# Thorunn Rafnar

## List of Publications by Citations



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| 83 papers | 12,472 citations | 48 h-index | 89 g-index |
| 89 ext. papers | 14,403 ext. citations | 22.5 avg, IF | 4.31 L-index |

| # | Paper | IF | Citations |
|---|---|---|---|
| 83 | A variant associated with nicotine dependence, lung cancer and peripheral arterial disease. *Nature*, **2008**, 452, 638-642 | 50.4 | 1239 |
| 82 | Genome-wide association study identifies a second prostate cancer susceptibility variant at 8q24. *Nature Genetics*, **2007**, 39, 631-7 | 36.3 | 739 |
| 81 | Common variants on chromosomes 2q35 and 16q12 confer susceptibility to estrogen receptor-positive breast cancer. *Nature Genetics*, **2007**, 39, 865-9 | 36.3 | 715 |
| 80 | Two variants on chromosome 17 confer prostate cancer risk, and the one in TCF2 protects against type 2 diabetes. *Nature Genetics*, **2007**, 39, 977-83 | 36.3 | 616 |
| 79 | Many sequence variants affecting diversity of adult human height. *Nature Genetics*, **2008**, 40, 609-15 | 36.3 | 522 |
| 78 | Sequence variants at the TERT-CLPTM1L locus associate with many cancer types. *Nature Genetics*, **2009**, 41, 221-7 | 36.3 | 509 |
| 77 | Large-scale whole-genome sequencing of the Icelandic population. *Nature Genetics*, **2015**, 47, 435-44 | 36.3 | 486 |
| 76 | A genome-wide association study of lung cancer identifies a region of chromosome 5p15 associated with risk for adenocarcinoma. *American Journal of Human Genetics*, **2009**, 85, 679-91 | 11 | 442 |
| 75 | A multi-stage genome-wide association study of bladder cancer identifies multiple susceptibility loci. *Nature Genetics*, **2010**, 42, 978-84 | 36.3 | 408 |
| 74 | Sequence variant on 8q24 confers susceptibility to urinary bladder cancer. *Nature Genetics*, **2008**, 40, 1307-12 | 36.3 | 332 |
| 73 | Detection of sharing by descent, long-range phasing and haplotype imputation. *Nature Genetics*, **2008**, 40, 1068-75 | 36.3 | 329 |
| 72 | Common variants on 9q22.33 and 14q13.3 predispose to thyroid cancer in European populations. *Nature Genetics*, **2009**, 41, 460-4 | 36.3 | 308 |
| 71 | Mutations in BRIP1 confer high risk of ovarian cancer. *Nature Genetics*, **2011**, 43, 1104-7 | 36.3 | 285 |
| 70 | Genome-wide association and replication studies identify four variants associated with prostate cancer susceptibility. *Nature Genetics*, **2009**, 41, 1122-6 | 36.3 | 281 |
| 69 | A genome-wide association study identifies susceptibility loci for ovarian cancer at 2q31 and 8q24. *Nature Genetics*, **2010**, 42, 874-9 | 36.3 | 277 |
| 68 | New common variants affecting susceptibility to basal cell carcinoma. *Nature Genetics*, **2009**, 41, 909-14 | 36.3 | 275 |
| 67 | ASIP and TYR pigmentation variants associate with cutaneous melanoma and basal cell carcinoma. *Nature Genetics*, **2008**, 40, 886-91 | 36.3 | 265 |

