Karen Gripp

List of Publications by Year
in descending order

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255 papers	16,607 citations	25034 57 h-index	19749 117 g-index
267 all docs	267 docs citations	267 times ranked	18230 citing authors

#	Article	IF	Citations
1	Mutations in PTPN11, encoding the protein tyrosine phosphatase SHP-2, cause Noonan syndrome. Nature Genetics, 2001, 29, 465-468.	21.4	1,555
2	Somatic mutations in PTPN11 in juvenile myelomonocytic leukemia, myelodysplastic syndromes and acute myeloid leukemia. Nature Genetics, 2003, 34, 148-150.	21.4	960
3	PTPN11 Mutations in Noonan Syndrome: Molecular Spectrum, Genotype-Phenotype Correlation, and Phenotypic Heterogeneity. American Journal of Human Genetics, 2002, 70, 1555-1563.	6.2	680
4	Patient-specific induced pluripotent stem-cell-derived models of LEOPARD syndrome. Nature, 2010, 465, 808-812.	27.8	672
5	Noonan syndrome. Lancet, The, 2013, 381, 333-342.	13.7	608
6	Hereditary hemorrhagic telangiectasia: genetics and molecular diagnostics in a new era. Frontiers in Genetics, 2015, 6, 1.	2.3	489
7	Identifying facial phenotypes of genetic disorders using deep learning. Nature Medicine, 2019, 25, 60-64.	30.7	449
8	Mutations in TGIF cause holoprosencephaly and link NODAL signalling to human neural axis determination. Nature Genetics, 2000, 25, 205-208.	21.4	368
9	Diversity and Functional Consequences of Germline and Somatic PTPN11 Mutations in Human Disease. American Journal of Human Genetics, 2006, 78, 279-290.	6.2	352
10	Noonan syndrome and clinically related disorders. Best Practice and Research in Clinical Endocrinology and Metabolism, 2011, 25, 161-179.	4.7	303
11	Ribosome Levels Selectively Regulate Translation and Lineage Commitment in Human Hematopoiesis. Cell, 2018, 173, 90-103.e19.	28.9	296
12	Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. Genetics in Medicine, 2021, 23, 1506-1513.	2.4	290
13	Genetic evidence for lineage-related and differentiation stage-related contribution of somatic PTPN11 mutations to leukemogenesis in childhood acute leukemia. Blood, 2004, 104, 307-313.	1.4	265
14	Impaired PIEZO1 function in patients with a novel autosomal recessive congenital lymphatic dysplasia. Nature Communications, 2015, 6, 8329.	12.8	239
15	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	27.0	205
16	Mutations in KCNH1 and ATP6V1B2 cause Zimmermann-Laband syndrome. Nature Genetics, 2015, 47, 661-667.	21.4	177
17	Tumor predisposition in Costello syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2005, 137C, 72-77.	1.6	169
18	Disorders of dysregulated signal traffic through the RASâ€MAPK pathway: phenotypic spectrum and molecular mechanisms. Annals of the New York Academy of Sciences, 2010, 1214, 99-121.	3.8	167

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19	HRAS mutation analysis in Costello syndrome: Genotype and phenotype correlation. American Journal of Medical Genetics, Part A, 2006, 140A, 1-7.	1.2	164
20	Double Inactivation of NF1 in Tibial Pseudarthrosis. American Journal of Human Genetics, 2006, 79, 143-148.	6.2	145
21	Genotype-Phenotype Correlation in NF1: Evidence for a More Severe Phenotype Associated with Missense Mutations Affecting NF1 Codons 844â€“848. American Journal of Human Genetics, 2018, 102, 69-87.	6.2	144
22	High Incidence of Noonan Syndrome Features Including Short Stature and Pulmonic Stenosis in Patients carrying NF1 Missense Mutations Affecting p.Arg1809: Genotypeâ€“Phenotype Correlation. Human Mutation, 2015, 36, 1052-1063.	2.5	143
23	Five additional Costello syndrome patients with rhabdomyosarcoma: Proposal for a tumor screening protocol. American Journal of Medical Genetics Part A, 2002, 108, 80-87.	2.4	138
24	Costello syndrome: a Ras/mitogen activated protein kinase pathway syndrome (rasopathy) resulting from HRAS germline mutations. Genetics in Medicine, 2012, 14, 285-292.	2.4	135
