2017 Publications

- Kringel, D et al. (2017) Next-generation sequencing of the human TRPV1 gene and the regulating co-players LTB4R and LTB4R2 based on a custom AmpliSeq™panel. *PLOS One*, DOI: https://doi.org/10.1371/journal.pone.0180116 [Abstract]


• Overton, N et al. (2017) Genetic susceptibility to severe asthma with fungal sensitization. *International Journal of Immunogenetics*, DOI: 10.1111/iji.12312 Abstract


• Kahr, W et al. (2017) Loss of the Arp2/3 complex component ARPC1B causes platelet abnormalities and predisposes to inflammatory disease. *Nature Communications*, DOI: 10.1038/ncomms14816 Abstract


• KLoss, B et al. (2017) Exome Sequence Analysis of 14 Families With High Myopia. *iovs*, DOI: 10.1167/iovs.16-20883 Abstract

• Nielsen, K et al. (2017) Interactions between SNPs affecting inflammatory response genes are associated with multiple myeloma disease risk and survival. *Leukemia & Lymphoma*, DOI: http://dx.doi.org/10.1080/10428194.2017.1306643 Abstract
• Tabatabaeifar, S et al. (2017) Investigating a case of possible field cancerization in oral squamous cell carcinoma by the use of next-generation sequencing. Oral Oncology, DOI: http://dx.doi.org/10.1016/j.oraloncology.2017.03.018 Abstract
• Matosin, N et al. (2017) Effects of common GRM5 genetic variants on cognition, hippocampal volume and mGluR5 protein levels in schizophrenia. Brain Imaging and Behavior, DOI: 10.1007/s11682-017-9712-0 Abstract
• Ruiz, P et al. (2017) Integration of in silico methods and computational systems biology to explore endocrine-disrupting chemical binding with nuclear hormone receptors. Chemosphere, DOI: 10.2527/fas.2016.1355 Abstract
• Norgaard, L et al. (2017) Population genomics of the raccoon dog (Nyctereutes procyonoides) in Denmark: insights into invasion history and population development. Biological Invasions, DOI: 10.1007/s10530-017-1385-5 Abstract
• Hampel, K et al. (2017) Variant call concordance between two laboratory-developed, solid tumor targeted genomic profiling assays using distinct workflows and sequencing instruments. Experimental and Molecular Pathology, DOI: http://dx.doi.org/10.1016/j.yexmp.2017.02.002 Abstract
• Evans, J et al. (2017) Beyond the MHC: A canine model of dermatomyositis shows a complex pattern of genetic risk involving novel loci. PLOS Genetics, DOI: http://dx.doi.org/10.1371/journal.pgen.1006604 Abstract
• Ference, B et al. (2017) Variation in PCSK9 and HMGCR and Risk of Cardiovascular Disease and Diabetes. The New England Journal of Medicine, DOI: 10.1056/NEJMoa1604304 Abstract
• Guo, T et al. (2017) Sexually dimorphic association of the pleiotropic GALNT2 SNPs and haplotypes and serum lipid traits in Jing and Han populations. International Journal of Clinical and Experimental Pathology. ISSN: 1936-2625/IJCEP0028135 Abstract
• Li, L et al. (2017) Regulatory Variants Modulate Protein Kinase C α (PRKCA) Gene Expression in Human Heart. *Pharmaceutical Research*, doi: 10.1007/s11095-017-2102-x Abstract

• Byun, E et al. (2017) Cytokine Polymorphisms are Associated with Daytime Napping in Adults Living with HIV. *Sleep Medicine*, doi: http://dx.doi.org/10.1016/j.sleep.2016.12.021 Abstract

• Wu, S et al. (2017) Association between NF-kB Pathway Gene Variants and sICAM1 Levels in Taiwanese. *PLOS one*, doi: http://dx.doi.org/10.1371/journal.pone.0169516 Abstract


• Sokolowski, M et al. (2017) Rare CNVs in Suicide Attempt include Schizophrenia-Associated Loci and Neurodevelopmental Genes: A Pilot Genome-Wide and Family-Based Study. *PLOS one*, doi: http://dx.doi.org/10.1371/journal.pone.0168531 Abstract


