The RASopathies

Annual Review of Genomics and Human Genetics 14, 355-369

DOI: 10.1146/annurev-genom-091212-153523

Citation Report

#	Article	IF	CITATIONS
1	Mammalian MYC Proteins and Cancer. New Journal of Science, 2014, 2014, 1-27.	1.0	170
2	Identification of a novel de novo deletion in <i>RAF1</i> associated with biventricular hypertrophy in Noonan syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 2069-2073.	0.7	5
3	A <i>Drosophila</i> screen identifies neurofibromatosis-1 genetic modifiers involved in systemic and synaptic growth. Rare Diseases (Austin, Tex), 2014, 2, e28341.	1.8	8
4	Activating mutations in RRAS underlie a phenotype within the RASopathy spectrum and contribute to leukaemogenesis. Human Molecular Genetics, 2014, 23, 4315-4327.	1.4	114
5	K-Ras ^{V14I} recapitulates Noonan syndrome in mice. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 16395-16400.	3.3	67
6	Next-generation sequencing identifies rare variants associated with Noonan syndrome. Proceedings of the United States of America, 2014, 111, 11473-11478.	3.3	158
7	A <i>PTPN11</i> allele encoding a catalytically impaired SHP2 protein in a patient with a Noonan syndrome phenotype. American Journal of Medical Genetics, Part A, 2014, 164, 2351-2355.	0.7	12
8	Structureâ€energyâ€based predictions and network modelling of <scp>RAS</scp> opathy and cancer missense mutations. Molecular Systems Biology, 2014, 10, 727.	3.2	74
9	Growth hormone replacement therapy in Costello syndrome. Growth Hormone and IGF Research, 2014, 24, 271-275.	0.5	5
10	What's in a disease name?. British Journal of Dermatology, 2014, 170, 1005-1007.	1.4	3
11	Critical care of patients with paediatric valvar cardiac disease. Cardiology in the Young, 2014, 24, 1071-1076.	0.4	1
12	N-of-1 genomic medicine for the rare pediatric genetic diseases. Expert Opinion on Orphan Drugs, 2014, 2, 1279-1290.	0.5	7
13	Update from the 2013 international neurofibromatosis conference. American Journal of Medical Genetics, Part A, 2014, 164, 2969-2978.	0.7	17
14	Contribution of <i>RIT1</i> mutations to the pathogenesis of Noonan syndrome: Four new cases and further evidence of heterogeneity. American Journal of Medical Genetics, Part A, 2014, 164, 2310-2316.	0.7	42
15	Dragging Ras Back in the Ring. Cancer Cell, 2014, 25, 272-281.	7.7	707
16	Elucidating ERK2 function in the heart. Journal of Molecular and Cellular Cardiology, 2014, 72, 336-338.	0.9	1
17	Diagnostic application of high resolution single nucleotide polymorphism array analysis for children with brain tumors. Cancer Genetics, 2014, 207, 111-123.	0.2	40
18	Malformation syndromes associated with disorders of sex development. Nature Reviews Endocrinology, 2014, 10, 476-487.	4.3	64

ιτατιώνι Ρεβώ

#	Article	IF	CITATIONS
19	Understanding intellectual disability through RASopathies. Journal of Physiology (Paris), 2014, 108, 232-239.	2.1	15
20	Rare copy number variations containing genes involved in RASopathies: deletion of SHOC2 and duplication of PTPN11. Molecular Cytogenetics, 2014, 7, 28.	0.4	23
21	Bedside to bench in juvenile myelomonocytic leukemia: insights into leukemogenesis from a rare pediatric leukemia. Blood, 2014, 124, 2487-2497.	0.6	98
22	Overcoming challenges in the acquisition of biospecimens for rare diseases. Expert Opinion on Orphan Drugs, 2014, 2, 1-4.	0.5	10
23	A Novel <i>SHOC2</i> Variant in Rasopathy. Human Mutation, 2014, 35, n/a-n/a.	1.1	28
24	Adult mice expressing aBrafQ241R mutation on an ICR/CD-1 background exhibit a cardio-facio-cutaneous syndrome phenotype. Human Molecular Genetics, 2015, 24, 7349-7360.	1.4	17
25	ERK1 and ERK2 Regulate Chondrocyte Terminal Differentiation During Endochondral Bone Formation. Journal of Bone and Mineral Research, 2015, 30, 765-774.	3.1	60
26	Inherited <i>CHST11/MIR3922</i> deletion is associated with a novel recessive syndrome presenting with skeletal malformation and malignant lymphoproliferative disease. Molecular Genetics & amp; Genomic Medicine, 2015, 3, 413-423.	0.6	11
27	126 novel mutations in Italian patients with neurofibromatosis type 1. Molecular Genetics & Genomic Medicine, 2015, 3, 513-525.	0.6	25
28	Oculoectodermal syndrome is a mosaic RASopathy associated with <i>KRAS</i> alterations. American Journal of Medical Genetics, Part A, 2015, 167, 1429-1435.	0.7	47
29	Distinct roles of the RasGAP family proteins in C. elegans associative learning and memory. Scientific Reports, 2015, 5, 15084.	1.6	18
30	Critical roles for murine Reck in the regulation of vascular patterning and stabilization. Scientific Reports, 2015, 5, 17860.	1.6	22
32	Cerebral volumetric abnormalities in Neurofibromatosis type 1: associations with parent ratings of social and attention problems, executive dysfunction, and autistic mannerisms. Journal of Neurodevelopmental Disorders, 2015, 7, 32.	1.5	41
33	Noonan syndromeâ€iike disorder with loose anagen hair: A second case with neuroblastoma. American Journal of Medical Genetics, Part A, 2015, 167, 1902-1907.	0.7	14
34	Craniosynostosis and Noonan syndrome with <i>KRAS</i> mutations: Expanding the phenotype with a case report and review of the literature. American Journal of Medical Genetics, Part A, 2015, 167, 2657-2663.	0.7	38
35	Mosaic partial deletion of <i>PTPN12</i> in a child with interrupted aortic arch type A. American Journal of Medical Genetics, Part A, 2015, 167, 2674-2683.	0.7	3
36	Copy number variants including RAS pathway genes—How much RASopathy is in the phenotype?. American Journal of Medical Genetics, Part A, 2015, 167, 2685-2690.	0.7	14
37	Behavioral phenotype in Costello syndrome with atypical mutation: A case report. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2015, 168, 66-71.	1.1	5

#	Article	IF	CITATIONS
38	A Novel Noonan Syndrome RAF1 Mutation: Lethal Course in a Preterm Infant. Mental Illness, 2015, 7, 5955.	0.8	8
39	Genetics of Bladder Malignant Tumors in Childhood. Current Genomics, 2015, 17, 14-32.	0.7	14
40	In vivo synaptic transmission and morphology in mouse models of Tuberous sclerosis, Fragile X syndrome, Neurofibromatosis type 1, and Costello syndrome. Frontiers in Cellular Neuroscience, 2015, 9, 234.	1.8	24
41	A Genetic Porcine Model of Cancer. PLoS ONE, 2015, 10, e0128864.	1.1	128
42	Differential allelic expression of SOS1 and hyperexpression of the activating SOS1 c.755C variant in a Noonan syndrome family. European Journal of Human Genetics, 2015, 23, 1531-1537.	1.4	14
43	Juvenile myelomonocytic leukemia due to a germline CBL Y371C mutation: 35-year follow-up of a large family. Human Genetics, 2015, 134, 775-787.	1.8	21
44	The 16p11.2 Deletion Mouse Model of Autism Exhibits Altered Cortical Progenitor Proliferation and Brain Cytoarchitecture Linked to the ERK MAPK Pathway. Journal of Neuroscience, 2015, 35, 3190-3200.	1.7	147
46	MEK Inhibitors Reverse cAMP-Mediated Anxiety in Zebrafish. Chemistry and Biology, 2015, 22, 1335-1346.	6.2	31
47	A Catalog of Genetic Syndromes in Childhood Cancer. Pediatric Blood and Cancer, 2015, 62, 2071-2075.	0.8	12
48	Nevus Anemicus: A Distinctive Cutaneous Finding in Neurofibromatosis Type 1. Pediatric Dermatology, 2015, 32, 342-347.	0.5	20
49	Compromised MAPK signaling in human diseases: an update. Archives of Toxicology, 2015, 89, 867-882.	1.9	782
50	Biome depletion in conjunction with evolutionary mismatches could play a role in the etiology of neurofibromatosis 1. Medical Hypotheses, 2015, 84, 305-314.	0.8	1
51	A novel single nucleotide splice site mutation in FHL1 confirms an Emery-Dreifuss plus phenotype with pulmonary artery hypoplasia and facial dysmorphology. European Journal of Medical Genetics, 2015, 58, 222-229.	0.7	11
52	Inhibition of RAS Activation Due to a Homozygous Ezrin Variant in Patients with Profound Intellectual Disability. Human Mutation, 2015, 36, 270-278.	1.1	18
53	Functional Redundancy of ERK1 and ERK2 MAP Kinases during Development. Cell Reports, 2015, 12, 913-921.	2.9	86
54	Fate and Plasticity of the Epidermis in Response to Congenital Activation of BRAF. Journal of Investigative Dermatology, 2015, 135, 481-489.	0.3	1
55	LEOPARD Syndrome Without Hearing Loss or Pulmonary Stenosis: A Report of 2 Cases. Actas Dermo-sifiliográficas, 2015, 106, e19-e22.	0.2	1
56	Dynamics of Inductive ERK Signaling in the Drosophila Embryo. Current Biology, 2015, 25, 1784-1790.	1.8	62

#	Article	IF	CITATIONS
57	SÃndrome LEOPARD sin sordera ni estenosis pulmonar: a propósito de 2 casos. Actas Dermo-sifiliográficas, 2015, 106, e19-e22.	0.2	4
58	Regulation of RAF protein kinases in ERK signalling. Nature Reviews Molecular Cell Biology, 2015, 16, 281-298.	16.1	506
59	Molecular Diversity and Associated Phenotypic Spectrum of Germline <i>CBL</i> Mutations. Human Mutation, 2015, 36, 787-796.	1.1	36
60	Dentate Gyrus Development Requires ERK Activity to Maintain Progenitor Population and MAPK Pathway Feedback Regulation. Journal of Neuroscience, 2015, 35, 6836-6848.	1.7	30
61	"RASopathic―astrocytes constrain neural plasticity. Science, 2015, 348, 636-637.	6.0	3
62	13q31.1 microdeletion: A prenatal case report with macrocephaly and macroglossia. European Journal of Medical Genetics, 2015, 58, 526-530.	0.7	6
63	The impact of the genetic background in the Noonan syndrome phenotype induced by K-RasV14I. Rare Diseases (Austin, Tex), 2015, 3, e1045169.	1.8	12
64	A Novel Mutation onRAF1in Association with Fetal Findings Suggestive of Noonan Syndrome. Fetal and Pediatric Pathology, 2015, 34, 361-364.	0.4	4
65	Thyroid C-Cell Biology and Oncogenic Transformation. Recent Results in Cancer Research, 2015, 204, 1-39.	1.8	39
66	RASopathies: unraveling mechanisms with animal models. DMM Disease Models and Mechanisms, 2015, 8, 769-782.	1.2	66
67	Haploinsufficiency and triploinsensitivity of the same 6p25.1p24.3 region in a family. BMC Medical Genomics, 2015, 8, 38.	0.7	7
68	Peripheral blood cells from children with RASopathies show enhanced spontaneous colonies growth in vitro and hyperactive RAS signaling. Blood Cancer Journal, 2015, 5, e324-e324.	2.8	3
69	SHP2 sails from physiology to pathology. European Journal of Medical Genetics, 2015, 58, 509-525.	0.7	182
70	Portrait of the PI3K/AKT pathway in colorectal cancer. Biochimica Et Biophysica Acta: Reviews on Cancer, 2015, 1855, 104-121.	3.3	205
72	1p13.2 deletion displays clinical features overlapping Noonan syndrome, likely related to NRAS gene haploinsufficiency. Genetics and Molecular Biology, 2016, 39, 349-357.	0.6	5
73	Layer specific and general requirements for ERK/MAPK signaling in the developing neocortex. ELife, 2016, 5, .	2.8	54
74	The yin–yang of kinase activation and unfolding explains the peculiarity of Val600 in the activation segment of BRAF. ELife, 2016, 5, e12814.	2.8	34
75	ERK1 and ERK2 Map Kinases: Specific Roles or Functional Redundancy?. Frontiers in Cell and Developmental Biology, 2016, 4, 53.	1.8	209

