms in this enzyme can cause reduced immune system function, exercise intolerance and fatigue. |
| SOCS1 | Suppressor of Cytokine Signaling 1 is an intracellular protein that is a member of the STAT (Signal Transducer and Activator of Transcription) family that is necessary to curb pro-inflammatory cytokine signaling. Mutations in SOCS1 are predicted to prolong inflammatory responses, thereby requiring assistance with inflammatory control. |
| STAT4 | The Signal Transducer and Activator of Transcription 4 (STAT4) gene encodes a transcription factor that responds to extracellular growth factors and cytokines. Mutations in the STAT4 gene are associated with inflammatory disorders like lupus and rheumatoid arthritis. |
| TNF | Tumor necrosis factor, TNF, is an important pro-inflammatory signaling molecule. Polymorphisms in the protein coding part of this gene are associated with more severe pro-inflammatory responses and require supplementation for inflammatory control. |
| TRAF-1 | TRAF1 (TNF Receptor Activation Factor 1) is produced by T cells and functions as an “off switch” for Toll like receptors and Janus Kinase. Polymorphisms of this gene are associated with chronic inflammation and can be associated with chronic Epstein Barr infections. |

# Disclaimers

**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

**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

**DISCLAIMER:**

Report contents and report recommendations are created and approved by GX Sciences. Sole responsibility for the proper use of the information on the GX Sciences report rests with the user, or those professionals with whom the user may consult. Nutrigenomic Testing and Neurobiologix Dietary Supplements are not "Designated Health Services" covered by Medicare or Medicaid and may not be reimbursed under any state or Federal health care program.

**DISCLAIMER:**

These products are not approved by the Food and Drug Administration and are not intended to diagnose, treat, cure or prevent a disease. These recommendations are for report purposes only and an individual is not required to use such products. These are recommendations only and do not replace the advisement of your own healthcare practitioner.

# GX Sciences SNP References

## DETOXIFICATION SNP References

### AHCY

• Motzek, A., Kneževi?, J., Switzeny, O. J., Cooper, A., Baric, I., Beluzi?, R., … Zechner, U. (2016). Abnormal hypermethylation at imprinting control regions in patients with S-adenosylhomocysteine hydrolase (AHCY) deficiency. PLoS ONE, 11(3). https://doi.org/10.1371/journal.pone.0151261 • Vugrek, O., Beluži?, R., & Naki?, N. (2009). S-adenosylhomocysteine hydrolase (AHCY) deficiency: Two novel mutations with lethal outcome. Human Mutation, 30(4). https://doi.org/10.1002/humu.20985

### CTH

• Huezo-Diaz, P et al. 2012. “Association of Cth Genetic Variant with Veno-Occlusive Disease in Children Receiving Intravenous Busulfan before Hematopoietic Stem Cell Transplantation.” Blood 120(21). http://www.embase.com/search/results?subaction=viewrecord&from=export&id=L70963192%5Cnhttp://abstracts.hematologylibrary.org/cgi/content/abstract/120/21/3025?maxtoshow=&hits=80&RESULTFORMAT=&searchid=1&FIRSTINDEX=1280&displaysectionid=Poster+Session&fdate. • Wang, J., & Hegele, R. a. (2003). Genomic basis of cystathioninuria (MIM 219500) revealed by multiple mutations in cystathionine gamma-lyase (CTH). Human Genetics, 112(4), 404–408. https://doi.org/10.1007/s00439-003-0906-8

### GSTP1

• Strange RC, Fryer AA (1999). "The glutathione S-transferases: influence of polymorphism on cancer susceptibility". IARC Sci. Publ. (148): 231–49. PMID 10493261. • Gerhard DS, Wagner L, Feingold EA, et al. (2004). "The status, quality, and expansion of the NIH full-length cDNA project: the Mammalian Gene Collection (MGC).". Genome Res. 14 (10B): 2121–7.PMID 15489334 • Buch SC, Notani PN, Bhisey RA (2002). "Polymorphism at GSTM1, GSTM3 and GSTT1 gene loci and susceptibility to oral cancer in an Indian population.". Carcinogenesis 23 (5): 803–7. PMID 12016153 • Sekine I, Minna JD, Nishio K, Tamura T, Saijo N (2006). "A literature review of molecular markers predictive of clinical response to cytotoxic chemotherapy in patients with lung cancer". J Thorac Oncol 1 (1): 31–7.doi:10.1097/01243894-200601000-00008. PMID 17409824. • Kellen E, Hemelt M, Broberg K, Golka K, Kristensen VN, Hung RJ, Matullo G, Mittal RD, Porru S, Povey A, Schulz WA, Shen J, Buntinx F, Zeegers MP, Taioli E (2007). "Pooled analysis and meta-analysis of the glutathione S-transferase P1 Ile 105Val polymorphism and bladder cancer: a HuGE-GSEC review". Am. J. Epidemiol. 165 (11): 1221–30. doi:10.1093/aje/kwm003. PMID 17404387.

## INFLAMMATORY SNP References

### AOC1

• McGrath, A. P., Hilmer, K. M., Collyer, C. A., Shepard, E. M., Elmore, B. O., Brown, D. E., … Guss, J. M. (2009). Structure and inhibition of human diamine oxidase. Biochemistry, 48(41), 9810–9822. https://doi.org/10.1021/bi9014192 • McGrath, A. P., Hilmer, K. M., Collyer, C. A., Shepard, E. M., Elmore, B. O., Brown, D. E., … Guss, J. M. (2009). Structure and Inhibition of Human Diamine Oxidase - Biochemistry (ACS Publications). Biochemistry, 48(41), 9810–9822. https://doi.org/10.1021/bi9014192 • Solismaa, A., Kampman, O., Lyytikäinen, L. P., Seppälä, N., Viikki, M., Mononen, N., … Leinonen, E. (2017). Histaminergic gene polymorphisms associated with sedation in clozapine-treated patients. European Neuropsychopharmacology, 27(5), 442–449. https://doi.org/10.1016/j.euroneuro.2017.03.009 • Schwelberger, H. G. (2007). The origin of mammalian plasma amine oxidases. In Journal of Neural Transmission (Vol. 114, pp. 757–762). https://doi.org/10.1007/s00702-007-0684-x