25	PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. JCI Insight, 2016, 1, .	5.0	134
26	De novo truncating mutations in ASXL3 are associated with a novel clinical phenotype with similarities to Bohring-Opitz syndrome. Genome Medicine, 2013, 5, 11.	8.2	128
27	ClinGenâ€™s RASopathy Expert Panel consensus methods for variant interpretation. Genetics in Medicine, 2018, 20, 1334-1345.	2.4	126
28	Diversity, parental germline origin, and phenotypic spectrum of de novoHRASmissense changes in Costello syndrome. Human Mutation, 2007, 28, 265-272.	2.5	123
29	Bone Mineral Density in Children and Adolescents with Neurofibromatosis Type 1. Journal of Pediatrics, 2007, 150, 83-88.	1.8	122
30	Diamondâ€“Blackfan anemia with mandibulofacial dystostosis is heterogeneous, including the novel DBA genes TSR2 and RPS28. American Journal of Medical Genetics, Part A, 2014, 164, 2240-2249.	1.2	121
31	A novel rasopathy caused by recurrent de novo missense mutations in PPP1CB closely resembles Noonan syndrome with loose anagen hair. American Journal of Medical Genetics, Part A, 2016, 170, 2237-2247.	1.2	117
32	Descriptive analysis of tibial pseudarthrosis in patients with neurofibromatosis 1. , 1999, 84, 413-419.		114
33	Activating mutations in RRAS underlie a phenotype within the RASopathy spectrum and contribute to leukaemogenesis. Human Molecular Genetics, 2014, 23, 4315-4327.	2.9	114
34	Germline gain-of-function mutations in AFF4 cause a developmental syndrome functionally linking the super elongation complex and cohesin. Nature Genetics, 2015, 47, 338-344.	21.4	109
35	Decreased Elastin Deposition and High Proliferation of Fibroblasts from Costello Syndrome Are Related to Functional Deficiency in the 67-kD Elastin-Binding Protein. American Journal of Human Genetics, 2000, 66, 859-872.	6.2	108
36	SHP-2 and myeloid malignancies. Current Opinion in Hematology, 2004, 11, 44-50.	2.5	106

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37	Familial Predisposition to Developmental Dysplasia of the Hip. Journal of Pediatric Orthopaedics, 2009, 29, 463-466.	1.2	105
38	Further delineation of cardiac abnormalities in Costello syndrome. American Journal of Medical Genetics Part A, 2002, 111, 115-129.	2.4	104
39	Unexpected death and critical illness in Praderâ€“Willi syndrome: Report of ten individuals. American Journal of Medical Genetics Part A, 2004, 124A, 158-164.	2.4	102
40	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. Brain, 2017, 140, 2610-2622.	7.6	102
41	Clinical, pathological, and molecular analyses of cardiovascular abnormalities in Costello syndrome: A Ras/MAPK pathway syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 486-507.	1.2	99
42	Gastric Rupture and Necrosis in Praderâ€Willi Syndrome. Journal of Pediatric Gastroenterology and Nutrition, 2007, 45, 272-274.	1.8	93
43	High incidence of progressive postnatal cerebellar enlargement in Costello syndrome: Brain overgrowth associated with HRAS mutations as the likely cause of structural brain and spinal cord abnormalities. American Journal of Medical Genetics, Part A, 2010, 152A, 1161-1168.	1.2	89
44	Mutations in the humanTWIST gene. Human Mutation, 2000, 15, 150-155.	2.5	86
45	RASA1 somatic mutation and variable expressivity in capillary malformation/arteriovenous malformation (CM/AVM) syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 1450-1454.	1.2	85
46	Somatic mosaicism for anHRAS mutation causes Costello syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 2163-2169.	1.2	79
47	Further delineation of the phenotype resulting fromBRAForMEK1germline mutations helps differentiate cardio-facio-cutaneous syndrome from Costello syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 1472-1480.	1.2	79
48	Myeloid Dysregulation in a Human Induced Pluripotent Stem Cell Model of PTPN11 -Associated Juvenile Myelomonocytic Leukemia. Cell Reports, 2015, 13, 504-515.	6.4	79