	Сітатіо	n Report	
#	Article	IF	CITATIONS
76	Human Engineered Cardiac Tissues Created Using Induced Pluripotent Stem Cells Reveal Functional Characteristics of BRAF-Mediated Hypertrophic Cardiomyopathy. PLoS ONE, 2016, 11, e0146697.	1.1	72
77	A Novel HRAS Mutation Independently Contributes to Left Ventricular Hypertrophy in a Family with a Known MYH7 Mutation. PLoS ONE, 2016, 11, e0168501.	1.1	13
78	Practice and Educational Gaps in Genodermatoses. Dermatologic Clinics, 2016, 34, 303-310.	1.0	11
79	Behavioral functioning in cardiofaciocutaneous syndrome: Risk factors and impact on parenting experience. American Journal of Medical Genetics, Part A, 2016, 170, 1974-1988.	0.7	17
80	Biochemical Classification of Disease-associated Mutants of RAS-like Protein Expressed in Many Tissues (RIT1). Journal of Biological Chemistry, 2016, 291, 15641-15652.	1.6	14
81	Neurovascular Stains in Two Girls with Neurofibromatosis 1. Pediatric Dermatology, 2016, 33, e158-9.	0.5	0
82	Retrospective study of prenatal ultrasound findings in newborns with a Noonan spectrum disorder. Prenatal Diagnosis, 2016, 36, 418-423.	1.1	25
83	Hypertrophic neuropathy in Noonan syndrome with multiple lentigines. American Journal of Medical Genetics, Part A, 2016, 170, 1570-1572.	0.7	12
84	Nutritional aspects of Noonan syndrome and Noonanâ€related disorders. American Journal of Medical Genetics, Part A, 2016, 170, 1525-1531.	0.7	18
85	Respiratory system involvement in Costello syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 1849-1857.	0.7	11
86	Neuropsychological Functioning in Individuals with Noonan Syndrome: a Systematic Literature Review with Educational and Treatment Recommendations. Journal of Pediatric Neuropsychology, 2016, 2, 14-33.	0.3	25
87	The MAPK Signaling Cascades. , 2016, , 122-127.		14
88	Casitas B-cell lymphoma (Cbl) proteins protect mammary epithelial cells from proteotoxicity of active c-Src accumulation. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, E8228-E8237.	3.3	9
89	De novo RRAGC mutation activates mTORC1 signaling in syndromic fetal dilated cardiomyopathy. Human Genetics, 2016, 135, 909-917.	1.8	28
90	RSK3 is required for concentric myocyte hypertrophy in an activated Raf1 model for Noonan syndrome. Journal of Molecular and Cellular Cardiology, 2016, 93, 98-105.	0.9	7
91	The Conundrum of Genetic "Drivers―in Benign Conditions. Journal of the National Cancer Institute, 2016, 108, djw036.	3.0	113
92	Metabolites involved in glycolysis and amino acid metabolism are altered in short children born small for gestational age. Pediatric Research, 2016, 80, 299-305.	1.1	6
93	Genotype and phenotype in patients with Noonan syndrome and a RIT1 mutation. Genetics in Medicine, 2016, 18, 1226-1234.	1.1	77

		CITATION REPORT		
#	Article		IF	CITATIONS
94	The Genetic Basis of Hydrocephalus. Annual Review of Neuroscience, 2016, 39, 409-43	ō.	5.0	93
95	Germline De Novo Mutations in GNB1 Cause Severe Neurodevelopmental Disability, Hy Seizures. American Journal of Human Genetics, 2016, 98, 1001-1010.	potonia, and	2.6	102
96	Impaired synaptic plasticity in RASopathies: a mini-review. Journal of Neural Transmissio 1133-1138.	n, 2016, 123,	1.4	14
97	Enhancing Literacy in Cardiovascular Genetics: A Scientific Statement From the America Association. Circulation: Cardiovascular Genetics, 2016, 9, 448-467.	in Heart	5.1	64
98	Systemic Lupus Erythematosus: Is It One Disease?. ReumatologÃa ClÃnica (English Editi 274-281.	on), 2016, 12,	0.2	10
99	Klippel–Trenaunay syndrome belongs to the <i>PIK3CA</i> â€related overgrowth spec Experimental Dermatology, 2016, 25, 17-19.	ctrum (PROS).	1.4	143
100	MAPK kinase signalling dynamics regulate cell fate decisions and drug resistance. Curre Structural Biology, 2016, 41, 151-158.	nt Opinion in	2.6	72
101	RASopathies Are Associated With Delayed Puberty; Are They Associated With Precociou Pediatrics, 2016, 138, .	ıs Puberty Too?.	1.0	10
102	Copy number variants and rasopathies: germline KRAS duplication in a patient with sync including pigmentation abnormalities. Orphanet Journal of Rare Diseases, 2016, 11, 102	drome 1.	1.2	7
103	Cutaneous skeletal hypophosphatemia syndrome (CSHS) is a multilineage somatic mos Journal of the American Academy of Dermatology, 2016, 75, 420-427.	aic RASopathy.	0.6	44
104	Pathogenetics of the RASopathies. Human Molecular Genetics, 2016, 25, R123-R132.		1.4	87
105	Expansion of the RASopathies. Current Genetic Medicine Reports, 2016, 4, 57-64.		1.9	85
106	The function of Shoc2: A scaffold and beyond. Communicative and Integrative Biology, e1188241.	2016, 9,	0.6	26
107	The PI3K signaling pathway as a pharmacological target in Autism related disorders and Molecular and Cellular Therapies, 2016, 4, 2.	Schizophrenia.	0.2	51
108	Cell type-specific roles of RAS-MAPK signaling in learning and memory: Implications in neurodevelopmental disorders. Neurobiology of Learning and Memory, 2016, 135, 13-2	1.	1.0	39
109	K-Ras ^{V14I} -induced Noonan syndrome predisposes to tumour developmen of Pathology, 2016, 239, 206-217.	t in mice. Journal	2.1	12
111	Objective studies of the face of Noonan, Cardioâ€facioâ€cutaneous, and Costello synd comparison of three disorders of the Ras/MAPK signaling pathway. American Journal of Genetics, Part A, 2016, 170, 2570-2577.	romes: A Medical	0.7	25
112	RAS and downstream RAF-MEK and PI3K-AKT signaling in neuronal development, function dysfunction. Biological Chemistry, 2016, 397, 215-222.	on and	1.2	60

#	Article	IF	CITATIONS
113	Mutations in RIT1 cause Noonan syndrome with possible juvenile myelomonocytic leukemia but are not involved in acute lymphoblastic leukemia. European Journal of Human Genetics, 2016, 24, 1124-1131.	1.4	23
114	An Introduction to Signal Transduction. , 2016, , 53-183.		1
115	HSF1: Guardian of Proteostasis in Cancer. Trends in Cell Biology, 2016, 26, 17-28.	3.6	166
116	Interaction between a Domain of the Negative Regulator of the Ras-ERK Pathway, SPRED1 Protein, and the GTPase-activating Protein-related Domain of Neurofibromin Is Implicated in Legius Syndrome and Neurofibromatosis Type 1. Journal of Biological Chemistry, 2016, 291, 3124-3134.	1.6	49
117	Acute lymphoblastic leukemia in the context of RASopathies. European Journal of Medical Genetics, 2016, 59, 173-178.	0.7	35
118	Lupus eritematoso sistémico: ¿es una sola enfermedad?. ReumatologÃa ClÃnica, 2016, 12, 274-281.	0.2	25
119	Differential Effects of HRAS Mutation on LTP-Like Activity Induced by Different Protocols of Repetitive Transcranial Magnetic Stimulation. Brain Stimulation, 2016, 9, 33-38.	0.7	11
120	Case Report: Congenital Erythroleukemia in a Premature Infant with Dysmorphic Features. Pediatric and Developmental Pathology, 2016, 19, 334-337.	0.5	1
121	The lymphatic phenotype in Noonan and Cardiofaciocutaneous syndrome. European Journal of Human Genetics, 2016, 24, 690-696.	1.4	66
122	Recent advances in RASopathies. Journal of Human Genetics, 2016, 61, 33-39.	1.1	290
123	Multiple odontogenic cysts in a patient with Neurofibromatosis–Noonan syndrome. Journal of Oral and Maxillofacial Surgery, Medicine, and Pathology, 2016, 28, 51-54.	0.2	2
124	How necessary is to analyze <i>PTPN11</i> gene in fetuses with first trimester cystic hygroma and normal karyotype?. Journal of Maternal-Fetal and Neonatal Medicine, 2017, 30, 938-941.	0.7	13
125	The Spatiotemporal Limits of Developmental Erk Signaling. Developmental Cell, 2017, 40, 185-192.	3.1	158
126	Macrocyclic MEK1/2 inhibitor with efficacy in a mouse model of cardiomyopathy caused by lamin A/C gene mutation. Bioorganic and Medicinal Chemistry, 2017, 25, 1004-1013.	1.4	19
127	The NF1 gene in tumor syndromes and melanoma. Laboratory Investigation, 2017, 97, 146-157.	1.7	144
128	Autism spectrum disorder and other neurobehavioural comorbidities in rare disorders of the Ras/ <scp>MAPK</scp> pathway. Developmental Medicine and Child Neurology, 2017, 59, 544-549.	1.1	40
129	Constitutional bone impairment in Noonan syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 692-698.	0.7	15
130	Divergent effects of intrinsically active MEK variants on developmental Ras signaling. Nature Genetics, 2017, 49, 465-469.	9.4	51

