# David A Van Heel

## List of Publications by Citations



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|---|---|---|---|
| 91 papers | 11,384 citations | 52 h-index | 96 g-index |
| 96 ext. papers | 13,268 ext. citations | 17.5 avg, IF | 5.3 L-index |

| # | Paper | IF | Citations |
|---|---|---|---|
| 91 | Multiple common variants for celiac disease influencing immune gene expression. *Nature Genetics*, **2010**, 42, 295-302 | 36.3 | 727 |
| 90 | Diagnosis and management of adult coeliac disease: guidelines from the British Society of Gastroenterology. *Gut*, **2014**, 63, 1210-28 | 19.2 | 660 |
| 89 | Shared and distinct genetic variants in type 1 diabetes and celiac disease. *New England Journal of Medicine*, **2008**, 359, 2767-77 | 59.2 | 546 |
| 88 | Dense genotyping identifies and localizes multiple common and rare variant association signals in celiac disease. *Nature Genetics*, **2011**, 43, 1193-201 | 36.3 | 535 |
| 87 | Newly identified genetic risk variants for celiac disease related to the immune response. *Nature Genetics*, **2008**, 40, 395-402 | 36.3 | 524 |
| 86 | A genome-wide association study for celiac disease identifies risk variants in the region harboring IL2 and IL21. *Nature Genetics*, **2007**, 39, 827-9 | 36.3 | 518 |
| 85 | A genome-wide meta-analysis identifies 22 loci associated with eight hematological parameters in the HaemGen consortium. *Nature Genetics*, **2009**, 41, 1182-90 | 36.3 | 433 |
| 84 | Pervasive sharing of genetic effects in autoimmune disease. *PLoS Genetics*, **2011**, 7, e1002254 | 6 | 413 |
| 83 | Genetic insights into common pathways and complex relationships among immune-mediated diseases. *Nature Reviews Genetics*, **2013**, 14, 661-73 | 30.1 | 394 |
| 82 | Comprehensive, quantitative mapping of T cell epitopes in gluten in celiac disease. *Science Translational Medicine*, **2010**, 2, 41ra51 | 17.5 | 321 |
| 81 | Muramyl dipeptide and toll-like receptor sensitivity in NOD2-associated Crohn's disease. *Lancet, The*, **2005**, 365, 1794-6 | 40 | 264 |
| 80 | Trans-eQTLs reveal that independent genetic variants associated with a complex phenotype converge on intermediate genes, with a major role for the HLA. *PLoS Genetics*, **2011**, 7, e1002197 | 6 | 261 |
| 79 | Association between a complex insertion/deletion polymorphism in NOD1 (CARD4) and susceptibility to inflammatory bowel disease. *Human Molecular Genetics*, **2005**, 14, 1245-50 | 5.6 | 260 |
| 78 | Inflammatory skin and bowel disease linked to ADAM17 deletion. *New England Journal of Medicine*, **2011**, 365, 1502-8 | 59.2 | 247 |
| 77 | Association Between Telomere Length and Risk of Cancer and Non-Neoplastic Diseases: A Mendelian Randomization Study. *JAMA Oncology*, **2017**, 3, 636-651 | 13.4 | 236 |
| 76 | Inflammatory bowel disease is associated with a TNF polymorphism that affects an interaction between the OCT1 and NF(-kappa)B transcription factors. *Human Molecular Genetics*, **2002**, 11, 1281-9 | 5.6 | 214 |
| 75 | Recent advances in coeliac disease. *Gut*, **2006**, 55, 1037-46 | 19.2 | 210 |

