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	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
2	Hypertrophic Cardiomyopathy in RASopathies. Heart Failure Clinics, 2022, 18, 19-29.	2.1	33
3	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
4	Youngâ€onset diabetes patients in Thailand: Data from Thai Typeâ€‰1 Diabetes and Diabetes diagnosed Age before 30â€‰years Registry, Care and Network (T1DDAR CN). Journal of Diabetes Investigation, 2022, 13, 796-809.	2.4	3
5	Metabolic profiling of Costello syndrome: Insights from a single-center cohort. European Journal of Medical Genetics, 2022, 65, 104439.	1.3	5
6	GestaltMatcher facilitates rare disease matching using facial phenotype descriptors. Nature Genetics, 2022, 54, 349-357.	21.4	73
7	The seventh international <scp>RASopathies</scp> symposium: Pathways to a cureâ€”expanding knowledge, enhancing research, and therapeutic discovery. American Journal of Medical Genetics, Part A, 2022, 188, 1915-1927.	1.2	10
8	Craniosynostosis is a feature of Costello syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 1280-1286.	1.2	3
9	Natural history of NF1 c.2970_2972del p.(Met992del): confirmation of a low risk of complications in a longitudinal study. European Journal of Human Genetics, 2022, 30, 291-297.	2.8	5
10	Bone tissue homeostasis and risk of fractures in Costello syndrome: A 4â€year followâ€up study. American Journal of Medical Genetics, Part A, 2022, 188, 422-430.	1.2	5
11	Potassium Channel KCNH1 Activating Variants Cause Altered Functional and Morphological Ciliogenesis. Molecular Neurobiology, 2022, 59, 4825-4838.	4.0	4
12	Juberg-Hayward syndrome is a cohesinopathy, caused by mutation in ESCO2. European Journal of Orthodontics, 2021, 43, 45-50.	2.4	3
13	A genotype-first approach to exploring Mendelian cardiovascular traits with clear external manifestations. Genetics in Medicine, 2021, 23, 94-102.	2.4	16
14	TypeÂ1 diabetes management and outcomes: A multicenter study in Thailand. Journal of Diabetes Investigation, 2021, 12, 516-526.	2.4	13
15	Missense substitutions at a conserved 14-3-3 binding site in HDAC4 cause a novel intellectual disability syndrome. Human Genetics and Genomics Advances, 2021, 2, 100015.	1.7	6
16	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	6.2	71
17	41st Annual David W. Smith workshop on malformations and morphogenesis: Abstracts of the 2020 annual meeting. American Journal of Medical Genetics, Part A, 2021, 185, 1328-1337.	1.2	0
18	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

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19	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
20	TAOK1 is associated with neurodevelopmental disorder and essential for neuronal maturation and cortical development. Human Mutation, 2021, 42, 445-459.	2.5	26
21	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
22	Congenital polyvalvular disease expands the cardiac phenotype of the RASopathies. American Journal of Medical Genetics, Part A, 2021, 185, 1486-1493.	1.2	3
23	Typical 22q11.2 deletion syndrome appears to confer a reduced risk of schwannoma. Genetics in Medicine, 2021, 23, 1779-1782.	2.4	3
24	Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. Genetics in Medicine, 2021, 23, 1506-1513.	2.4	290
25	Reliability of Handheld Dynamometry to Measure Focal Muscle Weakness in Neurofibromatosis Types 1 and 2. Neurology, 2021, 97, S99-S110.	1.1	2
26	Arteriovenous Malformationsâ€"Current Understanding of the Pathogenesis with Implications for Treatment. International Journal of Molecular Sciences, 2021, 22, 9037.	4.1	25
27	Epilepsy and BRAF Mutations: Phenotypes, Natural History and Genotype-Phenotype Correlations. Genes, 2021, 12, 1316.	2.4	13
28	Pregnancy Outcomes among Women with Gravesâ€™ Hyperthyroidism: A Retrospective Cohort Study. Journal of Clinical Medicine, 2021, 10, 4495.	2.4	8
29	Response to Hamosh etÂal.. American Journal of Human Genetics, 2021, 108, 1809-1810.	6.2	0
30	A novel P3H1 mutation is associated with osteogenesis imperfecta type VIII and dental anomalies. Oral Surgery, Oral Medicine, Oral Pathology and Oral Radiology, 2021, 132, e198-e207.	0.4	5
31	SPRED2 loss-of-function causes a recessive Noonan syndrome-like phenotype. American Journal of Human Genetics, 2021, 108, 2112-2129.	6.2	23
32	Are Some Randomized Clinical Trials Impossible?. Journal of Pediatric Orthopaedics, 2021, 41, e90-e93.	1.2	5
33	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
34	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
35	Medically actionable comorbidities in adults with Costello syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 130-136.	1.2	6
36	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