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| 66 | Large-scale association analysis identifies new lung cancer susceptibility loci and heterogeneity in genetic susceptibility across histological subtypes. *Nature Genetics*, **2017**, 49, 1126-1132 | 36.3 | 246 |
| 65 | Rare variants of large effect in BRCA2 and CHEK2 affect risk of lung cancer. *Nature Genetics*, **2014**, 46, 736-41 | 36.3 | 228 |
| 64 | A rare variant in MYH6 is associated with high risk of sick sinus syndrome. *Nature Genetics*, **2011**, 43, 316-20 | 36.3 | 228 |
| 63 | Common variants at 19p13 are associated with susceptibility to ovarian cancer. *Nature Genetics*, **2010**, 42, 880-4 | 36.3 | 210 |
| 62 | A germline variant in the TP53 polyadenylation signal confers cancer susceptibility. *Nature Genetics*, **2011**, 43, 1098-103 | 36.3 | 203 |
| 61 | Nonsense mutation in the LGR4 gene is associated with several human diseases and other traits. *Nature*, **2013**, 497, 517-20 | 50.4 | 192 |
| 60 | Genome-wide association study identifies sequence variants on 6q21 associated with age at menarche. *Nature Genetics*, **2009**, 41, 734-8 | 36.3 | 169 |
| 59 | Discovery of common variants associated with low TSH levels and thyroid cancer risk. *Nature Genetics*, **2012**, 44, 319-22 | 36.3 | 167 |
| 58 | Variants with large effects on blood lipids and the role of cholesterol and triglycerides in coronary disease. *Nature Genetics*, **2016**, 48, 634-9 | 36.3 | 162 |
| 57 | Genome-wide association study identifies a sequence variant within the DAB2IP gene conferring susceptibility to abdominal aortic aneurysm. *Nature Genetics*, **2010**, 42, 692-7 | 36.3 | 155 |
| 56 | Replication of lung cancer susceptibility loci at chromosomes 15q25, 5p15, and 6p21: a pooled analysis from the International Lung Cancer Consortium. *Journal of the National Cancer Institute*, **2010**, 102, 959-71 | 9.7 | 153 |
| 55 | A study based on whole-genome sequencing yields a rare variant at 8q24 associated with prostate cancer. *Nature Genetics*, **2012**, 44, 1326-9 | 36.3 | 151 |
| 54 | A sequence variant at 4p16.3 confers susceptibility to urinary bladder cancer. *Nature Genetics*, **2010** , 42, 415-9 | 36.3 | 138 |
| 53 | Genetic correction of PSA values using sequence variants associated with PSA levels. *Science Translational Medicine*, **2010**, 2, 62ra92 | 17.5 | 125 |
| 52 | Identification of low-frequency variants associated with gout and serum uric acid levels. *Nature Genetics*, **2011**, 43, 1127-30 | 36.3 | 117 |
| 51 | European genome-wide association study identifies SLC14A1 as a new urinary bladder cancer susceptibility gene. *Human Molecular Genetics*, **2011**, 20, 4268-81 | 5.6 | 105 |
| 50 | Genome-wide association study identifies multiple susceptibility loci for multiple myeloma. *Nature Communications*, **2016**, 7, 12050 | 17.4 | 101 |
| 49 | Loss-of-function variants in ATM confer risk of gastric cancer. *Nature Genetics*, **2015**, 47, 906-10 | 36.3 | 100 |

