

Karen Gripp

List of Publications by Year in descending order

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Version: 2024-02-01

255
papers

16,607
citations

30551

56
h-index

23173

116
g-index

267
all docs

267
docs citations

267
times ranked

19581
citing authors

#	ARTICLE	IF	CITATIONS
1	Genotype-cardiac phenotype correlations in a large single-center cohort of patients affected by RASopathies: Clinical implications and literature review. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 431-445.	0.7	25
2	Hypertrophic Cardiomyopathy in RASopathies. <i>Heart Failure Clinics</i> , 2022, 18, 19-29.	1.0	33
3	Novel diagnostic DNA methylation epigenatures expand and refine the epigenetic landscapes of Mendelian disorders. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100075.	1.0	42
4	Young-onset diabetes patients in Thailand: Data from Thai Type 1 Diabetes and Diabetes diagnosed Age before 30 years Registry, Care and Network (T1DDAR CN). <i>Journal of Diabetes Investigation</i> , 2022, 13, 796-809.	1.1	3
5	Metabolic profiling of Costello syndrome: Insights from a single-center cohort. <i>European Journal of Medical Genetics</i> , 2022, 65, 104439.	0.7	5
6	GestaltMatcher facilitates rare disease matching using facial phenotype descriptors. <i>Nature Genetics</i> , 2022, 54, 349-357.	9.4	73
7	The seventh international <sc>RASopathies</sc> symposium: Pathways to a cure" expanding knowledge, enhancing research, and therapeutic discovery. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1915-1927.	0.7	10
8	Craniosynostosis is a feature of Costello syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1280-1286.	0.7	3
9	Natural history of NF1 c.2970_2972del p.(Met992del): confirmation of a low risk of complications in a longitudinal study. <i>European Journal of Human Genetics</i> , 2022, 30, 291-297.	1.4	5
10	Bone tissue homeostasis and risk of fractures in Costello syndrome: A 4-year follow-up study. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 422-430.	0.7	5
11	Potassium Channel KCNH1 Activating Variants Cause Altered Functional and Morphological Ciliogenesis. <i>Molecular Neurobiology</i> , 2022, 59, 4825-4838.	1.9	4
12	Juberg-Hayward syndrome is a cohesinopathy, caused by mutation in ESCO2. <i>European Journal of Orthodontics</i> , 2021, 43, 45-50.	1.1	3
13	A genotype-first approach to exploring Mendelian cardiovascular traits with clear external manifestations. <i>Genetics in Medicine</i> , 2021, 23, 94-102.	1.1	16
14	Type 1 diabetes management and outcomes: A multicenter study in Thailand. <i>Journal of Diabetes Investigation</i> , 2021, 12, 516-526.	1.1	13
15	Missense substitutions at a conserved 14-3-3 binding site in HDAC4 cause a novel intellectual disability syndrome. <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100015.	1.0	6
16	A dyadic approach to the delineation of diagnostic entities in clinical genomics. <i>American Journal of Human Genetics</i> , 2021, 108, 8-15.	2.6	71
17	41st Annual David W. Smith workshop on malformations and morphogenesis: Abstracts of the 2020 annual meeting. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1328-1337.	0.7	0
18	When to test fetuses for RASopathies? Proposition from a systematic analysis of 352 multicenter cases and a postnatal cohort. <i>Genetics in Medicine</i> , 2021, 23, 1116-1124.	1.1	17

