Marco Tartaglia

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/1310592/publications.pdf

Version: 2024-02-01

362 papers	20,178 citations	14644 66 h-index	15249 126 g-index
374 all docs	374 docs citations	374 times ranked	21373 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.	9.4	1,555
2	Somatic mutations in PTPN11 in juvenile myelomonocytic leukemia, myelodysplastic syndromes and acute myeloid leukemia. Nature Genetics, 2003, 34, 148-150.	9.4	960
3	PTPN11 Mutations in Noonan Syndrome: Molecular Spectrum, Genotype-Phenotype Correlation, and Phenotypic Heterogeneity. American Journal of Human Genetics, 2002, 70, 1555-1563.	2.6	680
4	Patient-specific induced pluripotent stem-cell-derived models of LEOPARD syndrome. Nature, 2010, 465, 808-812.	13.7	672
5	Gain-of-function RAF1 mutations cause Noonan and LEOPARD syndromes with hypertrophic cardiomyopathy. Nature Genetics, 2007, 39, 1007-1012.	9.4	624
6	Noonan syndrome. Lancet, The, 2013, 381, 333-342.	6.3	608
7	Gain-of-function SOS1 mutations cause a distinctive form of Noonan syndrome. Nature Genetics, 2007, 39, 75-79.	9.4	523
8	Mutation of SHOC2 promotes aberrant protein N-myristoylation and causes Noonan-like syndrome with loose anagen hair. Nature Genetics, 2009, 41, 1022-1026.	9.4	358
9	Diversity and Functional Consequences of Germline and Somatic PTPN11 Mutations in Human Disease. American Journal of Human Genetics, 2006, 78, 279-290.	2.6	352
10	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	2.6	337
11	Somatically acquired JAK1 mutations in adult acute lymphoblastic leukemia. Journal of Experimental Medicine, 2008, 205, 751-758.	4.2	318
12	NOONAN SYNDROME AND RELATED DISORDERS: Genetics and Pathogenesis. Annual Review of Genomics and Human Genetics, 2005, 6, 45-68.	2.5	306
13	Noonan syndrome and clinically related disorders. Best Practice and Research in Clinical Endocrinology and Metabolism, 2011, 25, 161-179.	2.2	303
14	A restricted spectrum of NRAS mutations causes Noonan syndrome. Nature Genetics, 2010, 42, 27-29.	9.4	271
15	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.	0.6	265
16	GermlineBRAFmutations in Noonan, LEOPARD, and cardiofaciocutaneous syndromes: Molecular diversity and associated phenotypic spectrum. Human Mutation, 2009, 30, 695-702.	1.1	251
17	The mutational spectrum of PTPN11 in juvenile myelomonocytic leukemia and Noonan syndrome/myeloproliferative disease. Blood, 2005, 106, 2183-2185.	0.6	247
18	Heterozygous Germline Mutations in the CBL Tumor-Suppressor Gene Cause a Noonan Syndrome-like Phenotype. American Journal of Human Genetics, 2010, 87, 250-257.	2.6	221

#	Article	IF	Citations
19	ACE2 gene variants may underlie interindividual variability and susceptibility to COVID-19 in the Italian population. European Journal of Human Genetics, 2020, 28, 1602-1614.	1.4	208
20	Germline Missense Mutations Affecting KRAS Isoform B Are Associated with a Severe Noonan Syndrome Phenotype. American Journal of Human Genetics, 2006, 79, 129-135.	2.6	205
21	Noonan Syndrome: Clinical Aspects and Molecular Pathogenesis. Molecular Syndromology, 2010, 1, 2-26.	0.3	197
22	Noonan syndrome-associated SHP2/PTPN11 mutants cause EGF-dependent prolonged GAB1 binding and sustained ERK2/MAPK1 activation. Human Mutation, 2004, 23, 267-277.	1.1	177
23	Noonan syndrome and related disorders: dysregulated RAS-mitogen activated protein kinase signal transduction. Human Molecular Genetics, 2006, 15, R220-R226.	1.4	177
24	Mutations in KCNH1 and ATP6V1B2 cause Zimmermann-Laband syndrome. Nature Genetics, 2015, 47, 661-667.	9.4	177
25	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 356-370.	2.6	171