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37	Juberg-Hayward syndrome and Roberts syndrome are allelic, caused by mutations in ESCO2. Archives of Oral Biology, 2020, 119, 104918.	1.8	2
38	Mutations in FAM50A suggest that Armfield XLID syndrome is a spliceosomopathy. Nature Communications, 2020, 11, 3698.	12.8	38
39	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	6.2	48
40	KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. Brain, 2020, 143, 3242-3261.	7.6	57
41	The novel duplication HRAS c.186_206dup p.(Glu62_Arg68dup): clinical and functional aspects. European Journal of Human Genetics, 2020, 28, 1548-1554.	2.8	6
42	SAT-434 Phenotype and Genotype Analysis of Patients with Resistance to Thyroid Hormone Î²: A Single-Center Experience. Journal of the Endocrine Society, 2020, 4, .	0.2	0
43	CantÃº syndrome versus Zimmermann-Laband syndrome: Report of nine individuals with ABCC9 variants. European Journal of Medical Genetics, 2020, 63, 103996.	1.3	7
44	De novo heterozygous missense and lossâ€ofâ€function variants in CDC42BPB are associated with a neurodevelopmental phenotype. American Journal of Medical Genetics, Part A, 2020, 182, 962-973.	1.2	8
45	Stress and Coping in Caregivers of Children with RASopathies: Assessment of the Impact of Caregiver Conferences. Journal of Pediatric Genetics, 2020, 09, 235-242.	0.7	0
46	PTPN11 Gain-of-Function Mutations Affect the Developing Human Brain, Memory, and Attention. Cerebral Cortex, 2019, 29, 2915-2923.	2.9	19
47	A Novel <i>GNAS</i> Mutation Causing Isolated Infantile Cushingâ€™s Syndrome. Hormone Research in Paediatrics, 2019, 92, 196-202.	1.8	3
48	Cardiac transplantation in children with Noonan syndrome. Pediatric Transplantation, 2019, 23, e13535.	1.0	12
49	Near final adult height, and body mass index in overweight/obese and normal-weight children with idiopathic central precocious puberty and treated with gonadotropin-releasing hormone analogs. Journal of Pediatric Endocrinology and Metabolism, 2019, 32, 1369-1375.	0.9	10
50	WNT1-associated osteogenesis imperfecta with atrophic frontal lobes and arachnoid cysts. Journal of Human Genetics, 2019, 64, 291-296.	2.3	10
51	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	27.0	205
52	Costello syndrome: Clinical phenotype, genotype, and management guidelines. American Journal of Medical Genetics, Part A, 2019, 179, 1725-1744.	1.2	70
53	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
54	Localization and age distribution of telangiectases in children and adolescents with hereditary hemorrhagic telangiectasia: A retrospective cohort study. Journal of the American Academy of Dermatology, 2019, 81, 950-955.	1.2	14