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| 48 | Variant ASGR1 Associated with a Reduced Risk of Coronary Artery Disease. *New England Journal of Medicine*, **2016**, 374, 2131-41 | 59.2 | 94 |
| 47 | Common variants on 1p36 and 1q42 are associated with cutaneous basal cell carcinoma but not with melanoma or pigmentation traits. *Nature Genetics*, **2008**, 40, 1313-8 | 36.3 | 93 |
| 46 | A genome-wide association study yields five novel thyroid cancer risk loci. *Nature Communications*, **2017**, 8, 14517 | 17.4 | 80 |
| 45 | Ancestry-shift refinement mapping of the C6orf97-ESR1 breast cancer susceptibility locus. *PLoS Genetics*, **2010**, 6, e1001029 | 6 | 72 |
| 44 | Meta-analysis of Icelandic and UK data sets identifies missense variants in SMO, IL11, COL11A1 and 13 more new loci associated with osteoarthritis. *Nature Genetics*, **2018**, 50, 1681-1687 | 36.3 | 67 |
| 43 | Cross-Cancer Genome-Wide Analysis of Lung, Ovary, Breast, Prostate, and Colorectal Cancer Reveals Novel Pleiotropic Associations. *Cancer Research*, **2016**, 76, 5103-14 | 10.1 | 66 |
| 42 | Whole genome characterization of sequence diversity of 15,220 Icelanders. *Scientific Data*, **2017**, 4, 170184 | 8.5 | 64 |
| 41 | A Missense Variant in PTPN22 is a Risk Factor for Drug-induced Liver Injury. *Gastroenterology*, **2019**, 156, 1707-1716.e2 | 13.3 | 59 |
| 40 | Identification of multiple risk loci and regulatory mechanisms influencing susceptibility to multiple myeloma. *Nature Communications*, **2018**, 9, 3707 | 17.4 | 57 |
| 39 | Whole-genome sequencing identifies rare genotypes in COMP and CHADL associated with high risk of hip osteoarthritis. *Nature Genetics*, **2017**, 49, 801-805 | 36.3 | 56 |
| 38 | Insights into imprinting from parent-of-origin phased methylomes and transcriptomes. *Nature Genetics*, **2018**, 50, 1542-1552 | 36.3 | 56 |
| 37 | Variants in ELL2 influencing immunoglobulin levels associate with multiple myeloma. *Nature Communications*, **2015**, 6, 7213 | 17.4 | 54 |
| 36 | New basal cell carcinoma susceptibility loci. *Nature Communications*, **2015**, 6, 6825 | 17.4 | 49 |
| 35 | A common variant at 8q24.21 is associated with renal cell cancer. *Nature Communications*, **2013**, 4, 2776 | 17.4 | 48 |
| 34 | A loss-of-function variant in ALOX15 protects against nasal polyps and chronic rhinosinusitis. *Nature Genetics*, **2019**, 51, 267-276 | 36.3 | 44 |
| 33 | Sequence variants in the PTCH1 gene associate with spine bone mineral density and osteoporotic fractures. *Nature Communications*, **2016**, 7, 10129 | 17.4 | 41 |
| 32 | BRCA2, but not BRCA1, mutations account for familial ovarian cancer in Iceland: a population-based study. *European Journal of Cancer*, **2004**, 40, 2788-93 | 7.5 | 41 |
| 31 | Epigenetic and genetic components of height regulation. *Nature Communications*, **2016**, 7, 13490 | 17.4 | 39 |

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| 30 | Germline sequence variants in TGM3 and RGS22 confer risk of basal cell carcinoma. *Human Molecular Genetics*, **2014**, 23, 3045-53 | 5.6 | 39 |
| 29 | Sequence variants in ARHGAP15, COLQ and FAM155A associate with diverticular disease and diverticulitis. *Nature Communications*, **2017**, 8, 15789 | 17.4 | 37 |
| 28 | Clinical characteristics of patients with colorectal cancer with double somatic mismatch repair mutations compared with Lynch syndrome. *Journal of Medical Genetics*, **2019**, 56, 462-470 | 5.8 | 31 |
| 27 | Variants associating with uterine leiomyoma highlight genetic background shared by various cancers and hormone-related traits. *Nature Communications*, **2018**, 9, 3636 | 17.4 | 31 |
| 26 | Genome-wide association meta-analysis yields 20 loci associated with gallstone disease. *Nature Communications*, **2018**, 9, 5101 | 17.4 | 29 |
| 25 | Evaluation of association of HNF1B variants with diverse cancers: collaborative analysis of data from 19 genome-wide association studies. *PLoS ONE*, **2010**, 5, e10858 | 3.7 | 24 |
| 24 | Genome-wide associations for benign prostatic hyperplasia reveal a genetic correlation with serum levels of PSA. *Nature Communications*, **2018**, 9, 4568 | 17.4 | 24 |
| 23 | FLT3 stop mutation increases FLT3 ligand level and risk of autoimmune thyroid disease. *Nature*, **2020**, 584, 619-623 | 50.4 | 23 |
| 22 | Genome-wide significant association between a sequence variant at 15q15.2 and lung cancer risk. *Cancer Research*, **2011**, 71, 1356-61 | 10.1 | 21 |
| 21 | Identification of a novel susceptibility locus at 13q34 and refinement of the 20p12.2 region as a multi-signal locus associated with bladder cancer risk in individuals of European ancestry. *Human Molecular Genetics*, **2016**, 25, 1203-14 | 5.6 | 20 |
| 20 | A Splice Region Variant in LDLR Lowers Non-high Density Lipoprotein Cholesterol and Protects against Coronary Artery Disease. *PLoS Genetics*, **2015**, 11, e1005379 | 6 | 17 |
| 19 | Large-scale integration of the plasma proteome with genetics and disease. *Nature Genetics*, **2021**, 53, 1712-1721 | 36.3 | 17 |
| 18 | Association of BRCA2 K3326* With Small Cell Lung Cancer and Squamous Cell Cancer of the Skin. *Journal of the National Cancer Institute*, **2018**, 110, 967-974 | 9.7 | 16 |
| 17 | The Icelandic Cancer Project--a population-wide approach to studying cancer. *Nature Reviews Cancer*, **2004**, 4, 488-92 | 31.3 | 12 |
| 16 | Sequence variant at 4q25 near PITX2 associates with appendicitis. *Scientific Reports*, **2017**, 7, 3119 | 4.9 | 10 |
| 15 | A method for detecting long non-coding RNAs with tiled RNA expression microarrays. *PLoS ONE*, **2014**, 9, e99899 | 3.7 | 9 |
| 14 | Predicted loss and gain of function mutations in ACO1 are associated with erythropoiesis. *Communications Biology*, **2020**, 3, 189 | 6.7 | 8 |
| 13 | Fourteen sequence variants that associate with multiple sclerosis discovered by meta-analysis informed by genetic correlations. *Npj Genomic Medicine*, **2017**, 2, 24 | 6.2 | 8 |