26	A Specific Mutational Signature Associated with DNA 8-Oxoguanine Persistence in MUTYH-defective Colorectal Cancer. EBioMedicine, 2017, 20, 39-49.	2.7	170
27	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.	1.8	167
28	Missense Mutation in the Transcription Factor NKX2â€“5: A Novel Molecular Event in the Pathogenesis of Thyroid Dysgenesis. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 1428-1433.	1.8	157
29	NF1 Gene Mutations Represent the Major Molecular Event Underlying Neurofibromatosis-Noonan Syndrome. American Journal of Human Genetics, 2005, 77, 1092-1101.	2.6	139
30	Functional Dysregulation of CDC42 Causes Diverse Developmental Phenotypes. American Journal of Human Genetics, 2018, 102, 309-320.	2.6	138
31	A novel disorder involving dyshematopoiesis, inflammation, and HLH due to aberrant CDC42 function. Journal of Experimental Medicine, 2019, 216, 2778-2799.	4.2	132
32	Germ-line and somatic PTPN11 mutations in human disease. European Journal of Medical Genetics, 2005, 48, 81-96.	0.7	128
33	Detection of Bovine Mitochondrial DNA in Ruminant Feeds: A Molecular Approach to Test for the Presence of Bovine-Derived Materials. Journal of Food Protection, 1998, 61, 513-518.	0.8	123
34	Diversity, parental germline origin, and phenotypic spectrum of de novoHRASmissense changes in Costello syndrome. Human Mutation, 2007, 28, 265-272.	1.1	123
35	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
36	p.Arg1809Cys substitution in neurofibromin is associated with a distinctive NF1 phenotype without neurofibromas. European Journal of Human Genetics, 2015, 23, 1068-1071.	1.4	113

#	Article	IF	Citations
37	SHP-2 and myeloid malignancies. Current Opinion in Hematology, 2004, 11, 44-50.	1.2	106
38	Organoids as a new model for improving regenerative medicine and cancer personalized therapy in renal diseases. Cell Death and Disease, 2019, 10, 201.	2.7	105
39	Modeling medulloblastoma in vivo and with human cerebellar organoids. Nature Communications, 2020, 11, 583.	5.8	105
40	Mutations Impairing GSK3-Mediated MAF Phosphorylation Cause Cataract, Deafness, Intellectual Disability, Seizures, and a Down Syndrome-like Facies. American Journal of Human Genetics, 2015, 96, 816-825.	2.6	102
41	Genotypic and phenotypic characterization of Noonan syndrome: New data and review of the literature. American Journal of Medical Genetics, Part A, 2005, 134A, 165-170.	0.7	101
42	Cardiomyopathies in Noonan syndrome and the other RASopathies. Progress in Pediatric Cardiology, 2015, 39, 13-19.	0.2	99
43	SOS1 mutations in Noonan syndrome: molecular spectrum, structural insights on pathogenic effects, and genotype-phenotype correlations. Human Mutation, 2011, 32, 760-772.	1.1	97
44	Mutation of the receptor tyrosine phosphatase PTPRC (CD45) in T-cell acute lymphoblastic leukemia. Blood, 2012, 119, 4476-4479.	0.6	96
45	Mutations in PAX2 Associate with Adult-Onset FSGS. Journal of the American Society of Nephrology: JASN, 2014, 25, 1942-1953.	3.0	96
46	Decreased Proliferation and Altered Differentiation in Osteoblasts from Genetically and Clinically Distinct Craniosynostotic Disorders. American Journal of Pathology, 1999, 154, 1465-1477.	1.9	93
47	Disruption of the histone acetyltransferase MYST4 leads to a Noonan syndromeâ€“like phenotype and hyperactivated MAPK signaling in humans and mice. Journal of Clinical Investigation, 2011, 121, 3479-3491.	3.9	89
48	RAS signaling dysregulation in human embryonal Rhabdomyosarcoma. Genes Chromosomes and Cancer, 2009, 48, 975-982.	1.5	88
49	Noncanonical GLI1 signaling promotes stemness features and in vivo growth in lung adenocarcinoma. Oncogene, 2017, 36, 4641-4652.	2.6	86
50	Phosphatase-defective LEOPARD syndrome mutations in PTPN11 gene have gain-of-function effects during Drosophila development. Human Molecular Genetics, 2009, 18, 193-201.	1.4	82