### ATG16L1

• Role of autophagy genetic variants for the risk of Candida infections Diana C. Rosentul, Theo S. Plantinga, Marius Farcas, Marije Oosting, Omar J.M. Hamza, William K. Scott, Barbara D. Alexander, John C. Yang, Gregory M. Laird, Leo A.B. Joosten, Jos W. M. van der Meer, John R. Perfect, Bart-Jan Kullberg, Andre J.A.M. van der Ven, Melissa D. Johnson, Mihai G. Netea Med Mycol. 2014 May; 52(4): 333–341. Published online 2014 Apr 8.doi: 10.1093/mmy/myt035 • The T300A Crohn's disease risk polymorphism impairs function of the WD40 domain of ATG16L1 Emilio Boada-Romero, Inmaculada Serramito-Gómez, María P. Sacristán, David L. Boone, Ramnik J. Xavier, Felipe X. Pimentel-Muiños Nat Commun. 2016; 7: 11821. Published online 2016 Jun 8. doi: 10.1038/ncomms11821 • Atg16L1 T300A variant decreases selective autophagy resulting in altered cytokine signaling and decreased antibacterial defense Kara G. Lassen, Petric Kuballa, Kara L. Conway, Khushbu K. Patel, Christine E. Becker, Joanna M. Peloquin, Eduardo J. Villablanca, Jason M. Norman, Ta-Chiang Liu, Robert J. Heath, Morgan L. Becker, Lola Fagbami, Heiko Horn, Johnathan Mercer, Omer H. Yilmaz, Jacob D. Jaffe, Alykhan F. Shamji, Atul K. Bhan, Steven A. Carr, Mark J. Daly, Herbert W. Virgin, Stuart L. Schreiber, Thaddeus S. Stappenbeck, Ramnik J. Xavier Proc Natl Acad Sci U S A. 2014 May 27; 111(21): 7741–7746. Published online 2014 May 12. doi: 10.1073/pnas.1407001111 • The autophagy gene Atg16l1 differentially regulates Treg and TH2 cells to control intestinal inflammation Agnieszka M Kabat, Oliver J Harrison, Thomas Riffelmacher, Amin E Moghaddam, Claire F Pearson, Adam Laing, Lucie Abeler-Dörner, Simon P Forman, Richard K Grencis, Quentin Sattentau, Anna Katharina Simon, Johanna Pott, Kevin J Maloy eLife. 2016; 5: e12444. Published online 2016 Feb 24. doi: 10.7554/eLife.12444 • T300A polymorphism of ATG16L1 and susceptibility to inflammatory bowel diseases: A meta-analysis Jia-Fei Cheng, Yue-Ji Ning, Wei Zhang, Zong-Hai Lu, Lin Lin World J Gastroenterol. 2010 Mar 14; 16(10): 1258–1266. Published online 2010 Mar 14. doi: 10.3748/wjg.v16.i10.1258 • ATG16L1: A multifunctional susceptibility factor in Crohn disease Mohammad Salem, Mette Ammitzboell, Kris Nys, Jakob Benedict Seidelin, Ole Haagen Nielsen Autophagy. 2015 Apr; 11(4): 585–594. Published online 2015 Apr 23. doi: 10.1080/15548627.2015.1017187 • The Crohn's disease: associated ATG16L1 variant and Salmonella invasion Jeannette S Messer, Stephen F Murphy, Mark F Logsdon, James P Lodolce, Wesley A Grimm, Sarah J Bartulis, Tiphanie P Vogel, Melisa Burn, David L Boone BMJ Open. 2013; 3(6): e002790. Published online 2013 Jun 4. doi: 10.1136/bmjopen-2013-002790 • NOD2 and ATG16L1 polymorphisms affect monocyte responses in Crohn's disease Dylan M Glubb, Richard B Gearry, Murray L Barclay, Rebecca L Roberts, John Pearson, Jacqui I Keenan, Judy McKenzie, Robert W Bentley World J Gastroenterol. 2011 Jun 21; 17(23): 2829–2837. Published online 2011 Jun 21. doi: 10.3748/wjg.v17.i23.2829 • Integrated Genomics of Crohn's Disease Risk Variant Identifies a Role for CLEC12A in Antibacterial Autophagy Jakob Begun, Kara G. Lassen, Humberto B. Jijon, Leigh A. Baxt, Gautam Goel, Robert J. Heath, Aylwin Ng, Jenny M. Tam, Szu-Yu Kuo, Eduardo J. Villablanca, Lola Fagbami, Marije Oosting, Vinod Kumar, Monica Schenone, Steven A. Carr, Leo A.B. Joosten, Jatin M. Vyas, Mark J. Daly, Mihai G. Netea, Gordon D. Brown, Cisca Wijmenga, Ramnik J. Xavier Cell Rep. 2015 Jun 30; 11(12): 1905–1918. Published online 2015 Jun 18. doi: 10.1016/j.celrep.2015.05.045 • Polymorphisms in Autophagy Genes Are Associated with Paget Disease of Bone Ricardo Usategui-Martín, Judith García-Aparicio, Luis Corral-Gudino, Ismael Calero-Paniagua, Javier Del Pino-Montes, Rogelio González Sarmiento PLoS One. 2015; 10(6): e0128984. Published online 2015 Jun 1. doi: 10.1371/journal.pone.0128984 • Impact of T300A Variant of ATG16L1 on Antibacterial Response, Risk of Culture Positive Infections, and Clinical Course of Crohn's Disease Mohammad Salem, Ole Haagen Nielsen, Kris Nys, Shiva Yazdanyar, Jakob Benedict Seidelin Clin Transl Gastroenterol. 2015 Nov; 6(11): e122. Published online 2015 Nov 5. doi: 10.1038/ctg.2015.47 • NOD2/CARD15, ATG16L1 and IL23R gene polymorphisms and childhood-onset of Crohn's disease Maria Gazouli, Ioanna Pachoula, Ioanna Panayotou, Gerassimos Mantzaris, George Chrousos, Nicholas P Anagnou, Eleftheria Roma-Giannikou World J Gastroenterol. 2010 Apr 14; 16(14): 1753–1758. Published online 2010 Apr 14. doi: 10.3748/wjg.v16.i14.1753 • Impaired Autophagy of an Intracellular Pathogen Induced by a Crohn's Disease Associated ATG16L1 Variant Petric Kuballa, Alan Huett, John D. Rioux, Mark J. Daly, Ramnik J. Xavier PLoS ONE. 2008; 3(10): e3391. Published online 2008 Oct 13. doi: 10.1371/journal.pone.0003391 • Crohn disease ATG16L1 polymorphism increases susceptibility to infection with Helicobacter pylori in humans Deepa Raju, Séamus Hussey, Nicola L. Jones Autophagy. 2012 Sep 1; 8(9): 1387–1388. doi: 10.4161/auto.21007 • Crohn disease: A current perspective on genetics, autophagy and immunity Thaddeus S. Stappenbeck, John D. Rioux, Atsushi Mizoguchi, Tatsuya Saitoh, Alan Huett, Arlette Darfeuille-Michaud, Tom Wileman, Noboru Mizushima, Simon Carding, Shizuo Akira, Miles Parkes, Ramnik J. Xavier Autophagy. 2011 Apr 1; 7(4): 355–374. doi: 10.4161/auto.7.4.13074 • Interaction of the major inflammatory bowel disease susceptibility alleles in Crohn's disease patients Veronika Csöngei, Luca Járomi, Enik? Sáfrány, Csilla Sipeky, Lili Magyari, Bernadett Faragó, Judit Bene, Noémi Polgár, Lilla Lakner, Patrícia Sarlós, Márta Varga, Béla Melegh World J Gastroenterol. 2010 Jan 14; 16(2): 176–183. Published online 2010 Jan 14. doi: 10.3748/wjg.v16.i2.176

### ATG5

• Anton, Raymond F et al. 2008. “Pharmacogenomics” ed. Yoshiaki Tsuji. Nature Genetics 16(1): 268–78. http://www.ncbi.nlm.nih.gov/pubmed/26671228 (May 14, 2018). • White, Kirsten A M et al. 2016. “Variants in Autophagy-Related Genes and Clinical Characteristics in Melanoma: A Population-Based Study.” Cancer Medicine 5(11): 3336–45. http://www.ncbi.nlm.nih.gov/pubmed/27748080. • Martin, Lisa J et al. 2012. “Functional Variant in the Autophagy-Related 5 Gene Promotor Is Associated with Childhood Asthma” ed. Dominik Hartl. PLoS ONE 7(4): e33454. http://dx.plos.org/10.1371/journal.pone.0033454. • Yuan, Jiali et al. 2017. “Polymorphisms in Autophagy Related Genes and the Coal Workers' Pneumoconiosis in a Chinese Population.” Gene 632: 36–42. http://www.ncbi.nlm.nih.gov/pubmed/28844669.

### C3

• Apolipoprotein C3 gene variants and the risk of coronary heart disease: A meta-analysis.Li Y, Li C, Gao J.Meta Gene. 2016 Apr 23;9:104-9. doi: 10.1016/j.mgene.2016.04.004. eCollection 2016 Sep. Review. • Polymorphisms in complement genes and risk of preeclampsia in Taiyuan, China.Wu W, Yang H, Feng Y, Zhang P, Li S, Wang X, Peng T, Wang F, Xie B, Guo P, Li M, Wang Y, Zhao N, Wang D, Wang S, Zhang Y.Inflamm Res. 2016 Oct;65(10):837-45. doi: 10.1007/s00011-016-0968-4. Epub 2016 Jul 12. • Interactions of Environmental Factors and APOA1-APOC3-APOA4-APOA5 Gene Cluster Gene Polymorphisms with Metabolic Syndrome.Wu Y, Yu Y, Zhao T, Wang S, Fu Y, Qi Y, Yang G, Yao W, Su Y, Ma Y, Shi J, Jiang J, Kou C.PLoS One. 2016 Jan 29;11(1):e0147946. doi: 10.1371/journal.pone.0147946. eCollection 2016. • Erratum to: C3 Polymorphism Influences Circulating Levels of C3, ASP and Lipids in Schizophrenic Patients.Nsaibia MJ, Lapointe M, Mabrouk H, Douki W, Gaha L, Pérusse L, Bouchard C, Jrad BB, Cianflone K.Neurochem Res. 2016 Apr;41(4):944. doi: 10.1007/s11064-016-1866-4. No abstract available. • Serological and Genetic Evidence for Altered Complement System Functionality in Systemic Lupus Erythematosus: Findings of the GAPAID Consortium.Prechl J,

Papp K, Hérincs Z, Péterfy H, Lóránd V, Szittner Z, Estonba A, Rovero P, Paolini I, Del Amo J, Uribarri M, Alcaro MC, Ruiz-Larrañaga O, Migliorini P, Czirják L.PLoS One. 2016 Mar 7;11(3):e0150685. doi: 10.1371/journal.pone.0150685. eCollection 2016. • Association of polymorphisms in complement component 3 with age-related macular degeneration in an Iranian population.Bonyadi M, Mohammadian T, Jabbarpoor Bonyadi MH, Fotouhi N, Soheilian M, Javadzadeh A, Moein H, Yaseri M.Ophthalmic Genet. 2016 Mar 30:1-6. [Epub ahead of print] • Replication of association of the apolipoprotein A1-C3-A4 gene cluster with the risk of gout.Rasheed H, Phipps-Green AJ, Topless R, Smith MD, Hill C, Lester S, Rischmueller M, Janssen M, Jansen TL, Joosten LA, Radstake TR, Riches PL, Tausche AK, Lioté F, So A, van Rij A, Jones GT, McCormick SP, Harrison AA, Stamp LK, Dalbeth N, Merriman TR.Rheumatology (Oxford). 2016 Aug;55(8):1421-30. doi: 10.1093/rheumatology/kew057. Epub 2016 Apr 18. • Rare Genetic Variants Associated With Development of Age-Related Macular Degeneration.Saksens NT, Geerlings MJ, Bakker B, Schick T, Daha MR, Fauser S, Boon CJ, de Jong EK, Hoyng CB, den Hollander AI.JAMA Ophthalmol. 2016 Mar;134(3):287-93. doi: 10.1001/jamaophthalmol.2015.5592.

