

# Thorunn Rafnar

## List of Publications by Year in descending order

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Version: 2024-02-01

84  
papers

15,832  
citations

30070

54  
h-index

53230

85  
g-index

89  
all docs

89  
docs citations

89  
times ranked

21818  
citing authors

#	ARTICLE	IF	CITATIONS
1	A variant associated with nicotine dependence, lung cancer and peripheral arterial disease. <i>Nature</i> , 2008, 452, 638-642.	27.8	1,399
2	Genome-wide association study identifies a second prostate cancer susceptibility variant at 8q24. <i>Nature Genetics</i> , 2007, 39, 631-637.	21.4	818
3	Common variants on chromosomes 2q35 and 16q12 confer susceptibility to estrogen receptor-positive breast cancer. <i>Nature Genetics</i> , 2007, 39, 865-869.	21.4	774
4	Two variants on chromosome 17 confer prostate cancer risk, and the one in TCF2 protects against type 2 diabetes. <i>Nature Genetics</i> , 2007, 39, 977-983.	21.4	670
5	Large-scale whole-genome sequencing of the Icelandic population. <i>Nature Genetics</i> , 2015, 47, 435-444.	21.4	663
6	Many sequence variants affecting diversity of adult human height. <i>Nature Genetics</i> , 2008, 40, 609-615.	21.4	615
7	Sequence variants at the TERT-CLPTM1L locus associate with many cancer types. <i>Nature Genetics</i> , 2009, 41, 221-227.	21.4	572
8	A multi-stage genome-wide association study of bladder cancer identifies multiple susceptibility loci. <i>Nature Genetics</i> , 2010, 42, 978-984.	21.4	493
9	A Genome-wide Association Study of Lung Cancer Identifies a Region of Chromosome 5p15 Associated with Risk for Adenocarcinoma. <i>American Journal of Human Genetics</i> , 2009, 85, 679-691.	6.2	489
10	Large-scale association analysis identifies new lung cancer susceptibility loci and heterogeneity in genetic susceptibility across histological subtypes. <i>Nature Genetics</i> , 2017, 49, 1126-1132.	21.4	472
11	Detection of sharing by descent, long-range phasing and haplotype imputation. <i>Nature Genetics</i> , 2008, 40, 1068-1075.	21.4	409
12	Sequence variant on 8q24 confers susceptibility to urinary bladder cancer. <i>Nature Genetics</i> , 2008, 40, 1307-1312.	21.4	377
13	Rare variants of large effect in BRCA2 and CHEK2 affect risk of lung cancer. <i>Nature Genetics</i> , 2014, 46, 736-741.	21.4	360
14	Common variants on 9q22.33 and 14q13.3 predispose to thyroid cancer in European populations. <i>Nature Genetics</i> , 2009, 41, 460-464.	21.4	353
15	Large-scale integration of the plasma proteome with genetics and disease. <i>Nature Genetics</i> , 2021, 53, 1712-1721.	21.4	340
16	Mutations in BRIP1 confer high risk of ovarian cancer. <i>Nature Genetics</i> , 2011, 43, 1104-1107.	21.4	338
17	A genome-wide association study identifies susceptibility loci for ovarian cancer at 2q31 and 8q24. <i>Nature Genetics</i> , 2010, 42, 874-879.	21.4	321
18	Genome-wide association and replication studies identify four variants associated with prostate cancer susceptibility. <i>Nature Genetics</i> , 2009, 41, 1122-1126.	21.4	313