51	Cognitive profile of disorders associated with dysregulation of the RAS/MAPK signaling cascade. American Journal of Medical Genetics, Part A, 2009, 149A, 140-146.	0.7	82
52	Diverse driving forces underlie the invariant occurrence of the T42A, E139D, I282V and T468M SHP2 amino acid substitutions causing Noonan and LEOPARD syndromes. Human Molecular Genetics, 2008, 17, 2018-2029.	1.4	79
53	Multiple giant cell lesions in patients with Noonan syndrome and cardio-facio-cutaneous syndrome. European Journal of Human Genetics, 2009, 17, 420-425.	1.4	79
54	Mutations in ZBTB20 cause Primrose syndrome. Nature Genetics, 2014, 46, 815-817.	9.4	79

#	Article	IF	Citations
55	Myeloid Dysregulation in a Human Induced Pluripotent Stem Cell Model of PTPN11 -Associated Juvenile Myelomonocytic Leukemia. Cell Reports, 2015, 13, 504-515.	2.9	79
56	Somatic PTPN11 mutations in childhood acute myeloid leukaemia. British Journal of Haematology, 2005, 129, 333-339.	1.2	78
57	Genotyping of an Italian papillary thyroid carcinoma cohort revealed high prevalence of BRAF mutations, absence of RAS mutations and allowed the detection of a new mutation of BRAF oncoprotein (BRAFV599Ins). Clinical Endocrinology, 2006, 64, 105-109.	1.2	77
58	A Restricted Spectrum of Mutations in the SMAD4 Tumor-Suppressor Gene Underlies Myhre Syndrome. American Journal of Human Genetics, 2012, 90, 161-169.	2.6	77
59	Paternal Germline Origin and Sex-Ratio Distortion in Transmission of PTPN11 Mutations in Noonan Syndrome. American Journal of Human Genetics, 2004, 75, 492-497.	2.6	76
60	PTPN2 negatively regulates oncogenic JAK1 in T-cell acute lymphoblastic leukemia. Blood, 2011, 117, 7090-7098.	0.6	76
61	The Interplay between CD27dull and CD27bright B Cells Ensures the Flexibility, Stability, and Resilience of Human B Cell Memory. Cell Reports, 2020, 30, 2963-2977.e6.	2.9	76
62	Cardiac defects, morbidity and mortality in patients affected by RASopathies. CARNET study results. International Journal of Cardiology, 2017, 245, 92-98.	0.8	75
63	Spectrum of MEK1 and MEK2 gene mutations in cardio-facio-cutaneous syndrome and genotypeâ€“phenotype correlations. European Journal of Human Genetics, 2009, 17, 733-740.	1.4	74
64	RASopathies: Clinical Diagnosis in the First Year of Life. Molecular Syndromology, 2010, 1, 282-289.	0.3	73
65	Mutations in KCNK4 that Affect Gating Cause a Recognizable Neurodevelopmental Syndrome. American Journal of Human Genetics, 2018, 103, 621-630.	2.6	73
66	RAF1 mutations in childhood-onset dilated cardiomyopathy. Nature Genetics, 2014, 46, 635-639.	9.4	69
67	A Competitive Polymerase Chain Reactionâ€“Based Approach for the Identification and Semiquantification of Mitochondrial DNA in Differently Heat-Treated Bovine Meat and Bone Meal. Journal of Food Protection, 2003, 66, 103-109.	0.8	67
68	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. Human Mutation, 2015, 36, 1080-1087.	1.1	67
69	Transgenic Drosophila models of Noonan syndrome causing PTPN11 gain-of-function mutations. Human Molecular Genetics, 2006, 15, 543-553.	1.4	66
70	Biallelic Mutations in TBCD , Encoding the Tubulin Folding Cofactor D, Perturb Microtubule Dynamics and Cause Early-Onset Encephalopathy. American Journal of Human Genetics, 2016, 99, 962-973.	2.6	66
71	Assessing the geneâ€“disease association of 19 genes with the RASopathies using the ClinGen gene curation framework. Human Mutation, 2018, 39, 1485-1493.	1.1	66
72	Acquired PTPN11 mutations occur rarely in adult patients with myelodysplastic syndromes and chronic myelomonocytic leukemia. Leukemia Research, 2005, 29, 459-462.	0.4	64