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| 12 | Genome-wide association identifies seven loci for pelvic organ prolapse in Iceland and the UK Biobank. *Communications Biology*, **2020**, 3, 129 | 6.7 | 7 |
| 11 | The CRTAC1 Protein in Plasma Is Associated With Osteoarthritis and Predicts Progression to Joint Replacement: A Large-Scale Proteomics Scan in Iceland. *Arthritis and Rheumatology*, **2021**, 73, 2025-2034 | 9.5 | 6 |
| 10 | Identification of 22 susceptibility loci associated with testicular germ cell tumors. *Nature Communications*, **2021**, 12, 4487 | 17.4 | 5 |
| 9 | Profile of common prostate cancer risk variants in an unscreened Romanian population. *Journal of Cellular and Molecular Medicine*, **2018**, 22, 1574-1582 | 5.6 | 4 |
| 8 | A PRPH splice-donor variant associates with reduced sural nerve amplitude and risk of peripheral neuropathy. *Nature Communications*, **2019**, 10, 1777 | 17.4 | 3 |
| 7 | Common and Rare Sequence Variants Influencing Tumor Biomarkers in Blood. *Cancer Epidemiology Biomarkers and Prevention*, **2020**, 29, 225-235 | 4 | 3 |
| 6 | Comprehensive population-wide detection of Lynch syndrome in Iceland.. *Journal of Clinical Oncology*, **2016**, 34, 1542-1542 | 2.2 | 2 |
| 5 | Predicting the probability of death using proteomics. *Communications Biology*, **2021**, 4, 758 | 6.7 | 2 |
| 4 | Loss-of-Function Variants in the Tumor-Suppressor Gene Confer Increased Cancer Risk. *Cancer Research*, **2021**, 81, 1954-1964 | 10.1 | 2 |
| 3 | Germline variants at SOHLH2 influence multiple myeloma risk. *Blood Cancer Journal*, **2021**, 11, 76 | 7 | 1 |
| 2 | Functional dissection of inherited non-coding variation influencing multiple myeloma risk.. *Nature Communications*, **2022**, 13, 151 | 17.4 | 0 |
| 1 | Genetic variants associated with platelet count are predictive of human disease and physiological markers. *Communications Biology*, **2021**, 4, 1132 | 6.7 | 0 |