49	A Restricted Spectrum of Mutations in the SMAD4 Tumor-Suppressor Gene Underlies Myhre Syndrome. American Journal of Human Genetics, 2012, 90, 161-169.	6.2	77
50	Paternal Germline Origin and Sex-Ratio Distortion in Transmission of PTPN11 Mutations in Noonan Syndrome. American Journal of Human Genetics, 2004, 75, 492-497.	6.2	76
51	Mutations in KCNK4 that Affect Gating Cause a Recognizable Neurodevelopmental Syndrome. American Journal of Human Genetics, 2018, 103, 621-630.	6.2	73
52	GestaltMatcher facilitates rare disease matching using facial phenotype descriptors. Nature Genetics, 2022, 54, 349-357.	21.4	73
53	Phenotype of the fibroblast growth factor receptor 2 Ser351Cys mutation: Pfeiffer syndrome type III. American Journal of Medical Genetics Part A, 1998, 78, 356-360.	2.4	72
54	Analysis of skeletal dysplasias in the Utah population. American Journal of Medical Genetics, Part A, 2012, 158A, 1046-1054.	1.2	72

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55	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	6.2	71
56	Costello syndrome: Clinical phenotype, genotype, and management guidelines. American Journal of Medical Genetics, Part A, 2019, 179, 1725-1744.	1.2	70
57	Maternal uniparental disomy of chromosome 20: a novel imprinting disorder of growth failure. Genetics in Medicine, 2016, 18, 309-315.	2.4	69
58	Assessing the geneâ€“disease association of 19 genes with the RASopathies using the ClinGen gene curation framework. Human Mutation, 2018, 39, 1485-1493.	2.5	66
59	Maleâ€toâ€male transmission of Costello syndrome: G12S HRAS germline mutation inherited from a father with somatic mosaicism. American Journal of Medical Genetics, Part A, 2009, 149A, 315-321.	1.2	62
60	Contribution of malformations and genetic disorders to mortality in a children's hospital. American Journal of Medical Genetics Part A, 2004, 126A, 393-397.	2.4	61
61	Costello syndrome associated with novel germline HRAS mutations: An attenuated phenotype?. American Journal of Medical Genetics, Part A, 2008, 146A, 683-690.	1.2	61
62	Truncating mutations in the last exon of NOTCH3 cause lateral meningocele syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 271-281.	1.2	59
63	KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. Brain, 2020, 143, 3242-3261.	7.6	57
64	Deaths due to choking in Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 484-487.	1.2	56
65	Longâ€term survival in TARP syndrome and confirmation of RBM10 as the diseaseâ€causing gene. American Journal of Medical Genetics, Part A, 2011, 155, 2516-2520.	1.2	56
66	The musculoskeletal phenotype of the RASopathies. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2011, 157, 90-103.	1.6	56
67	The Cyclic AMP Pathway Is a Sex-Specific Modifier of Glioma Risk in Type I Neurofibromatosis Patients. Cancer Research, 2015, 75, 16-21.	0.9	56
68	Phenotypic analysis of individuals with Costello syndrome due to HRAS p.G13C. , 2011, 155, 706-716.		55
69	Approaches to Treating NF1 Tibial Pseudarthrosis. Journal of Pediatric Orthopaedics, 2013, 33, 269-275.	1.2	55
70	Nephroblastomatosis or Wilms tumor in a fourth patient with a somatic PIK3CA mutation. American Journal of Medical Genetics, Part A, 2016, 170, 2559-2569.	1.2	55
71	Asfotase-Î± improves bone growth, mineralization and strength in mouse models of neurofibromatosis type-1. Nature Medicine, 2014, 20, 904-910.	30.7	54
72	Lateral meningocele syndrome: Three new patients and review of the literature. American Journal of Medical Genetics Part A, 1997, 70, 229-239.	2.4	52

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73	Second case of bladder carcinoma in a patient with Costello syndrome. , 2000, 90, 256-259.		52
74	Prenatal features of Costello syndrome: ultrasonographic findings and atrial tachycardia. Prenatal Diagnosis, 2009, 29, 682-690.	2.3	52
75	Cardiac anomalies in Axenfeldâ€“Rieger syndrome due to a novel FOXC1 mutation. American Journal of Medical Genetics, Part A, 2013, 161, 114-119.	1.2	52
