## Alisa M Goldstein

List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	Germline p16 mutations in familial melanoma. Nature Genetics, 1994, 8, 15-21.	21.4	1,170
2	Chordoma: incidence and survival patterns in the United States, 1973-1995. Cancer Causes and Control, 2001, 12, 1-11.	1.8	820
3	Germline mutations in the p16INK4a binding domain of CDK4 in familial melanoma. Nature Genetics, 1996, 12, 97-99.	21.4	756
4	Increased Risk of Pancreatic Cancer in Melanoma-Prone Kindreds with <i>p16</i> <sup>INK4</sup> Mutations. New England Journal of Medicine, 1995, 333, 970-975.	27.0	608
5	Re-evaluation of the linkage relationship between chromosome 11p loci and the gene for bipolar affective disorder in the Old Order Amish. Nature, 1989, 342, 238-243.	27.8	448
6	Geographical Variation in the Penetrance of CDKN2A Mutations for Melanoma. Journal of the National Cancer Institute, 2002, 94, 894-903.	6.3	435
7	Genome-wide association study identifies three loci associated with melanoma risk. Nature Genetics, 2009, 41, 920-925.	21.4	422
8	High-risk Melanoma Susceptibility Genes and Pancreatic Cancer, Neural System Tumors, and Uveal Melanoma across GenoMEL. Cancer Research, 2006, 66, 9818-9828.	0.9	373
9	Features associated with germline CDKN2A mutations: a GenoMEL study of melanoma-prone families from three continents. Journal of Medical Genetics, 2006, 44, 99-106.	3.2	350
10	T (brachyury) gene duplication confers major susceptibility to familial chordoma. Nature Genetics, 2009, 41, 1176-1178.	21.4	284
11	Rare missense variants in POT1 predispose to familial cutaneous malignant melanoma. Nature Genetics, 2014, 46, 482-486.	21.4	283
12	Mutations associated with familial melanoma impair p16INK4 function. Nature Genetics, 1995, 10, 114-116.	21.4	273
13	CDKN2AMutations in Multiple Primary Melanomas. New England Journal of Medicine, 1998, 338, 879-887.	27.0	255
14	Genome-wide association study identifies three new melanoma susceptibility loci. Nature Genetics, 2011, 43, 1108-1113.	21.4	230
15	Genome-wide meta-analysis identifies five new susceptibility loci for cutaneous malignant melanoma. Nature Genetics, 2015, 47, 987-995.	21.4	218
16	Common sequence variants on 20q11.22 confer melanoma susceptibility. Nature Genetics, 2008, 40, 838-840.	21.4	209
17	Genotype-Phenotype Relationships in U.S. Melanoma-Prone Families With CDKN2A and CDK4 Mutations. Journal of the National Cancer Institute, 2000, 92, 1006-1010.	6.3	172
18	Analysis of Heritability and Shared Heritability Based on Genome-Wide Association Studies for Thirteen Cancer Types. Journal of the National Cancer Institute, 2015, 107, djv279.	6.3	152

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19	Joint analysis of three genome-wide association studies of esophageal squamous cell carcinoma in Chinese populations. Nature Genetics, 2014, 46, 1001-1006.	21.4	148
20	Genome-wide association study identifies a new melanoma susceptibility locus at 1q21.3. Nature Genetics, 2011, 43, 1114-1118.	21.4	140
21	Genome-wide association meta-analyses combining multiple risk phenotypes provide insights into the genetic architecture of cutaneous melanoma susceptibility. Nature Genetics, 2020, 52, 494-504.	21.4	138
22	Molecular Characterization of the Human Stomach Microbiota in Gastric Cancer Patients. Frontiers in Cellular and Infection Microbiology, 2017, 7, 302.	3.9	136
23	Familial melanoma, pancreatic cancer and germline CDKN2A mutations. Human Mutation, 2004, 23, 630-630.	2.5	130
24	Genetic Epidemiology of Cutaneous Melanoma. Archives of Dermatology, 2001, 137, 1493-6.	1.4	118
