Katherine A Rauen

List of Publications by Year
in descending order

67 papers	5,649 citations	116194 36 h-index	93651 72 g-index
78 all docs	78 docs citations	78 times ranked	7144 citing authors

#	Article	IF	Citations
1	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, .	1.2	9
2	Ophthalmic manifestations in Costello syndrome caused by Ras pathway dysregulation during development. Ophthalmic Genetics, 2022, 43, 48-57.	0.5	4
3	Defining RASopathy. DMM Disease Models and Mechanisms, 2022, 15, .	1.2	26
4	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.	0.7	10
5	A novel DPH5-related diphthamide-deficiency syndrome causing embryonic lethality or profound neurodevelopmental disorder. Genetics in Medicine, 2022, 24, 1567-1582.	1.1	5
6	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.	0.7	4
7	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.	0.8	6
8	Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. Genetics in Medicine, 2021, 23, 1506-1513.	1.1	290
9	Juvenile xanthogranuloma in Noonan syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 3048-3052.	0.7	2
10	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.	2.6	140
11	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.	0.7	40
12	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.	1.3	8
13	RAS pathway influences the number of melanocytic nevi in cardiofaciocutaneous and Costello syndromes. Journal of the American Academy of Dermatology, 2020, 82, 1091-1093.	0.6	5
14	The duality of human oncoproteins: drivers of cancer and congenital disorders. Nature Reviews Cancer, 2020, 20, 383-397.	12.8	44
15	Costello syndrome: Clinical phenotype, genotype, and management guidelines. American Journal of Medical Genetics, Part A, 2019, 179, 1725-1744.	0.7	70
16	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.	1.1	62
17	ClinGenâ€™s RASopathy Expert Panel consensus methods for variant interpretation. Genetics in Medicine, 2018, 20, 1334-1345.	1.1	126
18	RASopathies are associated with a distinct personality profile. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 434-446.	1.1	9

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19	Age and ASD symptoms in Costello syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1027-1028.	0.7	2
20	Mek1 Y130C mice recapitulate aspects of the human Cardio-Facio-Cutaneous syndrome. DMM Disease Models and Mechanisms, 2018, 11, .	1.2	19
21	Assessing the geneâ€“disease association of 19 genes with the RASopathies using the ClinGen gene curation framework. Human Mutation, 2018, 39, 1485-1493.	1.1	66
22	Reverse Pathway Genetic Approach Identifies Epistasis in Autism Spectrum Disorders. PLoS Genetics, 2017, 13, e1006516.	1.5	38
23	Autonomous and Non-autonomous Defects Underlie Hypertrophic Cardiomyopathy in BRAF-Mutant hiPSC-Derived Cardiomyocytes. Stem Cell Reports, 2016, 7, 355-369.	2.3	33
24	Pathogenetics of the RASopathies. Human Molecular Genetics, 2016, 25, R123-R132.	1.4	87
25	Expansion of the RASopathies. Current Genetic Medicine Reports, 2016, 4, 57-64.	1.9	85
26	Human iPS Cell-Derived Neurons Uncover the Impact of Increased Ras Signaling in Costello Syndrome. Journal of Neuroscience, 2016, 36, 142-152.	1.7	29
27	Dysregulation of astrocyte extracellular signaling in Costello syndrome. Science Translational Medicine, 2015, 7, 286ra66.	5.8	70
28	RASopathies: unraveling mechanisms with animal models. DMM Disease Models and Mechanisms, 2015, 8, 769-782.	1.2	66
29	Craniofacial and dental development in Costello syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 1425-1430.	0.7	21
30	Autism traits in the RASopathies. Journal of Medical Genetics, 2014, 51, 10-20.	1.5	134
31	Germline loss-of-function mutations in LZTR1 predispose to an inherited disorder of multiple schwannomas. Nature Genetics, 2014, 46, 182-187.	9.4	242
32	Abnormal Ras signaling in Costello syndrome (CS) negatively regulates enamel formation. Human Molecular Genetics, 2014, 23, 682-692.	1.4	36
33	Cardio-Facio-Cutaneous Syndrome: Clinical Features, Diagnosis, and Management Guidelines. Pediatrics, 2014, 134, e1149-e1162.	1.0	148
34	Deletion of MAP2K2/MEK2: a novel mechanism for a RASopathy?. Clinical Genetics, 2014, 85, 138-146.	1.0	30
35	Abstract 19583: Autonomous and Non-Autonomous Defects Underlie Hypertrophic Cardiomyopathy in BRAF-Mutant hiPSC-Derived Cardiomyocytes. Circulation, 2014, 130, .	1.6	0
36	The RASopathies. Annual Review of Genomics and Human Genetics, 2013, 14, 355-369.	2.5	673