76	Contributing factors of mortality in Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 196-205.	1.2	50
77	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	6.2	48
78	Mandibulofacial Dysostosis with Microcephaly: Mutation and Database Update. Human Mutation, 2016, 37, 148-154.	2.5	45
79	Gain-of-Function Mutations in KCNN3 Encoding the Small-Conductance Ca2+-Activated K+ Channel SK3 Cause Zimmermann-Laband Syndrome. American Journal of Human Genetics, 2019, 104, 1139-1157.	6.2	45
80	Clinical Presentation and Natural History of Hypertrophic Cardiomyopathy in RASopathies. Heart Failure Clinics, 2018, 14, 225-235.	2.1	44
81	Costello syndrome and related disorders. Current Opinion in Pediatrics, 2007, 19, 636-644.	2.0	42
82	Expanding the clinical and molecular findings in RASA1 capillary malformation-arteriovenous malformation. European Journal of Human Genetics, 2018, 26, 1521-1536.	2.8	42
83	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders. Human Genetics and Genomics Advances, 2022, 3, 100075.	1.7	42
84	Multiple increased osteoclast functions in individuals with neurofibromatosis type 1. American Journal of Medical Genetics, Part A, 2011, 155, 1050-1059.	1.2	41
85	TWIST gene mutation in a patient with radial aplasia and craniosynostosis: Further evidence for heterogeneity of Baller-Gerold syndrome. , 1999, 82, 170-176.		40
86	Hepatoblastoma and heart transplantation in a patient with cardio-facio-cutaneous syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 1481-1488.	1.2	39
87	The role of objective facial analysis using FDNA in making diagnoses following whole exome analysis. Report of two patients with mutations in the BAF complex genes. American Journal of Medical Genetics, Part A, 2016, 170, 1754-1762.	1.2	39
88	Phenotypic and molecular characterisation of CDK13-related congenital heart defects, dysmorphic facial features and intellectual developmental disorders. Genome Medicine, 2017, 9, 73.	8.2	39
89	Familial congenital non-immune hydrops, chylothorax, and pulmonary lymphangiectasia. American Journal of Medical Genetics, Part A, 2006, 140A, 368-372.	1.2	38
90	Duplication of Glu37 in the switch I region of HRAS impairs effector/GAP binding and underlies Costello syndrome by promoting enhanced growth factor-dependent MAPK and AKT activation. Human Molecular Genetics, 2010, 19, 790-802.	2.9	38

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91	Peripheral muscle weakness in RASopathies. Muscle and Nerve, 2012, 46, 394-399.	2.2	38
92	Expanding the SHOC2 mutation associated phenotype of noonan syndrome with loose anagen hair: Structural brain anomalies and myelofibrosis. American Journal of Medical Genetics, Part A, 2013, 161, 2420-2430.	1.2	38
93	X-linked hypomyelination with spondylometaphyseal dysplasia (H-SMD) associated with mutations in AIFM1. Neurogenetics, 2017, 18, 185-194.	1.4	38
94	Phenotype of CM-AVM2 caused by variants in EPHB4: how much overlap with hereditary hemorrhagic telangiectasia (HHT)?. Genetics in Medicine, 2019, 21, 2007-2014.	2.4	38
95	Mutations in FAM50A suggest that Armfield XLID syndrome is a spliceosomopathy. Nature Communications, 2020, 11, 3698.	12.8	38
96	Adaptive skills, cognitive, and behavioral characteristics of Costello syndrome. American Journal of Medical Genetics Part A, 2004, 128A, 396-400.	2.4	37
97	Evidence of Increased Bone Resorption in Neurofibromatosis Type 1 Using Urinary Pyridinium Crosslink Analysis. Pediatric Research, 2008, 63, 697-701.	2.3	37
98	Chiari malformation and tonsillar ectopia in twin brothers and father with autosomal dominant spondylo-epiphyseal dysplasia tarda. Skeletal Radiology, 1997, 26, 131-133.	2.0	36
99	The use of anterolateral bowing of the lower leg in the diagnostic criteria for neurofibromatosis type 1. Genetics in Medicine, 2007, 9, 409-412.	2.4	36