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19	Syndromic disorders caused by gain-of-function variants in KCNH1, KCNK4, and KCNN3—a subgroup of K ⁺ channelopathies. <i>European Journal of Human Genetics</i> , 2021, 29, 1384-1395.	1.4	21
20	TAOK1 is associated with neurodevelopmental disorder and essential for neuronal maturation and cortical development. <i>Human Mutation</i> , 2021, 42, 445-459.	1.1	26
21	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. <i>Genetics in Medicine</i> , 2021, 23, 1028-1040.	1.1	34
22	Congenital polyvalvular disease expands the cardiac phenotype of the RASopathies. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1486-1493.	0.7	3
23	Typical 22q11.2 deletion syndrome appears to confer a reduced risk of schwannoma. <i>Genetics in Medicine</i> , 2021, 23, 1779-1782.	1.1	3
24	Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. <i>Genetics in Medicine</i> , 2021, 23, 1506-1513.	1.1	290
25	Reliability of Handheld Dynamometry to Measure Focal Muscle Weakness in Neurofibromatosis Types 1 and 2. <i>Neurology</i> , 2021, 97, S99-S110.	1.5	2
26	Arteriovenous Malformations—Current Understanding of the Pathogenesis with Implications for Treatment. <i>International Journal of Molecular Sciences</i> , 2021, 22, 9037.	1.8	25
27	Epilepsy and BRAF Mutations: Phenotypes, Natural History and Genotype-Phenotype Correlations. <i>Genes</i> , 2021, 12, 1316.	1.0	13
28	Pregnancy Outcomes among Women with Graves™ Hyperthyroidism: A Retrospective Cohort Study. <i>Journal of Clinical Medicine</i> , 2021, 10, 4495.	1.0	8
29	Response to Hamosh et al.. <i>American Journal of Human Genetics</i> , 2021, 108, 1809-1810.	2.6	0
30	A novel P3H1 mutation is associated with osteogenesis imperfecta type VIII and dental anomalies. <i>Oral Surgery, Oral Medicine, Oral Pathology and Oral Radiology</i> , 2021, 132, e198-e207.	0.2	5
31	SPRED2 loss-of-function causes a recessive Noonan syndrome-like phenotype. <i>American Journal of Human Genetics</i> , 2021, 108, 2112-2129.	2.6	23
32	Are Some Randomized Clinical Trials Impossible?. <i>Journal of Pediatric Orthopaedics</i> , 2021, 41, e90-e93.	0.6	5
33	Targeting Oncogenic Src Homology 2 Domain-Containing Phosphatase 2 (SHP2) by Inhibiting Its Protein-Protein Interactions. <i>Journal of Medicinal Chemistry</i> , 2021, 64, 15973-15990.	2.9	17
34	Missense variants in <i>CTNNB1</i> can be associated with vitreoretinopathy—Seven new cases of <i>CTNNB1</i> -associated neurodevelopmental disorder including a previously unreported retinal phenotype. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1542.	0.6	15
35	Medically actionable comorbidities in adults with Costello syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 130-136.	0.7	6
36	De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder. <i>Genetics in Medicine</i> , 2020, 22, 538-546.	1.1	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.	0.8	2
38	Mutations in FAM50A suggest that Armfield XLID syndrome is a spliceosomopathy. Nature Communications, 2020, 11, 3698.	5.8	38
39	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	2.6	48
40	<i>KMT2B</i>-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. Brain, 2020, 143, 3242-3261.	3.7	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.	1.4	6
42	SAT-434 Phenotype and Genotype Analysis of Patients with Resistance to Thyroid Hormone β : A Single-Center Experience. Journal of the Endocrine Society, 2020, 4, .	0.1	0
43	CantÃ syndrome versus Zimmermann-Laband syndrome: Report of nine individuals with ABCC9 variants. European Journal of Medical Genetics, 2020, 63, 103996.	0.7	7
44	De novo heterozygous missense and loss-of-function variants in <i>CDC42BPB</i> are associated with a neurodevelopmental phenotype. American Journal of Medical Genetics, Part A, 2020, 182, 962-973.	0.7	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.	0.3	0
46	PTPN11 Gain-of-Function Mutations Affect the Developing Human Brain, Memory, and Attention. Cerebral Cortex, 2019, 29, 2915-2923.	1.6	19
47	A Novel <i>GNAS</i> Mutation Causing Isolated Infantile Cushing's Syndrome. Hormone Research in Paediatrics, 2019, 92, 196-202.	0.8	3
48	Cardiac transplantation in children with Noonan syndrome. Pediatric Transplantation, 2019, 23, e13535.	0.5	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.	0.4	10
50	WNT1-associated osteogenesis imperfecta with atrophic frontal lobes and arachnoid cysts. Journal of Human Genetics, 2019, 64, 291-296.	1.1	10
51	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	13.9	205
52	Costello syndrome: Clinical phenotype, genotype, and management guidelines. American Journal of Medical Genetics, Part A, 2019, 179, 1725-1744.	0.7	70
53	Gain-of-Function Mutations in KCNN3 Encoding the Small-Conductance Ca ²⁺ -Activated K ⁺ Channel SK3 Cause Zimmermann-Laband Syndrome. American Journal of Human Genetics, 2019, 104, 1139-1157.	2.6	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.	0.6	14