#	Article	IF	CITATIONS
131	Time-resolved Phosphoproteome Analysis of Paradoxical RAF Activation Reveals Novel Targets of ERK. Molecular and Cellular Proteomics, 2017, 16, 663-679.	2.5	26
132	Deciphering the RAS/ERK pathway <i>in vivo</i> . Biochemical Society Transactions, 2017, 45, 27-36.	1.6	45
133	Aberrant <i>HRAS</i> transcript processing underlies a distinctive phenotype within the RASopathy clinical spectrum. Human Mutation, 2017, 38, 798-804.	1.1	14
134	BRaf signaling principles unveiled by large-scale human mutation analysis with a rapid lentivirus-based gene replacement method. Genes and Development, 2017, 31, 537-552.	2.7	20
135	Oligodendrocyte Nf1 Controls Aberrant Notch Activation and Regulates Myelin Structure and Behavior. Cell Reports, 2017, 19, 545-557.	2.9	42
136	Neurobiology of Autism Spectrum Disorders. , 2017, , 29-93.		1
137	Sustained activation of ERK1/2 MAPK in Schwann cells causes corneal neurofibroma. Journal of Neuroscience Research, 2017, 95, 1712-1729.	1.3	22
138	Genotype and phenotype spectrum of NRAS germline variants. European Journal of Human Genetics, 2017, 25, 823-831.	1.4	36
139	New Genetic Diagnoses of Short Stature Provide Insights into Local Regulation of Childhood Growth. Hormone Research in Paediatrics, 2017, 88, 22-37.	0.8	29
140	Cochlear implantation and clinical features in patients with Noonan syndrome and Noonan syndrome with multiple lentigines caused by a mutation in PTPN11. International Journal of Pediatric Otorhinolaryngology, 2017, 97, 228-234.	0.4	10
141	Recommendations for Cancer Surveillance in Individuals with RASopathies and Other Rare Genetic Conditions with Increased Cancer Risk. Clinical Cancer Research, 2017, 23, e83-e90.	3.2	122
142	Application of Next-Generation Sequencing in Noonan Spectrum Disorders. , 2017, , 155-176.		0
143	Turning the tide in myelodysplastic/myeloproliferative neoplasms. Nature Reviews Cancer, 2017, 17, 425-440.	12.8	117
144	Parallel imaging of Drosophila embryos for quantitative analysis of genetic perturbations of the Ras pathway. DMM Disease Models and Mechanisms, 2017, 10, 923-929.	1.2	12
145	Mir-132/212 is required for maturation of binocular matching of orientation preference and depth perception. Nature Communications, 2017, 8, 15488.	5.8	31
146	Cellular interplay via cytokine hierarchy causes pathological cardiac hypertrophy in RAF1-mutant Noonan syndrome. Nature Communications, 2017, 8, 15518.	5.8	23
147	Testing for Noonan syndrome after increased nuchal translucency. Prenatal Diagnosis, 2017, 37, 750-753.	1.1	36
148	At first sight or second glance: clinical presentation of mosaic manifestations of autosomal dominant skin disorders – a case series. Journal of the European Academy of Dermatology and Venereology, 2017, 31, 1912-1915.	1.3	5

#	Article	IF	CITATIONS
149	Further evidence that variants in <i>PPP1CB</i> cause a rasopathy similar to Noonan syndrome with loose anagen hair. American Journal of Medical Genetics, Part A, 2017, 173, 565-567.	0.7	20
150	Single-Step Affinity Purification of ERK Signaling Complexes Using the Streptavidin-Binding Peptide (SBP) Tag. Methods in Molecular Biology, 2017, 1487, 113-126.	0.4	13
151	Modeling RASopathies with Genetically Modified Mouse Models. Methods in Molecular Biology, 2017, 1487, 379-408.	0.4	13
152	In vivo severity ranking of Ras pathway mutations associated with developmental disorders. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, 510-515.	3.3	44
153	How activating mutations affect MEK1 regulation and function. Journal of Biological Chemistry, 2017, 292, 18814-18820.	1.6	15
154	Mechanisms and causality in molecular diseases. History and Philosophy of the Life Sciences, 2017, 39, 35.	0.6	3
155	Loss of Extracellular Signal-Regulated Kinase 1/2 in the Retinal Pigment Epithelium Leads to RPE65 Decrease and Retinal Degeneration. Molecular and Cellular Biology, 2017, 37, .	1.1	11
156	<scp>C</scp> hronic impairment of ERK signaling in glutamatergic neurons of the forebrain does not affect spatial memory retention and LTP in the same manner as acute blockade of the ERK pathway. Hippocampus, 2017, 27, 1239-1249.	0.9	12
157	Hiperpigmentaciones. EMC - DermatologÃa, 2017, 51, 1-14.	0.1	0
158	Case report: Left ventricular noncompaction cardiomyopathy and RASopathies. European Journal of Medical Genetics, 2017, 60, 680-684.	0.7	3
159	Noonan syndrome: lessons learned from genetically modified mouse models. Expert Review of Endocrinology and Metabolism, 2017, 12, 367-378.	1.2	2
160	KRAS Alleles: The Devil Is in the Detail. Trends in Cancer, 2017, 3, 686-697.	3.8	257
161	RAC1 Missense Mutations in Developmental Disorders with Diverse Phenotypes. American Journal of Human Genetics, 2017, 101, 466-477.	2.6	119
162	Copy number variants of Ras/ <scp>MAPK</scp> pathway genes in patients with isolated cryptorchidism. Andrology, 2017, 5, 923-930.	1.9	4
163	Multiple central giant cell tumour lesions are exclusively linked to syndromes related to RAS/MAPK pathway anomalies. International Journal of Oral and Maxillofacial Surgery, 2017, 46, 1354-1355.	0.7	4
164	A Braf kinase-inactive mutant induces lung adenocarcinoma. Nature, 2017, 548, 239-243.	13.7	85
165	Cardiac defects, morbidity and mortality in patients affected by RASopathies. CARNET study results. International Journal of Cardiology, 2017, 245, 92-98.	0.8	75
166	RAF1 variants causing biventricular hypertrophic cardiomyopathy in two preterm infants: further phenotypic delineation and review of literature. Clinical Dysmorphology, 2017, 26, 195-199.	0.1	21

#	Article	IF	CITATIONS
167	Piconewtonâ€Scale Analysis of Rasâ€BRaf Signal Transduction with Singleâ€Molecule Force Spectroscopy. Small, 2017, 13, 1701972.	5.2	3
168	Cellular Phenotypes in Human iPSC-Derived Neurons from a Genetic Model of Autism Spectrum Disorder. Cell Reports, 2017, 21, 2678-2687.	2.9	109
169	Mechanisms underlying cognitive deficits in a mouse model for Costello Syndrome are distinct from other RASopathy mouse models. Scientific Reports, 2017, 7, 1256.	1.6	26
170	A review of craniofacial and dental findings of the RASopathies. Orthodontics and Craniofacial Research, 2017, 20, 32-38.	1.2	54
171	Human astrocytes are distinct contributors to the complexity of synaptic function. Brain Research Bulletin, 2017, 129, 66-73.	1.4	32
172	Severe Intellectual Disability and Enhanced Gamma-Aminobutyric Acidergic Synaptogenesis in a Novel Model of Rare RASopathies. Biological Psychiatry, 2017, 81, 179-192.	0.7	30
173	Role of astrocyte–synapse interactions in CNS disorders. Journal of Physiology, 2017, 595, 1903-1916.	1.3	158
174	A unique case of multiple non-ossifying fibromas with polyostotic monomelic distribution and aggressive clinical course. Skeletal Radiology, 2017, 46, 233-236.	1.2	3
175	Variability in clinical and neuropsychological features of individuals with <i>MAP2K1</i> mutations. American Journal of Medical Genetics, Part A, 2017, 173, 452-459.	0.7	12
176	Modeling Syndromic Congenital Heart Defects in Zebrafish. Current Topics in Developmental Biology, 2017, 124, 1-40.	1.0	36
177	Protein Hardware for Signaling. , 2017, , 425-442.		0
178	Impaired Osteogenesis of Disease-Specific Induced Pluripotent Stem Cells Derived from a CFC Syndrome Patient. International Journal of Molecular Sciences, 2017, 18, 2591.	1.8	6
179	Functional convergence of histone methyltransferases EHMT1 and KMT2C involved in intellectual disability and autism spectrum disorder. PLoS Genetics, 2017, 13, e1006864.	1.5	116
180	Learning Disability in RASopathies. , 2017, , .		0
181	Pathophysiology of Cardiomyopathies. , 2017, , 1563-1575.e4.		0
182	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. Genetics in Medicine, 2018, 20, 1175-1185.	1.1	133
183	ClinGen's RASopathy Expert Panel consensus methods for variant interpretation. Genetics in Medicine, 2018, 20, 1334-1345.	1.1	126
184	RASopathies are associated with a distinct personality profile. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 434-446.	1.1	9

#	Article	IF	CITATIONS
185	Spred1 Safeguards Hematopoietic Homeostasis against Diet-Induced Systemic Stress. Cell Stem Cell, 2018, 22, 713-725.e8.	5.2	33
186	Characteristic MR Imaging Findings of the Neonatal Brain in RASopathies. American Journal of Neuroradiology, 2018, 39, 1146-1152.	1.2	12
187	Structural fingerprints, interactions, and signaling networks of RAS family proteins beyond RAS isoforms. Critical Reviews in Biochemistry and Molecular Biology, 2018, 53, 130-156.	2.3	34
188	Loss of Capicua alters early T cell development and predisposes mice to T cell lymphoblastic leukemia/lymphoma. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E1511-E1519.	3.3	35
189	Autism Spectrum Disorder in an Unselected Cohort of Children with Neurofibromatosis Type 1 (NF1). Journal of Autism and Developmental Disorders, 2018, 48, 2278-2285.	1.7	37
190	Noonan syndrome with multiple lentigines and associated craniosynostosis. Clinical and Experimental Dermatology, 2018, 43, 357-359.	0.6	7
191	Mice with an Oncogenic HRAS Mutation are Resistant to High-Fat Diet-Induced Obesity and Exhibit Impaired Hepatic Energy Homeostasis. EBioMedicine, 2018, 27, 138-150.	2.7	33
192	The novel <i>RAF1</i> mutation p.(Cly361Ala) located outside the kinase domain of the CR3 region in two patients with Noonan syndrome, including one with a rare brain tumor. American Journal of Medical Genetics, Part A, 2018, 176, 470-476.	0.7	17
193	NGS testing for cardiomyopathy: Utility of adding RASopathy-associated genes. Human Mutation, 2018, 39, 954-958.	1.1	11
194	<i>Mek1 Y130C</i> mice recapitulate aspects of the human Cardio-Facio-Cutaneous syndrome. DMM Disease Models and Mechanisms, 2018, 11, .	1.2	19
195	Clinical Presentation and Natural History of Hypertrophic Cardiomyopathy in RASopathies. Heart Failure Clinics, 2018, 14, 225-235.	1.0	44
196	Defining the role of the RSK isoforms in cancer. Seminars in Cancer Biology, 2018, 48, 53-61.	4.3	71
197	Psychopathological features in Noonan syndrome. European Journal of Paediatric Neurology, 2018, 22, 170-177.	0.7	26
198	Epidermal nevus syndromes: New insights into whorls and swirls. Pediatric Dermatology, 2018, 35, 21-29.	O.5	44
199	Targeting the Architecture of Deregulated Protein Complexes in Cancer. Advances in Protein Chemistry and Structural Biology, 2018, 111, 101-132.	1.0	5
200	ERK/MAPK signaling and autism spectrum disorders. Progress in Brain Research, 2018, 241, 63-112.	0.9	69
201	RAS variant signalling. Biochemical Society Transactions, 2018, 46, 1325-1332.	1.6	61
202	Genetic diagnosis of neurofibromatosis type 1: targeted next- generation sequencing with Multiple Ligation-Dependent Probe Amplification analysis. Journal of Biomedical Science, 2018, 25, 72.	2.6	19