#	Article	IF	Citations
73	Behavioral Profile in RASopathies. American Journal of Medical Genetics, Part A, 2014, 164, 934-942.	0.7	64
74	CHK1-targeted therapy to deplete DNA replication-stressed, p53-deficient, hyperdiploid colorectal cancer stem cells. Gut, 2018, 67, 903-917.	6.1	64
75	Acute Lymphoblastic Leukemia-associated JAK1 Mutants Activate the Janus Kinase/STAT Pathway via Interleukin-9 Receptor Î± Homodimers. Journal of Biological Chemistry, 2009, 284, 6773-6781.	1.6	63
76	Phenotypic and genotypic characterisation of Noonan-like/multiple giant cell lesion syndrome. Journal of Medical Genetics, 2005, 42, e11-e11.	1.5	62
77	Heterozygous germline mutations in A2ML1 are associated with a disorder clinically related to Noonan syndrome. European Journal of Human Genetics, 2015, 23, 317-324.	1.4	61
78	Dominant Noonan syndrome-causing LZTR1 mutations specifically affect the Kelch domain substrate-recognition surface and enhance RAS-MAPK signaling. Human Molecular Genetics, 2019, 28, 1007-1022.	1.4	58
79	Biochemical and molecular characterization of the novel BRAFV599Ins mutation detected in a classic papillary thyroid carcinoma. Oncogene, 2006, 25, 4235-4240.	2.6	56
80	Childhood onset tubular aggregate myopathy associated with de novo STIM1 mutations. Journal of Neurology, 2014, 261, 870-876.	1.8	56
81	Phenotypic analysis of individuals with Costello syndrome due to HRAS p.G13C. , 2011, 155, 706-716.		55
82	Further delineation of an entity caused by CREBBP and EP300 mutations but not resembling Rubinsteinâ€“Taybi syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 862-876.	0.7	52
83	The impact of next-generation sequencing on the diagnosis of pediatric-onset hereditary spastic paraplegias: new genotype-phenotype correlations for rare HSP-related genes. Neurogenetics, 2018, 19, 111-121.	0.7	52
84	Jackson-Weiss syndrome: identification of two novel FGFR2 missense mutations shared with Crouzon and Pfeiffer craniosynostotic disorders. Human Genetics, 1997, 101, 47-50.	1.8	51
85	LYRM7mutations cause a multifocal cavitating leukoencephalopathy with distinct MRI appearance. Brain, 2016, 139, 782-794.	3.7	51
86	TBCE Mutations Cause Early-Onset Progressive Encephalopathy with Distal Spinal Muscular Atrophy. American Journal of Human Genetics, 2016, 99, 974-983.	2.6	49
87	RAS signaling pathway mutations and hypertrophic cardiomyopathy: getting into and out of the thick of it. Journal of Clinical Investigation, 2011, 121, 844-847.	3.9	49
88	Activating PTPN11 mutations play a minor role in pediatric and adult solid tumors. Cancer Genetics and Cytogenetics, 2006, 166, 124-129.	1.0	48
89	Cancer Stem Cell-Based Models of Colorectal Cancer Reveal Molecular Determinants of Therapy Resistance. Stem Cells Translational Medicine, 2016, 5, 511-523.	1.6	48
90	Aberrant Function of the C-Terminal Tail of HIST1H1E Accelerates Cellular Senescence and Causes Premature Aging. American Journal of Human Genetics, 2019, 105, 493-508.	2.6	48

#	Article	IF	Citations
91	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	2.6	48
92	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	2.6	48
93	Trp290Cys mutation in exon IIIa of the fibroblast growth factor receptor 2 (FGFR2) gene is associated with Pfeiffer syndrome. Human Genetics, 1997, 99, 602-606.	1.8	47
94	Craniosynostosis in patients with Noonan syndrome caused by germline KRAS mutations. American Journal of Medical Genetics, Part A, 2009, 149A, 1036-1040.	0.7	46
95	Absence of PTPN11 mutations in 28 cases of cardiofaciocutaneous (CFC) syndrome. Human Genetics, 2002, 111, 421-427.	1.8	45
96	Cooperating JAK1 and JAK3 mutants increase resistance to JAK inhibitors. Blood, 2014, 124, 3924-3931.	0.6	44