25	Risk factors for esophageal and gastric cancers in Shanxi Province, China: A case–control study. Cancer Epidemiology, 2011, 35, e91-e99.	1.9	112
26	Melanoma prone families with <i>CDK4</i> germline mutation: phenotypic profile and associations with <i>MC1R</i> variants. Journal of Medical Genetics, 2013, 50, 264-270.	3.2	112
27	A variant in FTO shows association with melanoma risk not due to BMI. Nature Genetics, 2013, 45, 428-432.	21.4	111
28	The Effect on Melanoma Risk of Genes Previously Associated With Telomere Length. Journal of the National Cancer Institute, 2014, 106, .	6.3	109
29	Characterization of Large Structural Genetic Mosaicism in Human Autosomes. American Journal of Human Genetics, 2015, 96, 487-497.	6.2	101
30	Genome-wide association study of gastric adenocarcinoma in Asia: a comparison of associations between cardia and non-cardia tumours. Gut, 2016, 65, 1611-1618.	12.1	99
31	Germline Mutations in PALB2, BRCA1, and RAD51C, Which Regulate DNA Recombination Repair, in Patients With Gastric Cancer. Gastroenterology, 2017, 152, 983-986.e6.	1.3	98
32	Second cancers after medulloblastoma: population-based results from the United States and Sweden. Cancer Causes and Control, 1997, 8, 865-871.	1.8	96
33	A genomeâ€wide search for loci contributing to smoking and alcoholism. Genetic Epidemiology, 1999, 17, S55-60.	1.3	91
34	Lung Cancer Prognosis Before and After Recurrence in a Population-Based Setting. Journal of the National Cancer Institute, 2015, 107, djv059.	6.3	86
35	Female chromosome X mosaicism is age-related and preferentially affects the inactivated X chromosome. Nature Communications, 2016, 7, 11843.	12.8	86
36	Allelic imbalance, including deletion of PTEN/MMAC1, at the Cowden disease locus on 10q22-23, in		85

hamartomas from patients with cowden syndrome and germlinePTEN mutation., 1998, 21, 61-69.

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37	Mutational signatures in esophageal squamous cell carcinoma from eight countries with varying incidence. Nature Genetics, 2021, 53, 1553-1563.	21.4	71
38	Genomic Landscape of Somatic Alterations in Esophageal Squamous Cell Carcinoma and Gastric Cancer. Cancer Research, 2016, 76, 1714-1723.	0.9	68
39	Identification of new susceptibility loci for gastric non-cardia adenocarcinoma: pooled results from two Chinese genome-wide association studies. Gut, 2017, 66, 581-587.	12.1	68
40	Genetic polymorphisms in the 9p21 region associated with risk of multiple cancers. Carcinogenesis, 2014, 35, 2698-2705.	2.8	67
41	Cell-type–specific eQTL of primary melanocytes facilitates identification of melanoma susceptibility genes. Genome Research, 2018, 28, 1621-1635.	5.5	67
42	The Genetics of Melanoma: Recent Advances. Annual Review of Genomics and Human Genetics, 2013, 14, 257-279.	6.2	66
43	Clinical findings in two Africanâ€American families with the nevoid basal cell carcinoma syndrome (NBCC). American Journal of Medical Genetics Part A, 1994, 50, 272-281.	2.4	64
44	Sporadic multiple primary melanoma cases:CDKN2Agermline mutations with a founder effect. Genes Chromosomes and Cancer, 2001, 32, 195-202.	2.8	63
45	Haplotype analysis of two recurrentCDKN2A mutations in 10 melanoma families: Evidence for common founders and independent mutations. , 1998, 11, 424-431.		61
46	High prevalence of the G101W germline mutation in theCDKN2A(P16ink4a) gene in 62 Italian malignant melanoma families. American Journal of Medical Genetics Part A, 2002, 107, 214-221.	2.4	60
47	Nested PCR Biases in Interpreting Microbial Community Structure in 16S rRNA Gene Sequence Datasets. PLoS ONE, 2015, 10, e0132253.	2.5	60