100	Longitudinal assessment of cognitive characteristics in Costello syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 3185-3193.	1.2	36
101	NALCN channelopathies. Neurology, 2016, 87, 1131-1139.	1.1	36
102	Genotype and phenotype spectrum of NRAS germline variants. European Journal of Human Genetics, 2017, 25, 823-831.	2.8	36
103	Genetic Variants Associated with Port-Wine Stains. PLoS ONE, 2015, 10, e0133158.	2.5	35
104	Significant overlap and possible identity of macrocephaly capillary malformation and megalencephaly polymicrogyriaâ€polydactyly hydrocephalus syndromes. American Journal of Medical Genetics, Part A, 2009, 149A, 868-876.	1.2	34
105	Neurocognitive, adaptive, and behavioral functioning of individuals with Costello syndrome: A review. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2011, 157, 115-122.	1.6	34
106	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. Genetics in Medicine, 2021, 23, 1028-1040.	2.4	34
107	Hypertrophic Cardiomyopathy in RASopathies. Heart Failure Clinics, 2022, 18, 19-29.	2.1	33
108	Response to longâ€term growth hormone therapy in patients affected by RASopathies and growth hormone deficiency: Patterns of growth, puberty and final height data. American Journal of Medical Genetics, Part A, 2015, 167, 2786-2794.	1.2	32

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109	Bilateral microtia and cleft palate in cousins with Diamond-Blackfan anemia. American Journal of Medical Genetics Part A, 2001, 101, 268-274.	2.4	31
110	Speechâ€“language characteristics of children with neurofibromatosis type 1. American Journal of Medical Genetics, Part A, 2010, 152A, 284-290.	1.2	30
111	The diagnosis of Costello syndrome: Nomenclature in Ras/MAPK pathway disorders. American Journal of Medical Genetics, Part A, 2008, 146A, 1218-1220.	1.2	29
112	Novel SMAD4 mutation causing Myhre syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 1835-1840.	1.2	29
113	PIAS4 is associated with macro/microcephaly in the novel interstitial 19p13.3 microdeletion/microduplication syndrome. European Journal of Human Genetics, 2015, 23, 1615-1626.	2.8	29
114	Multiscale, Converging Defects of Macro-Porosity, Microstructure and Matrix Mineralization Impact Long Bone Fragility in NF1. PLoS ONE, 2014, 9, e86115.	2.5	29
115	Autosomal Recessive Hypophosphatasia Manifestingin Uterowith Long Bone Deformity but Showing Spontaneous Postnatal Improvement. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 3443-3448.	3.6	28
116	Longitudinal course of cognitive, adaptive, and behavioral characteristics in Costello syndrome. American Journal of Medical Genetics, Part A, 2009, 149A, 2666-2672.	1.2	28
117	Low Bone Mineral Content and Challenges in Interpretation of Dual-Energy X-Ray Absorptiometry in Children With Mucopolysaccharidosis Types I, II, and VI. Journal of Clinical Densitometry, 2014, 17, 200-206.	1.2	27
118	Evaluation of somatic mutations in tibial pseudarthrosis samples in neurofibromatosis type 1. Journal of Medical Genetics, 2015, 52, 256-261.	3.2	27
119	Pediatric 25-hydroxyvitamin D concentrationsin neurofibromatosis type 1. Journal of Pediatric Endocrinology and Metabolism, 2011, 24, 169-74.	0.9	26
120	Expanding the neurodevelopmental phenotype of PURA syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 56-67.	1.2	26
121	TAOK1 is associated with neurodevelopmental disorder and essential for neuronal maturation and cortical development. Human Mutation, 2021, 42, 445-459.	2.5	26
122	Elevated catecholamine metabolites in patients with Costello syndrome. American Journal of Medical Genetics Part A, 2004, 128A, 48-51.	2.4	25
123	Paternal uniparental disomy of chromosome 14: Confirmation of a clinically-recognizable phenotype. American Journal of Medical Genetics Part A, 2004, 130A, 88-91.	2.4	25
124	Copy Number Variation Analysis in 98 Individuals with PHACE Syndrome. Journal of Investigative Dermatology, 2013, 133, 677-684.	0.7	25