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55	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
56	NF1 Somatic Mutation in Dystrophic Scoliosis. Journal of Molecular Neuroscience, 2019, 68, 11-18.	2.3	15
57	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
58	Contributing factors of mortality in Praderâ€“Willi syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 196-205.	1.2	50
59	Identifying facial phenotypes of genetic disorders using deep learning. Nature Medicine, 2019, 25, 60-64.	30.7	449
60	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
61	ClinGenâ€™s RASopathy Expert Panel consensus methods for variant interpretation. Genetics in Medicine, 2018, 20, 1334-1345.	2.4	126
62	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
63	Racial/ethnic disparities and incidence of malignant peripheral nerve sheath tumors: results from the Surveillance, Epidemiology, and End Results Program, 2000â€“2014. Journal of Neuro-Oncology, 2018, 139, 69-75.	2.9	6
64	Osteogenesis imperfecta with ectopic mineralizations in dentin and cementum and a COL1A2 mutation. Journal of Human Genetics, 2018, 63, 811-820.	2.3	12
65	Vitamin D deficiency and its relationship with cardiac iron and function in patients with transfusion-dependent thalassemia at Chiang Mai University Hospital. Pediatric Hematology and Oncology, 2018, 35, 52-59.	0.8	7
66	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
67	Dietary intervention rescues myopathy associated with neurofibromatosis type 1. Human Molecular Genetics, 2018, 27, 577-588.	2.9	21
68	Ribosome Levels Selectively Regulate Translation and Lineage Commitment in Human Hematopoiesis. Cell, 2018, 173, 90-103.e19.	28.9	296
69	Clinical Presentation and Natural History of Hypertrophic Cardiomyopathy in RASopathies. Heart Failure Clinics, 2018, 14, 225-235.	2.1	44
70	Quantitative Ultrasound and Tibial Dysplasia in Neurofibromatosis Type 1. Journal of Clinical Densitometry, 2018, 21, 179-184.	1.2	7
71	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
72	Expanding the neurodevelopmental phenotype of PURA syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 56-67.	1.2	26

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73	Mutations in KCNK4 that Affect Gating Cause a Recognizable Neurodevelopmental Syndrome. American Journal of Human Genetics, 2018, 103, 621-630.	6.2	73
74	Role of Leucine 341 in Thyroid Hormone Receptor Beta Revealed by a Novel Mutation Causing Thyroid Hormone Resistance. Thyroid, 2018, 28, 1723-1726.	4.5	4
75	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
76	Use of Flow Cytometry for Diagnosis of Epilepsy Associated With Homozygous PIGW Variants. Pediatric Neurology, 2018, 85, 67-70.	2.1	4
77	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
78	Epistaxis in children and adolescents with hereditary hemorrhagic telangiectasia. Laryngoscope, 2018, 128, 1714-1719.	2.0	17
79	Predictive Value and Interrater Reliability of Radiographic Factors in Neurofibromatosis Patients With Dystrophic Scoliosis. Spine Deformity, 2018, 6, 560-567.	1.5	6
80	Expanding the clinical and molecular findings in RASA1 capillary malformation-arteriovenous malformation. European Journal of Human Genetics, 2018, 26, 1521-1536.	2.8	42
81	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
82	Promoting appropriate genetic testing: the impact of a combined test review and consultative service. Genetics in Medicine, 2017, 19, 1049-1054.	2.4	19
83	Attenuated phenotype of Costello syndrome and early death in a patient with an <scp>HRAS</scp> mutation (c.<scp>179G</scp>>T; p.<scp>Gly60Val</scp>) affecting signalling dynamics. Clinical Genetics, 2017, 92, 332-337.	2.0	8
84	A novel patient with an attenuated Costello syndrome phenotype due to an HRAS mutation affecting codon 146â€"Literature review and update. American Journal of Medical Genetics, Part A, 2017, 173, 1109-1114.	1.2	7
85	AberrantHRAStranscript processing underlies a distinctive phenotype within the RASopathy clinical spectrum. Human Mutation, 2017, 38, 798-804.	2.5	14
86	Phenotypic spectrum of Costello syndrome individuals harboring the rare HRAS mutation p.Gly13Asp. , 2017, 173, 1309-1318.		24
87	Ageâ€related differences in prevalence of autism spectrum disorder symptoms in children and adolescents with Costello syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 1294-1300.	1.2	10
88	Utilization of Whole-Exome Next-Generation Sequencing Variant Read Frequency for Detection of Lesion-Specific, Somatic Loss of Heterozygosity in a Neurofibromatosis Type 1 Cohort with Tibial Pseudarthrosis. Journal of Molecular Diagnostics, 2017, 19, 468-474.	2.8	6
89	Genotype and phenotype spectrum of NRAS germline variants. European Journal of Human Genetics, 2017, 25, 823-831.	2.8	36
90	Constitutional LZTR1 mutation presenting with a unilateral vestibular schwannoma in a teenager. Clinical Genetics, 2017, 92, 540-543.	2.0	9