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19	ASIP and TYR pigmentation variants associate with cutaneous melanoma and basal cell carcinoma. <i>Nature Genetics</i> , 2008, 40, 886-891.	21.4	306
20	New common variants affecting susceptibility to basal cell carcinoma. <i>Nature Genetics</i> , 2009, 41, 909-914.	21.4	303
21	A rare variant in MYH6 is associated with high risk of sick sinus syndrome. <i>Nature Genetics</i> , 2011, 43, 316-320.	21.4	275
22	A germline variant in the TP53 polyadenylation signal confers cancer susceptibility. <i>Nature Genetics</i> , 2011, 43, 1098-1103.	21.4	251
23	Nonsense mutation in the LGR4 gene is associated with several human diseases and other traits. <i>Nature</i> , 2013, 497, 517-520.	27.8	236
24	Common variants at 19p13 are associated with susceptibility to ovarian cancer. <i>Nature Genetics</i> , 2010, 42, 880-884.	21.4	235
25	Variants with large effects on blood lipids and the role of cholesterol and triglycerides in coronary disease. <i>Nature Genetics</i> , 2016, 48, 634-639.	21.4	214
26	Discovery of common variants associated with low TSH levels and thyroid cancer risk. <i>Nature Genetics</i> , 2012, 44, 319-322.	21.4	208
27	Genome-wide association study identifies sequence variants on 6q21 associated with age at menarche. <i>Nature Genetics</i> , 2009, 41, 734-738.	21.4	199
28	Genome-wide association study identifies a sequence variant within the DAB2IP gene conferring susceptibility to abdominal aortic aneurysm. <i>Nature Genetics</i> , 2010, 42, 692-697.	21.4	181
29	A study based on whole-genome sequencing yields a rare variant at 8q24 associated with prostate cancer. <i>Nature Genetics</i> , 2012, 44, 1326-1329.	21.4	178
30	Replication of Lung Cancer Susceptibility Loci at Chromosomes 15q25, 5p15, and 6p21: A Pooled Analysis From the International Lung Cancer Consortium. <i>Journal of the National Cancer Institute</i> , 2010, 102, 959-971.	6.3	174
31	A sequence variant at 4p16.3 confers susceptibility to urinary bladder cancer. <i>Nature Genetics</i> , 2010, 42, 415-419.	21.4	169
32	Loss-of-function variants in ATM confer risk of gastric cancer. <i>Nature Genetics</i> , 2015, 47, 906-910.	21.4	155
33	Genome-wide association study identifies multiple susceptibility loci for multiple myeloma. <i>Nature Communications</i> , 2016, 7, 12050.	12.8	146
34	Genetic Correction of PSA Values Using Sequence Variants Associated with PSA Levels. <i>Science Translational Medicine</i> , 2010, 2, 62ra92.	12.4	140
35	Variant <i>ASGR1</i> Associated with a Reduced Risk of Coronary Artery Disease. <i>New England Journal of Medicine</i> , 2016, 374, 2131-2141.	27.0	137
36	Identification of low-frequency variants associated with gout and serum uric acid levels. <i>Nature Genetics</i> , 2011, 43, 1127-1130.	21.4	134

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37	European genome-wide association study identifies SLC14A1 as a new urinary bladder cancer susceptibility gene. <i>Human Molecular Genetics</i> , 2011, 20, 4268-4281.	2.9	134
38	Meta-analysis of Icelandic and UK data sets identifies missense variants in SMO, IL11, COL11A1 and 13 more new loci associated with osteoarthritis. <i>Nature Genetics</i> , 2018, 50, 1681-1687.	21.4	131
39	A genome-wide association study yields five novel thyroid cancer risk loci. <i>Nature Communications</i> , 2017, 8, 14517.	12.8	117
40	Common variants on 1p36 and 1q42 are associated with cutaneous basal cell carcinoma but not with melanoma or pigmentation traits. <i>Nature Genetics</i> , 2008, 40, 1313-1318.	21.4	111
41	Variants in ELL2 influencing immunoglobulin levels associate with multiple myeloma. <i>Nature Communications</i> , 2015, 6, 7213.	12.8	101
42	Cross-Cancer Genome-Wide Analysis of Lung, Ovary, Breast, Prostate, and Colorectal Cancer Reveals Novel Pleiotropic Associations. <i>Cancer Research</i> , 2016, 76, 5103-5114.	0.9	100
43	Whole genome characterization of sequence diversity of 15,220 Icelanders. <i>Scientific Data</i> , 2017, 4, 170115.	5.3	98
44	A Missense Variant in PTPN22 is a Risk Factor for Drug-induced Liver Injury. <i>Gastroenterology</i> , 2019, 156, 1707-1716.e2.	1.3	97
45	Insights into imprinting from parent-of-origin phased methylomes and transcriptomes. <i>Nature Genetics</i> , 2018, 50, 1542-1552.	21.4	94
46	Identification of multiple risk loci and regulatory mechanisms influencing susceptibility to multiple myeloma. <i>Nature Communications</i> , 2018, 9, 3707.	12.8	86
47	A loss-of-function variant in ALOX15 protects against nasal polyps and chronic rhinosinusitis. <i>Nature Genetics</i> , 2019, 51, 267-276.	21.4	83
48	Ancestry-Shift Refinement Mapping of the C6orf97-ESR1 Breast Cancer Susceptibility Locus. <i>PLoS Genetics</i> , 2010, 6, e1001029.	3.5	82
49	FLT3 stop mutation increases FLT3 ligand level and risk of autoimmune thyroid disease. <i>Nature</i> , 2020, 584, 619-623.	27.8	81
50	Whole-genome sequencing identifies rare genotypes in COMP and CHADL associated with high risk of hip osteoarthritis. <i>Nature Genetics</i> , 2017, 49, 801-805.	21.4	75
51	Variants associating with uterine leiomyoma highlight genetic background shared by various cancers and hormone-related traits. <i>Nature Communications</i> , 2018, 9, 3636.	12.8	74
52	Genome-wide association meta-analysis yields 20 loci associated with gallstone disease. <i>Nature Communications</i> , 2018, 9, 5101.	12.8	73
53	Sequence variants in ARHGAP15, COLQ and FAM155A associate with diverticular disease and diverticulitis. <i>Nature Communications</i> , 2017, 8, 15789.	12.8	67
54	Clinical characteristics of patients with colorectal cancer with double somatic mismatch repair mutations compared with Lynch syndrome. <i>Journal of Medical Genetics</i> , 2019, 56, 462-470.	3.2	61