125	Microcephaly, intractable seizures and developmental delay caused by biallelic variants in <scp>TBCD</scp>: further delineation of a new chaperoneâ€mediated tubulinopathy. Clinical Genetics, 2017, 91, 725-738.	2.0	25
126	Arteriovenous Malformationsâ€”Current Understanding of the Pathogenesis with Implications for Treatment. International Journal of Molecular Sciences, 2021, 22, 9037.	4.1	25

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127	Genotypeâ€cardiac phenotype correlations in a large singleâ€center cohort of patients affected by RASopathies: Clinical implications and literature review. American Journal of Medical Genetics, Part A, 2022, 188, 431-445.	1.2	25
128	Phenotypic spectrum of Costello syndrome individuals harboring the rare HRAS mutation p.Gly13Asp. , 2017, 173, 1309-1318.		24
129	De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder. Genetics in Medicine, 2020, 22, 538-546.	2.4	24
130	Paternal uniparental disomy with segmental loss of heterozygosity of chromosome 11 are hallmark characteristics of syndromic and sporadic embryonal rhabdomyosarcoma. American Journal of Medical Genetics, Part A, 2016, 170, 3197-3206.	1.2	23
131	Expansion and further delineation of the SETD5 phenotype leading to global developmental delay, variable dysmorphic features, and reduced penetrance. Clinical Genetics, 2018, 93, 752-761.	2.0	23
132	Biallelic sequence variants in INTS1 in patients with developmental delays, cataracts, and craniofacial anomalies. European Journal of Human Genetics, 2019, 27, 582-593.	2.8	23
133	SPRED2 loss-of-function causes a recessive Noonan syndrome-like phenotype. American Journal of Human Genetics, 2021, 108, 2112-2129.	6.2	23
134	Diaphragmatic hernia-exomphalos-hypertelorism syndrome: A new case and further evidence of autosomal recessive inheritance. , 1997, 68, 441-444.		22
135	A diagnostic approach to identifying submicroscopic 7p21 deletions in Saethre-Chotzen syndrome: Fluorescence in situ hybridization and dosage-sensitive Southern blot analysis. Genetics in Medicine, 2001, 3, 102-108.	2.4	22
136	Exome Analysis in Clinical Practice: Expanding the Phenotype of Bartsocas-Papas Syndrome. , 2013, 161, 1058-1063.		22
137	Decreased bone mineral density in Costello syndrome. Molecular Genetics and Metabolism, 2014, 111, 41-45.	1.1	22
138	Apparently new syndrome of congenital cataracts, sensorineural deafness, Down syndrome-like facial appearance, short stature, and mental retardation. American Journal of Medical Genetics Part A, 1996, 61, 382-386.	2.4	21
139	Dietary intervention rescues myopathy associated with neurofibromatosis type 1. Human Molecular Genetics, 2018, 27, 577-588.	2.9	21
140	Pain in individuals with RASopathies: Prevalence and clinical characterization in a sample of 80 affected patients. American Journal of Medical Genetics, Part A, 2019, 179, 940-947.	1.2	21
141	Syndromic disorders caused by gain-of-function variants in KCNH1, KCNK4, and KCNN3â€"a subgroup of K+ channelopathies. European Journal of Human Genetics, 2021, 29, 1384-1395.	2.8	21
142	A novel HRAS substitution (c.266C>G; p.S89C) resulting in decreased downstream signaling suggests a new dimension of RAS pathway dysregulation in human development. American Journal of Medical Genetics, Part A, 2012, 158A, 2106-2118.	1.2	20
143	Orthopedic manifestations and implications for individuals with Costello syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 1940-1949.	1.2	20
144	An attenuated phenotype of Costello syndrome in three unrelated individuals with a HRAS c.179G>A (p.Gly60Asp) mutation correlates with uncommon functional consequences. American Journal of Medical Genetics, Part A, 2015, 167, 2085-2097.	1.2	20

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145	Nasal dimple as part of the 22q11.2 deletion syndrome. , 1997, 69, 290-292.		19