#	Article	IF	CITATIONS
204	SHOC2–MRAS–PP1 complex positively regulates RAF activity and contributes to Noonan syndrome pathogenesis. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E10576-E10585.	3.3	59
205	Assessing the gene–disease association of 19 genes with the RASopathies using the ClinGen gene curation framework. Human Mutation, 2018, 39, 1485-1493.	1.1	66
206	Fast Dynamic in vivo Monitoring of Erk Activity at Single Cell Resolution in DREKA Zebrafish. Frontiers in Cell and Developmental Biology, 2018, 6, 111.	1.8	33
207	Study of Ras/ <scp>MAPK</scp> pathway gene variants in Chilean patients with Cryptorchidism. Andrology, 2018, 6, 579-584.	1.9	2
208	De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features. American Journal of Human Genetics, 2018, 103, 154-162.	2.6	56
209	Outstanding questions in developmental ERK signaling. Development (Cambridge), 2018, 145, .	1.2	48
210	Nonreentrant atrial tachycardia occurs independently of hypertrophic cardiomyopathy in RASopathy patients. American Journal of Medical Genetics, Part A, 2018, 176, 1711-1722.	0.7	21
211	RAS GTPase-dependent pathways in developmental diseases: old guys, new lads, and current challenges. Current Opinion in Cell Biology, 2018, 55, 42-51.	2.6	18
212	RIT1 controls actin dynamics via complex formation with RAC1/CDC42 and PAK1. PLoS Genetics, 2018, 14, e1007370.	1.5	25
213	Whole Genome Sequencing Improves Outcomes of Genetic Testing in Patients With Hypertrophic Cardiomyopathy. Journal of the American College of Cardiology, 2018, 72, 419-429.	1.2	138
214	A quantitative model of developmental RTK signaling. Developmental Biology, 2018, 442, 80-86.	0.9	15
215	The MAPK Erk5 is necessary for proper skeletogenesis through a molecular axis that involves Smurfs-Smads-Sox9. Development (Cambridge), 2018, 145, .	1.2	29
216	Small molecule inhibition of RAS/MAPK signaling ameliorates developmental pathologies of Kabuki Syndrome. Scientific Reports, 2018, 8, 10779.	1.6	50
217	Social skills in children with RASopathies: a comparison of Noonan syndrome and neurofibromatosis type 1. Journal of Neurodevelopmental Disorders, 2018, 10, 21.	1.5	25

218 Attention Deficit Hyperactivity Disorder in Neurofibromatosis Type 1: Evaluation with a Continuous

#	Article	IF	CITATIONS
222	Chondroblastoma-like tumor of the skull in a patient with cardio-facio-cutaneous syndrome. Pathology Research and Practice, 2018, 214, 1510-1513.	1.0	1
224	A novel homozygous missense mutation in the SH3-binding motif of STAMBP causing microcephaly-capillary malformation syndrome. Journal of Human Genetics, 2018, 63, 957-963.	1.1	10
225	Ras/ERK-signalling promotes tRNA synthesis and growth via the RNA polymerase III repressor Maf1 in Drosophila. PLoS Genetics, 2018, 14, e1007202.	1.5	27
226	Co-occurrence of Noonan and Cardiofaciocutaneous Syndrome Features in a Patient with KRAS Variant. Journal of Pediatric Genetics, 2018, 07, 158-163.	0.3	4
227	Mouse models as a tool for discovering new neurological diseases. Neurobiology of Learning and Memory, 2019, 165, 106902.	1.0	17
228	Distinct Clinical and Pathological Features of Melorheostosis Associated With Somatic <i>MAP2K1</i> Mutations. Journal of Bone and Mineral Research, 2019, 34, 145-156.	3.1	22
229	Ras-Specific GTPase-Activating Proteins—Structures, Mechanisms, and Interactions. Cold Spring Harbor Perspectives in Medicine, 2019, 9, a031500.	2.9	40
230	Conserved NDR/LATS kinase controls RAS GTPase activity to regulate cell growth and chronological lifespan. Molecular Biology of the Cell, 2019, 30, 2598-2616.	0.9	14
231	Identification of lysine methylation in the core GTPase domain by GoMADScan. PLoS ONE, 2019, 14, e0219436.	1.1	6
232	Mutations in RABL3 alter KRAS prenylation and are associated with hereditary pancreatic cancer. Nature Genetics, 2019, 51, 1308-1314.	9.4	47
233	Genotype-phenotype correlation analysis in Japanese patients with Noonan syndrome. Endocrine Journal, 2019, 66, 983-994.	0.7	12
234	Antenatal diagnosis of cardio-facio-cutaneous syndrome: Prenatal characteristics and contribution of fetal facial dysmorphic signs in utero. About a case and review of literature. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2019, 240, 232-241.	0.5	9
235	Application of Proteomics Profiling for Biomarker Discovery in Hypertrophic Cardiomyopathy. Journal of Cardiovascular Translational Research, 2019, 12, 569-579.	1.1	17
236	Diagnostic Utility of Next-Generation Sequencing for Disorders of Somatic Mosaicism: A Five-Year Cumulative Cohort. American Journal of Human Genetics, 2019, 105, 734-746.	2.6	23
237	Pharmacological Targeting of STK19 Inhibits Oncogenic NRAS-Driven Melanomagenesis. Cell, 2019, 176, 1113-1127.e16.	13.5	74
238	Sixâ€yearâ€old male with a chief complaint of telangiectasias. Pediatric Dermatology, 2019, 36, 147-148.	0.5	0
239	Costello syndrome: Clinical phenotype, genotype, and management guidelines. American Journal of Medical Genetics, Part A, 2019, 179, 1725-1744.	0.7	70
240	SHOC2 complex-driven RAF dimerization selectively contributes to ERK pathway dynamics. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 13330-13339.	3.3	33

#	Article	IF	CITATIONS
241	Functional characterisation of a novel class of in-frame insertion variants of KRAS and HRAS. Scientific Reports, 2019, 9, 8239.	1.6	12
242	Activating Mutations of RRAS2 Are a Rare Cause of Noonan Syndrome. American Journal of Human Genetics, 2019, 104, 1223-1232.	2.6	43
243	Functional robustness of adult spermatogonial stem cells after induction of hyperactive Hras. PLoS Genetics, 2019, 15, e1008139.	1.5	6
244	Fetal cardiac abnormalities: Genetic etiologies to be considered. Prenatal Diagnosis, 2019, 39, 758-780.	1.1	11
245	A Premature Stop Codon in RAF1 Is the Priority Candidate Causative Mutation of the Inherited Chicken Wingless-2 Developmental Syndrome. Genes, 2019, 10, 353.	1.0	2
246	Gastrointestinal Manifestations of Noonan Syndrome. , 2019, , 85-91.		0
247	Noonan Syndrome: Phenotypic Variations and Molecular Genetics. , 2019, , 1-14.		1
248	The Noonan Syndrome-linked Raf1L613V mutation drives increased glial number in the mouse cortex and enhanced learning. PLoS Genetics, 2019, 15, e1008108.	1.5	22
249	Activating KRAS mutations in arteriovenous malformations of the brain: frequency and clinicopathologic correlation. Human Pathology, 2019, 89, 33-39.	1.1	45
250	New Noonan syndrome model mice with RIT1 mutation exhibit cardiac hypertrophy and susceptibility to β-adrenergic stimulation-induced cardiac fibrosis. EBioMedicine, 2019, 42, 43-53.	2.7	23
251	Excitatory neuron–specific SHP2-ERK signaling network regulates synaptic plasticity and memory. Science Signaling, 2019, 12, .	1.6	30
253	Mutations in the RAS pathway as potential precision medicine targets in treatment of rhabdomyosarcoma. Biochemical and Biophysical Research Communications, 2019, 512, 524-530.	1.0	12
254	<p>Social cognitive training for adults with Noonan syndrome: a feasibility study</p> . Neuropsychiatric Disease and Treatment, 2019, Volume 15, 611-626.	1.0	8
255	Nuclear ERK: Mechanism of Translocation, Substrates, and Role in Cancer. International Journal of Molecular Sciences, 2019, 20, 1194.	1.8	121
256	Rasopathies case report: concurrence of two pathogenic variations de novo in NF1 and KRAS genes in a patient. BMC Pediatrics, 2019, 19, 92.	0.7	2
257	The spectrum of genetic variants and phenotypic features of Southeast Asian patients with Noonan syndrome. Molecular Genetics & Genomic Medicine, 2019, 7, e00581.	0.6	8
258	Rosette-Forming Glioneuronal Tumor in Opticochiasmatic Region—Novel Entity in New Location. World Neurosurgery, 2019, 125, 253-256.	0.7	4
259	Clinical and mutation profile of pediatric patients with RASopathy-associated hypertrophic cardiomyopathy: results from a Chinese cohort. Orphanet Journal of Rare Diseases, 2019, 14, 29.	1.2	27

#	Article	IF	CITATIONS
260	Genetic predisposition to cancer: Surveillance and intervention. Seminars in Pediatric Surgery, 2019, 28, 150858.	0.5	5
261	Cancer Risk in Congenital Heart Disease—What Is the Evidence?. Canadian Journal of Cardiology, 2019, 35, 1750-1761.	0.8	21
262	Architecture of autoinhibited and active BRAF–MEK1–14-3-3 complexes. Nature, 2019, 575, 545-550.	13.7	197
264	A report on a girl of Noonan syndrome 9 presenting with bilateral lower limbs lymphedema. Chinese Medical Journal, 2019, 132, 480-482.	0.9	4
265	Selective Regulation of B-Raf Dependent K-Ras/Mitogen-Activated Protein by Natural Occurring Multi-kinase Inhibitors in Cancer Cells. Frontiers in Oncology, 2019, 9, 1220.	1.3	25
266	The impact of RASopathy-associated mutations on CNS development in mice and humans. Molecular Brain, 2019, 12, 96.	1.3	30
267	Optimizing photoswitchable MEK. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 25756-25763.	3.3	30
268	Caring for children with neurofibromatosis type 1. Nursing, 2019, 49, 30-36.	0.2	2
269	miRNA Genetic Variants Alter Their Secondary Structure and Expression in Patients With RASopathies Syndromes. Frontiers in Genetics, 2019, 10, 1144.	1.1	11
270	RAS genes in colorectal carcinoma: pathogenesis, testing guidelines and treatment implications. Journal of Clinical Pathology, 2019, 72, 135-139.	1.0	28
271	Congenital Hydrocephalus. , 2019, , 87-113.		2
272	Metformin for Treatment of Fragile X Syndrome and Other Neurological Disorders. Annual Review of Medicine, 2019, 70, 167-181.	5.0	52
273	C-type natriuretic peptide improves growth retardation in a mouse model of cardio-facio-cutaneous syndrome. Human Molecular Genetics, 2019, 28, 74-83.	1.4	10
274	A QTL on chromosome 3q23 influences processing speed in humans. Genes, Brain and Behavior, 2019, 18, e12530.	1.1	1
275	Family-based germline sequencing in children with cancer. Oncogene, 2019, 38, 1367-1380.	2.6	33
276	Hematopoietic and neural crest defects in zebrafish <i>shoc2</i> mutants: a novel vertebrate model for Noonan-like syndrome. Human Molecular Genetics, 2019, 28, 501-514.	1.4	12
277	The Nuclear Translocation of Mitogen-Activated Protein Kinases: Molecular Mechanisms and Use as Novel Therapeutic Target. Neuroendocrinology, 2019, 108, 121-131.	1.2	48
278	Of local translation control and lipid signaling in neurons. Advances in Biological Regulation, 2019, 71, 194-205.	1.4	8