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91	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. Brain, 2017, 140, 2610-2622.	7.6	102
92	X-linked hypomyelination with spondylometaphyseal dysplasia (H-SMD) associated with mutations in AIFM1. Neurogenetics, 2017, 18, 185-194.	1.4	38
93	Analysis of copy number variants in 11 pairs of monozygotic twins with neurofibromatosis type 1. American Journal of Medical Genetics, Part A, 2017, 173, 647-653.	1.2	12
94	Cytotoxicity of Zardaverine in Embryonal Rhabdomyosarcoma from a Costello Syndrome Patient. Frontiers in Oncology, 2017, 7, 42.	2.8	7
95	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
96	Subtle inflammation: a possible mechanism of future cardiovascular risk in obese children. Korean Journal of Pediatrics, 2017, 60, 359.	1.9	7
97	The Splicing Efficiency of Activating HRAS Mutations Can Determine Costello Syndrome Phenotype and Frequency in Cancer. PLoS Genetics, 2016, 12, e1006039.	3.5	18
98	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
99	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
100	36th Annual David W. Smith Workshop on Malformations and Morphogenesis: Abstracts of the 2015 annual meeting. , 2016, 170, 1665-1726.		1
101	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
102	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
103	Mandibulofacial Dysostosis with Microcephaly: Mutation and Database Update. Human Mutation, 2016, 37, 148-154.	2.5	45
104	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
105	Paternal uniparental disomy 11p15.5 in the pancreatic nodule of an infant with Costello syndrome: Shared mechanism for hyperinsulinemic hypoglycemia in neonates with Costello and Beckwithâ€“Wiedemann syndrome and somatic loss of heterozygosity in Costello syndrome driving clonal expansion. American Journal of Medical Genetics, Part A, 2016, 170, 559-564.	1.2	11
106	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
107	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
108	NALCN channelopathies. Neurology, 2016, 87, 1131-1139.	1.1	36

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109	Maternal uniparental disomy of chromosome 20: a novel imprinting disorder of growth failure. Genetics in Medicine, 2016, 18, 309-315.	2.4	69
110	PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. JCI Insight, 2016, 1, .	5.0	134
111	A HIV-infected adolescent with polycystic ovary syndrome. Journal of Pediatric Infectious Diseases, 2015, 06, 045-049.	0.2	1
112	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
113	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
114	Noonan syndromeâ€like disorder with loose anagen hair: A second case with neuroblastoma. American Journal of Medical Genetics, Part A, 2015, 167, 1902-1907.	1.2	14
115	Differentiating between copyâ€numberâ€variation and gainâ€ofâ€function mutation. American Journal of Medical Genetics, Part A, 2015, 167, 2684-2684.	1.2	0
116	Dystrophic Spinal Deformities in a Neurofibromatosis Type 1 Murine Model. PLoS ONE, 2015, 10, e0119093.	2.5	13
117	Genetic Variants Associated with Port-Wine Stains. PLoS ONE, 2015, 10, e0133158.	2.5	35
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	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
120	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
121	Hereditary hemorrhagic telangiectasia: genetics and molecular diagnostics in a new era. Frontiers in Genetics, 2015, 6, 1.	2.3	489
122	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
123	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
124	Mutations in KCNH1 and ATP6V1B2 cause Zimmermann-Laband syndrome. Nature Genetics, 2015, 47, 661-667.	21.4	177
125	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
126	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