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| 74 | Inflammatory bowel disease susceptibility loci defined by genome scan meta-analysis of 1952 affected relative pairs. *Human Molecular Genetics*, **2004**, 13, 763-70 | 5.6 | 198 |
| 73 | Genetic analysis of innate immunity in Crohn's disease and ulcerative colitis identifies two susceptibility loci harboring CARD9 and IL18RAP. *American Journal of Human Genetics*, **2008**, 82, 1202-10 | 11 | 196 |
| 72 | Health and population effects of rare gene knockouts in adult humans with related parents. *Science* , **2016**, 352, 474-7 | 33.3 | 185 |
| 71 | Identification of rare sequence variation underlying heritable pulmonary arterial hypertension. *Nature Communications*, **2018**, 9, 1416 | 17.4 | 182 |
| 70 | Novel association in chromosome 4q27 region with rheumatoid arthritis and confirmation of type 1 diabetes point to a general risk locus for autoimmune diseases. *American Journal of Human Genetics* , **2007**, 81, 1284-8 | 11 | 171 |
| 69 | Negligible impact of rare autoimmune-locus coding-region variants on missing heritability. *Nature*, **2013**, 498, 232-5 | 50.4 | 156 |
| 68 | Coeliac disease-associated risk variants in TNFAIP3 and REL implicate altered NF-kappaB signalling. *Gut*, **2009**, 58, 1078-83 | 19.2 | 147 |
| 67 | A meta-analysis of genome-wide association scans identifies IL18RAP, PTPN2, TAGAP, and PUS10 as shared risk loci for Crohn's disease and celiac disease. *PLoS Genetics*, **2011**, 7, e1001283 | 6 | 142 |
| 66 | HLA-DQA1-HLA-DRB1 variants confer susceptibility to pancreatitis induced by thiopurine immunosuppressants. *Nature Genetics*, **2014**, 46, 1131-4 | 36.3 | 130 |
| 65 | Evolutionary and functional analysis of celiac risk loci reveals SH2B3 as a protective factor against bacterial infection. *American Journal of Human Genetics*, **2010**, 86, 970-7 | 11 | 130 |
| 64 | A structural and immunological basis for the role of human leukocyte antigen DQ8 in celiac disease. *Immunity*, **2007**, 27, 23-34 | 32.3 | 128 |
| 63 | Synergistic enhancement of Toll-like receptor responses by NOD1 activation. *European Journal of Immunology*, **2005**, 35, 2471-6 | 6.1 | 127 |
| 62 | Associations with tight junction genes PARD3 and MAGI2 in Dutch patients point to a common barrier defect for coeliac disease and ulcerative colitis. *Gut*, **2008**, 57, 463-7 | 19.2 | 119 |
| 61 | Effective detection of human leukocyte antigen risk alleles in celiac disease using tag single nucleotide polymorphisms. *PLoS ONE*, **2008**, 3, e2270 | 3.7 | 113 |
| 60 | Widespread non-additive and interaction effects within HLA loci modulate the risk of autoimmune diseases. *Nature Genetics*, **2015**, 47, 1085-90 | 36.3 | 112 |
| 59 | Common and different genetic background for rheumatoid arthritis and coeliac disease. *Human Molecular Genetics*, **2009**, 18, 4195-203 | 5.6 | 109 |
| 58 | Analysis of HLA and non-HLA alleles can identify individuals at high risk for celiac disease. *Gastroenterology*, **2009**, 137, 834-40, 840.e1-3 | 13.3 | 103 |
| 57 | T cells in peripheral blood after gluten challenge in coeliac disease. *Gut*, **2005**, 54, 1217-23 | 19.2 | 101 |