2016 Publications

• Wieneke, H et al. (2016) Polymorphisms in the GNAS Gene as Predictors of Ventricular Tachyarrhythmias and Sudden Cardiac Death: Results From the DISCOVERY Trial and Oregon Sudden Unexpected Death Study. *Journal of the American Heart Association*, doi: 10.1161/JAHA.116.003905 Abstract


• Bulayeva, K et al. (2016) Descriptions and Methods of Study in Selected Genetic Isolates of Dagestan. *Springer Publishing*, 978-3-319-31962-9 Abstract


• Matsunami, K et al. (2016) Genome-Wide Association Study Identifies ZNF354C Variants Associated with Depression from Interferon-Based Therapy for Chronic Hepatitis C. *PLOS one*, http://dx.doi.org/10.1371/journal.pone.0164418 Abstract


• Rao, A et al. (2016) Rare deleterious mutations are associated with disease in bipolar disorder families. *Molecular Psychiatry*, doi: 10.1038/mp.2016.181 Abstract


• Reis, L et al. (2016) Analysis of CYP1B1 in pediatric and adult glaucoma and other ocular phenotypes. *Molecular Vision*, Abstract


• Liang, K et al. (2016) UGT2B28 genomic variation is associated with hepatitis B e-antigen seroconversion in response to antiviral therapy. *Scientific Reports*, doi: 10.1038/srep34088 [Abstract]


- Ubeagha-Awemu, E et al. (2016) High density genome wide genotyping-by-sequencing and association identifies common and low frequency SNPs, and novel candidate genes influencing cow milk traits. *Scientific Reports*, doi: 10.1038/srep31109 Abstract
- Goffinet, L et al. (2016) Cystathionine β-synthase genetic variant rs2124459 is associated with a reduced risk of cleft palate in French and Belgian populations. *Journal of Medical Genetics*, doi: 10.1136/jmedgenet-2016-104111 Abstract
- Jiao, X et al. (2016) A Common Ancestral Mutation in CRYBB3 Identified in Multiple Consanguineous Families with Congenital Cataracts. *PLOS one*, doi: http://dx.doi.org/10.1371/journal.pone.0157005 Abstract


Ko, Y et al. (2016) CRP and SAA1 Haplotypes Are Associated with Both C-Reactive Protein and Serum Amyloid A Levels: Role of Suppression Effects. *Mediators of Inflammation*, doi: http://dx.doi.org/10.1155/2016/5830361 Abstract


• Kringel, D. et al. (2016) Emergent biomarker derived from next-generation sequencing to identify pain patients requiring uncommonly high opioid doses. *Quality of Life Research*, doi: 10.1038/tjp.2016.28 Abstract


• Ko, Y. et al. (2016) CRP and SAA1 haplotypes are associated with both C-reactive protein and serum amyloid A levels: Role of Suppression effects. Abstract


• Cropp, C. et al. (2016) Rare variant discovery in known cancer genes from whole-exome sequencing of African American hereditary prostate cancer families. *Cancer Epidemiology Biomarkers & Prevention*, doi: 10.1158/1538-7755.DISP15-B40 Abstract

• Geisel, M. et al. (2016) Update of the effect estimates for common variants associated with carotid intima media thickness within four independent samples: The Bonn IMT Family


Ma, X. et al. (2016) Polymorphism rs7278468 is associated with Age-related cataract through decreasing transcriptional activity of theCRYAA promoter. *Scientific Reports*, doi: 10.1038/srep23206 [Abstract](#)


• Nguyen, L. et al. (2016) Effects of TEX11 and Polymorphisms on Reproduction and Growth Traits in Australian Beef Cattle. PAABG Abstract
• Fortes, M. et al. (2016) Non-Synonomous Polymorphism in Helb is Associated With Male and Female Reproductive Traits in Cattle. PAABG Abstract
• Weber, J. et al. (2016) Sentieon DNA pipeline for variant detection - Software-only solution, over 20x faster than GATK 3.3 with indenticle results. PeerJPreprints Abstract


