Katherine A Rauen

List of Publications by Year
in descending order

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67	5,649	101543	82547
papers	citations	36 h-index	72 g-index

78	78	78	6615
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	The RASopathies. Annual Review of Genomics and Human Genetics, 2013, 14, 355-369.	6.2	673
2	The RASopathies: developmental syndromes of Ras/MAPK pathway dysregulation. Current Opinion in Genetics and Development, 2009, 19, 230-236.	3.3	640
3	Germline Mutations in Genes Within the MAPK Pathway Cause Cardio-facio-cutaneous Syndrome. Science, 2006, 311, 1287-1290.	12.6	505
4	Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. Genetics in Medicine, 2021, 23, 1506-1513.	2.4	290
5	Germline loss-of-function mutations in LZTR1 predispose to an inherited disorder of multiple schwannomas. Nature Genetics, 2014, 46, 182-187.	21.4	242
6	HRAS mutations in Costello syndrome: Detection of constitutional activating mutations in codon 12 and 13 and loss of wild-type allele in malignancy. American Journal of Medical Genetics, Part A, 2006, 140A, 8-16.	1.2	157
7	Cardio-Facio-Cutaneous Syndrome: Clinical Features, Diagnosis, and Management Guidelines. Pediatrics, 2014, 134, e1149-e1162.	2.1	148
8	Project Baby Bear: Rapid precision care incorporating rWGS in 5 California childrenâ€™s hospitals demonstrates improved clinical outcomes and reduced costs of care. American Journal of Human Genetics, 2021, 108, 1231-1238.	6.2	140
9	Autism traits in the RASopathies. Journal of Medical Genetics, 2014, 51, 10-20.	3.2	134
10	Expression of the coxsackie adenovirus receptor in normal prostate and in primary and metastatic prostate carcinoma: potential relevance to gene therapy. Cancer Research, 2002, 62, 3812-8.	0.9	128
11	ClinGenâ€™s RASopathy Expert Panel consensus methods for variant interpretation. Genetics in Medicine, 2018, 20, 1334-1345.	2.4	126
12	Diversity, parental germline origin, and phenotypic spectrum of de novoHRASmissense changes in Costello syndrome. Human Mutation, 2007, 28, 265-272.	2.5	123
13	Neurological complications of cardioâ€facioâ€cutaneous syndrome. Developmental Medicine and Child Neurology, 2007, 49, 894-899.	2.1	106
14	HRAS and the Costello syndrome. Clinical Genetics, 2007, 71, 101-108.	2.0	104
15	Clinical, pathological, and molecular analyses of cardiovascular abnormalities in Costello syndrome: A Ras/MAPK pathway syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 486-507.	1.2	99
16	Kinase-activating and kinase-impaired cardio-facio-cutaneous syndrome alleles have activity during zebrafish development and are sensitive to small molecule inhibitors. Human Molecular Genetics, 2009, 18, 2543-2554.	2.9	89
17	Pathogenetics of the RASopathies. Human Molecular Genetics, 2016, 25, R123-R132.	2.9	87
18	Expansion of the RASopathies. Current Genetic Medicine Reports, 2016, 4, 57-64.	1.9	85