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| 56 | Investigating the Causal Relationship of C-Reactive Protein with 32 Complex Somatic and Psychiatric Outcomes: A Large-Scale Cross-Consortium Mendelian Randomization Study. *PLoS Medicine*, **2016**, 13, e1001976 | 11.6 | 100 |
| 55 | Fine mapping in the MHC region accounts for 18% additional genetic risk for celiac disease. *Nature Genetics*, **2015**, 47, 577-8 | 36.3 | 99 |
| 54 | Improving coeliac disease risk prediction by testing non-HLA variants additional to HLA variants. *Gut*, **2014**, 63, 415-22 | 19.2 | 92 |
| 53 | Synergy between TLR9 and NOD2 innate immune responses is lost in genetic Crohn's disease. *Gut*, **2005**, 54, 1553-7 | 19.2 | 92 |
| 52 | Trans-ethnic and Ancestry-Specific Blood-Cell Genetics in 746,667 Individuals from 5 Global Populations. *Cell*, **2020**, 182, 1198-1213.e14 | 56.2 | 88 |
| 51 | Analysis of the IBD5 locus and potential gene-gene interactions in Crohn's disease. *Gut*, **2003**, 52, 541-6 | 19.2 | 85 |
| 50 | Genetics in coeliac disease. *Bailliere's Best Practice and Research in Clinical Gastroenterology*, **2005**, 19, 323-39 | 2.5 | 78 |
| 49 | Mutations in CSTA, encoding Cystatin A, underlie exfoliative ichthyosis and reveal a role for this protease inhibitor in cell-cell adhesion. *American Journal of Human Genetics*, **2011**, 89, 564-71 | 11 | 68 |
| 48 | Modulation of dendritic cell phenotype and function in an in vitro model of the intestinal epithelium. *European Journal of Immunology*, **2006**, 36, 864-74 | 6.1 | 67 |
| 47 | A common CTLA4 haplotype associated with coeliac disease. *European Journal of Human Genetics*, **2005**, 13, 440-4 | 5.3 | 65 |
| 46 | Replication of celiac disease UK genome-wide association study results in a US population. *Human Molecular Genetics*, **2009**, 18, 4219-25 | 5.6 | 60 |
| 45 | DNAJC21 Mutations Link a Cancer-Prone Bone Marrow Failure Syndrome to Corruption in 60S Ribosome Subunit Maturation. *American Journal of Human Genetics*, **2016**, 99, 115-24 | 11 | 55 |
| 44 | Crohn's disease: genetic susceptibility, bacteria, and innate immunity. *Lancet, The*, **2001**, 357, 1902-4 | 40 | 55 |
| 43 | Evaluating drug targets through human loss-of-function genetic variation. *Nature*, **2020**, 581, 459-464 | 50.4 | 53 |
| 42 | Further evidence of IBD5/CARD15 (NOD2) epistasis in the susceptibility to ulcerative colitis. *American Journal of Human Genetics*, **2003**, 73, 1465-6 | 11 | 53 |
| 41 | NOD2 (CARD15), the first susceptibility gene for Crohn's disease. *Gut*, **2001**, 49, 752-4 | 19.2 | 53 |
| 40 | Estimating the human mutation rate from autozygous segments reveals population differences in human mutational processes. *Nature Communications*, **2017**, 8, 303 | 17.4 | 52 |
| 39 | Antagonists and non-toxic variants of the dominant wheat gliadin T cell epitope in coeliac disease. *Gut*, **2006**, 55, 485-91 | 19.2 | 49 |

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| 38 | Lack of association of MYO9B genetic variants with coeliac disease in a British cohort. *Gut*, **2006**, 55, 969-72 | 19.2 | 49 |
| 37 | Selective NOD1 agonists cause shock and organ injury/dysfunction in vivo. *American Journal of Respiratory and Critical Care Medicine*, **2007**, 175, 595-603 | 10.2 | 49 |
| 36 | Calcium channel TRPV6 expression in human duodenum: different relationships to the vitamin D system and aging in men and women. *Journal of Bone and Mineral Research*, **2006**, 21, 1770-7 | 6.3 | 45 |
| 35 | Comparative methylomics reveals gene-body H3K36me3 in Drosophila predicts DNA methylation and CpG landscapes in other invertebrates. *Genome Research*, **2011**, 21, 1841-50 | 9.7 | 44 |
| 34 | The IBD6 Crohnß disease locus demonstrates complex interactions with CARD15 and IBD5 disease-associated variants. *Human Molecular Genetics*, **2003**, 12, 2569-75 | 5.6 | 44 |
| 33 | Associations of autozygosity with a broad range of human phenotypes. *Nature Communications*, **2019**, 10, 4957 | 17.4 | 40 |
| 32 | The genetics of chronic inflammatory diseases. *Human Molecular Genetics*, **2009**, 18, R101-6 | 5.6 | 40 |
| 31 | Genetics and pathogenesis of coeliac disease. *Seminars in Immunology*, **2009**, 21, 346-54 | 10.7 | 40 |
| 30 | Translational mini-review series on the immunogenetics of gut disease: immunogenetics of coeliac disease. *Clinical and Experimental Immunology*, **2008**, 153, 162-73 | 6.2 | 38 |
| 29 | Formalising recall by genotype as an efficient approach to detailed phenotyping and causal inference. *Nature Communications*, **2018**, 9, 711 | 17.4 | 35 |
| 28 | Detection, imputation, and association analysis of small deletions and null alleles on oligonucleotide arrays. *American Journal of Human Genetics*, **2008**, 82, 1316-33 | 11 | 32 |
| 27 | Association study of IL2/IL21 and FcgRIIa: significant association with the IL2/IL21 region in Scandinavian coeliac disease families. *Genes and Immunity*, **2008**, 9, 364-7 | 4.4 | 30 |
| 26 | Interleukin 15: its role in intestinal inflammation. *Gut*, **2006**, 55, 444-5 | 19.2 | 29 |
| 25 | Association study of the IL18RAP locus in three European populations with coeliac disease. *Human Molecular Genetics*, **2009**, 18, 1148-55 | 5.6 | 27 |
| 24 | The power of genetic diversity in genome-wide association studies of lipids. *Nature*, **2021**, | 50.4 | 24 |
| 23 | Exome Sequencing and Rare Variant Analysis Reveals Multiple Filaggrin Mutations in Bangladeshi Families with Atopic Eczema and Additional Risk Genes. *Journal of Investigative Dermatology*, **2018**, 138, 2674-2677 | 4.3 | 19 |
| 22 | Recent advances in coeliac disease genetics. *Gut*, **2009**, 58, 473-6 | 19.2 | 19 |
| 21 | Detection of muramyl dipeptide-sensing pathway defects in patients with Crohnß disease. *Inflammatory Bowel Diseases*, **2006**, 12, 598-605 | 4.5 | 19 |