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37	Craniofacial and dental development in cardioâ€facioâ€cutaneous syndrome: the importance of Ras signaling homeostasis. Clinical Genetics, 2013, 83, 539-544.	1.0	20
38	Continual and partial MEK inhibition ameliorates cardio-facio-cutaneous phenotypes in zebrafish. DMM Disease Models and Mechanisms, 2012, 5, 546-52.	1.2	44
39	Peripheral muscle weakness in RASopathies. Muscle and Nerve, 2012, 46, 394-399.	1.0	38
40	Dermatological phenotype in Costello syndrome: consequences of Ras dysregulation in development. British Journal of Dermatology, 2012, 166, 601-607.	1.4	70
41	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.	1.0	21
42	Dermatological findings in 61 mutation-positive individuals with cardiofaciocutaneous syndrome. British Journal of Dermatology, 2011, 164, no-no.	1.4	74
43	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.	0.7	99
44	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.	0.7	7
45	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.	0.7	41
46	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.	0.7	42
47	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.	0.7	40
48	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.	0.7	37
49	Mutational and Functional Analysis in Human Ras/MAP Kinase Genetic Syndromes. Methods in Molecular Biology, 2010, 661, 433-447.	0.4	10
50	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.	1.4	89
51	The RASopathies: developmental syndromes of Ras/MAPK pathway dysregulation. Current Opinion in Genetics and Development, 2009, 19, 230-236.	1.5	640
52	Biochemical Characterization of Novel Germline BRAF and MEK Mutations in Cardioâ€Facioâ€Cutaneous Syndrome. Methods in Enzymology, 2008, 438, 277-289.	0.4	29
53	Mutation Analysis of BRAF, MEK1 and MEK2 in 15 Ovarian Cancer Cell Lines: Implications for Therapy. PLoS ONE, 2007, 2, e1279.	1.1	77
54	Diversity, parental germline origin, and phenotypic spectrum of de novoHRASmissense changes in Costello syndrome. Human Mutation, 2007, 28, 265-272.	1.1	123

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55	HRAS and the Costello syndrome. Clinical Genetics, 2007, 71, 101-108.	1.0	104
56	Interstitial deletions of chromosome 6q: genotype-phenotype correlation utilizing array CGH. Clinical Genetics, 2007, 71, 260-266.	1.0	45
57	Neurological complications of cardioâ€facioâ€cutaneous syndrome. Developmental Medicine and Child Neurology, 2007, 49, 894-899.	1.1	106
58	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.	0.7	157
59	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.	0.7	34
60	Germline Mutations in Genes Within the MAPK Pathway Cause Cardio-facio-cutaneous Syndrome. Science, 2006, 311, 1287-1290.	6.0	505
61	Prader-Willi syndrome resulting from an unbalanced translocation: characterization by array comparative genomic hybridization. Clinical Genetics, 2004, 65, 477-482.	1.0	26
62	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.	1.0	38
63	Fertility in a female with mosaic trisomy 8. Fertility and Sterility, 2003, 79, 206-208.	0.5	16
64	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
65	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.	0.4	128
66	Tandem duplication mosaicism: characterization of a mosaic dup(5q) and review. Clinical Genetics, 2001, 60, 366-370.	1.0	11
67	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