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19	Mutation Analysis of BRAF, MEK1 and MEK2 in 15 Ovarian Cancer Cell Lines: Implications for Therapy. PLoS ONE, 2007, 2, e1279.	2.5	77
20	Dermatological findings in 61 mutation-positive individuals with cardiofaciocutaneous syndrome. British Journal of Dermatology, 2011, 164, no-no.	1.5	74
21	Dermatological phenotype in Costello syndrome: consequences of Ras dysregulation in development. British Journal of Dermatology, 2012, 166, 601-607.	1.5	70
22	Dysregulation of astrocyte extracellular signaling in Costello syndrome. Science Translational Medicine, 2015, 7, 286ra66.	12.4	70
23	Costello syndrome: Clinical phenotype, genotype, and management guidelines. American Journal of Medical Genetics, Part A, 2019, 179, 1725-1744.	1.2	70
24	RASopathies: unraveling mechanisms with animal models. DMM Disease Models and Mechanisms, 2015, 8, 769-782.	2.4	66
25	Assessing the geneâ€“disease association of 19 genes with the RASopathies using the ClinGen gene curation framework. Human Mutation, 2018, 39, 1485-1493.	2.5	66
26	Expanding the clinical phenotype of individuals with a 3-bp in-frame deletion of the NF1 gene (c.2970_2972del): an update of genotypeâ€“phenotype correlation. Genetics in Medicine, 2019, 21, 867-876.	2.4	62
27	Additional patient with del(12)(q21.2q22): Further evidence for a candidate region for cardio-facio-cutaneous syndrome?. American Journal of Medical Genetics Part A, 2002, 110, 51-56.	2.4	53
28	Interstitial deletions of chromosome 6q: genotypeâ€“phenotype correlation utilizing array CGH. Clinical Genetics, 2007, 71, 260-266.	2.0	45
29	Continual and partial MEK inhibition ameliorates cardio-facio-cutaneous phenotypes in zebrafish. DMM Disease Models and Mechanisms, 2012, 5, 546-52.	2.4	44
30	The duality of human oncoproteins: drivers of cancer and congenital disorders. Nature Reviews Cancer, 2020, 20, 383-397.	28.4	44
31	Skeletal muscle pathology in Costello and cardioâ€facioâ€cutaneous syndromes: Developmental consequences of germline Ras/MAPK activation on myogenesis. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2011, 157, 104-114.	1.6	42
32	Costello and cardioâ€facioâ€cutaneous syndromes: Moving toward clinical trials in RASopathies. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2011, 157, 136-146.	1.6	41
33	Effects of germline mutations in the Ras/MAPK signaling pathway on adaptive behavior: Cardiofaciocutaneous syndrome and Noonan syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 591-600.	1.2	40
34	Advancing <scp>RAS/RASopathy</scp> therapies: An NCIâ€sponsored intramural and extramural collaboration for the study of <scp>RASopathies</scp>. American Journal of Medical Genetics, Part A, 2020, 182, 866-876.	1.2	40
35	Exclusion of PTPN11 mutations in Costello syndrome: further evidence for distinct genetic etiologies for Noonan, cardio-facio-cutaneous and Costello syndromes. Clinical Genetics, 2003, 63, 423-426.	2.0	38
36	Peripheral muscle weakness in RASopathies. Muscle and Nerve, 2012, 46, 394-399.	2.2	38

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37	Reverse Pathway Genetic Approach Identifies Epistasis in Autism Spectrum Disorders. PLoS Genetics, 2017, 13, e1006516.	3.5	38
38	Cardio-facio-cutaneous syndrome phenotype in an individual with an interstitial deletion of 12q: Identification of a candidate region for CFC syndrome. American Journal of Medical Genetics Part A, 2000, 93, 219-222.	2.4	37
39	Molecular and functional analysis of a novel MEK2 mutation in cardioâ€facioâ€cutaneous syndrome: Transmission through four generations. American Journal of Medical Genetics, Part A, 2010, 152A, 807-814.	1.2	37
40	Abnormal Ras signaling in Costello syndrome (CS) negatively regulates enamel formation. Human Molecular Genetics, 2014, 23, 682-692.	2.9	36
41	Distinguishing Costello versus cardio-facio-cutaneous syndrome:BRAF mutations in patients with a Costello phenotype. American Journal of Medical Genetics, Part A, 2006, 140A, 1681-1683.	1.2	34
42	Autonomous and Non-autonomous Defects Underlie Hypertrophic Cardiomyopathy in BRAF-Mutant hiPSC-Derived Cardiomyocytes. Stem Cell Reports, 2016, 7, 355-369.	4.8	33
43	Deletion of MAP2K2/MEK2: a novel mechanism for a RASopathy?. Clinical Genetics, 2014, 85, 138-146.	2.0	30
44	Biochemical Characterization of Novel Germline BRAF and MEK Mutations in Cardioâ€Facioâ€Cutaneous Syndrome. Methods in Enzymology, 2008, 438, 277-289.	1.0	29
45	Human iPS Cell-Derived Neurons Uncover the Impact of Increased Ras Signaling in Costello Syndrome. Journal of Neuroscience, 2016, 36, 142-152.	3.6	29
46	Prader-Willi syndrome resulting from an unbalanced translocation: characterization by array comparative genomic hybridization. Clinical Genetics, 2004, 65, 477-482.	2.0	26
47	Defining RASopathy. DMM Disease Models and Mechanisms, 2022, 15, .	2.4	26
48	Germline mutation in BRAF codon 600 is compatible with human development: de novo p.V600G mutation identified in a patient with CFC syndrome. Clinical Genetics, 2011, 79, 468-474.	2.0	21
49	Craniofacial and dental development in Costello syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 1425-1430.	1.2	21
50	Craniofacial and dental development in cardioâ€facioâ€cutaneous syndrome: the importance of Ras signaling homeostasis. Clinical Genetics, 2013, 83, 539-544.	2.0	20
51	Mek1 Y130C mice recapitulate aspects of the human Cardio-Facio-Cutaneous syndrome. DMM Disease Models and Mechanisms, 2018, 11, .	2.4	19
52	Fertility in a female with mosaic trisomy 8. Fertility and Sterility, 2003, 79, 206-208.	1.0	16
53	Tandem duplication mosaicism: characterization of a mosaic dup(5q) and review. Clinical Genetics, 2001, 60, 366-370.	2.0	11
54	Mutational and Functional Analysis in Human Ras/MAP Kinase Genetic Syndromes. Methods in Molecular Biology, 2010, 661, 433-447.	0.9	10