		CITATION REPORT		
#	Article	IF	-	Citations
279	Feedback regulation of RTK signaling in development. Developmental Biology, 2019, 447, 71-8	9. 0	0.9	53
280	Is Nanoclustering essential for all oncogenic KRas pathways? Can it explain why wild-type KRas inhibit its oncogenic variant?. Seminars in Cancer Biology, 2019, 54, 114-120.	can 4	.3	35
281	Activating MRAS mutations cause Noonan syndrome associated with hypertrophic cardiomyop Human Molecular Genetics, 2020, 29, 1772-1783.	athy. 1	.4	30
282	Microarray and RASopathyâ€disorder testing in fetuses with increased nuchal translucency. Ultrasound in Obstetrics and Gynecology, 2020, 55, 383-390.	0).9	31
283	Enhancing cognition through pharmacological and environmental interventions: Examples from preclinical models of neurodevelopmental disorders. Neuroscience and Biobehavioral Reviews, 110, 28-45.	1 2020, 2	.9	14
284	Mutational spectrum by phenotype: panelâ€based NGS testing of patients with clinical suspici RASopathy and children with multiple caféâ€auâ€lait macules. Clinical Genetics, 2020, 97, 2	on of 64-275. ¹	.0	13
285	NRAS associated RASopathy and embryonal rhabdomyosarcoma. American Journal of Medical (Part A, 2020, 182, 195-200.	Jenetics, 0).7	9
286	Identification of Gene Mutations in Primary Pediatric Cardiomyopathy by Whole Exome Sequer Pediatric Cardiology, 2020, 41, 165-174.	ncing. 0).6	7
287	Abnormal Fetal Growth. , 2020, , 913-938.			1
288	Making sense of giant cell lesions of the jaws (GCLJ): lessons learned from nextâ€generation sequencing. Journal of Pathology, 2020, 250, 126-133.	2	.1	27
289	Medically actionable comorbidities in adults with Costello syndrome. American Journal of Medi Genetics, Part A, 2020, 182, 130-136.	cal o).7	6
290	An Update on Neurofibromatosis Type 1-Associated Gliomas. Cancers, 2020, 12, 114.	1	.7	50
291	Advancing <scp>RAS/RASopathy</scp> therapies: An NClâ€sponsored intramural and extramu collaboration for the study of <scp>RASopathies</scp> . American Journal of Medical Genetics, 2020, 182, 866-876.	ral Part A, O).7	40
292	Single-domain antibodies for functional targeting of the signaling scaffold Shoc2. Molecular Immunology, 2020, 118, 110-116.	1	.0	4
293	Comparison of hair manifestations in cardioâ€facioâ€cutaneous and Costello syndromes highl influence of the RAS pathway on hair growth. Journal of the European Academy of Dermatolog Venereology, 2020, 34, 601-607.	ghts the y and 1	.3	8
294	Dominant-negative antagonists of the Ras–ERK pathway: DA-Raf and its related proteins gen alternative splicing of Raf. Experimental Cell Research, 2020, 387, 111775.	erated by 1	.2	6
295	Clinical trial design in neurofibromatosis type 1 as a model for other tumor predisposition syndromes. Neuro-Oncology Advances, 2020, 2, i134-i140.	0).4	5
296	Understanding the Biological Activities of Vitamin D in Type 1 Neurofibromatosis: New Insights Disease Pathogenesis and Therapeutic Design. Cancers, 2020, 12, 2965.	into 1	.7	12

#	Article	IF	CITATIONS
297	Association between ARID2 and RAS-MAPK pathway in intellectual disability and short stature. Journal of Medical Genetics, 2021, 58, 767-777.	1.5	4
298	SOS GEFs in health and disease. Biochimica Et Biophysica Acta: Reviews on Cancer, 2020, 1874, 188445.	3.3	44
299	Current therapy of advanced colorectal cancer according to RAS/RAF mutational status. Cancer and Metastasis Reviews, 2020, 39, 1143-1157.	2.7	19
300	A Review of Psychological, Social, and Behavioral Functions in the RASopathies. Journal of Pediatric Neuropsychology, 2020, 6, 131-142.	0.3	2
301	Hyperinsulinemic Hypoglycemia in a Patient with Costello Syndrome: An Etiology to Consider in Hypoglycemia. Molecular Syndromology, 2020, 11, 207-216.	0.3	5
302	Severe Lymphatic Disorder Resolved With MEK Inhibition in a Patient With Noonan Syndrome and SOS1 Mutation. Pediatrics, 2020, 146, .	1.0	56
303	Ras Pathways on Prox1 and Lymphangiogenesis: Insights for Therapeutics. Frontiers in Cardiovascular Medicine, 2020, 7, 597374.	1.1	23
304	Integrated molecular drivers coordinate biological and clinical states in melanoma. Nature Genetics, 2020, 52, 1373-1383.	9.4	36
305	M-Ras is Muscle-Ras, Moderate-Ras, Mineral-Ras, Migration-Ras, and Many More-Ras. Experimental Cell Research, 2020, 397, 112342.	1.2	7
306	Structural bioinformatics enhances mechanistic interpretation of genomic variation, demonstrated through the analyses of 935 distinct RAS family mutations. Bioinformatics, 2021, 37, 1367-1375.	1.8	6
307	Genodermatoses with malignant potential. Clinics in Dermatology, 2020, 38, 432-454.	0.8	9
308	Genetic Testing for Inherited Cardiovascular Diseases: A Scientific Statement From the American Heart Association. Circulation Genomic and Precision Medicine, 2020, 13, e000067.	1.6	200
309	Treatment of Dystonia Using Trihexyphenidyl in Costello Syndrome. Brain Sciences, 2020, 10, 450.	1.1	4
310	Clinical and Genetic Characteristics of Noonan Syndrome and Noonan-like Diseases. Russian Journal of Genetics, 2020, 56, 540-547.	0.2	0
311	Development of Noonan syndrome by deregulation of allosteric SOS autoactivation. Journal of Biological Chemistry, 2020, 295, 13651-13663.	1.6	6
312	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	2.6	48
313	Costello syndrome model mice with a Hras G12S mutation are susceptible to develop house dust mite-induced atopic dermatitis. Cell Death and Disease, 2020, 11, 617.	2.7	2
314	Neuron type-specific expression of a mutant KRAS impairs hippocampal-dependent learning and memory. Scientific Reports, 2020, 10, 17730.	1.6	2

	CHAHON R	LFURI	1
#	Article	IF	CITATIONS
315	The Emerging Role of Ras Pathway Signaling in Pediatric Cancer. Cancer Research, 2020, 80, 5155-5163.	0.4	17
316	Autoimmune Thyroid Disease in Specific Genetic Syndromes in Childhood and Adolescence. Frontiers in Endocrinology, 2020, 11, 543.	1.5	51
317	Clinical Implications of Mosaicism and Low-Level Mosaicism in Neurocutaneous Disorders. Current Genetic Medicine Reports, 2020, 8, 132-139.	1.9	0
318	Spectrum of driver mutations and clinical impact of circulating tumor DNA analysis in non–small cell lung cancer: Analysis of over 8000 cases. Cancer, 2020, 126, 3219-3228.	2.0	106
319	Certain ortho-hydroxylated brominated ethers are promiscuous kinase inhibitors that impair neuronal signaling and neurodevelopmental processes. Journal of Biological Chemistry, 2020, 295, 6120-6137.	1.6	7
320	†̃Kinesinopathies': emerging role of the kinesin family member genes in birth defects. Journal of Medical Genetics, 2020, 57, 797-807.	1.5	27
321	Sinus pericranii, skull defects, and structural brain anomalies in TRAF7 â€related disorder. Birth Defects Research, 2020, 112, 1085-1092.	0.8	5
322	RAS-targeted therapies: is the undruggable drugged?. Nature Reviews Drug Discovery, 2020, 19, 533-552.	21.5	569
323	ERK signalling: a master regulator of cell behaviour, life and fate. Nature Reviews Molecular Cell Biology, 2020, 21, 607-632.	16.1	535
324	Atypical cardiac defects in patients with RASopathies: Updated data on CARNET study. Birth Defects Research, 2020, 112, 725-731.	0.8	17
325	Selective Translation of Cell Fate Regulators Mediates Tolerance to Broad Oncogenic Stress. Cell Stem Cell, 2020, 27, 270-283.e7.	5.2	14
326	Swimming toward solutions: Using fish and frogs as models for understanding <scp>RASopathies</scp> . Birth Defects Research, 2020, 112, 749-765.	0.8	10
327	Compound craniosynostosis, intellectual disability, and Noonanâ€like facial dysmorphism associated with 7q32.3â€q35 deletion. Birth Defects Research, 2020, 112, 740-748.	0.8	4
328	A conserved, N-terminal tyrosine signal directs Ras for inhibition by Rabex-5. PLoS Genetics, 2020, 16, e1008715.	1.5	7
329	How Anionic Lipids Affect Spatiotemporal Properties of KRAS4B on Model Membranes. Journal of Physical Chemistry B, 2020, 124, 5434-5453.	1.2	18
330	Impairments in communication and social interaction in children with neurofibromatosis type 1: Characteristics and role of ADHD and language delay. Applied Neuropsychology: Child, 2022, 11, 220-225.	0.7	1
331	Introduction to the special issue on " <scp>RASopathies</scp> : Misregulation of signaling― Birth Defects Research, 2020, 112, 703-707.	0.8	0
332	Inference of Multisite Phosphorylation Rate Constants and Their Modulation by Pathogenic Mutations. Current Biology, 2020, 30, 877-882.e6.	1.8	14

#	Article	IF	CITATIONS
333	KRAS: Structure, function, and development of anticancer drugs. , 2020, , 359-389.		0
334	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
335	Cardiofaciocutaneous syndrome with KRAS gene mutation presenting as chylopericardium. American Journal of Medical Genetics, Part A, 2020, 182, 532-535.	0.7	3
336	Mitochondrial functions and rare diseases. Molecular Aspects of Medicine, 2020, 71, 100842.	2.7	39
337	RASopathies. Clinics in Dermatology, 2020, 38, 455-461.	0.8	22
338	The duality of human oncoproteins: drivers of cancer and congenital disorders. Nature Reviews Cancer, 2020, 20, 383-397.	12.8	44
339	Germline and sporadic cancers driven by the RAS pathway: parallelsÂandÂcontrasts. Annals of Oncology, 2020, 31, 873-883.	0.6	35
340	Autism Spectrum Disorder: Signaling Pathways and Prospective Therapeutic Targets. Cellular and Molecular Neurobiology, 2021, 41, 619-649.	1.7	36
341	Syndromic Autism Revisited: Review of the Literature and Lessons Learned. Pediatric Neurology, 2021, 114, 21-25.	1.0	21
342	<scp>RASopathies</scp> : A significant cause of polyhydramnios?. Prenatal Diagnosis, 2021, 41, 362-367.	1.1	8
343	De novo <scp>HRAS</scp> gene mutation associated with Costello syndrome identified by nonâ€invasive cellâ€free fetal <scp>DNA</scp> screening. Prenatal Diagnosis, 2021, 41, 11-14.	1.1	5
344	Activation of the MAPK pathway (RASopathies) and partial growth hormone insensitivity. Molecular and Cellular Endocrinology, 2021, 519, 111040.	1.6	15
345	Clinical and molecular spectra of BRAF-associated RASopathy. Journal of Human Genetics, 2021, 66, 389-399.	1.1	15
346	The RAL signaling network: Cancer and beyond. International Review of Cell and Molecular Biology, 2021, 361, 21-105.	1.6	11
347	Familial cardioâ€facioâ€eutaneous 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
348	Overlapping Molecular Pathways Leading to Autism Spectrum Disorders, Fragile X Syndrome, and Targeted Treatments. Neurotherapeutics, 2021, 18, 265-283.	2.1	15
349	Phenotypic Variability of an Inherited Pathogenic Variant in CIC Gene: A New Case Report in Two-Generation Family and Literature Review. Journal of Pediatric Neurology, 2021, 19, 193-201.	0.0	1
351	Autism Spectrum Disorder Symptom Profile Across the RASopathies. Frontiers in Psychiatry, 2020, 11, 585700.	1.3	9