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55	Pain in individuals with RASopathies: Prevalence and clinical characterization in a sample of 80 affected patients. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 940-947.	0.7	21
56	NF1 Somatic Mutation in Dystrophic Scoliosis. <i>Journal of Molecular Neuroscience</i> , 2019, 68, 11-18.	1.1	15
57	Phenotype of CM-AVM2 caused by variants in EPHB4: how much overlap with hereditary hemorrhagic telangiectasia (HHT)?. <i>Genetics in Medicine</i> , 2019, 21, 2007-2014.	1.1	38
58	Contributing factors of mortality in Prader-Willi syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 196-205.	0.7	50
59	Identifying facial phenotypes of genetic disorders using deep learning. <i>Nature Medicine</i> , 2019, 25, 60-64.	15.2	449
60	Biallelic sequence variants in INTS1 in patients with developmental delays, cataracts, and craniofacial anomalies. <i>European Journal of Human Genetics</i> , 2019, 27, 582-593.	1.4	23
61	ClinGen™s RASopathy Expert Panel consensus methods for variant interpretation. <i>Genetics in Medicine</i> , 2018, 20, 1334-1345.	1.1	126
62	Evaluation of racial disparities in pediatric optic pathway glioma incidence: Results from the Surveillance, Epidemiology, and End Results Program, 2000-2014. <i>Cancer Epidemiology</i> , 2018, 54, 90-94.	0.8	18
63	Racial/ethnic disparities and incidence of malignant peripheral nerve sheath tumors: results from the Surveillance, Epidemiology, and End Results Program, 2000-2014. <i>Journal of Neuro-Oncology</i> , 2018, 139, 69-75.	1.4	6
64	Osteogenesis imperfecta with ectopic mineralizations in dentin and cementum and a COL1A2 mutation. <i>Journal of Human Genetics</i> , 2018, 63, 811-820.	1.1	12
65	Vitamin D deficiency and its relationship with cardiac iron and function in patients with transfusion-dependent thalassemia at Chiang Mai University Hospital. <i>Pediatric Hematology and Oncology</i> , 2018, 35, 52-59.	0.3	7
66	Genotype-Phenotype Correlation in NF1: Evidence for a More Severe Phenotype Associated with Missense Mutations Affecting NF1 Codons 844-848. <i>American Journal of Human Genetics</i> , 2018, 102, 69-87.	2.6	144
67	Dietary intervention rescues myopathy associated with neurofibromatosis type 1. <i>Human Molecular Genetics</i> , 2018, 27, 577-588.	1.4	21
68	Ribosome Levels Selectively Regulate Translation and Lineage Commitment in Human Hematopoiesis. <i>Cell</i> , 2018, 173, 90-103.e19.	13.5	296
69	Clinical Presentation and Natural History of Hypertrophic Cardiomyopathy in RASopathies. <i>Heart Failure Clinics</i> , 2018, 14, 225-235.	1.0	44
70	Quantitative Ultrasound and Tibial Dysplasia in Neurofibromatosis Type 1. <i>Journal of Clinical Densitometry</i> , 2018, 21, 179-184.	0.5	7
71	Expansion and further delineation of the <i>SETD5</i> phenotype leading to global developmental delay, variable dysmorphic features, and reduced penetrance. <i>Clinical Genetics</i> , 2018, 93, 752-761.	1.0	23
72	Expanding the neurodevelopmental phenotype of <i>PURA</i> syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 56-67.	0.7	26