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55	The seventh international <scp>RASopathies</scp> symposium: Pathways to a cureâ€"expanding knowledge, enhancing research, and therapeutic discovery. American Journal of Medical Genetics, Part A, 2022, 188, 1915-1927.	1.2	10
56	RASopathies are associated with a distinct personality profile. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 434-446.	1.7	9
57	MEK-inhibitor-mediated rescue of skeletal myopathy caused by activating Hras mutation in a Costello syndrome mouse model. DMM Disease Models and Mechanisms, 2022, 15, .	2.4	9
58	Comparison of hair manifestations in cardioâ€facioâ€cutaneous and Costello syndromes highlights the influence of the RAS pathway on hair growth. Journal of the European Academy of Dermatology and Venereology, 2020, 34, 601-607.	2.4	8
59	Congenital diaphragmatic hernia in Smithâ€"Magenis syndrome: A possible locus at chromosome 17p11.2. American Journal of Medical Genetics, Part A, 2011, 155, 2816-2820.	1.2	7
60	Ras/<scp>MAPK</scp> dysregulation in development causes a skeletal myopathy in an activating <scp>Braf^{L597V}</scp> mouse model for cardioâ€facioâ€cutaneous syndrome. Developmental Dynamics, 2021, 250, 1074-1095.	1.8	6
61	RAS pathway influences the number of melanocytic nevi in cardiofaciocutaneous and Costello syndromes. Journal of the American Academy of Dermatology, 2020, 82, 1091-1093.	1.2	5
62	A novel DPH5-related diphthamide-deficiency syndrome causing embryonic lethality or profound neurodevelopmental disorder. Genetics in Medicine, 2022, 24, 1567-1582.	2.4	5
63	Familial cardioâ€facioâ€cutaneous syndrome: Vertical transmission of the <scp>BRAF</scp> p.<scp>G464R</scp> pathogenic variant and review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 469-475.	1.2	4
64	Ophthalmic manifestations in Costello syndrome caused by Ras pathway dysregulation during development. Ophthalmic Genetics, 2022, 43, 48-57.	1.2	4
65	Age and ASD symptoms in Costello syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1027-1028.	1.2	2
66	Juvenile xanthogranuloma in Noonan syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 3048-3052.	1.2	2
67	Abstract 19583: Autonomous and Non-Autonomous Defects Underlie Hypertrophic Cardiomyopathy in BRAF-Mutant hiPSC-Derived Cardiomyocytes. Circulation, 2014, 130, .	1.6	0