48	Mutation screening of theCDKN2A promoter in melanoma families. , 2000, 28, 45-57.		59
49	Epstein-Barr virus seroreactivity among unaffected individuals within high-risk nasopharyngeal carcinoma families in Taiwan. International Journal of Cancer, 2004, 111, 117-123.	5.1	56
50	The Value of Small Observations in the Era of Big Science. Cancer Epidemiology Biomarkers and Prevention, 2005, 14, 2472-2473.	2.5	55
51	Whole genome sequencing of skull-base chordoma reveals genomic alterations associated with recurrence and chordoma-specific survival. Nature Communications, 2021, 12, 757.	12.8	55
52	Characterization of T gene sequence variants and germline duplications in familial and sporadic chordoma. Human Genetics, 2014, 133, 1289-1297.	3.8	54
53	Assessing the Incremental Contribution of Common Genomic Variants to Melanoma Risk Prediction in Two Population-Based Studies. Journal of Investigative Dermatology, 2018, 138, 2617-2624.	0.7	52
54	Clinical features distinguish childhood chordoma associated with tuberous sclerosis complex (TSC) from chordoma in the general paediatric population. Journal of Medical Genetics, 2011, 48, 444-449.	3.2	51

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55	Germline mutations in <i>Protection of Telomeres 1</i> in two families with Hodgkin lymphoma. British Journal of Haematology, 2018, 181, 372-377.	2.5	48
56	Deletion analysis of the adenomatous polyposis coli andPTCH gene loci in patients with sporadic and nevoid basal cell carcinoma syndrome-associated medulloblastoma. Cancer, 1999, 85, 2662-2667.	4.1	46
57	Cutaneous phenotype andMC1R variants as modifying factors for the development of melanoma inCDKN2A G101W mutation carriers from 4 countries. International Journal of Cancer, 2007, 121, 825-831.	5.1	45
58	PLCE1 mRNA and Protein Expression and Survival of Patients with Esophageal Squamous Cell Carcinoma and Gastric Adenocarcinoma. Cancer Epidemiology Biomarkers and Prevention, 2014, 23, 1579-1588.	2.5	42
59	Impact of E27X, a novel CDKN2A germ line mutation, on p16 and p14ARF expression in Italian melanoma families displaying pancreatic cancer and neuroblastoma. Human Molecular Genetics, 2006, 15, 2682-2689.	2.9	41
60	Whole exome sequencing in families at high risk for Hodgkin lymphoma: identification of a predisposing mutation in the KDR gene. Haematologica, 2016, 101, 853-860.	3.5	40
61	Rare germline variants in known melanoma susceptibility genes in familial melanoma. Human Molecular Genetics, 2017, 26, 4886-4895.	2.9	37
62	Distribution of Epstein-Barr viral load in serum of individuals from nasopharyngeal carcinoma high-risk families in Taiwan. International Journal of Cancer, 2006, 118, 780-784.	5.1	36
63	Familial eosinophilia: Clinical and laboratory results on a U.S. Kindred. American Journal of Medical Genetics Part A, 1998, 76, 229-237.	2.4	35
64	Whole-Exome Sequencing of Nasopharyngeal Carcinoma Families Reveals Novel Variants Potentially Involved in Nasopharyngeal Carcinoma. Scientific Reports, 2019, 9, 9916.	3.3	32
65	Nevoid basal cell carcinoma syndrome with medulloblastoma in an African-American boy: A rare case illustrating gene-environment interaction. American Journal of Medical Genetics Part A, 1997, 69, 309-314.	2.4	30
66	Integrative molecular characterisation of gallbladder cancer reveals micro-environment-associated subtypes. Journal of Hepatology, 2021, 74, 1132-1144.	3.7	30
67	Gastric microbiota features associated with cancer risk factors and clinical outcomes: A pilot study in gastric cardia cancer patients from Shanxi, China. International Journal of Cancer, 2017, 141, 45-51.	5.1	29