#	Article	IF	Citations
352	Cutaneous Findings in Neurofibromatosis Type 1. Cancers, 2021, 13, 463.	1.7	18
353	Hypertrophic Cardiomyopathy in Infants from the Perspective of Cardiomyocyte Maturation. Korean Circulation Journal, 2021, 51, 733.	0.7	9
354	KRAS/BRAF mutations in brain arteriovenous malformations: A systematic review and meta-analysis. Interventional Neuroradiology, 2021, 27, 159101992098281.	0.7	14
355	Neurotransmitters, neuromodulators, synapses. , 2021, , 21-48.		0
356	Gene expression, regulation, and epigenetics in brain. , 2021, , 99-128.		0
357	Noonan Syndrome with Multiple Lentigines and <i>PTPN11</i> Mutation: A Case with Intracerebral Hemorrhage. Molecular Syndromology, 2021, 12, 1-7.	0.3	1
358	Mechanistic insights into the effect of phosphorylation on Ras conformational dynamics and its interactions with cell signaling proteins. Computational and Structural Biotechnology Journal, 2021, 19, 1184-1199.	1.9	51
360	RASopathies. , 2021, , 389-398.		0
361	Drug resistance in targeted cancer therapies with RAF inhibitors. , 2021, 4, 665-683.		9
362	Ras/ <scp>MAPK</scp> dysregulation in development causes a skeletal myopathy in an activating <scp><i>Braf</i>^{<i>L597V</i>}</scp> mouse model for cardioâ€facioâ€cutaneous syndrome. Developmental Dynamics, 2021, 250, 1074-1095.	0.8	6
363	Embryonic Expression of NrasG 12 D Leads to Embryonic Lethality and Cardiac Defects. Frontiers in Cell and Developmental Biology, 2021, 9, 633661.	1.8	4
364	Promotion of cancer cell stemness by Ras. Biochemical Society Transactions, 2021, 49, 467-476.	1.6	14
365	The Ins and Outs of RAS Effector Complexes. Biomolecules, 2021, 11, 236.	1.8	27
366	TBL1XR1 Ensures Balanced Neural Development Through NCOR Complex-Mediated Regulation of the MAPK Pathway. Frontiers in Cell and Developmental Biology, 2021, 9, 641410.	1.8	7
367	Manic and Depressive Symptoms in Children Diagnosed with Noonan Syndrome. Brain Sciences, 2021, 11, 233.	1.1	3
368	Hyperactive MEK1 Signaling in Cortical GABAergic Neurons Promotes Embryonic Parvalbumin Neuron Loss and Defects in Behavioral Inhibition. Cerebral Cortex, 2021, 31, 3064-3081.	1.6	10
369	First prenatal case of Noonan syndrome with SOS2 mutation: Implications of early diagnosis for genetic counseling. American Journal of Medical Genetics, Part A, 2021, 185, 1897-1902.	0.7	1
370	Prominent roles of ribosomal S6 kinase 4 (RSK4) in cancer. Pathology Research and Practice, 2021, 219, 153374.	1.0	14

# 371	ARTICLE Congenital polyvalvular disease expands the cardiac phenotype of the RASopathies. American Journal	IF 0.7	CITATIONS 3
372	Cutaneous Squamous Cell Carcinoma in the Age of Immunotherapy. Cancers, 2021, 13, 1148.	1.7	19
373	Clinical and Molecular Diagnosis of Beckwith-Wiedemann Syndrome with Single- or Multi-Locus Imprinting Disturbance. International Journal of Molecular Sciences, 2021, 22, 3445.	1.8	14
374	Myriad of pigmented lesions in a patient with Costello syndrome. Journal of the European Academy of Dermatology and Venereology, 2021, 35, e514-e516.	1.3	0
376	Drosophila RASopathy models identify disease subtype differences and biomarkers of drug efficacy. IScience, 2021, 24, 102306.	1.9	12
377	Molecular mechanisms underlying cellular effects of human MEK1 mutations. Molecular Biology of the Cell, 2021, 32, 974-983.	0.9	6
378	ERK1/2: An Integrator of Signals That Alters Cardiac Homeostasis and Growth. Biology, 2021, 10, 346.	1.3	17
379	Enlarged spinal nerve roots in RASopathies: Report of two cases. European Journal of Medical Genetics, 2021, 64, 104187.	0.7	2
380	MiR-140-5p targets Prox1 to regulate the proliferation and differentiation of neural stem cells through the ERK/MAPK signaling pathway. Annals of Translational Medicine, 2021, 9, 671-671.	0.7	13
382	Role of extracellular signal-regulated kinase 1/2 signaling underlying cardiac hypertrophy. Cardiology Journal, 2021, 28, 473-482.	0.5	12
383	Costello syndrome with special cutaneous manifestations and HRAS G12D mutation: A case report and literature review. Molecular Genetics & amp; Genomic Medicine, 2021, 9, e1690.	0.6	1
384	Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. Genetics in Medicine, 2021, 23, 1506-1513.	1.1	290
385	Juvenile xanthogranuloma in Noonan syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 3048-3052.	0.7	2
386	DNA Methylation in Babies Born to Nonsmoking Mothers Exposed to Secondhand Smoke during Pregnancy: An Epigenome-Wide Association Study. Environmental Health Perspectives, 2021, 129, 57010.	2.8	15
387	RAS mutations drive proliferative chronic myelomonocytic leukemia via a KMT2A-PLK1 axis. Nature Communications, 2021, 12, 2901.	5.8	44
388	Pediatric Rhabdomyosarcoma: Epidemiology and Genetic Susceptibility. Journal of Clinical Medicine, 2021, 10, 2028.	1.0	25
389	Hypertrophic Cardiomyopathy and Primary Restrictive Cardiomyopathy: Similarities, Differences and Phenocopies. Journal of Clinical Medicine, 2021, 10, 1954.	1.0	16
390	In vivo Functional Genomics for Undiagnosed Patients: The Impact of Small GTPases Signaling Dysregulation at Pan-Embryo Developmental Scale. Frontiers in Cell and Developmental Biology, 2021, 9, 642235.	1.8	3

#	Article	IF	CITATIONS
391	SOCS, SPRED, and NR4a: Negative regulators of cytokine signaling and transcription in immune tolerance. Proceedings of the Japan Academy Series B: Physical and Biological Sciences, 2021, 97, 277-291.	1.6	8
392	Clinical Interpretation Challenges of Germline-Shared Somatic Variants in Cancer. Laboratory Medicine, 2021, , .	0.8	0
393	RASopathies: From germline mutations to somatic and multigenic diseases. Biomedical Journal, 2021, 44, 422-432.	1.4	28
394	Elucidation of Abnormal Extracellular Regulated Kinase (ERK) Signaling and Associations with Syndromic and Non-syndromic Autism. Current Drug Targets, 2021, 22, 1071-1086.	1.0	5
396	MEK inhibition ameliorates social behavior phenotypes in a Spred1 knockout mouse model for RASopathy disorders. Molecular Autism, 2021, 12, 53.	2.6	7
397	Divergent Mechanisms Activating RAS and Small GTPases Through Post-translational Modification. Frontiers in Molecular Biosciences, 2021, 8, 707439.	1.6	13
398	Noonan syndromeâ€like phenotype in a patient with heterozygous <scp><i>ERF</i></scp> truncating variant. Congenital Anomalies (discontinued), 2021, 61, 226-230.	0.3	1
399	The Importance of Being PI3K in the RAS Signaling Network. Genes, 2021, 12, 1094.	1.0	28
400	Ubiquitin ligases in cancer: Functions and clinical potentials. Cell Chemical Biology, 2021, 28, 918-933.	2.5	36
401	Diagnostic difficulties and possibilities of NF1-like syndromes in childhood. BMC Pediatrics, 2021, 21, 331.	0.7	6
402	Ras isoform-specific expression, chromatin accessibility, and signaling. Biophysical Reviews, 2021, 13, 489-505.	1.5	14
403	Comprehensive Proteomics Profiling Reveals Circulating Biomarkers of Hypertrophic Cardiomyopathy. Circulation: Heart Failure, 2021, 14, e007849.	1.6	26
404	Ultra-rare renal diseases diagnosed with whole-exome sequencing: Utility in diagnosis and management. BMC Medical Genomics, 2021, 14, 177.	0.7	6
405	A Narrative Review of the Ocular Manifestations in Noonan Syndrome. Seminars in Ophthalmology, 2022, 37, 215-221.	0.8	7
406	Psychopathology and Adaptive Functioning in Children, Adolescents, and Young Adults with Noonan Syndrome. Journal of Developmental and Behavioral Pediatrics, 2021, Publish Ahead of Print, .	0.6	4
407	Case Report: Blepharophimosis and Ptosis as Leading Dysmorphic Features of Rare Congenital Malformation Syndrome With Developmental Delay – New Cases With TRAF7 Variants. Frontiers in Medicine, 2021, 8, 708717.	1.2	4
408	Multiple versus solitary giant cell lesions of the jaw: Similar or distinct entities?. Bone, 2021, 149, 115935.	1.4	10
409	Shedding light on developmental ERK signaling with genetically encoded biosensors. Development (Cambridge), 2021, 148, .	1.2	17

#	Δρτιςι ε	IF	CITATIONS
110	Clinical and Cytometric Study of Immune Involvement in a Heterogeneous Cohort of Subjects With		-
410	RASopathies and mTORopathies. Frontiers in Pediatrics, 2021, 9, 703613.	0.9	5
411	Epilepsy and BRAF Mutations: Phenotypes, Natural History and Genotype-Phenotype Correlations. Genes, 2021, 12, 1316.	1.0	13
412	Mitochondrial Fragmentation Triggers Ineffective Hematopoiesis in Myelodysplastic Syndromes. Cancer Discovery, 2022, 12, 250-269.	7.7	14
413	Late moyamoya-like angiopathy syndrome revealing MAP2K1 Noonan syndrome. Revue Neurologique, 2022, 178, 263-265.	0.6	0
414	The mediating role of ADHD symptoms between executive function and social skills in children with neurofibromatosis type 1. Child Neuropsychology, 2022, 28, 318-336.	0.8	2
415	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
416	See Spots? Look Deeper!. Pediatrics in Review, 2021, 42, 141-145.	0.2	0
417	Development and physiological functions of the lymphatic system: insights from human genetic studies of primary lymphedema. Physiological Reviews, 2021, 101, 1809-1871.	13.1	32
418	RASopathies: The musculoskeletal consequences and their etiology and pathogenesis. Bone, 2021, 152, 116060.	1.4	12
419	Emerging RAS-directed therapies for cancer. , 2021, 4, 543-558.		8
420	Germline Predisposition in AML. Hematologic Malignancies, 2021, , 55-70.	0.2	0
421	Compound heterozygosity for <scp>PTPN11</scp> variants in a subject with Noonan syndrome provides insights into the mechanism of <scp>SHP2</scp> â€related disorders. Clinical Genetics, 2021, 99, 457-461.	1.0	2
422	Genetic Aspects of Peripheral Nervous System Tumors. , 2021, , 331-345.		0
424	Genomic Applications in Inherited Genetic Disorders. , 2015, , 535-551.		1
425	Xanthohumol and Structurally Related Prenylflavonoids for Cancer Chemoprevention and Control. , 2020, , 319-350.		5
427	Clinical Disorders of Primary Malfunctioning of the Lymphatic System. Advances in Anatomy, Embryology and Cell Biology, 2014, 214, 187-204.	1.0	19
428	Posttranslational Modifications of Small G Proteins. , 2014, , 99-131.		5
429	Prenatal genetic considerations in congenital ventriculomegaly and hydrocephalus. Child's Nervous System, 2020, 36, 1645-1660.	0.6	23