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55	New basal cell carcinoma susceptibility loci. Nature Communications, 2015, 6, 6825.	12.8	59
56	Sequence variants in the PTCH1 gene associate with spine bone mineral density and osteoporotic fractures. Nature Communications, 2016, 7, 10129.	12.8	58
57	A common variant at 8q24.21 is associated with renal cell cancer. Nature Communications, 2013, 4, 2776.	12.8	56
58	Epigenetic and genetic components of height regulation. Nature Communications, 2016, 7, 13490.	12.8	52
59	BRCA2, but not BRCA1, mutations account for familial ovarian cancer in Iceland: a population-based study. European Journal of Cancer, 2004, 40, 2788-2793.	2.8	50
60	Germline sequence variants in TGM3 and RGS22 confer risk of basal cell carcinoma. Human Molecular Genetics, 2014, 23, 3045-3053.	2.9	48
61	Genome-wide associations for benign prostatic hyperplasia reveal a genetic correlation with serum levels of PSA. Nature Communications, 2018, 9, 4568.	12.8	44
62	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, 1203-1214.	2.9	38
63	Predicted loss and gain of function mutations in ACO1 are associated with erythropoiesis. Communications Biology, 2020, 3, 189.	4.4	30
64	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.	6.3	29
65	Evaluation of Association of HNF1B Variants with Diverse Cancers: Collaborative Analysis of Data from 19 Genome-Wide Association Studies. PLoS ONE, 2010, 5, e10858.	2.5	28
66	Identification of 22 susceptibility loci associated with testicular germ cell tumors. Nature Communications, 2021, 12, 4487.	12.8	27
67	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.	5.6	27
68	Genome-Wide Significant Association Between a Sequence Variant at 15q15.2 and Lung Cancer Risk. Cancer Research, 2011, 71, 1356-1361.	0.9	26
69	A Splice Region Variant in LDLR Lowers Non-high Density Lipoprotein Cholesterol and Protects against Coronary Artery Disease. PLoS Genetics, 2015, 11, e1005379.	3.5	24
70	Genome-wide association identifies seven loci for pelvic organ prolapse in Iceland and the UK Biobank. Communications Biology, 2020, 3, 129.	4.4	20
71	Fourteen sequence variants that associate with multiple sclerosis discovered by meta-analysis informed by genetic correlations. Npj Genomic Medicine, 2017, 2, 24.	3.8	16
72	The Icelandic Cancer Project – a population-wide approach to studying cancer. Nature Reviews Cancer, 2004, 4, 488-492.	28.4	15

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73	Loss-of-Function Variants in the Tumor-Suppressor Gene <i>PTPN14</i> Confer Increased Cancer Risk. <i>Cancer Research</i> , 2021, 81, 1954-1964.	0.9	15
74	Sequence variant at 4q25 near PITX2 associates with appendicitis. <i>Scientific Reports</i> , 2017, 7, 3119.	3.3	14
75	A Method for Detecting Long Non-Coding RNAs with Tiled RNA Expression Microarrays. <i>PLoS ONE</i> , 2014, 9, e99899.	2.5	12
76	Predicting the probability of death using proteomics. <i>Communications Biology</i> , 2021, 4, 758.	4.4	10
77	Functional dissection of inherited non-coding variation influencing multiple myeloma risk. <i>Nature Communications</i> , 2022, 13, 151.	12.8	10
78	Common and rare sequence variants influencing tumor biomarkers in blood. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2019, 29, cebp.1060.2018.	2.5	9
79	A PRPH splice-donor variant associates with reduced sural nerve amplitude and risk of peripheral neuropathy. <i>Nature Communications</i> , 2019, 10, 1777.	12.8	7
80	Genetic variants associated with platelet count are predictive of human disease and physiological markers. <i>Communications Biology</i> , 2021, 4, 1132.	4.4	7
81	Germline variants at SOHLH2 influence multiple myeloma risk. <i>Blood Cancer Journal</i> , 2021, 11, 76.	6.2	6
82	Profile of common prostate cancer risk variants in an unscreened Romanian population. <i>Journal of Cellular and Molecular Medicine</i> , 2018, 22, 1574-1582.	3.6	4
83	Comprehensive population-wide detection of Lynch syndrome in Iceland.. <i>Journal of Clinical Oncology</i> , 2016, 34, 1542-1542.	1.6	3
84	Genetic architecture of band neutrophil fraction in Iceland. <i>Communications Biology</i> , 2022, 5, .	4.4	1