146	Biomarkers of bone remodeling in children with mucopolysaccharidosis types I, II, and VI. Journal of Pediatric Rehabilitation Medicine, 2014, 7, 159-165.	0.5	19
147	Promoting appropriate genetic testing: the impact of a combined test review and consultative service. Genetics in Medicine, 2017, 19, 1049-1054.	2.4	19
148	PTPN11 Gain-of-Function Mutations Affect the Developing Human Brain, Memory, and Attention. Cerebral Cortex, 2019, 29, 2915-2923.	2.9	19
149	Lateral meningocele syndrome and Hajduâ€“Cheney syndrome: Different disorders with overlapping phenotypes. American Journal of Medical Genetics, Part A, 2011, 155, 1773-1774.	1.2	18
150	Normative growth charts for individuals with Costello syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 2692-2699.	1.2	18
151	Assessing genotypeâ€“phenotype correlation in Costello syndrome using a severity score. Genetics in Medicine, 2013, 15, 554-557.	2.4	18
152	The Splicing Efficiency of Activating HRAS Mutations Can Determine Costello Syndrome Phenotype and Frequency in Cancer. PLoS Genetics, 2016, 12, e1006039.	3.5	18
153	Evaluation of racial disparities in pediatric optic pathway glioma incidence: Results from the Surveillance, Epidemiology, and End Results Program, 2000â€“2014. Cancer Epidemiology, 2018, 54, 90-94.	1.9	18
154	Aphallia as part of urorectal septum malformation sequence in an infant of a diabetic mother. American Journal of Medical Genetics Part A, 1999, 82, 363-367.	2.4	17
155	Brief Report: The Prevalence of Neurofibromatosis Type 1 among Children with Autism Spectrum Disorder Identified by the Autism and Developmental Disabilities Monitoring Network. Journal of Autism and Developmental Disorders, 2016, 46, 3369-3376.	2.7	17
156	Epistaxis in children and adolescents with hereditary hemorrhagic telangiectasia. Laryngoscope, 2018, 128, 1714-1719.	2.0	17
157	When to test fetuses for RASopathies? Proposition from a systematic analysis of 352 multicenter cases and a postnatal cohort. Genetics in Medicine, 2021, 23, 1116-1124.	2.4	17
158	Targeting Oncogenic Src Homology 2 Domain-Containing Phosphatase 2 (SHP2) by Inhibiting Its Proteinâ€“Protein Interactions. Journal of Medicinal Chemistry, 2021, 64, 15973-15990.	6.4	17
159	Molecular confirmation of HRAS p.G12S in siblings with Costello syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 2263-2268.	1.2	16
160	Axenfeldâ€Rieger syndrome: Further clinical and array delineation of four unrelated patients with a 4q25 microdeletion. American Journal of Medical Genetics, Part A, 2014, 164, 1695-1701.	1.2	16
161	A genotype-first approach to exploring Mendelian cardiovascular traits with clear external manifestations. Genetics in Medicine, 2021, 23, 94-102.	2.4	16
162	Healthâ€related quality of life measures in genetic disorders: An outcome variable for consideration in clinical trials. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2009, 151C, 255-260.	1.6	15

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163	Natural history and life-threatening complications in Myhre syndrome and review of the literature. European Journal of Pediatrics, 2016, 175, 1307-1315.	2.7	15
164	Further delineation of AymÃ©â€Gripp syndrome and use of automated facial analysis tool. American Journal of Medical Genetics, Part A, 2018, 176, 1648-1656.	1.2	15
165	NF1 Somatic Mutation in Dystrophic Scoliosis. Journal of Molecular Neuroscience, 2019, 68, 11-18.	2.3	15
166	Missense variants in CTNNB1 can be associated with vitreoretinopathyâ€"Seven new cases of CTNNB1â€associated neurodevelopmental disorder including a previously unreported retinal phenotype. Molecular Genetics & Genomic Medicine, 2021, 9, e1542.	1.2	15
167	Grade 1 microtia, wide anterior fontanel and novel type tracheoâ€esophageal fistula in methimazole embryopathy. American Journal of Medical Genetics, Part A, 2011, 155, 526-533.	1.2	14
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