## CD14

• Association between CD14 Gene C-260T Polymorphism and Inflammatory Bowel Disease: A Meta-Analysis Zhengting Wang, Jiajia Hu, Rong Fan, Jie Zhou, Jie Zhong PLoS One. 2012; 7(9): e45144. Published online 2012 Sep 26. doi: 10.1371/journal.pone.0045144 • Clinical application of human ?-defensin and CD14 gene polymorphism in evaluating the status of chronic inflammation Wings TY Loo, Lan-jun Bai, Chang-bin Fan, Yuan Yue, Yi-ding Dou, Min Wang, Hao Liang, Mary NB Cheung, Louis WC Chow, Jin-le Li, Ye Tian, Liu Qing J Transl Med. 2012; 10(Suppl 1): S9. Published online 2012 Sep 19. doi: 10.1186/1479-5876-10-S1-S9 • Genetic association between inflammatory genes (IL-1?, CD14, LGALS2, PSMA6) and risk of ischemic stroke: A meta-analysis Shubham Misra, Pradeep Kumar, Amit Kumar, Ram Sagar, Kamalesh Chakravarty, Kameshwar Prasad Meta Gene. 2016 Jun; 8: 21–29. Published online 2016 Jan 19. doi: 10.1016/j.mgene.2016.01.003 • PLoS One. 2013; 8(5): e64747. Published online 2013 May 31. doi: 10.1371/journal.pone.0064747 • CD14 ?159 C>T Gene Polymorphism with Increased Risk of Tuberculosis: Evidence from a Meta-Analysis MY. Areeshi, Raju K. Mandal, Aditya K. Panda, Shekhar C. Bisht, Shafiul Haque • Helicobacter pylori Infection Enhances Gastric Mucosal Inflammation in Individuals Carrying the 260-T Allele of the CD14 Gene Eun Jung Kim, Woo Chul Chung, Kang-Moon Lee, Chang Nyol Paik, Sang Bae Kim, You Suk Oh, Yang Woon Lee, Sung-Goo Kang, Seung June Noh Gut Liver. 2013 May; 7(3): 317–322. Published online 2013 May 13. doi: 10.5009/gnl.2013.7.3.317 • Association between CD14 Gene Polymorphisms and Cancer Risk: A Meta-Analysis Jun Wang, Xufeng Guo, Shijie Yu, Jia Song, Jixiang Zhang, Zhuo Cao, Jing Wang, Min Liu, Weiguo Dong PLoS One. 2014; 9(6): e100122. Published online 2014 Jun 30. doi: 10.1371/journal.pone.0100122 • Racial differences in the association of CD14 polymorphisms with serum total IgE levels and allergen skin test reactivity ZongYao Wang, John S Sundy, Catherine M Foss, Huiman X Barnhart, Scott M Palmer, Sallie D Allgood, Evan Trudeau, Katie M Alexander, Marc C Levesque J Asthma Allergy. 2013; 6: 81–92. Published online 2013 Jun 25. doi: 10.2147/JAA.S42695 • CD14-expressing cancer cells establish the inflammatory and proliferative tumor microenvironment in bladder cancer Ming T. Cheah, James Y. Chen, Debashis Sahoo, Humberto Contreras-Trujillo, Anne K. Volkmer, Ferenc A. Scheeren, Jens-Peter Volkmer, Irving L. WeissmanProc Natl Acad Sci U S A. 2015 Apr 14; 112(15): 4725–4730. Published online 2015 Mar 30. doi: 10.1073/pnas.1424795112 • CD14++CD16+ monocytes are enriched by glucocorticoid treatment and are functionally attenuated in driving effector T cell responses Baoying Liu, Ashwin Dhanda, Sima Hirani, Emily L. Williams, H. Nida Sen, Fernando Martinez Estrada, Diamond Ling, Ian Thompson, Megan Casady, Zhiyu Li, Han Si, William Tucker, Lai Wei, Shayma Jawad, Amol Sura, Jennifer Dailey, Susan Hannes, Ping Chen, Jason L. Chien, Siamon Gordon, Richard W.J. Lee, Robert B. NussenblattJ Immunol. Author manuscript; available in PMC 2016 Jun 1.Published in final edited form as: J Immunol. 2015 Jun 1; 194(11): 5150–5160. Published online 2015 Apr 24.doi: 10.4049/jimmunol.1402409 • Association between CD14 Promoter -159C/T Polymorphism and the Risk of Sepsis and Mortality: A Systematic Review and Meta-Analysis An-qiang Zhang, Cai-li Yue, Wei Gu, Juan Du, Hai-yan Wang, Jianxin Jiang PLoS One. 2013; 8(8): e71237. Published online 2013 Aug 19. doi: 10.1371/journal.pone.0071237

## CTLA4

• Investigation of Soluble and Transmembrane CTLA-4 Isoforms in Serum and Microvesicles Laura Esposito, Kara M. D. Hunter, Jan Clark, Daniel B. Rainbow, Helen Stevens, Jennifer Denesha, Simon Duley, Sarah Dawson, Gillian Coleman, Sarah Nutland, Gwynneth L. Bell, Carla Moran, Marcin Pekalski, John A. Todd, Linda S. Wicker J Immunol. 2014 Jul 15; 193(2): 889–900. Published online 2014 Jun 13. doi: 10.4049/jimmunol.1303389 • Overexpression and Secretion of the Soluble CTLA-4 Splice Variant in Various Autoimmune Diseases and in Cases with Overlapping Autoimmunity Suad AlFadhli Genet Test Mol Biomarkers. 2013 Apr; 17(4): 336–341. doi: 10.1089/gtmb.2012.0391 • CTLA-4 as a genetic determinant in autoimmune Addison's disease A S B Wolff, A L Mitchell, H J Cordell, A Short, B Skinningsrud, W Ollier, K Badenhoop, G Meyer, A Falorni, O Kampe, D Undlien, S H S Pearce, E S Husebye Genes Immun. 2015 Sep; 16(6): 430–436. Published online 2015 Jul 23. doi: 10.1038/gene.2015.27 • The associations between the polymorphisms in the CTLA-4 gene and the risk of Graves' disease in the Chinese population Liang Du, Jiqiao Yang, Jichong Huang, Yaxian Ma, Haichuan Wang, Tianyuan Xiong, Zhangpeng Xiang, Yonggang Zhang, Jin Huang BMC Med Genet. 2013; 14: 46. Published online 2013 Apr 19. doi: 10.1186/1471-2350-14-46 • The Role of CTLA-4 Exon-1 49 A/G Polymorphism and Soluble CTLA-4 Protein Level in Egyptian Patients with Behçet's Disease Sahar M. Abdel Galil, Hoda A. Hagrass Biomed Res Int. 2014; 2014: 513915. Published online 2014 Jun 2. doi: 10.1155/2014/513915 • Cytotoxic T-Lymphocyte Associated Antigen 4 Polymorphisms and Asthma Risk: A Meta-Analysis Wei Nie, Jiquan Chen, Qingyu Xiu PLoS One. 2012; 7(7): e42062. Published online 2012 Jul 26. doi: 10.1371/journal.pone.0042062 • Association of Cytotoxic T-Lymphocyte Antigen 4 (CTLA4) and Thyroglobulin (TG) GeneticVariants with Autoimmune Hypothyroidism Hinal Patel, Mohmmad Shoab Mansuri, Mala Singh, Rasheedunnisa Begum, Minal Shastri, Ambikanandan Misra PLoS One. 2016; 11(3): e0149441. Published online 2016 Mar 10. doi: 10.1371/journal.pone.0149441 • Common Variants on Cytotoxic T Lymphocyte Antigen-4 Polymorphisms Contributes to Type 1 Diabetes Susceptibility: Evidence Based on 58 Studies Jingnan Wang, Lianyong Liu, Junhua Ma, Fei Sun, Zefei Zhao, Mingjun Gu PLoS One. 2014; 9(1): e85982. Published online 2014 Jan 23. doi: 10.1371/journal.pone.0085982 • Association between CTLA-4 60G/A and -1661A/G Polymorphisms and the Risk of Cancers: A Meta-Analysis Qing Yan, Pin Chen, Ailin Lu, Peng Zhao, Aihua Gu PLoS One. 2013; 8(12): e83710. Published online 2013 Dec 23. doi: 10.1371/journal.pone.0083710 • The CTLA-4 gene polymorphisms are associated with CTLA-4 protein expression levels in multiple sclerosis patients and with susceptibility to disease Lidia Karabon, Agata Kosmaczewska, Malgorzata Bilinska, Edyta Pawlak, Lidia Ciszak, Anna Jedynak, Anna Jonkisz, Leszek Noga, Anna Pokryszko-Dragan, Magdalena Koszewicz, Irena Frydecka Immunology. 2009 Sep; 128(1 Pt 2): e787–e796. doi: 10.1111/j.1365-2567.2009.03083.x • Biochemical analysis of CTLA-4 immunoreactive material from human blood Matt Tector, Bhupendra O Khatri, Karen Kozinski, Kate Dennert, Martin K Oaks BMC Immunol. 2009; 10: 51. Published online 2009 Sep 22. doi: 10.1186/1471-2172-10-51 • Intrinsic and extrinsic control of peripheral T-cell tolerance by costimulatory molecules of the CD28/B7 family Hélène Bour-Jordan, Jonathan H. Esensten, Marc Martinez-Llordella, Cristina Penaranda, Melanie Stumpf, Jeffrey A. Bluestone Immunol Rev. Author manuscript; available in PMC 2012 May 1. Published in final edited form as: Immunol Rev. 2011 May; 241(1): 180–205. doi: 10.1111/j.1600-065X.2011.01011.x • Vitamin D Antagonises the Suppressive Effect of Inflammatory Cytokines on CTLA-4 Expression and Regulatory Function Louisa E. Jeffery, Omar S. Qureshi, David Gardner, Tie Z. Hou, Zoe Briggs, Blagoje Soskic, Jennifer Baker, Karim Raza, David M. Sansom PLoS One. 2015; 10(7): e0131539. Published online 2015 Jul 2. doi: 10.1371/journal.pone.0131539 • Association between CTLA-4 gene polymorphism and ankylosing spondylitis: a case-control study Nai-Guo Wang, Da-Chuan Wang, Bing-Yi Tan, Feng Wang, Ze-Nong Yuan Int J Clin Exp Pathol. 2015; 8(6): 7421–7425. Published online 2015 Jun 1. • CTLA-4 Polymorphisms and Systemic Lupus Erythematosus: A Comprehensive Meta-Analysis Jie Liu, Hong-Xin Zhang Genet Test Mol Biomarkers. 2013 Mar; 17(3): 226–231. doi: 10.1089/gtmb.2012.0302 • Recipient CTLA-4*CT60-AA genotype is a prognostic factor for acute graft-versus-host disease in hematopoietic stem cell transplantation for thalassemia Sandro Orrù, Nicola Orrù, Emmanouil Manolakos, Roberto Littera, Giovanni Caocci, Giovanna Giorgiani, Alice Bertaina, Daria Pagliara, Claudio Giardini, Sonia Nesci, Franco Locatelli, Carlo Carcassi, Giorgio La Nasa Hum Immunol. 2012 Mar; 73(3-2): 282–286. doi: 10.1016/j.humimm.2011.12.014 • Association of CT60 cytotoxic T lymphocyte antigen-4 gene polymorphism with thyroid autoantibody production in patients with Hashimoto's and postpartum thyroiditis K Zaletel, B Krhin, S Gaberš?ek, A Bi?ek, T Paji?, S Hojker Clin Exp Immunol. 2010 Jul; 161(1): 41–47. doi: 10.1111/j.1365-2249.2010.04113.x • Treg and CTLA-4: Two intertwining pathways to immune tolerance Lucy S.K. Walker J Autoimmun. 2013 Sep; 45(100): 49–57. doi: 10.1016/j.jaut.2013.06.006 • CTLA-4 and MDR1 polymorphisms increase the risk for ulcerative colitis: A meta-analysis Jia-Jun Zhao, Di Wang, Hui Yao, Da-Wei Sun, Hong-Yu Li World J Gastroenterol. 2015 Sep 14; 21(34): 10025–10040. Published online 2015 Sep 14. doi: 10.3748/wjg.v21.i34.10025

