

# Thorunn Rafnar

## List of Publications by Year in descending order

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Version: 2024-02-01

84  
papers

15,832  
citations

34493

54  
h-index

60403

85  
g-index

89  
all docs

89  
docs citations

89  
times ranked

24052  
citing authors

#	ARTICLE	IF	CITATIONS
1	Functional dissection of inherited non-coding variation influencing multiple myeloma risk. <i>Nature Communications</i> , 2022, 13, 151.	5.8	10
2	Genetic architecture of band neutrophil fraction in Iceland. <i>Communications Biology</i> , 2022, 5, .	2.0	1
3	Loss-of-Function Variants in the Tumor-Suppressor Gene <i>PTPN14</i> Confer Increased Cancer Risk. <i>Cancer Research</i> , 2021, 81, 1954-1964.	0.4	15
4	Germline variants at <i>SOHLH2</i> influence multiple myeloma risk. <i>Blood Cancer Journal</i> , 2021, 11, 76.	2.8	6
5	Predicting the probability of death using proteomics. <i>Communications Biology</i> , 2021, 4, 758.	2.0	10
6	Identification of 22 susceptibility loci associated with testicular germ cell tumors. <i>Nature Communications</i> , 2021, 12, 4487.	5.8	27
7	Genetic variants associated with platelet count are predictive of human disease and physiological markers. <i>Communications Biology</i> , 2021, 4, 1132.	2.0	7
8	The <i>CRTAC1</i> Protein in Plasma Is Associated With Osteoarthritis and Predicts Progression to Joint Replacement: A Large-scale Proteomics Scan in Iceland. <i>Arthritis and Rheumatology</i> , 2021, 73, 2025-2034.	2.9	27
9	Large-scale integration of the plasma proteome with genetics and disease. <i>Nature Genetics</i> , 2021, 53, 1712-1721.	9.4	340
10	<i>FLT3</i> stop mutation increases <i>FLT3</i> ligand level and risk of autoimmune thyroid disease. <i>Nature</i> , 2020, 584, 619-623.	13.7	81
11	Genome-wide association identifies seven loci for pelvic organ prolapse in Iceland and the UK Biobank. <i>Communications Biology</i> , 2020, 3, 129.	2.0	20
12	Predicted loss and gain of function mutations in <i>ACO1</i> are associated with erythropoiesis. <i>Communications Biology</i> , 2020, 3, 189.	2.0	30
13	A <i>PRPH</i> splice-donor variant associates with reduced sural nerve amplitude and risk of peripheral neuropathy. <i>Nature Communications</i> , 2019, 10, 1777.	5.8	7
14	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.	1.5	61
15	A Missense Variant in <i>PTPN22</i> is a Risk Factor for Drug-induced Liver Injury. <i>Gastroenterology</i> , 2019, 156, 1707-1716.e2.	0.6	97
16	Common and rare sequence variants influencing tumor biomarkers in blood. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2019, 29, cebp.1060.2018.	1.1	9
17	A loss-of-function variant in <i>ALOX15</i> protects against nasal polyps and chronic rhinosinusitis. <i>Nature Genetics</i> , 2019, 51, 267-276.	9.4	83
18	Profile of common prostate cancer risk variants in an unscreened Romanian population. <i>Journal of Cellular and Molecular Medicine</i> , 2018, 22, 1574-1582.	1.6	4