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73	Mutations in KCNK4 that Affect Gating Cause a Recognizable Neurodevelopmental Syndrome. <i>American Journal of Human Genetics</i> , 2018, 103, 621-630.	2.6	73
74	Role of Leucine 341 in Thyroid Hormone Receptor Beta Revealed by a Novel Mutation Causing Thyroid Hormone Resistance. <i>Thyroid</i> , 2018, 28, 1723-1726.	2.4	4
75	Assessing the gene-disease association of 19 genes with the RASopathies using the ClinGen gene curation framework. <i>Human Mutation</i> , 2018, 39, 1485-1493.	1.1	66
76	Use of Flow Cytometry for Diagnosis of Epilepsy Associated With Homozygous PIGW Variants. <i>Pediatric Neurology</i> , 2018, 85, 67-70.	1.0	4
77	Further delineation of Aymã©Gripp syndrome and use of automated facial analysis tool. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1648-1656.	0.7	15
78	Epistaxis in children and adolescents with hereditary hemorrhagic telangiectasia. <i>Laryngoscope</i> , 2018, 128, 1714-1719.	1.1	17
79	Predictive Value and Interrater Reliability of Radiographic Factors in Neurofibromatosis Patients With Dystrophic Scoliosis. <i>Spine Deformity</i> , 2018, 6, 560-567.	0.7	6
80	Expanding the clinical and molecular findings in RASA1 capillary malformation-arteriovenous malformation. <i>European Journal of Human Genetics</i> , 2018, 26, 1521-1536.	1.4	42
81	Microcephaly, intractable seizures and developmental delay caused by biallelic variants in <i>TBCD</i> : further delineation of a new chaperone-mediated tubulinopathy. <i>Clinical Genetics</i> , 2017, 91, 725-738.	1.0	25
82	Promoting appropriate genetic testing: the impact of a combined test review and consultative service. <i>Genetics in Medicine</i> , 2017, 19, 1049-1054.	1.1	19
83	Attenuated phenotype of Costello syndrome and early death in a patient with an <i>HRAS</i> mutation (c.179G>T; p.Gly60Val) affecting signalling dynamics. <i>Clinical Genetics</i> , 2017, 92, 332-337.	1.0	8
84	A novel patient with an attenuated Costello syndrome phenotype due to an <i>HRAS</i> mutation affecting codon 146 Literature review and update. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1109-1114.	0.7	7
85	Aberrant <i>HRAS</i> transcript processing underlies a distinctive phenotype within the RASopathy clinical spectrum. <i>Human Mutation</i> , 2017, 38, 798-804.	1.1	14
86	Phenotypic spectrum of Costello syndrome individuals harboring the rare <i>HRAS</i> mutation p.Gly13Asp. , 2017, 173, 1309-1318.		24
87	Age-related differences in prevalence of autism spectrum disorder symptoms in children and adolescents with Costello syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1294-1300.	0.7	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. <i>Journal of Molecular Diagnostics</i> , 2017, 19, 468-474.	1.2	6
89	Genotype and phenotype spectrum of <i>NRAS</i> germline variants. <i>European Journal of Human Genetics</i> , 2017, 25, 823-831.	1.4	36
90	Constitutional <i>LZTR1</i> mutation presenting with a unilateral vestibular schwannoma in a teenager. <i>Clinical Genetics</i> , 2017, 92, 540-543.	1.0	9