## DRD2

• Sasabe, Toshikazu et al. 2007. “Association Analysis of the Dopamine Receptor D2 (DRD2) SNP Rs1076560 in Alcoholic Patients.” Neuroscience Letters 412(2): 139–42. http://www.ncbi.nlm.nih.gov/pubmed/17196743. • Anton, Raymond F et al. 2008. “Pharmacogenomics” ed. Yoshiaki Tsuji. Nature Genetics 16(1): 268–78. http://www.ncbi.nlm.nih.gov/pubmed/26671228 (May 14, 2018). • Clarke, Toni-Kim et al. 2014. “The Dopamine Receptor D2 ( DRD2 ) SNP Rs1076560 Is Associated with Opioid Addiction.” Annals of Human Genetics 78(1): 33–39. http://www.ncbi.nlm.nih.gov/pubmed/24359476.

## FUT2

• Kimura K; Wakamatsu A; Suzuki Y; et al. (2006). "Diversification of transcriptional modulation: large-scale identification and characterization of putative alternative promoters of human genes". Genome Res. 16 (1): 55–65. PMID 16344560. • Reguigne-Arnould I; Couillin P; Mollicone R; et al. (1995). "Relative positions of two clusters of human alpha-L-fucosyltransferases in 19q (FUT1-FUT2) and 19p (FUT6-FUT3-FUT5) within the microsatellite genetic map of chromosome 19". Cytogenet. Cell Genet. 71 (2): 158–62. doi:10.1159/000134098. PMID 7656588. • Ball SP, Tongue N, Gibaud A, Le Pendu J, Mollicone R, Gerard G, Oriol R (Feb 1992). "The human chromosome 19 linkage group FUT1 (H), FUT2 (SE), LE, LU, PEPD, C3, APOC2, D19S7 and D19S9". Ann Hum Genet 55 (Pt 3): 225–33. PMID 1763885. • "Entrez Gene: FUT2 fucosyltransferase 2 (secretor status included)". • Koda Y, Soejima M, Wang B, Kimura H (1997). "Structure and expression of the gene encoding secretor-type galactoside 2-alpha-L-fucosyltransferase (FUT2)". Eur. J. Biochem. 246 (3): 750–5. • Strausberg RL; Feingold EA; Grouse LH; et al. (2003). "Generation and initial analysis of more than 15,000 full-length human and mouse cDNA sequences". Proc. Natl. Acad. Sci. U.S.A. 99 (26): 16899–903.PMID 12477932.

## HLA-DQA1

• Schiffenbauer J, Didier DK, Klearman M, et al. (1987). "Complete sequence of the HLA DQ alpha and DQ beta cDNA from a DR5/DQw3 cell line". J. Immunol. 139 (1): 228–33. PMID 3584986. • Kao HT, Gregersen PK, Tang JC, et al. (1989). "Molecular analysis of the HLA class II genes in two DRw6-related haplotypes, DRw13 DQw1 and DRw14 DQw3.". J. Immunol. 142 (5): 1743–7. PMID 2493052. • Jonsson AK, Hyldig-Nielsen JJ, Servenius B, et al. (1987). "Class II genes of the human major histocompatibility complex. Comparisons of the DQ and DX alpha and beta genes". J. Biol. Chem. 262 (18): 8767–77. PMID 3036828. • Schmidt H, Williamson D, Ashley-Koch A (2007). "HLA-DR15 haplotype and multiple sclerosis: a HuGE review.". Am. J. Epidemiol. 165 (10): 1097–109. doi:10.1093/aje/kwk118. PMID 17329717. • Marsh SG, Bodmer JG (1993). "HLA class II nucleotide sequences, 1992.". Tissue Antigens 40 (5): 229–43. PMID 1362295. • Todd JA, Fukui Y, Kitagawa T, Sasazuki T (1990). "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. 87 (3): 1094–8. . PMID 2300572. • Liu CP, Bach FH, Wu SK (1988). "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. 140 (10): 3631–9. PMID 3129499. • Horn GT, Bugawan TL, Long CM, et al. (1988). "Sequence analysis of HLA class II genes from insulin-dependent diabetic individuals". Hum. Immunol. 21 (4): 249–63. PMID 3372263.