97	Clinical Presentation and Natural History of Hypertrophic Cardiomyopathy in RASopathies. Heart Failure Clinics, 2018, 14, 225-235.	1.0	44
98	Specific combinations of biallelic POLR3A variants cause Wiedemann-Rautenstrauch syndrome. Journal of Medical Genetics, 2018, 55, 837-846.	1.5	44
99	Prenatal features of Noonan syndrome: prevalence and prognostic value. Prenatal Diagnosis, 2011, 31, 949-954.	1.1	43
100	CREBBP mutations in individuals without Rubinsteinâ€“Taybi syndrome phenotype. American Journal of Medical Genetics, Part A, 2016, 170, 2681-2693.	0.7	43
101	Recessive Inactivating Mutations in TBCK, Encoding a Rab GTPase-Activating Protein, Cause Severe Infantile Syndromic Encephalopathy. American Journal of Human Genetics, 2016, 98, 772-781.	2.6	43
102	Activating Mutations of RRAS2 Are a Rare Cause of Noonan Syndrome. American Journal of Human Genetics, 2019, 104, 1223-1232.	2.6	43
103	Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	4.7	43
104	Tyr1068-phosphorylated epidermal growth factor receptor (EGFR) predicts cancer stem cell targeting by erlotinib in preclinical models of wild-type EGFR lung cancer. Cell Death and Disease, 2015, 6, e1850-e1850.	2.7	42
105	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders. Human Genetics and Genomics Advances, 2022, 3, 100075.	1.0	42
106	Distinct Acute Lymphoblastic Leukemia (ALL)-associated Janus Kinase 3 (JAK3) Mutants Exhibit Different Cytokine-Receptor Requirements and JAK Inhibitor Specificities. Journal of Biological Chemistry, 2015, 290, 29022-29034.	1.6	41
107	Loss of function of the E3 ubiquitin-protein ligase UBE3B causes Kaufman oculocerebrofacial syndrome. Journal of Medical Genetics, 2013, 50, 493-499.	1.5	40
108	Frameshift mutations at the C-terminus of HIST1H1E result in a specific DNA hypomethylation signature. Clinical Epigenetics, 2020, 12, 7.	1.8	40

#	Article	IF	Citations
109	Structural, Functional, and Clinical Characterization of a NovelPTPN11Mutation Cluster Underlying Noonan Syndrome. Human Mutation, 2017, 38, 451-459.	1.1	39
110	Congenital heart disease and genetic syndromes: new insights into molecular mechanisms. Expert Review of Molecular Diagnostics, 2017, 17, 861-870.	1.5	39
111	Not only dominant, not only optic atrophy: expanding the clinical spectrum associated with OPA1 mutations. Orphanet Journal of Rare Diseases, 2017, 12, 89.	1.2	39
112	Copy number variants in autism spectrum disorders. Progress in Neuro-Psychopharmacology and Biological Psychiatry, 2019, 92, 421-427.	2.5	39
113	Exclusion of PTPN11 mutations in Costello syndrome: further evidence for distinct genetic etiologies for Noonan, cardio-facio-cutaneous and Costello syndromes. Clinical Genetics, 2003, 63, 423-426.	1.0	38
114	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.	1.4	38
115	Novel mutations in IBA57 are associated with leukodystrophy and variable clinical phenotypes. Journal of Neurology, 2017, 264, 102-111.	1.8	38
116	Synonymous GATA2 mutations result in selective loss of mutated RNA and are common in patients with GATA2 deficiency. Leukemia, 2020, 34, 2673-2687.	3.3	38
117	De Novo VPS4A Mutations Cause Multisystem Disease with Abnormal Neurodevelopment. American Journal of Human Genetics, 2020, 107, 1129-1148.	2.6	38
118	Mutations in Fibronectin Cause a Subtype of Spondylometaphyseal Dysplasia with â€œCorner Fracturesâ€• American Journal of Human Genetics, 2017, 101, 815-823.	2.6	37
119	A new bioavailable fenretinide formulation with antiproliferative, antimetabolic, and cytotoxic effects on solid tumors. Cell Death and Disease, 2019, 10, 529.	2.7	37
120	SCUBE3 loss-of-function causes a recognizable recessive developmental disorder due to defective bone morphogenetic protein signaling. American Journal of Human Genetics, 2021, 108, 115-133.	2.6	37