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91	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. <i>Brain</i> , 2017, 140, 2610-2622.	3.7	102
92	X-linked hypomyelination with spondylometaphyseal dysplasia (H-SMD) associated with mutations in AIFM1. <i>Neurogenetics</i> , 2017, 18, 185-194.	0.7	38
93	Analysis of copy number variants in 11 pairs of monozygotic twins with neurofibromatosis type 1. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 647-653.	0.7	12
94	Cytotoxicity of Zardaverine in Embryonal Rhabdomyosarcoma from a Costello Syndrome Patient. <i>Frontiers in Oncology</i> , 2017, 7, 42.	1.3	7
95	Phenotypic and molecular characterisation of CDK13-related congenital heart defects, dysmorphic facial features and intellectual developmental disorders. <i>Genome Medicine</i> , 2017, 9, 73.	3.6	39
96	Subtle inflammation: a possible mechanism of future cardiovascular risk in obese children. <i>Korean Journal of Pediatrics</i> , 2017, 60, 359.	1.9	7
97	The Splicing Efficiency of Activating HRAS Mutations Can Determine Costello Syndrome Phenotype and Frequency in Cancer. <i>PLoS Genetics</i> , 2016, 12, e1006039.	1.5	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. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1754-1762.	0.7	39
99	Nephroblastomatosis or Wilms tumor in a fourth patient with a somatic <i>PIK3CA</i> mutation. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2559-2569.	0.7	55
100	36th Annual David W. Smith Workshop on Malformations and Morphogenesis: Abstracts of the 2015 annual meeting. , 2016, 170, 1665-1726.		1
101	<i>RASA1</i> somatic mutation and variable expressivity in capillary malformation/arteriovenous malformation (CM/AVM) syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1450-1454.	0.7	85
102	A novel rasopathy caused by recurrent de novo missense mutations in <i>PPP1CB</i> closely resembles Noonan syndrome with loose anagen hair. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2237-2247.	0.7	117
103	Mandibulofacial Dysostosis with Microcephaly: Mutation and Database Update. <i>Human Mutation</i> , 2016, 37, 148-154.	1.1	45
104	Paternal uniparental disomy with segmental loss of heterozygosity of chromosome 11 are hallmark characteristics of syndromic and sporadic embryonal rhabdomyosarcoma. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 3197-3206.	0.7	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 Beckwith-Wiedemann syndrome and somatic loss of heterozygosity in Costello syndrome driving clonal expansion. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 559-564.	0.7	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. <i>Journal of Autism and Developmental Disorders</i> , 2016, 46, 3369-3376.	1.7	17
107	Natural history and life-threatening complications in Myhre syndrome and review of the literature. <i>European Journal of Pediatrics</i> , 2016, 175, 1307-1315.	1.3	15
108	NALCN channelopathies. <i>Neurology</i> , 2016, 87, 1131-1139.	1.5	36

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109	Maternal uniparental disomy of chromosome 20: a novel imprinting disorder of growth failure. <i>Genetics in Medicine</i> , 2016, 18, 309-315.	1.1	69
110	PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. <i>JCI Insight</i> , 2016, 1, .	2.3	134
111	A HIV-infected adolescent with polycystic ovary syndrome. <i>Journal of Pediatric Infectious Diseases</i> , 2015, 06, 045-049.	0.1	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. <i>Human Mutation</i> , 2015, 36, 1052-1063.	1.1	143
113	Response to long-term growth hormone therapy in patients affected by RASopathies and growth hormone deficiency: Patterns of growth, puberty and final height data. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2786-2794.	0.7	32
114	Noonan syndrome-like disorder with loose anagen hair: A second case with neuroblastoma. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1902-1907.	0.7	14
115	Differentiating between copy-number-variation and gain-of-function mutation. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2684-2684.	0.7	0
116	Dystrophic Spinal Deformities in a Neurofibromatosis Type 1 Murine Model. <i>PLoS ONE</i> , 2015, 10, e0119093.	1.1	13
117	Genetic Variants Associated with Port-Wine Stains. <i>PLoS ONE</i> , 2015, 10, e0133158.	1.1	35
118	Evaluation of somatic mutations in tibial pseudarthrosis samples in neurofibromatosis type 1. <i>Journal of Medical Genetics</i> , 2015, 52, 256-261.	1.5	27
119	Truncating mutations in the last exon of <i>NOTCH3</i> cause lateral meningocele syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 271-281.	0.7	59
120	The Cyclic AMP Pathway Is a Sex-Specific Modifier of Glioma Risk in Type I Neurofibromatosis Patients. <i>Cancer Research</i> , 2015, 75, 16-21.	0.4	56
121	Hereditary hemorrhagic telangiectasia: genetics and molecular diagnostics in a new era. <i>Frontiers in Genetics</i> , 2015, 6, 1.	1.1	489
122	An attenuated phenotype of Costello syndrome in three unrelated individuals with a <i>HRAS</i> c.179G>A (p.Gly60Asp) mutation correlates with uncommon functional consequences. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2085-2097.	0.7	20
123	PIAS4 is associated with macro/microcephaly in the novel interstitial 19p13.3 microdeletion/microduplication syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 1615-1626.	1.4	29
124	Mutations in <i>KCNH1</i> and <i>ATP6V1B2</i> cause Zimmermann-Laband syndrome. <i>Nature Genetics</i> , 2015, 47, 661-667.	9.4	177
125	Germline gain-of-function mutations in <i>AFF4</i> cause a developmental syndrome functionally linking the super elongation complex and cohesin. <i>Nature Genetics</i> , 2015, 47, 338-344.	9.4	109
126	Myeloid Dysregulation in a Human Induced Pluripotent Stem Cell Model of <i>PTPN11</i> -Associated Juvenile Myelomonocytic Leukemia. <i>Cell Reports</i> , 2015, 13, 504-515.	2.9	79