• Goldstein, O. et al. (2016) OPTN 691_692insAG is a founder mutation causing recessive ALS and increased risk in heterozygotes. *Neurology*, doi: http://dx.doi.org/10.1212/WNL.0000000000002334 [Abstract]


• Morgan, E et al. (2016) Identification of Genetic Markers for Ventricular Septal Defects in Arabian Horses. *Vetmed Abstract*


### 2015 Publications

• Thompson, P. et al. (2015) ENIGMA and the individual: Predicting factors that affect the brain in 35 countries worldwide. *Neurolmage*, http://dx.doi.org/10.1016/j.neuroimage.2015.11.057 [Abstract]


- Cotteni, E. et al. (2015) Genetic and Functional Investigation of Inherited Neuropathies. MRC Centre for Neuromuscular Diseases Abstract
• Popp, N. et al. (2015) Functional single nucleotide polymorphism in IL-17A 3’ untranslated region is targeted by miR-4480 in vitro and may be associated with age-related macular degeneration. *Environmental and Molecular Mutagenesis*, doi: 10.1002/em.21982 Abstract


• Weaver, K, et al. (2015) Acrofacial Dysostosis, Cincinnati Type, a Mandibulofacial Dysostosis Syndrome with Limb Anomalies, Is Caused by POLR1A Dysfunction. *AJHG*, doi: 10.1016/j.ajhg.2015.03.011 Abstract


• Cierny, D, et al. (2015) Genetic variants in interleukin 7 receptor a chain (IL-7Ra) are associated with multiple sclerosis risk and disability progression in Central European Slovak population. *Journal of Neuroimmunology*, doi: http://dx.doi.org/10.1016/j.jneuroim.2015.03.010 Abstract


• Zhang, L et al. (2015) Exome Sequencing of Normal and Isogenic Transformed Human Colonic Epithelial Cells (HCECs) Reveals Novel Genes Potentially Involved in the Early...
2014 Publications


• Lee, K et al. (2014) Cytokine polymorphisms are associated with fatigue in adults living with HIV/AIDS. *Brain, Behavior, and Immunity*, doi:10.1016/j.bbi.2014.02.017. [Abstract]


• Taylor, K et al. (2014) Recurrent activating ACVR1 mutations in diffuse intrinsic pontine glioma. *Nature Genetics*, doi:10.1038/ng.2925. [Abstract]


2013 Publications


• Eun, Y et al. (2013) Interleukin 22 Polymorphisms and Papillary Thyroid Cancer. *Journal of Endocrinological Investigation*. Abstract


• Hanson, E et al. (2013) Genetic Variants of Coagulation Factor XI Show Association with Ischemic Stroke Up to 70 Years of Age. *PLoS ONE*, 8(9):e75286, doi:10.1371/journal.pone.0075286. [Abstract](#)


• Kim, S et al. (2013) Missense Polymorphisms in XIAP-Associated Factor-1 (XAF1) and Risk of Papillary Thyroid Cancer: Correlation with Clinicopathological Features. *Anticancer Research*, 33(5):2205-2210. [Abstract](#)


• Kim, Y et al. (2013) Genome-Wide Association Study Identified New Variants Associated with the Risk of Chronic Hepatitis B. *Human Molecular Genetics*, doi:10.1093/hmg/ddt266. [Abstract](#)

• Kim, Y et al. (2013) Genome-wide association analysis identifies new susceptibility loci for Behçet's disease and epistasis between HLA-B*51 and ERAP1. *Nature Genetics*, doi:10.1038/ng.2520. [Abstract](#)


• Lencz, T et al. (2013) Genome-wide association study implicates NDST3 in schizophrenia and bipolar disorder. *Nature Communications*, 4(2739), doi:10.1038/ncomms3739. [Abstract](#)


• Li, M et al. (2013) Tetra-primer ARMS-PCR is an efficient SNP genotyping method: with an example from SIRT2. *Analytical Methods*, doi:10.1039/c3ay41370e. [Abstract](#)


• Wang, M et al. (2013) A Novel Approach to Detect Cumulative Genetic Effects and Genetic Interactions in Crohn’s Disease. *Inflammatory Bowel Diseases*, doi:10.1097/MIB.0b013e31828706a0. [Abstract](#)