68	Recent Tanning Bed Use. Archives of Dermatology, 2006, 142, 485-8.	1.4	28
69	Detecting gene-environment interactions using a case-control design. Genetic Epidemiology, 1997, 14, 1085-1089.	1.3	25
70	Cancer patterns in nasopharyngeal carcinoma multiplex families in Taiwan. International Journal of Cancer, 2009, 124, 1622-1625.	5.1	25
71	Multiple rare variants in high-risk pancreatic cancer-related genes may increase risk for pancreatic cancer in a subset of patients with and without germline CDKN2A mutations. Human Genetics, 2016, 135, 1241-1249.	3.8	24
72	An interstitial deletion within 9p21.3 and extending beyond <i>CDKN2A</i> predisposes to melanoma, neural system tumours and possible haematological malignancies. Journal of Medical Genetics, 2016, 53, 721-727.	3.2	23

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73	Body mass index and height and risk of cutaneous melanoma: Mendelian randomization analyses. International Journal of Epidemiology, 2020, 49, 1236-1245.	1.9	21
74	Histologic features of melanoma associated with CDKN2A genotype. Journal of the American Academy of Dermatology, 2015, 72, 496-507.e7.	1.2	19
75	Risks of Melanoma and Other Cancers in Melanoma-Prone Families over 4 Decades. Journal of Investigative Dermatology, 2018, 138, 1620-1626.	0.7	19
76	Insights into Genetic Susceptibility to Melanoma by Gene Panel Testing: Potential Pathogenic Variants in ACD, ATM, BAP1, and POT1. Cancers, 2020, 12, 1007.	3.7	19
77	Sebaceous Carcinoma Epidemiology and Genetics: Emerging Concepts and Clinical Implications for Screening, Prevention, and Treatment. Clinical Cancer Research, 2021, 27, 389-393.	7.0	19
78	Germline Variation at CDKN2A and Associations with Nevus Phenotypes amongÂMembers of Melanoma Families. Journal of Investigative Dermatology, 2017, 137, 2606-2612.	0.7	18
79	Estimating CDKN2A mutation carrier probability among global familial melanoma cases using GenoMELPREDICT. Journal of the American Academy of Dermatology, 2019, 81, 386-394.	1.2	17
80	Lack of phospholipase A2 mutations in neuroblastoma, melanoma and colon-cancer cell lines. , 1997, 72, 337-339.		16
81	Wholeâ€exome sequencing of nevoid basal cell carcinoma syndrome families and review of Human Gene Mutation Database <i>PTCH1</i> mutation data. Molecular Genetics & Genomic Medicine, 2018, 6, 1168-1180.	1.2	16
82	Pediatric melanoma in melanomaâ€prone families. Cancer, 2018, 124, 3715-3723.	4.1	16
83	Constitutional promoter methylation and risk of familial melanoma. Epigenetics, 2014, 9, 685-692.	2.7	15
84	Whole exome sequencing in families with CLL detects a variant in Integrin $\hat{I}^2$ 2 associated with disease susceptibility. Blood, 2016, 128, 2261-2263.	1.4	15
85	Prevalence of pathogenic/likely pathogenic variants in the 24 cancer genes of the ACMG Secondary Findings v2.0 list in a large cancer cohort and ethnicity-matched controls. Genome Medicine, 2018, 10, 99.	8.2	15
86	A systematic review of the prevalence of germline pathogenic variants in patients with pancreatic cancer. Journal of Gastroenterology, 2021, 56, 713-721.	5.1	15
87	Variants Associated with Susceptibility to Pancreatic Cancer and Melanoma Do Not Reciprocally Affect Risk. Cancer Epidemiology Biomarkers and Prevention, 2014, 23, 1121-1124.	2.5	14
88	Sebaceous Carcinoma Incidence and Survival Among Solid Organ Transplant Recipients in the United States, 1987-2017. JAMA Dermatology, 2020, 156, 1307.	4.1	14
89	Ambient Ultraviolet Radiation and Sebaceous Carcinoma Incidence in the United States, 2000–2016. JNCI Cancer Spectrum, 2020, 4, pkaa020.	2.9	14