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127	Impaired PIEZO1 function in patients with a novel autosomal recessive congenital lymphatic dysplasia. <i>Nature Communications</i> , 2015, 6, 8329.	5.8	239
128	Activity and participation in children with neurofibromatosis type 1. <i>Research in Developmental Disabilities</i> , 2015, 36, 213-221.	1.2	8
129	Activating mutations in RRAS underlie a phenotype within the RASopathy spectrum and contribute to leukaemogenesis. <i>Human Molecular Genetics</i> , 2014, 23, 4315-4327.	1.4	114
130	Asfotase- α improves bone growth, mineralization and strength in mouse models of neurofibromatosis type-1. <i>Nature Medicine</i> , 2014, 20, 904-910.	15.2	54
131	Goltz syndrome and <i>PORCN</i> mosaicism. <i>International Journal of Dermatology</i> , 2014, 53, 1481-1484.	0.5	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.	0.5	8
133	Diamond's Blackfan anemia with mandibulofacial dystostosis is heterogeneous, including the novel DBA genes <i>TSR2</i> and <i>RPS28</i> . <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2240-2249.	0.7	121
134	Cutis laxa with pulmonary emphysema, conjunctivochalasis, nasolacrimal duct obstruction, abnormal hair, and a novel <i>FBLN5</i> mutation. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2370-2377.	0.7	10
135	Neural tube defects and atypical deletion on 22q11.2. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2701-2706.	0.7	6
136	Biomarkers of bone remodeling in children with mucopolysaccharidosis types I, II, and VI. <i>Journal of Pediatric Rehabilitation Medicine</i> , 2014, 7, 159-165.	0.3	19
137	Postural control in children with and without neurofibromatosis type 1. <i>Human Movement Science</i> , 2014, 34, 157-163.	0.6	6
138	Axenfeld-Rieger syndrome: Further clinical and array delineation of four unrelated patients with a 4q25 microdeletion. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1695-1701.	0.7	16
139	Screening children with neurofibromatosis type 1 for autism spectrum disorder. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1706-1712.	0.7	9
140	Novel <i>SMAD4</i> mutation causing Myhre syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1835-1840.	0.7	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. <i>Journal of Clinical Densitometry</i> , 2014, 17, 200-206.	0.5	27
142	Decreased bone mineral density in Costello syndrome. <i>Molecular Genetics and Metabolism</i> , 2014, 111, 41-45.	0.5	22
143	Multiscale, Converging Defects of Macro-Porosity, Microstructure and Matrix Mineralization Impact Long Bone Fragility in NF1. <i>PLoS ONE</i> , 2014, 9, e86115.	1.1	29
144	Noonan syndrome. <i>Lancet, The</i> , 2013, 381, 333-342.	6.3	608

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