2012 Publications


• Choe, B et al. (2012) Polymorphisms of TGFBRI contribute to the progression of papillary thyroid carcinoma. *Molecular and Cellular Toxicology*, 8:1-8, doi:10.1007/s13273-012-0001-0. [Abstract]


• Kim, S et al. (2012) A missense polymorphism (rs11466653, Met326Thr) of toll-like receptor 10 (TLR10) is associated with tumor size of papillary thyroid carcinoma in the Korean population. Endocrine, doi:10.1007/s12020-012-9783-z. Abstract


• Koizumi, A et al. (2012) P.R4810K, a polymorphism of RNF213, the susceptibility gene for moyamoya disease, is associated with blood pressure. Environmental Health and Preventive Medicine, doi:10.1007/s12199-012-0299-1. Abstract


2011 Publications


• Adkins, R et al. (2011) Parental ages and levels of DNA methylation in the newborn are correlated. BMC Medical Genetics, 12:47, doi:10.1186/1471-2350-12-47. Abstract
• Bulayeva, K et al. (2011) Mapping Genes Related to Early Onset Major Depressive Disorder in Dagestan Genetic Isolates. Turkish Journal of Psychiatry. Abstract


• Carrol, E et al. (2011) The IL1RN Promoter rs4251961 Correlates with IL-1 Receptor Antagonist Concentrations in Human Infection and Is Differentially Regulated by GATA-1. *Journal of Immunology*, doi:10.4049/jimmunol.1002402. Abstract


• Kim, S et al. (2011) Promoter polymorphisms of the HLA-G gene, but not the HLA-E and HLA-F genes, is associated with non-segmental vitiligo patients in the Korean population. *Archives of Dermatological Research*, doi:10.1007/s00403-011-1160-x. [Abstract](#)


• Millan Sanchez, M et al. (2011) BDNF polymorphism predicts the rate of decline in skilled task performance and hippocampal volume in healthy individuals. Translational Psychiatry, doi:10.1038/tp.2011.47. Abstract


• Park, H et al. (2011) A polymorphism (rs2073287) of glutamate receptor, metabotropic 1 (GRM1) is associated with an increased risk of stroke in Korean population. Molecular & Cellular Toxicology, 7(3):243-250, doi:10.1007/s13273-011-0030-0. Abstract


• Sanchez, M et al. (2011) BDNF polymorphism predicts the rate of decline in skilled task performance and hippocampal volume in healthy individuals. *Translational Psychiatry*, 1(51), doi:10.1038/tp.2011.47. [Abstract]


• Teltsh, O et al. (2011) Oxytocin and vasopressin genes are significantly associated with schizophrenia in a large Arab-Israeli pedigree. *International Journal of Neuropsychopharmacology*, doi:10.1017/S1461145711001374. [Abstract]


### 2010 Publications


- Doshi, A et al. (2010) A Promoter Polymorphism of the Endothelial Nitric Oxide Synthase Gene is Associated With Reduced mRNA and Protein Expression in Failing Human Myocardium. *Journal of Cardiac Failure*. [Abstract](#)


• Kim, H et al. (2010) Analysis of copy number variation in 8,842 Korean individuals reveals 39 genes associated with hepatic biomarkers AST and ALT. BMB Reports, 43(8):547-553. Abstract


• Mizuki, N et al. (2010) Genome-wide association studies identify IL23R-IL12RB2 and IL10 as Behcet's disease susceptibility loci. *Nature Genetics*, doi:10.1038/ng.624. [Abstract]


• Smith, R et al. (2010) Nicotinic alpha-5 receptor subunit mRNA expression is associated with distant 5' upstream polymorphisms. *European Journal of Human Genetics*, doi:10.1038/ejhg.2010.120. [Abstract]


### 2009 Publications


• Chinoy, H et al. (2009) HLA*DPB1* associations differ between DRB1*03 positive anti-Jo-1 and anti-PM-Scl antibody positive idiopathic inflammatory myopathy. *Rheumatology*, doi:10.1093/rheumatology/kep248. [Abstract](#)