90	Rare Germline Copy Number Variations and Disease Susceptibility in Familial Melanoma. Journal of Investigative Dermatology, 2016, 136, 2436-2443.	0.7	13

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91	Phenotypic and Histopathological Tumor Characteristics According to CDKN2A Mutation Status among Affected Members ofAMelanoma Families. Journal of Investigative Dermatology, 2016, 136, 1066-1069.	0.7	13
92	Association of high-evidence gastric cancer susceptibility loci and somatic gene expression levels with survival. Carcinogenesis, 2017, 38, 1119-1128.	2.8	13
93	Cell-type-specific meQTLs extend melanoma GWAS annotation beyond eQTLs and inform melanocyte gene-regulatory mechanisms. American Journal of Human Genetics, 2021, 108, 1631-1646.	6.2	12
94	GWAS follow-up study of esophageal squamous cell carcinoma identifies potential genetic loci associated with family history of upper gastrointestinal cancer. Scientific Reports, 2017, 7, 4642.	3.3	11
95	Phenocopies in melanoma-prone families with germ-line CDKN2A mutations. Genetics in Medicine, 2018, 20, 1087-1090.	2.4	11
96	Use of Big Data to Estimate Prevalence of Defective DNA Repair Variants in the US Population. JAMA Dermatology, 2019, 155, 72.	4.1	11
97	Variation in Cutaneous Patterns of Melanomagenesis According to Germline CDKN2A/CDK4 Status in Melanoma-Prone Families. Journal of Investigative Dermatology, 2020, 140, 174-181.e3.	0.7	11
98	Frequency of Pathogenic Germline Variants in Cancer-Susceptibility Genes in the Childhood Cancer Survivor Study. JNCI Cancer Spectrum, 2021, 5, pkab007.	2.9	11
99	A Systematic Literature Review of Whole Exome and Genome Sequencing Population Studies of Genetic Susceptibility to Cancer. Cancer Epidemiology Biomarkers and Prevention, 2020, 29, 1519-1534.	2.5	10
100	First international workshop of the ATM and cancer risk group (4-5 December 2019). Familial Cancer, 2022, 21, 211-227.	1.9	10
101	Oesophageal squamous cell carcinoma in high-risk Chinese populations: Possible role for vascular epithelial growth factor A. European Journal of Cancer, 2014, 50, 2855-2865.	2.8	9
102	Unconditional analyses can increase efficiencyin assessing gene–environment interaction of the case-combined-control design. International Journal of Epidemiology, 2006, 35, 1067-1073.	1.9	8
103	Common genetic variants related to vitamin D status are not associated with esophageal squamous cell carcinoma risk in China. Cancer Epidemiology, 2015, 39, 157-159.	1.9	8
104	Using whole-exome sequencing and protein interaction networks to prioritize candidate genes for germline cutaneous melanoma susceptibility. Scientific Reports, 2020, 10, 17198.	3.3	8
105	ABO genotypes and the risk of esophageal and gastric cancers. BMC Cancer, 2021, 21, 589.	2.6	8
106	Clinical findings in families with chordoma with and without T gene duplications and in patients with sporadic chordoma reported to the Surveillance, Epidemiology, and End Results program. Journal of Neurosurgery, 2021, 134, 1399-1408.	1.6	8
107	Sib-pair linkage analyses of nuclear family data: Quantitative versus dichotomous disease classification. Genetic Epidemiology, 1997, 14, 827-832.	1.3	7
108	Low Epstein–Barr Virus Prevalence in Cardia Gastric Cancer Among a High-Incidence Chinese Population. Digestive Diseases and Sciences, 2021, 66, 1220-1226.	2.3	7

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109	A UVB-responsive common variant at chromosome band 7p21.1 confers tanning response and melanoma risk via regulation of the aryl hydrocarbon receptor, AHR. American Journal of Human Genetics, 2021, 108, 1611-1630.	6.2	7