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19	Genome-wide associations for benign prostatic hyperplasia reveal a genetic correlation with serum levels of PSA. <i>Nature Communications</i> , 2018, 9, 4568.	5.8	44
20	Genome-wide association meta-analysis yields 20 loci associated with gallstone disease. <i>Nature Communications</i> , 2018, 9, 5101.	5.8	73
21	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.	9.4	131
22	Insights into imprinting from parent-of-origin phased methylomes and transcriptomes. <i>Nature Genetics</i> , 2018, 50, 1542-1552.	9.4	94
23	Identification of multiple risk loci and regulatory mechanisms influencing susceptibility to multiple myeloma. <i>Nature Communications</i> , 2018, 9, 3707.	5.8	86
24	Variants associating with uterine leiomyoma highlight genetic background shared by various cancers and hormone-related traits. <i>Nature Communications</i> , 2018, 9, 3636.	5.8	74
25	Association of BRCA2 K3326* With Small Cell Lung Cancer and Squamous Cell Cancer of the Skin. <i>Journal of the National Cancer Institute</i> , 2018, 110, 967-974.	3.0	29
26	A genome-wide association study yields five novel thyroid cancer risk loci. <i>Nature Communications</i> , 2017, 8, 14517.	5.8	117
27	Sequence variants in ARHGAP15, COLQ and FAM155A associate with diverticular disease and diverticulitis. <i>Nature Communications</i> , 2017, 8, 15789.	5.8	67
28	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.	9.4	472
29	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.	9.4	75
30	Whole genome characterization of sequence diversity of 15,220 Icelanders. <i>Scientific Data</i> , 2017, 4, 170115.	2.4	98
31	Fourteen sequence variants that associate with multiple sclerosis discovered by meta-analysis informed by genetic correlations. <i>Npj Genomic Medicine</i> , 2017, 2, 24.	1.7	16
32	Sequence variant at 4q25 near PITX2 associates with appendicitis. <i>Scientific Reports</i> , 2017, 7, 3119.	1.6	14
33	Variant <i>ASGR1</i> Associated with a Reduced Risk of Coronary Artery Disease. <i>New England Journal of Medicine</i> , 2016, 374, 2131-2141.	13.9	137
34	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.	9.4	214
35	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.	0.4	100
36	Epigenetic and genetic components of height regulation. <i>Nature Communications</i> , 2016, 7, 13490.	5.8	52

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37	Genome-wide association study identifies multiple susceptibility loci for multiple myeloma. <i>Nature Communications</i> , 2016, 7, 12050.	5.8	146
38	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. <i>Human Molecular Genetics</i> , 2016, 25, 1203-1214.	1.4	38
39	Sequence variants in the PTCH1 gene associate with spine bone mineral density and osteoporotic fractures. <i>Nature Communications</i> , 2016, 7, 10129.	5.8	58
40	Comprehensive population-wide detection of Lynch syndrome in Iceland.. <i>Journal of Clinical Oncology</i> , 2016, 34, 1542-1542.	0.8	3
41	A Splice Region Variant in LDLR Lowers Non-high Density Lipoprotein Cholesterol and Protects against Coronary Artery Disease. <i>PLoS Genetics</i> , 2015, 11, e1005379.	1.5	24
42	Loss-of-function variants in ATM confer risk of gastric cancer. <i>Nature Genetics</i> , 2015, 47, 906-910.	9.4	155
43	Large-scale whole-genome sequencing of the Icelandic population. <i>Nature Genetics</i> , 2015, 47, 435-444.	9.4	663
44	Variants in ELL2 influencing immunoglobulin levels associate with multiple myeloma. <i>Nature Communications</i> , 2015, 6, 7213.	5.8	101
45	New basal cell carcinoma susceptibility loci. <i>Nature Communications</i> , 2015, 6, 6825.	5.8	59
46	A Method for Detecting Long Non-Coding RNAs with Tiled RNA Expression Microarrays. <i>PLoS ONE</i> , 2014, 9, e99899.	1.1	12
47	Germline sequence variants in TGM3 and RGS22 confer risk of basal cell carcinoma. <i>Human Molecular Genetics</i> , 2014, 23, 3045-3053.	1.4	48
48	Rare variants of large effect in BRCA2 and CHEK2 affect risk of lung cancer. <i>Nature Genetics</i> , 2014, 46, 736-741.	9.4	360
49	Nonsense mutation in the LGR4 gene is associated with several human diseases and other traits. <i>Nature</i> , 2013, 497, 517-520.	13.7	236
50	A common variant at 8q24.21 is associated with renal cell cancer. <i>Nature Communications</i> , 2013, 4, 2776.	5.8	56
51	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.	9.4	178
52	Discovery of common variants associated with low TSH levels and thyroid cancer risk. <i>Nature Genetics</i> , 2012, 44, 319-322.	9.4	208
53	Mutations in BRIP1 confer high risk of ovarian cancer. <i>Nature Genetics</i> , 2011, 43, 1104-1107.	9.4	338
54	A germline variant in the TP53 polyadenylation signal confers cancer susceptibility. <i>Nature Genetics</i> , 2011, 43, 1098-1103.	9.4	251