#	Article	IF	CITATIONS
430	Intracellular Signaling Networks in Fragile X Syndrome: Approaches to Drug Discovery and Therapeutics. , 2017, , 217-239.		1
431	Repeating or spacing learning sessions are strategies for memory improvement with shared molecular and neuronal components. Neurobiology of Learning and Memory, 2020, 172, 107233.	1.0	2
432	A Novel Locus and Candidate Gene for Familial Developmental Dyslexia on Chromosome 4q. Zeitschrift FÜr Kinder- Und Jugendpsychiatrie Und Psychotherapie, 2020, 48, 478-489.	0.4	5
433	The molecular functions of RIT1 and its contribution to human disease. Biochemical Journal, 2020, 477, 2755-2770.	1.7	11
434	Genetic heterogeneity of pediatric systemic lupus erythematosus with lymphoproliferation. Medicine (United States), 2020, 99, e20232.	0.4	13
437	Neurocutaneous Disorders. CONTINUUM Lifelong Learning in Neurology, 2018, 24, 96-129.	0.4	19
438	Exploiting codon usage identifies intensity-specific modifiers of Ras/MAPK signaling in vivo. PLoS Genetics, 2020, 16, e1009228.	1.5	7
439	Neurodevelopmental Aspects of RASopathies. Molecules and Cells, 2019, 42, 441-447.	1.0	47
440	Chondroblastoma-like mass of the temporal bone, secondary aneurysmal bone cyst, and intracerebral hemorrhage in a patient with cardiofaciocutaneous syndrome: case report. Journal of Neurosurgery: Pediatrics, 2019, 24, 153-158.	0.8	1
441	From microcephaly to megalencephaly: determinants of brain size. Dialogues in Clinical Neuroscience, 2018, 20, 267-282.	1.8	61
442	Risk of autoimmune diseases in patients with RASopathies: systematic study of humoral and cellular immunity. Orphanet Journal of Rare Diseases, 2021, 16, 410.	1.2	11
444	Ophthalmic manifestations in Costello syndrome caused by Ras pathway dysregulation during development. Ophthalmic Genetics, 2022, 43, 48-57.	0.5	4
445	Oncogenic <i>RAS</i> instructs morphological transformation of human epithelia via differential tissue mechanics. Science Advances, 2021, 7, eabg6467.	4.7	18
446	DA-Raf and the MEK inhibitor trametinib reverse skeletal myocyte differentiation inhibition or muscle atrophy caused by myostatin and GDF11 through the non-Smad Ras–ERK pathway. Journal of Biochemistry, 2022, 171, 109-122.	0.9	5
447	SPRED2 loss-of-function causes a recessive Noonan syndrome-like phenotype. American Journal of Human Genetics, 2021, 108, 2112-2129.	2.6	23
448	ERK/MAPK signalling in the developing brain: Perturbations and consequences. Neuroscience and Biobehavioral Reviews, 2021, 131, 792-805.	2.9	46
450	Mouse Models of RAS-Induced Tumors and Developmental Disorders. , 2014, , 211-231.		0
451-	PAS Cones and Cancer 2014 157 171		0 -

Article	IF	CITATIONS
General aspects of aetiology, diagnostics and therapy. , 2016, , 3-104.		0
Mitogen-Activated Protein Kinases 2016 1-4		0
		Ŭ
ERK1 and ERK2., 2016,, 1-9.		0
Overview of Molecular Pathology of Bone and Soft Tissue Tumors. , 2016, , 8-290-8-295.		Ο
Reconstructing ERK Signaling in the Drosophila Embryo from Fixed Images. Methods in Molecular Biology, 2017, 1487, 337-351.	0.4	0
Congenital Nevi. , 2018, , 17-33.		0
ERK1 and ERK2. , 2018, , 1624-1632.		0
Mitogen-Activated Protein Kinases. , 2018, , 3138-3141.		1
Genetic testing for lymphedema in RASopathies. The EuroBiotech Journal, 2018, 2, 10-12.	0.5	0
Genomic Applications in Inherited Genetic Disorders. , 2019, , 543-560.		0
Cardiofaciocutaneus syndrome: literature review and case report. Nervno-Myshechnye Bolezni, 2019, 8, 49-53.	0.2	0
Jaw and Bones of theÂHead and Face. , 2019, , 153-191.		0
Cuidado de niños con neurofibromatosis de tipo 1. Nursing (Ed Española), 2019, 36, 26-31.	0.0	0
Pediatric Myelodysplastic Syndrome With Germline RRAS Mutation: Expanding the Phenotype of RASopathies. Journal of Pediatric Hematology/Oncology, 2021, 43, e517-e520.	0.3	2
Translating the Role of mTOR- and RAS-Associated Signalopathies in Autism Spectrum Disorder: Models, Mechanisms and Treatment. Genes, 2021, 12, 1746.	1.0	4
Recombinant GH treatment in a case of Costello syndrome with a 5-year follow-up. Clinical Pediatric Endocrinology, 2020, 29, 195-199.	0.4	1
Cancer Risk and Spectrum in Individuals with RASopathies. , 2020, , 249-260.		0

482A Pathogenic Homozygous Mutation in The Pleckstrin Homology Domain of RASA1 Is Responsible for
Familial Tricuspid Atresia in An Iranian Consanguineous Family. Cell Journal, 2019, 21, 70-77.0.25

#

#	Article	IF	Citations
485	Standardized phenotype documentation, documentation of genotype phenotype correlations. , 2022, , 133-154.		0
486	Induced pluripotent stem cells for modeling Noonan, Noonan Syndrome with Multiple Lentigines, and Costello Syndromes. , 2022, , 65-110.		0
487	Novel Methylation Biomarkers for Colorectal Cancer Prognosis. Biomolecules, 2021, 11, 1722.	1.8	21
488	A brief history of RAS and the RAS Initiative. Advances in Cancer Research, 2022, 153, 1-27.	1.9	6
489	How can same-gene mutations promote both cancer and developmental disorders?. Science Advances, 2022, 8, eabm2059.	4.7	29
490	Cardiomyopathies in Children and Systemic Disorders When Is It Useful to Look beyond the Heart?. Journal of Cardiovascular Development and Disease, 2022, 9, 47.	0.8	5
491	Endosomal trafficking defects alter neural progenitor proliferation and cause microcephaly. Nature Communications, 2022, 13, 16.	5.8	15
492	Management of failed Chiari decompression and intrasyringeal hemorrhage in Noonan syndrome: illustrative cases. Journal of Neurosurgery Case Lessons, 2022, 3, .	0.1	0
493	Cardiofaciocutaneous syndrome – a longitudinal study of a case over 33 years: case report and review of the literature. Romanian Journal of Morphology and Embryology, 2022, 62, 563-568.	0.4	5
496	Delineating the autistic phenotype in children with neurofibromatosis type 1. Molecular Autism, 2022, 13, 3.	2.6	8
498	Social behavior in RASopathies and idiopathic autism. Journal of Neurodevelopmental Disorders, 2022, 14, 5.	1.5	1
499	Lymphatic Anomalies in Children: Update on Imaging Diagnosis, Genetics, and Treatment. American Journal of Roentgenology, 2022, 218, 1089-1101.	1.0	8
500	Targeting RAS oncogenesis with SOS1 inhibitors. Advances in Cancer Research, 2022, 153, 169-203.	1.9	13
501	Endothelial k-RasV12 Expression Induces Capillary Deficiency Attributable to Marked Tube Network Expansion Coupled to Reduced Pericytes and Basement Membranes. Arteriosclerosis, Thrombosis, and Vascular Biology, 2022, 42, 205-222.	1.1	11
502	Defining RASopathy. DMM Disease Models and Mechanisms, 2022, 15, .	1.2	26
503	Malformations of Cortical Development. , 2021, , 1-237.		1
504	RAS pathway regulation in melanoma. DMM Disease Models and Mechanisms, 2022, 15, .	1.2	11
505	An Assessment of the Therapeutic Landscape for the Treatment of Heart Disease in the RASopathies. Cardiovascular Drugs and Therapy, 2022, , 1.	1.3	3

#	Article	IF	CITATIONS
506	The RASopathies: from pathogenetics to therapeutics. DMM Disease Models and Mechanisms, 2022, 15, .	1.2	41
507	Giant Cell Lesions of the Jaws Involving RASopathy Syndromes. Acta Stomatologica Croatica, 2022, 56, 77-88.	0.4	2
508	Classification of <i>KRAS</i> -Activating Mutations and the Implications for Therapeutic Intervention. Cancer Discovery, 2022, 12, 913-923.	7.7	32
509	Extending the prenatal Noonan's phenotype by review of ultrasound and autopsy data. Prenatal Diagnosis, 2022, 42, 574-582.	1.1	4
511	Genomic and Epigenomic Landscape of Juvenile Myelomonocytic Leukemia. Cancers, 2022, 14, 1335.	1.7	5
512	Challenges in the diagnosis of neurofibromatosis type 1 (NF1) in young children facilitated by means of revised diagnostic criteria including genetic testing for pathogenic NF1 gene variants. Human Genetics, 2022, 141, 177-191.	1.8	29
513	RASopathies: Dermatologists' viewpoints. Indian Journal of Dermatology, Venereology and Leprology, 2021, 88, 452-463.	0.2	1
514	IQ-Switch is a QF-based innocuous, silencing-free, and inducible gene switch system in zebrafish. Communications Biology, 2021, 4, 1405.	2.0	3
516	A severe clinical phenotype of Noonan syndrome concomitant with incomplete Cantrell syndrome. Pediatrics International, 2022, 64, e15139.	0.2	0
517	Mitochondria and the future of RASopathies: the emergence of bioenergetics. Journal of Clinical Investigation, 2022, 132, 1-5.	3.9	14
533	Cancer Predisposition Genes in Adolescents and Young Adults (AYAs): a Review Paper from the Italian AYA Working Group. Current Oncology Reports, 2022, 24, 843-860.	1.8	6
534	Cross-species analysis of LZTR1 loss-of-function mutants demonstrates dependency to RIT1 orthologs. ELife, 2022, 11, .	2.8	8
535	An early diagnostic case of noonan syndrome with leukocytosis and pulmonary stenosis. Medicine, Case Reports and Study Protocols, 2022, 3, e0226.	0.0	0
536	Discovery of Raf Family Is a Milestone in Deciphering the Ras-Mediated Intracellular Signaling Pathway. International Journal of Molecular Sciences, 2022, 23, 5158.	1.8	16
537	Neurologic and neurodevelopmental complications in cardiofaciocutaneous syndrome are associated with genotype: A multinational cohort study. Genetics in Medicine, 2022, 24, 1556-1566.	1.1	15
538	Inflammatory response in hematopoietic stem and progenitor cells triggered by activating SHP2 mutations evokes blood defects. ELife, 2022, 11, .	2.8	9
539	Prenatal case of RIT1 mutation associated Noonan syndrome by whole exome sequencing (WES) and review of the literature. Taiwanese Journal of Obstetrics and Gynecology, 2022, 61, 535-538.	0.5	3
540	Epilepsy in a cohort of children with Noonan syndrome and related disorders. European Journal of Pediatrics, 2022, 181, 2919-2926.	1.3	4