121	Molecular Diversity and Associated Phenotypic Spectrum of GermlineCBLMutations. Human Mutation, 2015, 36, 787-796.	1.1	36
122	Genotype and phenotype spectrum of NRAS germline variants. European Journal of Human Genetics, 2017, 25, 823-831.	1.4	36
123	Wiedemannâ€“Rautenstrauch syndrome: A phenotype analysis. American Journal of Medical Genetics, Part A, 2017, 173, 1763-1772.	0.7	36
124	Counteracting Effects Operating on Src Homology 2 Domain-containing Protein-tyrosine Phosphatase 2 (SHP2) Function Drive Selection of the Recurrent Y62D and Y63C Substitutions in Noonan Syndrome*. Journal of Biological Chemistry, 2012, 287, 27066-27077.	1.6	35
125	Clinical, biochemical and molecular characterization of prosaposin deficiency. Clinical Genetics, 2016, 90, 220-229.	1.0	35
126	Understanding Growth Failure in Costello Syndrome: Increased Resting Energy Expenditure. Journal of Pediatrics, 2016, 170, 322-324.	0.9	35

#	Article	IF	Citations
127	Biallelic SQSTM1 mutations in early-onset, variably progressive neurodegeneration. Neurology, 2018, 91, e319-e330.	1.5	35
128	De Novo Variants Disturbing the Transactivation Capacity of POU3F3 Cause a Characteristic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 105, 403-412.	2.6	35
129	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nature Communications, 2020, 11, 595.	5.8	35
130	A mutation in PAK3 with a dual molecular effect deregulates the RAS/MAPK pathway and drives an X-linked syndromic phenotype. Human Molecular Genetics, 2014, 23, 3607-3617.	1.4	33
131	Gaucher disease due to saposin C deficiency is an inherited lysosomal disease caused by rapidly degraded mutant proteins. Human Molecular Genetics, 2014, 23, 5814-5826.	1.4	33
132	The phenotypic and molecular spectrum of PEHO syndrome and PEHO-like disorders. Brain, 2017, 140, e49-e49.	3.7	33
133	Hypertrophic Cardiomyopathy in RASopathies. Heart Failure Clinics, 2022, 18, 19-29.	1.0	33
134	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.	0.7	32
135	Structural and functional effects of disease-causing amino acid substitutions affecting residues Ala72 and Glu76 of the protein tyrosine phosphatase SHP-2. Proteins: Structure, Function and Bioinformatics, 2006, 66, 963-974.	1.5	31
136	The miRâ€139â€5p regulates proliferation of supratentorial paediatric lowâ€grade gliomas by targeting the PI3K/AKT/mTORC1 signalling. Neuropathology and Applied Neurobiology, 2018, 44, 687-706.	1.8	31
137	De Novo Missense Variants in FBXW11 Cause Diverse Developmental Phenotypes Including Brain, Eye, and Digit Anomalies. American Journal of Human Genetics, 2019, 105, 640-657.	2.6	31
138	Mutations in the <scp>IRBIT</scp> domain of <scp>ITPR1</scp> are a frequent cause of autosomal dominant nonprogressive congenital ataxia. Clinical Genetics, 2017, 91, 86-91.	1.0	30
139	Activating MRAS mutations cause Noonan syndrome associated with hypertrophic cardiomyopathy. Human Molecular Genetics, 2020, 29, 1772-1783.	1.4	30
140	Novel SMAD4 mutation causing Myhre syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 1835-1840.	0.7	29
141	Congenital immunodeficiency in an individual with Wiedemannâ€“Steiner syndrome due to a novel missense mutation in KMT2A. American Journal of Medical Genetics, Part A, 2016, 170, 2389-2393.	0.7	29
142	Defective kinesin binding of TUBB2A causes progressive spastic ataxia syndrome resembling sacsinopathy. Human Molecular Genetics, 2018, 27, 1892-1904.	1.4	29
143	Genomic duplication of PTPN11 is an uncommon cause of Noonan syndrome. American Journal of Medical Genetics, Part A, 2009, 149A, 2122-2128.	0.7	28