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55	Identification of low-frequency variants associated with gout and serum uric acid levels. <i>Nature Genetics</i> , 2011, 43, 1127-1130.	9.4	134
56	A rare variant in MYH6 is associated with high risk of sick sinus syndrome. <i>Nature Genetics</i> , 2011, 43, 316-320.	9.4	275
57	Genome-Wide Significant Association Between a Sequence Variant at 15q15.2 and Lung Cancer Risk. <i>Cancer Research</i> , 2011, 71, 1356-1361.	0.4	26
58	European genome-wide association study identifies SLC14A1 as a new urinary bladder cancer susceptibility gene. <i>Human Molecular Genetics</i> , 2011, 20, 4268-4281.	1.4	134
59	A sequence variant at 4p16.3 confers susceptibility to urinary bladder cancer. <i>Nature Genetics</i> , 2010, 42, 415-419.	9.4	169
60	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.	9.4	181
61	Common variants at 19p13 are associated with susceptibility to ovarian cancer. <i>Nature Genetics</i> , 2010, 42, 880-884.	9.4	235
62	A genome-wide association study identifies susceptibility loci for ovarian cancer at 2q31 and 8q24. <i>Nature Genetics</i> , 2010, 42, 874-879.	9.4	321
63	A multi-stage genome-wide association study of bladder cancer identifies multiple susceptibility loci. <i>Nature Genetics</i> , 2010, 42, 978-984.	9.4	493
64	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.	3.0	174
65	Genetic Correction of PSA Values Using Sequence Variants Associated with PSA Levels. <i>Science Translational Medicine</i> , 2010, 2, 62ra92.	5.8	140
66	Ancestry-Shift Refinement Mapping of the C6orf97-ESR1 Breast Cancer Susceptibility Locus. <i>PLoS Genetics</i> , 2010, 6, e1001029.	1.5	82
67	Evaluation of Association of HNF1B Variants with Diverse Cancers: Collaborative Analysis of Data from 19 Genome-Wide Association Studies. <i>PLoS ONE</i> , 2010, 5, e10858.	1.1	28
68	Sequence variants at the TERT-CLPTM1L locus associate with many cancer types. <i>Nature Genetics</i> , 2009, 41, 221-227.	9.4	572
69	Common variants on 9q22.33 and 14q13.3 predispose to thyroid cancer in European populations. <i>Nature Genetics</i> , 2009, 41, 460-464.	9.4	353
70	Genome-wide association study identifies sequence variants on 6q21 associated with age at menarche. <i>Nature Genetics</i> , 2009, 41, 734-738.	9.4	199
71	New common variants affecting susceptibility to basal cell carcinoma. <i>Nature Genetics</i> , 2009, 41, 909-914.	9.4	303
72	Genome-wide association and replication studies identify four variants associated with prostate cancer susceptibility. <i>Nature Genetics</i> , 2009, 41, 1122-1126.	9.4	313

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73	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.	2.6	489
74	A variant associated with nicotine dependence, lung cancer and peripheral arterial disease. <i>Nature</i> , 2008, 452, 638-642.	13.7	1,399
75	Many sequence variants affecting diversity of adult human height. <i>Nature Genetics</i> , 2008, 40, 609-615.	9.4	615
76	ASIP and TYR pigmentation variants associate with cutaneous melanoma and basal cell carcinoma. <i>Nature Genetics</i> , 2008, 40, 886-891.	9.4	306
77	Detection of sharing by descent, long-range phasing and haplotype imputation. <i>Nature Genetics</i> , 2008, 40, 1068-1075.	9.4	409
78	Sequence variant on 8q24 confers susceptibility to urinary bladder cancer. <i>Nature Genetics</i> , 2008, 40, 1307-1312.	9.4	377
79	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.	9.4	111
80	Genome-wide association study identifies a second prostate cancer susceptibility variant at 8q24. <i>Nature Genetics</i> , 2007, 39, 631-637.	9.4	818
81	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.	9.4	670
82	Common variants on chromosomes 2q35 and 16q12 confer susceptibility to estrogen receptor-positive breast cancer. <i>Nature Genetics</i> , 2007, 39, 865-869.	9.4	774
83	The Icelandic Cancer Project – a population-wide approach to studying cancer. <i>Nature Reviews Cancer</i> , 2004, 4, 488-492.	12.8	15
84	BRCA2, but not BRCA1, mutations account for familial ovarian cancer in Iceland: a population-based study. <i>European Journal of Cancer</i> , 2004, 40, 2788-2793.	1.3	50