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| 20 | Characterising a healthy adult with a rare HAO1 knockout to support a therapeutic strategy for primary hyperoxaluria. *ELife*, **2020**, 9, | 8.9 | 19 |
| 19 | Cohort Profile: East London Genes & Health (ELGH), a community-based population genomics and health study in British Bangladeshi and British Pakistani people. *International Journal of Epidemiology*, **2020**, 49, 20-21i | 7.8 | 19 |
| 18 | Identification of novel polymorphisms in the beta7 integrin gene: family-based association studies in inflammatory bowel disease. *Genes and Immunity*, **2001**, 2, 455-60 | 4.4 | 16 |
| 17 | NOD2 activity modulates the phenotype of LPS-stimulated dendritic cells to promote the development of T-helper type 2-like lymphocytes - Possible implications for NOD2-associated Crohn's disease. *Journal of Crohn's and Colitis*, **2007**, 1, 106-15 | 1.5 | 14 |
| 16 | Normal responses to specific NOD1-activating peptidoglycan agonists in the presence of the NOD2 frameshift and other mutations in Crohn's disease. *European Journal of Immunology*, **2006**, 36, 1629-35 | 6.1 | 12 |
| 15 | Evaluating potential drug targets through human loss-of-function genetic variation | | 12 |
| 14 | Human pancreatic secretory trypsin inhibitor stabilizes intestinal mucosa against noxious agents. *American Journal of Pathology*, **2007**, 171, 1462-73 | 5.8 | 11 |
| 13 | A direct multi-generational estimate of the human mutation rate from autozygous segments seen in thousands of parentally related individuals | | 11 |
| 12 | MC3R links nutritional state to childhood growth and the timing of puberty. *Nature*, **2021**, 599, 436-441 | 50.4 | 9 |
| 11 | Exome sequencing of 75 individuals from multiply affected coeliac families and large scale resequencing follow up. *PLoS ONE*, **2015**, 10, e0116845 | 3.7 | 8 |
| 10 | A single nucleotide polymorphism genetic risk score to aid diagnosis of coeliac disease: a pilot study in clinical care. *Alimentary Pharmacology and Therapeutics*, **2020**, 52, 1165-1173 | 6.1 | 8 |
| 9 | Association of TNF-alpha-857C with inflammatory bowel disease in the Australian population. *Scandinavian Journal of Gastroenterology*, **2003**, 38, 533-4 | 2.4 | 8 |
| 8 | Cohort Profile: East London Genes & Health (ELGH), a community based population genomics and health study of British-Bangladeshi and British-Pakistani people | | 6 |
| 7 | Detecting the risks of osteoporotic fractures in coeliac disease. *Gut*, **2003**, 52, 1229-30 | 19.2 | 5 |
| 6 | Deep phenotyping of a healthy human HAO1 knockout informs therapeutic development for primary hyperoxaluria type 1 | | 5 |
| 5 | Transferability of genetic loci and polygenic scores for cardiometabolic traits in British Pakistanis and Bangladeshis | | 3 |
| 4 | Colonoscopic appearances and diagnosis of intussusception due to large-bowel lipoma. *Endoscopy*, **1999**, 31, 508 | 3.4 | 3 |
| 3 | Fine-scale population structure and demographic history of British Pakistanis | | 1 |

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| 2 | Integrating polygenic risk scores in the prediction of type 2 diabetes risk and subtypes in British Pakistanis and Bangladeshis: A population-based cohort study.. *PLoS Medicine*, **2022**, 19, e1003981 | 11.6 | 1 |
| 1 | Fine-scale population structure and demographic history of British Pakistanis. *Nature Communications*, **2021**, 12, 7189 | 17.4 | 0 |