#	Article	IF	CITATIONS
541	Surgical treatment of obstructive hypertrophic cardiomyopathy in children with Noonan syndrome. Russian Pediatric Journal, 2022, 25, 96-105.	0.0	1
542	Neurodevelopmental disorders, immunity, and cancer are connected. IScience, 2022, 25, 104492.	1.9	10
543	The treatment of advanced non-small cell lung cancer harboring KRAS mutation: a new class of drugs for an old target—a narrative review. Translational Lung Cancer Research, 2021, .	1.3	4
544	Optogenetic actuator – ERK biosensor circuits identify MAPK network nodes that shape ERKÂdynamics. Molecular Systems Biology, 2022, 18, .	3.2	27
546	Perioperative anaphylaxis to fibrin sealants in children with Noonan syndrome. Annals of Allergy, Asthma and Immunology, 2022, 129, 11-12.	0.5	0
548	Cognitive, Behavioural, Speech, Language and Developmental Outcomes Associated with Pathogenic Variants in the ERF Gene. Journal of Craniofacial Surgery, 2022, 33, 1847-1852.	0.3	1
549	The genetic heterogeneity and drug resistance mechanisms of relapsed refractory multiple myeloma. Nature Communications, 2022, 13, .	5.8	22
550	Fibrous dysplasia in cardioâ€facio utaneous syndrome: A case report and review of literature. American Journal of Medical Genetics, Part A, 0, , .	0.7	1
551	Exome sequencing in a child with neurodevelopmental disorder and epilepsy: Variant analysis of the <scp>AHNAK2</scp> gene. Molecular Genetics & Genomic Medicine, 0, , .	0.6	1
552	Molecular genetic mechanisms of congenital heart disease. Current Opinion in Genetics and Development, 2022, 75, 101949.	1.5	10
554	Autism Symptoms in Children and Young Adults With Fragile X Syndrome, Angelman Syndrome, Tuberous Sclerosis Complex, and Neurofibromatosis Type 1: A Cross-Syndrome Comparison. Frontiers in Psychiatry, 0, 13, .	1.3	3
555	Molecular and clinical profile of patients referred as Noonan or Noonan-like syndrome in Greece: a cohort of 86 patients. European Journal of Pediatrics, 2022, 181, 3691-3700.	1.3	1
556	Clinical and pathological characteristics of familial melanoma with germline <scp><i>TERT</i></scp> promoter variants. Pigment Cell and Melanoma Research, 2022, 35, 573-586.	1.5	2
558	Ca2+ homeostasis maintained by TMCO1 underlies corpus callosum development via ERK signaling. Cell Death and Disease, 2022, 13, .	2.7	2
560	RASopathies due to de novo pathogenic variants: clinical features, genetic findings and outcomes in nine neonates born with congenital heart defects. BMC Medical Genomics, 2022, 15, .	0.7	1
562	Mechanistic Insights into the Long-range Allosteric Regulation of KRAS Via Neurofibromatosis Type 1 (NF1) Scaffold Upon SPRED1 Loading. Journal of Molecular Biology, 2022, 434, 167730.	2.0	17
563	Genetics of FASD: Confounding Rare Craniofacial and Neurodevelopmental Disorders May Identify Ethanol-Sensitizing Genetic Variants of FASD. Neuromethods, 2022, , 77-117.	0.2	1
564	The MAPK Signaling Cascades. , 2022, , .		0

#	Article	IF	CITATIONS
565	Case Report: Progressive central conducting lymphatic abnormalities in the RASopathies. Two case reports, including successful treatment by MEK inhibition. Frontiers in Genetics, 0, 13, .	1.1	9
566	Cancer ausing <i>MAP2K1</i> mutation in a mosaic patient with cardioâ€facioâ€cutaneous syndrome and immunodeficiency. Human Mutation, 2022, 43, 1852-1855.	1.1	1
567	Syndrome-Specific Neuroanatomical Phenotypes in Girls With Turner and Noonan Syndromes. Biological Psychiatry: Cognitive Neuroscience and Neuroimaging, 2024, 9, 146-155.	1.1	1
568	Poor prognosis, hypomethylation, and immune infiltrates are associated with downregulation of INMT in head and neck squamous cell carcinoma. Frontiers in Genetics, 0, 13, .	1.1	0
569	Bi-allelic loss-of-function variants in TMEM147 cause moderate to profound intellectual disability with facial dysmorphism and pseudo-Pelger-Huët anomaly. American Journal of Human Genetics, 2022, 109, 1909-1922.	2.6	5
570	Succesful MEK-inhibition of severe hypertrophic cardiomyopathy in RIT1-related Noonan Syndrome. European Journal of Medical Genetics, 2022, 65, 104630.	0.7	8
571	Prediction of Major Adverse Cardiovascular Events in Patients With Hypertrophic Cardiomyopathy Using Proteomics Profiling. Circulation Genomic and Precision Medicine, 0, , .	1.6	0
572	RASopathy Cohort of Patients Enrolled in a Brazilian Reference Center for Rare Diseases: A Novel Familial LZTR1 Variant and Recurrent Mutations. The Application of Clinical Genetics, 0, Volume 15, 153-170.	1.4	2
573	Genotype/Phenotype Correlation of Cases with <i>PTPN11</i> Gene Mutation: Eastern Black Sea Experience. Journal of Ankara University Faculty of Medicine, 2022, 75, 368-372.	0.0	0
574	Effects of Noonan Syndrome-Germline Mutations on Mitochondria and Energy Metabolism. Cells, 2022, 11, 3099.	1.8	5
575	Neurofibromatosis Type 1 and Hypospadias in a Male 46, XY with a Mutation in the NF1 Gene and a Mutation in NR5A1. Pharmacogenomics and Personalized Medicine, 0, Volume 15, 873-878.	0.4	0
576	Case report: The cardio-facio-cutaneous syndrome due to a novel germline mutation in MAP2K1: A multifaceted disease with immunodeficiency and short stature. Frontiers in Pediatrics, 0, 10, .	0.9	3
577	Obstetrical and neonatal outcomes of cardioâ€facioâ€cutaneous syndrome: Prenatal consequences of Ras/ <scp>MAPK</scp> dysregulation. American Journal of Medical Genetics, Part A, O, , .	0.7	0
578	Genetic conditions of short stature: A review of three classic examples. Frontiers in Endocrinology, 0, 13, .	1.5	5
579	Shoc2 controls ERK1/2-driven neural crest development by balancing components of the extracellular matrix. Developmental Biology, 2022, 492, 156-171.	0.9	1
580	Hydrocephalus and genetic disorders. , 2023, , 99-116.		0
581	The complex, dynamic SpliceOme of the small GTPase transcripts altered by technique, sex, genetics, tissue specificity, and RNA base editing. Frontiers in Cell and Developmental Biology, 0, 10, .	1.8	2
582	RASopathy mutations provide functional insight into the BRAF cysteine-rich domain and reveal the importance of autoinhibition in BRAF regulation. Molecular Cell, 2022, 82, 4262-4276.e5.	4.5	8

#	Article	IF	CITATIONS
584	Germline selection of <i>PTPN11</i> (HGNC:9644) variants make a major contribution to both Noonan syndrome's high birth rate and the transmission of sporadic cancer variants resulting in fetal abnormality. Human Mutation, 2022, 43, 2205-2221.	1.1	4
585	The molecular genetics of <scp>RASopathies</scp> : An update on novel disease genes and new disorders. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2022, 190, 425-439.	0.7	11
586	Endocrine system involvement in patients with RASopathies: A case series. Frontiers in Endocrinology, 0, 13, .	1.5	2
587	Binding of active Ras and its mutants to the Ras binding domain of PI-3-kinase: A quantitative approach to KD measurements. Analytical Biochemistry, 2023, 663, 115019.	1.1	1
588	Infantile epileptic spasms syndrome in children with cardiofaciocutanous syndrome: Clinical presentation and associations with genotype. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2022, 190, 501-509.	0.7	3
589	The heart in <scp>RASopathies</scp> . American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2022, 190, 440-451.	0.7	7
590	Neuropsychological features in RASopathies: A pilot study on parent training program involving families of children with Noonan syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 0, , .	0.7	0
591	Vascular malformation rupture in a patient affected by Costello syndrome. BMJ Case Reports, 2022, 15, e250948.	0.2	0
592	Management of nutritional and gastrointestinal issues in <scp>RASopathies</scp> : A narrative review. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 0, , .	0.7	5
593	Predisposing Genetic Variants and Potent Genetic Mutations in Cancer. , 2023, , 1-25.		0
594	Neurofibromatosis-1 microdeletiós szindróma Orvosi Hetilap, 2022, 163, 2041-2051.	0.1	0
595	The frequency of somatic mutations in cancer predicts the phenotypic relevance of germline mutations. Frontiers in Genetics, 0, 13, .	1.1	1
596	Craniofacial findings in syndromes associated with cafe-au-lait spots: a literature review. Revista Da Associação Médica Brasileira, 0, , .	0.3	0
597	Autosomal recessive Noonan-like syndrome caused by homozygosity for a previously unreported variant in SPRED2. European Journal of Medical Genetics, 2023, 66, 104695.	0.7	2
598	Stochastic phenotypes in RAS-dependent developmental diseases. Current Biology, 2023, 33, 807-816.e4.	1.8	3
599	FMRP activity and control of Csw/SHP2 translation regulate MAPK-dependent synaptic transmission. PLoS Biology, 2023, 21, e3001969.	2.6	3
600	Destabilizing NF1 variants act in a dominant negative manner through neurofibromin dimerization. Proceedings of the National Academy of Sciences of the United States of America, 2023, 120, .	3.3	3
601	Engineering Small Molecule Switches of Protein Function in Zebrafish Embryos. Journal of the American Chemical Society, 2023, 145, 2395-2403.	6.6	8

#	Article	IF	CITATIONS
602	Cancer risk in adult congenital heart disease. International Journal of Cardiology Congenital Heart Disease, 2023, 12, 100441.	0.2	0
603	Precision diagnostics in children. , 2023, 1, .		Ο
604	Prenatal diagnosis of euploid increased nuchal translucency on fetal ultrasound (II): RASopathy disorders – Prenatal ultrasound findings and genotype–phenotype correlations. Journal of Medical Ultrasound, 2023, 31, 13.	0.2	0
605	Kinase Inhibitors in Genetic Diseases. International Journal of Molecular Sciences, 2023, 24, 5276.	1.8	0
606	Case report: Gastroenterological management in a case of cardio-facio-cutaneous syndrome. Frontiers in Pediatrics, 0, 11, .	0.9	0
607	RAS pathway: The new frontier of brain mosaicism in epilepsy. Neurobiology of Disease, 2023, 180, 106074.	2.1	3
608	Neurofibromatosis Type 1: Pediatric Aspects and Review of Genotype–Phenotype Correlations. Cancers, 2023, 15, 1217.	1.7	5
609	Presence of neurologic signs in children with neurofibromatosis type 1. Minerva Pediatrics, 2023, 75, .	0.2	1
611	Case report: Revascularization failure in NF1-related moyamoya syndrome after selumetinib: A possible pathophysiological correlation?. Frontiers in Pediatrics, 0, 11, .	0.9	2
612	Cellular senescence and developmental defects. FEBS Journal, 2023, 290, 1303-1313.	2.2	5
614	Cancer in Costello syndrome: a systematic review and meta-analysis. British Journal of Cancer, 0, , .	2.9	0
615	Neurodevelopmental disorders, like cancer, are connected to impaired chromatin remodelers, PI3K/mTOR, and PAK1-regulated MAPK. Biophysical Reviews, 2023, 15, 163-181.	1.5	14
616	Connecting developmental defects and evolutionary findings. , 2023, , 327-357.		0
617	Target Hyperactive ERK Signaling for Cancer Therapy. , 2023, , 1-39.		0
618	BRAF-mediated brain tumors in adults and children: A review and the Australian and New Zealand experience. Frontiers in Oncology, 0, 13, .	1.3	1
619	<scp>RAF1</scp> deficiency causes a lethal syndrome that underscores <scp>RTK</scp> signaling during embryogenesis. EMBO Molecular Medicine, 0, , .	3.3	3
620	Setting sail: Maneuvering SHP2 activity and its effects in cancer. Advances in Cancer Research, 2023, , .	1.9	0
622	A rare mutation in a patient with Noonan syndrome with multiple lentigines. JAAD Case Reports, 2023, 36, 96-98.	0.4	0

#	Article	IF	CITATIONS
663	Common Monogenetic Conditions in Newborns. , 2024, , 690-698.		0