## HLA-DQA2

• A family study confirms that the HLA-DP associations with celiac disease are the result of an extended HLA-DR3 haplotype. Bolsover, W.J., Hall, M.A., Vaughan, R.W., Welsh, K.I., Ciclitira, P.J. Hum. Immunol. (1991) • Entrez Gene: HLA-DQA2 major histocompatibility complex, class II, DQ alpha 2" • HLA class II-associated genetic susceptibility in multiple sclerosis: a critical evaluation. Olerup, O., Hillert, J. Tissue Antigens (1991) • The cryptic HLA-DQA2 ("DX alpha") gene is expressed in human B cell lines. Yu, L.P., Sheehy, M.J. J. Immunol. (1991) • HLA-DQA and DQB alleles contribute to susceptibility to insulin-dependent diabetes mellitus. Wang, H., He, R. Chin. Med. Sci. J. (1993) • Evidence for a primary association of celiac disease to a particular HLA-DQ alpha/beta heterodimer. Sollid, L.M., Markussen, G., Ek, J., Gjerde, H., Vartdal, F., Thorsby, E. J. Exp. Med. (1989) • Polymorphic DQ alpha and DQ beta interactions dictate HLA class II determinants of allo-recognition. Kwok, W.W., Mickelson, E., Masewicz, S., Milner, E.C., Hansen, J., Nepom, G.T. J. Exp. Med. (1990) • Celiac disease is associated with an extended HLA-DR3 haplotype which includes HLA-DPw1. Hall, M.A., Lanchbury, J.S., Bolsover, W.J., Welsh, K.I., Ciclitira, P.J. Hum. Immunol. (1990) • Trans-encoded DQ alpha beta heterodimers confer susceptibility to myasthenia gravis disease. Khalil, I., Berrih-Aknin, S., Lepage, V., Loste, M.N., Gajdos, P., Hors, J., Charron, D., Degos, L. C. R. Acad. Sci. III, Sci. Vie (1993) • A combination of HLA-DQ beta Asp57-negative and HLA DQ alpha Arg52 confers susceptibility to insulin-dependent diabetes mellitus. Khalil, I., d'Auriol, L., Gobet, M., Morin, L., Lepage, V., Deschamps, I., Park, M.S., Degos, L., Galibert, F., Hors, J. J. Clin. Invest. (1990)

## HLA-DRB1

• Denny, Joshua C et al. 2013. “Systematic Comparison of Phenome-Wide Association Study of Electronic Medical Record Data and Genome-Wide Association Study Data.” Nature Biotechnology 31(12): 1102–11. http://www.ncbi.nlm.nih.gov/pubmed/24270849. • Anton, Raymond F et al. 2008. “Pharmacogenomics” ed. Yoshiaki Tsuji. Nature Genetics 16(1): 268–78. http://www.ncbi.nlm.nih.gov/pubmed/26671228 (May 14, 2018). • Gorman, Jennifer D et al. 2004. “Particular HLA-DRB1 Shared Epitope Genotypes Are Strongly Associated with Rheumatoid Vasculitis.” Arthritis & Rheumatism 50(11): 3476–84. http://www.ncbi.nlm.nih.gov/pubmed/15529352.

## HLA-DRB2

• Profaizer, T., Li¿½zï¿½r-Molnï¿½r, E., Close, D. W., Delgado, J. C., & Kumïï¿½novics, A. (2016). HLA genotyping in the clinical laboratory: Comparison of next-generation sequencing methods. HLA, 88(1–2), 14–24. https://doi.org/10.1111/tan.12850 • Hoeppli, R. E., Macdonald, K. G., Levings, M. K., & Cook, L. (2016). How antigen specificity directs regulatory T-cell function: Self, foreign and engineered specificity. HLA. https://doi.org/10.1111/tan.12822 • Duke, J. L., Lind, C., Mackiewicz, K., Ferriola, D., Papazoglou, A., Gasiewski, A., … Monos, D. S. (2016). Determining performance characteristics of an NGS-based HLA typing method for clinical applications. HLA, 87(3), 141–152. https://doi.org/10.1111/tan.12736

## HNMT

• Keeling, Brett H et al. 2010. “Histamine N-Methyltransferase Thr105Ile Is Not Associated with Parkinson’s Disease or Essential Tremor.” Parkinsonism & Related Disorders 16(2): 112–14. http://www.ncbi.nlm.nih.gov/pubmed/19773194. • Anton, Raymond F et al. 2008. “Pharmacogenomics” ed. Yoshiaki Tsuji. Nature Genetics 16(1): 268–78. http://www.ncbi.nlm.nih.gov/pubmed/26671228 (May 14, 2018). • “Rs12995000 (SNP) - Explore This Variant - Homo Sapiens - Ensembl Genome Browser 92.” https://useast.ensembl.org/Homo_sapiens/Variation/Explore?r=2:137963579-137964579;v=rs12995000;vdb=variation;vf=8000533 (May 14, 2018).

## IL-13

• Molecular pathogenesis of eosinophilic esophagitis.Blanchard C.Curr Opin Gastroenterol. 2015 Jul;31(4):321-7. doi: 10.1097/MOG.0000000000000186. Review. • The influence of genetic variability and proinflammatory status on the development of bone disease in patients with Gaucher disease.Gervas-Arruga J, Cebolla JJ, de Blas I, Roca M, Pocovi M, Giraldo P.PLoS One. 2015 May 15;10(5):e0126153. doi: 10.1371/journal.pone.0126153. eCollection 2015. • Association of IL-13 single nucleotide polymorphisms in Iranian patients to multiple sclerosis.Seyfizadeh N, Kazemi T, Farhoudi M, Aliparasti MR, Sadeghi-Bazargani H, Almasi S, Babaloo Z.J Clin Exp Immunol. 2014 Dec 5;3(3):124-9. eCollection 2014. • IL-28B is a key regulator of B- and T-cell vaccine responses against influenza.Egli A, Santer DM, O'Shea D, Barakat K, Syedbasha M, Vollmer M, Baluch A, Bhat R, Groenendyk J, Joyce MA, Lisboa LF, Thomas BS, Battegay M, Khanna N, Mueller T, Tyrrell DL, Houghton M, Humar A, Kumar D.PLoS Pathog. 2014 Dec 11;10(12):e1004556. doi: 10.1371/journal.ppat.1004556. eCollection 2014 Dec. • A meta-analysis of IL-13 polymorphisms and pediatric asthma risk.Liu Z, Li P, Wang J, Fan Q, Yan P, Zhang X, Han B.Med Sci Monit. 2014 Dec 11;20:2617-23. doi: 10.12659/MSM.891017. • Polymorphisms in IL-4/IL-13 pathway genes and glioma risk: an updated meta-analysis.Chen P, Chen C, Chen K, Xu T, Luo C.Tumour Biol. 2015 Jan;36(1):121-7. doi: 10.1007/s13277-014-2895-8. Epub 2014 Dec 4. • An Interleukin 13 Polymorphism Is Associated with Symptom Severity in Adult Subjects with Ever Asthma.Accordini S, et al. PLoS One, 2016. PMID 26986948, • Genome-Wide Methylation Study Identifies an IL-13-induced Epigenetic Signature in Asthmatic Airways.Nicodemus-Johnson J, et al. Am J Respir Crit Care Med, 2016 Feb 15. PMID 26474238, • IL-13 Augments Compressive Stress-Induced Tissue Factor Expression in Human Airway Epithelial Cells.Mitchel JA, et al. Am J Respir Cell Mol Biol, 2016 Apr. PMID 26407210, • Single Nucleotide Polymorphisms in IL-10, IL-12p40, and IL-13 Genes and Susceptibility to Glioma.Shamran HA, Ghazi HF, Al-Salman A, Al-Juboory AA, Taub DD, Price RL, Nagarkatti M, Nagarkatti PS, Singh UP.Int J Med Sci. 2015 Sep 19;12(10):790-6. doi: • Polymorphisms in the interleukin 4, interleukin 4 receptor and interleukin 13 genes and allergic phenotype: A case control study.Naro?na B, Hoffmann A, Sobkowiak P, Schoneich N, Br?borowicz A, Szczepankiewicz A.Adv Med Sci. 2016 Mar;61(1):40-5. doi: • From genetics to treatment of eosinophilic esophagitis.Cianferoni A, Spergel JM.Curr Opin Allergy Clin Immunol. 2015 Oct;15(5):417-25. doi: 10.1097/ACI.0000000000000020 • Commentary: IL-4 and IL-13 receptors and signaling.McCormick SM, Heller NM.Cytokine. 2015 Sep;75(1):38-50. doi: 10.1016/j.cyto.2015.05.023. Epub 2015 Jul 14. • Chronic graft-versus-host-disease in CD34(+)-humanized NSG mice is associated with human susceptibility HLA haplotypes for autoimmune disease.Sonntag K, Eckert F, Welker C, Müller H, Müller F, Zips D, Sipos B, Klein R, Blank G, Feuchtinger T, Schumm M, Handgretinger R, Schilbach K.J Autoimmun. 2015 Aug;62:55-66. doi: 10.1016/j.jaut.2015.06.006. Epub 2015 Jul 3.

## IL23R

• Duerr, R H et al. 2006. “A Genome-Wide Association Study Identifies IL23R as an Inflammatory Bowel Disease Gene.” Science 314(5804): 1461–63. http://www.ncbi.nlm.nih.gov/pubmed/17068223. • Silverberg, Mark S et al. 2009. “Ulcerative Colitis–risk Loci on Chromosomes 1p36 and 12q15 Found by Genome-Wide Association Study.” Nature Genetics 41(2): 216–20. http://www.ncbi.nlm.nih.gov/pubmed/19122664. • Anton, Raymond F et al. 2008. “Pharmacogenomics” ed. Yoshiaki Tsuji. Nature Genetics 16(1): 268–78. http://www.ncbi.nlm.nih.gov/pubmed/26671228 (May 14, 2018).