144	Expanding the clinical and molecular spectrum of PRMT7 mutations: 3 additional patients and review. Clinical Genetics, 2018, 93, 675-681.	1.0	28

#	Article	IF	Citations
145	Enhanced human brain associative plasticity in Costello syndrome. Journal of Physiology, 2010, 588, 3445-3456.	1.3	27
146	Protracted late infantile ceroid lipofuscinosis due to TPP1 mutations: Clinical, molecular and biochemical characterization in three sibs. Journal of the Neurological Sciences, 2015, 356, 65-71.	0.3	27
147	Identification of novel and hotspot mutations in the channel domain of ITPR1 in two patients with Gillespie syndrome. Gene, 2017, 628, 141-145.	1.0	27
148	Anti-Hypothalamus and Anti-Pituitary AutoÂantibodies in ROHHAD Syndrome: Additional Evidence Supporting an Autoimmune Etiopathogenesis. Hormone Research in Paediatrics, 2019, 92, 124-132.	0.8	27
149	Whole exome sequencing is necessary to clarify ID/DD cases with de novo copy number variants of uncertain significance: Two proofâ€ofâ€concept examples. American Journal of Medical Genetics, Part A, 2016, 170, 1772-1779.	0.7	26
150	Psychopathological features in Noonan syndrome. European Journal of Paediatric Neurology, 2018, 22, 170-177.	0.7	26
151	NBAS pathogenic variants: Defining the associated clinical and facial phenotype and genotypeâ€“phenotype correlations. Human Mutation, 2019, 40, 721-728.	1.1	26
152	Long Term Memory Profile of Disorders Associated with Dysregulation of the RAS-MAPK Signaling Cascade. Behavior Genetics, 2011, 41, 423-429.	1.4	25
153	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.	1.0	25
154	Role of DNA Methylation Profile in Diagnosing Astroblastoma: A Case Report and Literature Review. Frontiers in Genetics, 2019, 10, 391.	1.1	25
155	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. Genetics in Medicine, 2020, 22, 1338-1347.	1.1	25
156	A second cohort of CHD3 patients expands the molecular mechanisms known to cause Snijders Blok-Campeau syndrome. European Journal of Human Genetics, 2020, 28, 1422-1431.	1.4	25
157	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.	0.7	25
158	Hedgehog signaling reprograms hair follicle niche fibroblasts to a hyper-activated state. Developmental Cell, 2022, 57, 1758-1775.e7.	3.1	25
159	ALL-associated JAK1 mutations confer hypersensitivity to the antiproliferative effect of type I interferon. Blood, 2010, 115, 3287-3295.	0.6	24
160	Expanding the molecular diversity and phenotypic spectrum of glycerol 3â€phosphate dehydrogenase 1 deficiency. Journal of Inherited Metabolic Disease, 2016, 39, 689-695.	1.7	24
161	A novel mutation in <scp>NDUFB11</scp> unveils a new clinical phenotype associated with lactic acidosis and sideroblastic anemia. Clinical Genetics, 2017, 91, 441-447.	1.0	24
162	Neurobehavioral features in individuals with <scp>K</scp>abuki syndrome. Molecular Genetics & Genomic Medicine, 2018, 6, 322-331.	0.6	24

#	Article	IF	Citations
163	DNA Methylation Profiling for Diagnosing Undifferentiated Sarcoma with Capicua Transcriptional Receptor (CIC) Alterations. International Journal of Molecular Sciences, 2020, 21, 1818.	1.8	24
164	Differences in the prevalence of PTPN11 mutations in FAB M5 paediatric acute myeloid leukaemia. British Journal of Haematology, 2005, 130, 801-803.	1.2	23
165	Prevalence of Sequence Variants in the RAS-Mitogen Activated Protein Kinase Signaling Pathway in Pre-Adolescent Children With Hypertrophic Cardiomyopathy. Circulation: Cardiovascular Genetics, 2012, 5, 317-326.	5.1	23
166	Expanding the phenotypic spectrum of truncating POGZ mutations: Association with CNS malformations, skeletal abnormalities, and distinctive facial dysmorphism. American Journal of Medical Genetics, Part A, 2017, 173, 1965-1969.	0.7	23