• Juhasz, G et al. (2009) CNR1 Gene is Associated with High Neuroticism and Low Agreeableness and Interacts with Recent Negative Life Events to Predict Current Depressive Symptoms. Neuropsychopharmacology, 34,2019?2027; doi:10.1038/npp.2009.19. Abstract


Park, S et al. (2009) Involvement of tryptophan hydroxylase 2 (TPH2) gene polymorphisms in susceptibility to coronary artery lesions in Korean children with Kawasaki


• Sombekke, M et al. (2009) HLA-DRB1*1501 and Spinal Cord Magnetic Resonance Imaging Lesions in Multiple Sclerosis. *Archives of Neurology*, 66(12):1531-1536. [Abstract](http://example.com)

• Sun, Y et al. (2009) A Common CNV on Chr 6 Association With the Gene Expression Level of Endothelin 1 in Transformed B Lymphocytes From Three Racial Groups. *Circulation: Cardiovascular Genetics*. [Abstract](http://example.com)


• Xiong, D et al. (2009) Genome-wide Association and Follow-Up Replication Studies Identified ADAMTS18 and TGFBR3 as Bone Mass Candidate Genes in Different Ethnic Groups. American Journal of Human Genetics. Abstract


2008 Publications


• Sissung, T et al. (2008) ABCB1 Genetic Variation Influences the Toxicity and Clinical Outcome of Patients with Androgen-Independent Prostate Cancer Treated with Docetaxel. *Clinical Cancer Research*, 14:4543-4549.


2007 Publications


• Chinoy, H et al. (2007) Tumour necrosis factor- single nucleotide polymorphisms are not independent of HLA class I in UK Caucasians with adult onset idiopathic inflammatory myopathies. *Rheumatology*.


• Knafo, A et al. (2007) Individual Differences in Allocation of Funds in the Dictator Game Associated with Length of the Arginine Vasopressin 1a Receptor (AVPR1a) RS3 Promoter-region and Correlation between RS3 Length and Hippocampal mRNA. *Genes, Brain and Behavior*.


• Owen, K et al. (2007) Common Variation in the LMNA Gene (Encoding Lamin A/C) and Type 2 Diabetes. *Diabetes*, 56(3):579-883, doi: 0.2337/db06-0930.


• Zhang, H et al. (2007) The OPRD1 and OPRK1 loci in alcohol or drug dependence: OPRD1 variation modulates substance dependence risk. *Molecular Psychiatry*.

2006 Publications

• Bleecker, E et al. (2006) Salmeterol response is not affected by \( \beta_2 \)-adrenergic receptor genotype in subjects with persistent asthma. *Journal of Allergy and Clinical Immunology*, 118(4):809-816, doi:10.1016/j.jaci.2006.06.036.


• Chinoy, H et al. (2006) In adult onset myositis, the presence of interstitial lung disease and myositis specific/associated antibodies are governed by HLA class II haplotype, rather than by myositis subtype. *Arthritis Research & Therapy*, 8:R13.


• Zeggini, E et al. (2006) Variation Within the Gene Encoding the Upstream Stimulatory Factor 1 Does Not Influence Susceptibility to Type 2 Diabetes in Samples From Populations With Replicated Evidence of Linkage to Chromosome 1q. *Diabetes*, 55(9):2541-2548, doi:10.2337/db06-0088.


**2005 Publications**


• Carroll, W et al. (2005) Maternal glutathione S-transferase GSTP 1 genotype is a specific predictor of phenotype in children with asthma. *Pediatric Allergy and Immunology*, 16(1):32-39.


• Zeggini, E et al. (2005) Examining the relationships between the Pro12Ala variant in PPARG and Type 2 diabetes-related traits in UK samples. *Diabetic Medicine*, 22(12):1696-1700.

2004 Publications


• Strange, R et al. (2004) PTCH Polymorphism Is Associated With the Rate of Increase in Basal Cell Carcinoma Numbers During Follow-Up. *Environmental and Molecular Mutagenesis*, 44:469-476.


2003 Publications