110	A Genome-First Approach to Estimate Prevalence of Germline Pathogenic Variants and Risk of Pancreatic Cancer in Select Cancer Susceptibility Genes. Cancers, 2022, 14, 3257.	3.7	6
111	Constitutive Mitochondrial DNA Copy Number in Peripheral Blood of Melanoma Families with and without CDKN2A Mutations. Journal of Carcinogenesis & Mutagenesis, 2012, S4`, .	0.3	5
112	Evaluation of Rare and Common Variants from Suspected Familial or Sporadic Nasopharyngeal Carcinoma (NPC) Susceptibility Genes in Sporadic NPC. Cancer Epidemiology Biomarkers and Prevention, 2019, 28, 1682-1686.	2.5	5
113	Histologic features of melanoma associated with germline mutations of CDKN2A, CDK4, and POT1 in melanoma-prone families from the United States, Italy, and Spain. Journal of the American Academy of Dermatology, 2020, 83, 860-869.	1.2	5
114	Rare Germline Variants in Chordoma-Related Genes and Chordoma Susceptibility. Cancers, 2021, 13, 2704.	3.7	5
115	Integrated Analysis of Coexpression and Exome Sequencing to Prioritize Susceptibility Genes for Familial Cutaneous Melanoma. Journal of Investigative Dermatology, 2022, 142, 2464-2475.e5.	0.7	4
116	Variation in second cancer risk by melanoma subtype among survivors. Journal of the American Academy of Dermatology, 2023, 88, 433-434.	1.2	4
117	Clustering of high density lipoprotein cholesterol levels in premenopausal and postmenopausal female twins. Genetic Epidemiology, 1993, 10, 563-567.	1.3	3
118	Gene×Environment Interaction from Case ontrol and Case ase Approaches. Genetic Epidemiology, 2001, 21, S825-30.	1.3	3
119	Use of Weighted pâ€Values in Regional Inference Procedures. Genetic Epidemiology, 2001, 21, S484-9.	1.3	3
120	The Impact of Longitudinal Surveillance on Tumor Thickness for Melanoma-Prone Families with and without Pathogenic Germline Variants of <i>CDKN2A</i> and <i>CDK4</i> . Cancer Epidemiology Biomarkers and Prevention, 2021, 30, 676-681.	2.5	3
121	Elevated antibodies against Epstein–Barr virus among individuals predicted to carry nasopharyngeal carcinoma susceptibility variants. Journal of General Virology, 2018, 99, 1268-1273.	2.9	3
122	Rare germline variants in <i>PALB2</i> and <i>BRCA2</i> in familial and sporadic chordoma. Human Mutation, 2022, 43, 1396-1407.	2.5	3
123	A PROBLEM IN IDENTIFYING RISK FACTORS FOR DISEASE USING SURROGATE EXPOSURE VARIABLES THAT ARE UNDER GENETIC CONTROL. American Journal of Epidemiology, 1990, 132, 1171-1175.	3.4	2
124	Cancer patterns in nasopharyngeal carcinoma multiplex families over 15 years. Cancer, 2021, 127, 4171-4176.	4.1	2
125	Novel MAPK/AKT-impairing germline NRAS variant identified in a melanoma-prone family. Familial Cancer, 2022, 21, 347-355.	1.9	1
126	Familial eosinophilia: Clinical and laboratory results on a U.S. Kindred. American Journal of Medical Genetics Part A, 1998, 76, 229-237.	2.4	1

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127	Deletion analysis of the adenomatous polyposis coli and PTCH gene loci in patients with sporadic and nevoid basal cell carcinoma syndromeâ€associated medulloblastoma. Cancer, 1999, 85, 2662-2667.	4.1	1
128	Mutation screening of the CDKN2A promoter in melanoma families. Genes Chromosomes and Cancer, 2000, 28, 45.	2.8	1
129	Modified eQTL and Somatic DNA Segment Alterations in Esophageal Squamous Cell Carcinoma for Genes Related to Immunity, DNA Repair, and Inflammation. Cancers, 2022, 14, 1629.	3.7	1
130	Sibâ€pair linkage analyses of alcoholism: Dichotomous and quantitative measures. Genetic Epidemiology, 1999, 17, S205-10.	1.3	0
131	Novel loss-of-function variant in DENND5A impedes melanosomal cargo transport and predisposes to familial cutaneous melanoma. Genetics in Medicine, 2022, 24, 157-169.	2.4	0