## IL2RA

• Anton, Raymond F et al. 2008. “Pharmacogenomics” ed. Yoshiaki Tsuji. Nature Genetics 16(1): 268–78. http://www.ncbi.nlm.nih.gov/pubmed/26671228 (May 14, 2018). • Consortium, The International Multiple Sclerosis Genetics. 2007. “Risk Alleles for Multiple Sclerosis Identified by a Genomewide Study.” New England Journal of Medicine 357(9): 851–62. http://www.nejm.org/doi/abs/10.1056/NEJMoa073493. • Wang, L-M, D-M Zhang, Y-M Xu, and S-L Sun. 2011. “Interleukin 2 Receptor $?$ Gene Polymorphism and Risk of Multiple Sclerosis: A Meta-Analysis.” Journal of International Medical Research 39(5): 1625–35. http://www.ncbi.nlm.nih.gov/pubmed/22117963.

## IL-5

• Previously undescribed grass pollen antigens are the major inducers of T helper 2 cytokine-producing T cells in allergic individuals.Schulten V, Greenbaum JA, Hauser M, McKinney DM, Sidney J, Kolla R, Lindestam Arlehamn CS, Oseroff C, Alam R, Broide DH, Ferreira F, Grey HM, Sette A, Peters B.Proc Natl Acad Sci U S A. 2013 Feb 26;110(9):3459-64. doi: 10.1073/pnas.1300512110. Epub 2013 Feb 11. Erratum in: Proc Natl Acad Sci U S A. 2013 Mar 26;110(13):5269. Ferreira-Briza, Fatima [corrected to Ferreira, Fatima]. Proc Natl Acad Sci U S A. 2013 May 21;110(21):8750. • Clin Exp Allergy. 2015 Aug;45(8):1296-304. doi: 10.1111/cea.12543. • Cholesterol selectively regulates IL-5 induced mitogen activated protein kinase signaling in human eosinophils.Burnham ME, Esnault S, Roti Roti EC, Bates ME, Bertics PJ, Denlinger LC.PLoS One. 2014 Aug 14;9(8):e103122. doi: 10.1371/journal.pone.0103122. eCollection 2014. • Plasma IL-5 concentration and subclinical carotid atherosclerosis.Silveira A, McLeod O, Strawbridge RJ, Gertow K, Sennblad B, Baldassarre D, Veglia F, Deleskog A, Persson J, Leander K, Gigante B, Kauhanen J, Rauramaa R, Smit AJ, Mannarino E, Giral P, Gustafsson S, Söderberg S, Öhrvik J, Humphries SE, Tremoli E, de Faire U, Hamsten A.Atherosclerosis. 2015 Mar;239(1):125-30. doi: 10.1016/j.atherosclerosis.2014.12.046. Epub 2014 Dec 23 • IL-5 production by resident mucosal allergen-specific T cells in an explant model of allergic rhinitis.Skrindo I, Ballke C, Gran E, Johansen FE, Baekkevold ES, Jahnsen FL. • Sputum IL-5 concentration is associated with a sputum eosinophilia and attenuated by corticosteroid therapy in COPD.Bafadhel M, Saha S, Siva R, McCormick M, Monteiro W, Rugman P, Dodson P, Pavord ID, Newbold P, Brightling CE.Respiration. 2009;78(3):256-62. doi: 10.1159/000221902. Epub 2009 May 27. • Molecular and clinical rationale for therapeutic targeting of interleukin-5 and its receptor.Molfino NA, Gossage D, Kolbeck R, Parker JM, Geba GP.Clin Exp Allergy. 2012 May;42(5):712-37. doi: 10.1111/j.1365-2222.2011.03854.x. Epub 2011 Sep 23. Review. • Allergen-specific IL-5 responses in early childhood predict asthma at age eight.Weber-Chrysochoou C, Crisafulli D, Kemp AS, Britton WJ, Marks GB; CAPS Investigators.PLoS One. 2014 May 29;9(5):e97995. doi: 10.1371/journal.pone.0097995. eCollection 2014. • IL-5, IL-8 and MMP -9 levels in exhaled breath condensate of atopic and nonatopic asthmatic children.Turkeli A, Yilmaz O, Taneli F, Horasan GD, Kanik ET, Kizilkaya M, Gozukara C, Yuksel H.Respir Med. 2015 Jun;109(6):680-8. doi: 10.1016/j.rmed.2015.04.004. Epub 2015 Apr 15. • Mechanisms of human eosinophil migration induced by the combination of IL-5 and the endocannabinoid 2-arachidonoyl-glycerol.Larose MC, Turcotte C, Chouinard F, Ferland C, Martin C, Provost V, Laviolette M, Flamand N.J Allergy Clin Immunol. 2014 May;133(5):1480-2, 1482.e1-3. doi: 10.1016/j.jaci.2013.12.1081. Epub 2014 Feb 13. No abstract available. • Inhibition of interleukin-5 for the treatment of eosinophilic diseases.Corren J.Discov Med. 2012 Apr;13(71):305-12. Review.

## IL6

• Buxens, A. et al. 2011. “Can We Predict Top-Level Sports Performance in Power vs Endurance Events? A Genetic Approach.” Scandinavian Journal of Medicine and Science in Sports 21(4): 570–79. • Illig, T et al. 2004. “Significant Association of the Interleukin-6 Gene Polymorphisms C-174G and A-598G with Type 2 Diabetes.” The Journal of Clinical Endocrinology & Metabolism 89(10): 5053–58. http://www.ncbi.nlm.nih.gov/pubmed/15472205. • Anton, Raymond F et al. 2008. “Pharmacogenomics” ed. Yoshiaki Tsuji. Nature Genetics 16(1): 268–78. http://www.ncbi.nlm.nih.gov/pubmed/26671228 (May 14, 2018). • Fishman, D et al. 1998. “The Effect of Novel Polymorphisms in the Interleukin-6 (IL-6) Gene on IL-6 Transcription and Plasma IL-6 Levels, and an Association with Systemic-Onset Juvenile Chronic Arthritis.” Journal of Clinical Investigation 102(7): 1369–76. http://www.ncbi.nlm.nih.gov/pubmed/9769329. • Baumert, Philipp et al. 2016. “Genetic Variation and Exercise-Induced Muscle Damage: Implications for Athletic Performance, Injury and Ageing.” European Journal of Applied Physiology 116(9): 1595–1625. http://www.ncbi.nlm.nih.gov/pubmed/27294501 (February 25, 2018).

## NOD2

• Anton, Raymond F et al. 2008. “Pharmacogenomics” ed. Yoshiaki Tsuji. Nature Genetics 16(1): 268–78. http://www.ncbi.nlm.nih.gov/pubmed/26671228 (May 14, 2018). • Ogura, Yasunori et al. 2001. “A Frameshift Mutation in NOD2 Associated with Susceptibility to Crohn’s Disease.” Nature 411(6837): 603–6. http://www.ncbi.nlm.nih.gov/pubmed/11385577. • Nuij, V. J.A.A., M. P. Peppelenbosch, C. J. Woude, and G. M. Fuhler. 2017. “Genetic Polymorphism in ATG16L1 Gene Is Associated with Adalimumab Use in Inflammatory Bowel Disease.” Journal of Translational Medicine 15(1). • Hugot, Jean-Pierre et al. 2001. “Association of NOD2 Leucine-Rich Repeat Variants with Susceptibility to Crohn’s Disease.” Nature 411(6837): 599–603. http://www.ncbi.nlm.nih.gov/pubmed/11385576.