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127	Impaired PIEZO1 function in patients with a novel autosomal recessive congenital lymphatic dysplasia. Nature Communications, 2015, 6, 8329.	12.8	239
128	Activity and participation in children with neurofibromatosis type 1. Research in Developmental Disabilities, 2015, 36, 213-221.	2.2	8
129	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
130	Asfotase-Î± improves bone growth, mineralization and strength in mouse models of neurofibromatosis type-1. Nature Medicine, 2014, 20, 904-910.	30.7	54
131	Goltz syndrome and PORCN mosaicism. International Journal of Dermatology, 2014, 53, 1481-1484.	1.0	11
132	Early-Lethal Costello Syndrome Due to Rare HRAS Tandem Base Substitution (c.35_36GC>AA;) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 54 421-430.	1.0	8
133	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
134	Cutis laxa with pulmonary emphysema, conjunctivochalasis, nasolacrimal duct obstruction, abnormal hair, and a novel FBLN5 mutation. American Journal of Medical Genetics, Part A, 2014, 164, 2370-2377.	1.2	10
135	Neural tube defects and atypical deletion on 22q11.2. American Journal of Medical Genetics, Part A, 2014, 164, 2701-2706.	1.2	6
136	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
137	Postural control in children with and without neurofibromatosis type 1. Human Movement Science, 2014, 34, 157-163.	1.4	6
138	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
139	Screening children with neurofibromatosis type 1 for autism spectrum disorder. American Journal of Medical Genetics, Part A, 2014, 164, 1706-1712.	1.2	9
140	Novel SMAD4 mutation causing Myhre syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 1835-1840.	1.2	29
141	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
142	Decreased bone mineral density in Costello syndrome. Molecular Genetics and Metabolism, 2014, 111, 41-45.	1.1	22
143	Multiscale, Converging Defects of Macro-Porosity, Microstructure and Matrix Mineralization Impact Long Bone Fragility in NF1. PLoS ONE, 2014, 9, e86115.	2.5	29
144	Noonan syndrome. Lancet, The, 2013, 381, 333-342.	13.7	608

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145	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
146	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
147	Copy Number Variation Analysis in 98 Individuals with PHACE Syndrome. Journal of Investigative Dermatology, 2013, 133, 677-684.	0.7	25
148	Approaches to Treating NF1 Tibial Pseudarthrosis. Journal of Pediatric Orthopaedics, 2013, 33, 269-275.	1.2	55
149	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
150	Orthopedic manifestations and implications for individuals with Costello syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 1940-1949.	1.2	20
151	Keratoconus in Costello Syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 1132-1136.	1.2	5
152	Exome Analysis in Clinical Practice: Expanding the Phenotype of Bartsocas-Papas Syndrome. , 2013, 161, 1058-1063.		22
153	Assessing genotypeâ€“phenotype correlation in Costello syndrome using a severity score. Genetics in Medicine, 2013, 15, 554-557.	2.4	18
154	Skeletal abnormalities in lysosomal storage diseases. Pediatric Endocrinology Reviews, 2013, 10 Suppl 2, 406-16.	1.2	7
155	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
156	Special section. Syndromeâ€specific growth charts. American Journal of Medical Genetics, Part A, 2012, 158A, 2645-2646.	1.2	8
157	Mosaicism in Stickler syndrome. European Journal of Medical Genetics, 2012, 55, 418-422.	1.3	12
158	Normative growth charts for individuals with Costello syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 2692-2699.	1.2	18
159	Peripheral muscle weakness in RASopathies. Muscle and Nerve, 2012, 46, 394-399.	2.2	38
160	Transmission of the rare HRAS mutation (c. 173Câ€‰>â€‰T; p.T58I) further illustrates its attenuated phenotype. American Journal of Medical Genetics, Part A, 2012, 158A, 1095-1101.	1.2	13
161	Analysis of skeletal dysplasias in the Utah population. American Journal of Medical Genetics, Part A, 2012, 158A, 1046-1054.	1.2	72
162	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

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164	Noonan syndrome and clinically related disorders. Best Practice and Research in Clinical Endocrinology and Metabolism, 2011, 25, 161-179.	4.7	303
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