## NOS2

• Genetic variation in immunoregulatory pathways and atopic phenotypes in infancy. Hoffjan, S., Ostrovnaja, I., Nicolae, D., Newman, D.L., Nicolae, R., Gangnon, R., Steiner, L., Walker, K., Reynolds, R., Greene, D., Mirel, D., Gern, J.E., Lemanske, R.F., Ober, C. J. Allergy Clin. Immunol. (2004) [Pubmed] • Cross-talk between cyclooxygenase and nitric oxide pathways: prostaglandin E2 negatively modulates induction of nitric oxide synthase by interleukin 1. Tetsuka, T., Daphna-Iken, D., Srivastava, S.K., Baier, L.D., DuMaine, J., Morrison, A.R. Proc. Natl. Acad. Sci. U.S.A. (1994) [Pubmed] • Molecular cloning and expression of inducible nitric oxide synthase from human hepatocytes. Geller, D.A., Lowenstein, C.J., Shapiro, R.A., Nussler, A.K., Di Silvio, M., Wang, S.C., Nakayama, D.K., Simmons, R.L., Snyder, S.H., Billiar, T.R. Proc. Natl. Acad. Sci. U.S.A. (1993) [Pubmed] • Role of interferon regulatory factor 1 in induction of nitric oxide synthase. Martin, E., Nathan, C., Xie, Q.W. J. Exp. Med. (1994) [Pubmed] • Physiology and pathophysiology of nitric oxide. Ignarro, L.J. Kidney Int. Suppl. (1996) [Pubmed] • Regulation of the mammalian heart function by nitric oxide. Massion, P.B., Pelat, M., Belge, C., Balligand, J.L. Comp. Biochem. Physiol., Part A Mol. Integr. Physiol. (2005) [Pubmed] • Association of elevated glial expression of interleukin-1beta with improved survival in patients with glioblastomas multiforme. Cuny, E., Loiseau, H., Penchet, G., Ellie, E., Arsaut, J., Vital, A., Vincendeau, P., Demotes-Mainard, J. J. Neurosurg. (2002) [Pubmed] • Platelet phagocytosis and processing of beta-amyloid precursor protein as a mechanism of macrophage activation in atherosclerosis. De Meyer, G.R., De Cleen, D.M., Cooper, S., Knaapen, M.W., Jans, D.M., Martinet, W., Herman, A.G., Bult, H., Kockx, M.M. Circ. Res. (2002) [Pubmed] • Modulation of prostaglandin biosynthesis by nitric oxide and nitric oxide donors. Mollace, V., Muscoli, C., Masini, E., Cuzzocrea, S., Salvemini, D. Pharmacol. Rev. (2005) [Pubmed] • Expression of type II nitric oxide synthase in primary human astrocytes and microglia: role of IL-1beta and IL-1 receptor antagonist. Liu, J., Zhao, M.L., Brosnan, C.F., Lee, S.C. J. Immunol. (1996) [Pubmed] • Linkage and association with the NOS2A locus on chromosome 17q11 in multiple sclerosis. Barcellos, L.F., Begovich, A.B., Reynolds, R.L., Caillier, S.J., Brassat, D., Schmidt, S., Grams, S.E., Walker, K., Steiner, L.L., Cree, B.A., Stillman, A., Lincoln, R.R., Pericak-Vance, M.A., Haines, J.L., Erlich, H.A., Hauser, S.L., Oksenberg, J.R. Ann. Neurol. (2004) [Pubmed] • Association of a functional inducible nitric oxide synthase promoter variant with susceptibility to biopsy-proven giant cell arteritis. Gonzalez-Gay, M.A., Oliver, J., Sanchez, E., Garcia-Porrua, C., Paco, L., Lopez-Nevot, M.A., Ollier, W.E., Martin, J. J. Rheumatol. (2005) [Pubmed] • Nitric oxide synthase gene polymorphisms in Alzheimer's disease and dementia with Lewy bodies. Singleton, A.B., Gibson, A.M., McKeith, I.G., Ballard, C.G., Edwardson, J.A., Morris, C.M. Neurosci. Lett. (2001) [Pubmed] • Nitric oxide and macrophage function. MacMicking, J., Xie, Q.W., Nathan, C. Annu. Rev. Immunol. (1997) [Pubmed] • Nitric oxide in health and disease of the respiratory system. Ricciardolo, F.L., Sterk, P.J., Gaston, B., Folkerts, G. Physiol. Rev. (2004) [Pubmed] • Free radicals in the physiological control of cell function. Dröge, W. Physiol. Rev. (2002) [Pubmed] • Bidirectional regulation of osteoclast function by nitric oxide synthase isoforms. Brandi, M.L., Hukkanen, M., Umeda, T., Moradi-Bidhendi, N., Bianchi, S., Gross, S.S., Polak, J.M., MacIntyre, I. Proc. Natl. Acad. Sci. U.S.A. (1995)[Pubmed] • Role of nitric oxide in Sjögren's syndrome. Konttinen, Y.T., Platts, L.A., Tuominen, S., • Regulation of nitric oxide synthesis by proinflammatory cytokines in human umbilical vein endothelial cells. Elevations in tetrahydrobiopterin levels enhance endothelial nitric oxide synthase specific activity. Rosenkranz-Weiss, P., Sessa, W.C., Milstien, S., Kaufman, S., Watson, C.A., Pober, J.S. J. Clin. Invest. (1994) [Pubmed]

## SOCS1

• Anton, Raymond F et al. 2008. “Pharmacogenomics” ed. Yoshiaki Tsuji. Nature Genetics 16(1): 268–78. http://www.ncbi.nlm.nih.gov/pubmed/26671228 (May 14, 2018). • Vandenbroeck, K et al. 2012. “A Cytokine Gene Screen Uncovers SOCS1 as Genetic Risk Factor for Multiple Sclerosis.” Genes and Immunity 13(1): 21–28. http://www.ncbi.nlm.nih.gov/pubmed/21716315. • Pahlevan Kakhki, Majid et al. 2015. “Expression of Suppressor of Cytokine Signaling 1 (SOCS1) Gene Dramatically Increases in Relapsing–remitting Multiple Sclerosis.” Journal of the Neurological Sciences 350(1–2): 40–45. http://www.ncbi.nlm.nih.gov/pubmed/25701091.

## STAT4

• STAT4 controls GM-CSF production by both Th1 and Th17 cells during EAElan L. McWilliams, Rajani Rajbhandari, Susan Nozell, Etty Benveniste, Laurie E. HarringtonJ Neuroinflammation. 2015; 12: 128. Published online 2015 Jun 30. doi: 10.1186/s12974-015-0351-3 • Altered STAT4 isoform expression in patients with inflammatory bowel disease Rukhsana Jabeen, Lucy Miller, Weiguo Yao, Sandeep Gupta, Steven Steiner, Mark H. Kaplan Inflamm Bowel Dis. 2015 Oct; 21(10): 2383–2392. doi: 10.1097/MIB.0000000000000495 • A risk haplotype of STAT4 for systemic lupus erythematosus is over-expressed, correlates with anti-dsDNA and shows additive effects with two risk alleles of IRF5 Snaevar Sigurdsson, Gunnel Nordmark, Sophie Garnier, Elin Grundberg, Tony Kwan, Olof Nilsson, Maija-Leena Eloranta, Iva Gunnarsson, Elisabet Svenungsson, Gunnar Sturfelt, Anders A. Bengtsson, Andreas Jönsen, Lennart Truedsson, Solbritt Rantapää-Dahlqvist, Catharina Eriksson, Gunnar Alm, Harald H.H. Göring, Tomi Pastinen, Ann-Christine Syvänen, Lars Rönnblom Hum Mol Genet. 2008 Sep 15; 17(18): 2868–2876. Published online 2008 Jun 25. doi: 10.1093/hmg/ddn184 • High density genotyping of STAT4 gene reveals multiple haplotypic associations with Systemic Lupus Erythematosus in different racial groups Bahram Namjou, Andrea L. Sestak, Don L. Armstrong, Raphael Zidovetzki, Jennifer A. Kelly, Noam Jacob, Voicu Ciobanu, Kenneth M. Kaufman, Joshua O. Ojwang, Julie Ziegler, Francesco Quismorio, Andreas Reiff, Barry L. Myones, Joel M. Guthridge, Swapan K. Nath, Gail R. Bruner, Ruth Mehrian-Shai, Earl Silverman, Marisa Klein-Gitelman, Deborah McCurdy, Linda Wagner-Weiner, James J. Nocton, Chaim Putterman, Sang-Cheol Bae, Yun Jung Kim, Michelle Petri, John D. Reveille, Timothy J. Vyse, Gary S. Gilkeson, Diane L. Kamen, Marta E. Alarcón-Riquelme, Patrick M. Gaffney, Kathy L Moser, Joan T. Merrill, R. Hal Scofield, Judith A. James, Carl D. Langefeld, John B. Harley, Chaim O. Jacob Arthritis Rheum. 2009 Apr; 60(4): 1085–1095. doi: 10.1002/art.24387 • Association between a C8orf13–BLK Polymorphism and Polymyositis/Dermatomyositis in the Japanese Population: An Additive Effect with STAT4 on Disease Susceptibility Tomoko Sugiura, Yasushi Kawaguchi, Kanako Goto, Yukiko Hayashi, Takahisa Gono, Takefumi Furuya, Ichizo Nishino, Hisashi Yamanaka PLoS One. 2014; 9(3): e90019. Published online 2014 Mar 14. doi: 10.1371/journal.pone.0090019 • Polymorphisms in TBX21 and STAT4 Increase the Risk of Systemic Sclerosis: Evidence of Possible Gene–Gene Interaction and Alterations in Th1/Th2 Cytokines Pravitt Gourh, Sandeep K. Agarwal, Dipal Divecha, Shervin Assassi, Gene Paz, Rajpreet K. Arora-Singh, John D. Reveille, Sanjay Shete, Maureen D. Mayes, Frank C. Arnett, Filemon K. Tan Arthritis Rheum. 2009 Dec; 60(12): 3794–3806. doi: 10.1002/art.24958 • The TT Genotype of the STAT4 rs7574865 Polymorphism Is Associated with High Disease Activity and Disability in Patients with Early Arthritis Amalia Lamana, Alejandro Balsa, Blanca Rueda, Ana M. Ortiz, Laura Nuño, Maria Eugenia Miranda-Carus, Maria F. Gonzalez-Escribano, Miguel A. Lopez-Nevot, Dora Pascual-Salcedo, Javier Martin, Isidoro González-Álvaro PLoS One. 2012; 7(8): e43661. Published online 2012 Aug 24. doi: 10.1371/journal.pone.0043661 • Evidence for STAT4 as a Common Autoimmune Gene: rs7574865 Is Associated with Colonic Crohn's Disease and Early Disease Onset Jürgen Glas, Julia Seiderer, Melinda Nagy, Christoph Fries, Florian Beigel, Maria Weidinger, Simone Pfennig, Wolfram Klein, Jörg T. Epplen, Peter Lohse, Matthias Folwaczny, Burkhard Göke, Thomas Ochsenkühn, Julia Diegelmann, Bertram Müller-Myhsok, Darina Roeske, Stephan Brand PLoS One. 2010; 5(4): e10373. Published online 2010 Apr 29. doi: 10.1371/journal.pone.0010373 • Association between STAT4 Gene Polymorphisms and Autoimmune Thyroid Diseases in a Chinese Population Ni Yan, Shuai Meng, Jiaozhen Zhou, Jian Xu, Fatuma Said Muhali, Wenjuan Jiang, Liangfeng Shi, Xiaohong Shi, Jinan Zhang Int J Mol Sci. 2014 Jul; 15(7): 12280–12293. Published online 2014 Jul 11. doi: 10.3390/ijms150712280 • STAT4 Regulates Antiviral Gamma Interferon Responses and Recurrent Disease during Herpes Simplex Virus 2 Infection Alexandra Svensson, Petra Tunbäck, Inger Nordström, Andrey Shestakov, Leonid Padyukov, Kristina Eriksson J Virol. 2012 Sep; 86(17): 9409–9415. doi: 10.1128/JVI.00947-12 • Signal Transducer and Activator of Transcription 4 in Liver Diseases Yan Wang, Aijuan Qu, Hua Wang Int J Biol Sci. 2015; 11(4): 448–455. Published online 2015 Feb 27. doi: 10.7150/ijbs.11164 • The Minor Allele of rs7574865 in the STAT4 Gene Is Associated with Increased mRNA and Protein ExpressionAmalia Lamana, Mercedes López-Santalla, Raquel Castillo-González, Ana María Ortiz, Javier Martín, Rosario García-Vicuña, Isidoro González-ÁlvaroPLoS One. 2015; 10(11): e0142683. Published online 2015 Nov 16. doi: 10.1371/journal.pone.0142683

## TNF

• Association between TNF-? ?308 G/A polymorphism and COPD susceptibility: a meta-analysis updateLu Zhang, Hao Gu, Yihang Gu, Xiaoning Zeng Int J Chron Obstruct Pulmon Dis. 2016; 11: 1367–1379. Published online 2016 Jun 22. doi: 10.2147/COPD.S105394 • Tumor Necrosis Factor (TNF) –308G>A, Nitric Oxide Synthase 3 (NOS3) +894G>T Polymorphisms and Migraine Risk: A Meta-Analysis Min Chen, Wenjing Tang, Lei Hou, Ruozhuo Liu, Zhao Dong, Xun Han, Xiaofei Zhang, Dongjun Wan, Shengyuan Yu PLoS One. 2015; 10(6): e0129372. Published online 2015 Jun 22. doi: 10.1371/journal.pone.0129372 • Genetic polymorphism at codon 10 of the transforming growth factor-?1 gene in patients with alcoholic liver cirrhosis Jong Joon Lee, Soo Kyung Park, Oh Sang Kwon, In Sik Won, Dong Kyu Kim, Young Kul Jung, Yang Suh Ku, Yun Soo Kim, Duck Joo Choi, Ju Hyun Kim Korean J Hepatol. 2011 Mar; 17(1): 37–43. Published online 2011 Mar 21. doi: 10.3350/kjhep.2011.17.1.37 • Associations between TNF-?-308A/G Polymorphism and Susceptibility with Dermatomyositis: A Meta-Analysis Si Chen, Qian Wang, Ziyan Wu, Qingjun Wu, Ping Li, Yuan Li, Jing Li, Chuiwen Deng, Chanyuan Wu, Lei Gao, Fengchun Zhang, Yongzhe Li PLoS One. 2014; 9(8): e102841. Published online 2014 Aug 7. doi: 10.1371/journal.pone.0102841 • Association of tumor necrosis factor ? genetic polymorphism and sepsis susceptibility FRANCIELI DELONGUI, CÍNTIA MAGALHÃES CARVALHO GRION, MARIA ANGELICA EHARA WATANABE, HELENA KAMINAMI MORIMOTO, ANA MARIA BONAMETTI, JULIE MASSAYO MAEDA ODA, ANA PAULA KALLAUR, TIEMI MATSUO, EDNA MARIA VISSOCI REICHE Exp Ther Med. 2011 Mar-Apr; 2(2): 349–356. Published online 2011 Jan 20. doi: 10.3892/etm.2011.213 • TNF-?-308 polymorphism and risk of digestive system cancers: A meta-analysis Xu-Feng Guo, Jun Wang, Shi-Jie Yu, Jia Song, Meng-Yao Ji, Zhuo Cao, Ji-Xiang Zhang, Jing Wang, Wei-Guo Dong World J Gastroenterol. 2013 Dec 28; 19(48): 9461–9471. Published online 2013 Dec 28. doi: 10.3748/wjg.v19.i48.9461 • Pathogenetic and Therapeutic Applications of Tumor Necrosis Factor-? (TNF-?) in Major Depressive Disorder: A Systematic Review Ke Ma, Hongxiu Zhang, Zulqarnain Baloch Int J Mol Sci. 2016 May; 17(5): 733. Published online 2016 May 14. doi: 10.3390/ijms17050733 • TNF-? -308 G?>?A (rs1800629) Polymorphism is Associated with Celiac Disease: A Meta-analysis of 11 Case-Control Studies Saif Khan, Raju K. Mandal, Arshad Jawed, Sajad A. Dar, Mohd Wahid, Aditya K. Panda, Mohammed Y. Areeshi, Md. Ekhlaque Ahmed Khan, Shafiul Haque Sci Rep. 2016; 6: 32677. Published online 2016 Sep 6. doi: 10.1038/srep32677 • Tumour Necrosis Factor-? Gene Polymorphism Is Associated with Metastasis in Patients with Triple Negative Breast Cancer Hui-Hui Li, Hui Zhu, Li-Sheng Liu, Yong Huang, Jun Guo, Jie Li, Xin-Ping Sun, Chun-Xiao Chang, Zhe-Hai Wang, Kan Zhai Sci Rep. 2015; 5: 10244. Published online 2015 Jul 13. doi: 10.1038/srep10244 • Relation between inflammatory cytokine levels in serum and bronchoalveolar lavage fluid and gene polymorphism in young adult patients with bronchiectasis Gulhan Ayhan, Dilaver Tas, Ismail Yilmaz, Oguzhan Okutan, Ersin Demirer, Omer Ayten, Zafer Kartaloglu J Thorac Dis. 2014 Jun; 6(6): 684–693. doi: 10.3978/j.issn.2072-1439.2014.04.14 • Increased Tumor Necrosis Factor (TNF)-? and Its Promoter Polymorphisms Correlate with Disease Progression and Higher Susceptibility towards Vitiligo Naresh C. Laddha, Mitesh Dwivedi, Rasheedunnisa Begum PLoS One. 2012 Dec 20; 7(12): e52298. Published online 2012 Dec 20. doi: 10.1371/journal.pone.0052298 • TNF -308 G/A Polymorphism and Risk of Acne Vulgaris: A Meta-Analysis Jian-Kang Yang, Wen-Juan Wu, Jue Qi, Li He, Ya-Ping Zhang PLoS One. 2014; 9(2): e87806. Published online 2014 Feb 3. doi: 10.1371/journal.pone.0087806 • Meta-Analysis of TNF 308 G/A Polymorphism and Type 2 Diabetes Mellitus Ren-Nan Feng, Chen Zhao, Chang-Hao Sun, Ying Li PLoS One. 2011; 6(4): e18480. Published online 2011 Apr 8. doi: 10.1371/journal.pone.0018480 • Tumor Necrosis Factor Alpha rs1800629 Polymorphism and Risk of Cervical Lesions: A Meta-Analysis Min Li, Ying Han, Ting-Ting Wu, Yichen Feng, Hong-Bo Wang PLoS One. 2013; 8(8): e69201. Published online 2013 Aug 27. doi: 10.1371/journal.pone.0069201

## TRAF-1

• Panoulas, Vasileios F, Jacqueline P Smith, Peter Nightingale, and George D Kitas. 2009. “Association of the TRAF1/C5 Locus with Increased Mortality, Particularly from Malignancy or Sepsis, in Patients with Rheumatoid Arthritis.” Arthritis & Rheumatism 60(1): 39–46. http://www.ncbi.nlm.nih.gov/pubmed/19116907. • Plenge, Robert M et al. 2007. “TRAF1–C5 as a Risk Locus for Rheumatoid Arthritis — A Genomewide Study.” New England Journal of Medicine 357(12): 1199–1209. http://www.ncbi.nlm.nih.gov/pubmed/17804836. • Anton, Raymond F et al. 2008. “Pharmacogenomics” ed. Yoshiaki Tsuji. Nature Genetics 16(1): 268–78. http://www.ncbi.nlm.nih.gov/pubmed/26671228 (May 14, 2018).