167	Clinical spectrum of Kabukiâ€like syndrome caused by <scp>HNRNPK</scp> haploinsufficiency. Clinical Genetics, 2018, 93, 401-407.	1.0	23
168	Biallelic mutations in <scp>DYNC2LI1</scp> are a rare cause of Ellisâ€van Creveld syndrome. Clinical Genetics, 2018, 93, 632-639.	1.0	23
169	A Recurrent Gain-of-Function Mutation in CLCN6, Encoding the ClC-6 Clâ¯/H+-Exchanger, Causes Early-Onset Neurodegeneration. American Journal of Human Genetics, 2020, 107, 1062-1077.	2.6	23
170	VarGenius executes cohort-level DNA-seq variant calling and annotation and allows to manage the resulting data through a PostgreSQL database. BMC Bioinformatics, 2018, 19, 477.	1.2	23
171	SPRED2 loss-of-function causes a recessive Noonan syndrome-like phenotype. American Journal of Human Genetics, 2021, 108, 2112-2129.	2.6	23
172	Atrioventricular canal defect in patients with RASopathies. European Journal of Human Genetics, 2013, 21, 200-204.	1.4	22
173	Decreased bone mineral density in Costello syndrome. Molecular Genetics and Metabolism, 2014, 111, 41-45.	0.5	22
174	Bi-allelic Variants in the GPI Transamidase Subunit PIGK Cause a Neurodevelopmental Syndrome with Hypotonia, Cerebellar Atrophy, and Epilepsy. American Journal of Human Genetics, 2020, 106, 484-495.	2.6	22
175	Childhood-onset dystonia-causing KMT2B variants result in a distinctive genomic hypermethylation profile. Clinical Epigenetics, 2021, 13, 157.	1.8	22
176	Cyclosporine attenuates cardiomyocyte hypertrophy induced by RAF1 mutants in Noonan and LEOPARD syndromes. Journal of Molecular and Cellular Cardiology, 2011, 51, 4-15.	0.9	21
177	Hydrops fetalis in a preterm newborn heterozygous for the c.4A>G SHOC2 mutation. American Journal of Medical Genetics, Part A, 2014, 164, 1015-1020.	0.7	21
178	Novel SEC61Gâ€“EGFR Fusion Gene in Pediatric Ependymomas Discovered by Clonal Expansion of Stem Cells in Absence of Exogenous Mitogens. Cancer Research, 2017, 77, 5860-5872.	0.4	21
179	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.	0.7	21
180	Low-Grade Gliomas in Patients with Noonan Syndrome: Case-Based Review of the Literature. Diagnostics, 2020, 10, 582.	1.3	21

#	Article	IF	Citations
181	Acute lymphoblastic leukaemia in Noonan syndrome. British Journal of Haematology, 2006, 133, 448-450.	1.2	20
182	Visual Function in Noonan and LEOPARD Syndrome. Neuropediatrics, 2008, 39, 335-340.	0.3	20
183	Phenotypic variability associated with the invariant SHOC2 c.4A>G (p.Ser2Gly) missense mutation. American Journal of Medical Genetics, Part A, 2014, 164, 3120-3125.	0.7	20
184	Atrioventricular canal defect and genetic syndromes: The unifying role of sonic hedgehog. Clinical Genetics, 2019, 95, 268-276.	1.0	20
185	Musculo-skeletal phenotype of Costello syndrome and cardio-facio-cutaneous syndrome: insights on the functional assessment status. Orphanet Journal of Rare Diseases, 2021, 16, 43.	1.2	20
186	MEK Inhibition in a Newborn with RAF1-Associated Noonan Syndrome Ameliorates Hypertrophic Cardiomyopathy but Is Insufficient to Revert Pulmonary Vascular Disease. Genes, 2022, 13, 6.	1.0	20
187	POGZâ€related epilepsy: Case report and review of the literature. American Journal of Medical Genetics, Part A, 2019, 179, 1631-1636.	0.7	19
188	Upfront treatment with <scp>mTOR</scp> inhibitor everolimus in pediatric lowâ€grade gliomas: A singleâ€center experience. International Journal of Cancer, 2021, 148, 2522-2534.	2.3	19
189	Biallelic mutations in the homeodomain of NKX6-2 underlie a severe hypomyelinating leukodystrophy. Brain, 2017, 140, 2550-2556.	3.7	18
190	Expanding the clinical spectrum associated with PACS2 mutations. Clinical Genetics, 2019, 95, 525-531.	1.0	18
191	Clinical and functional characterization of a novel RASopathyâ€causingSHOC2mutation associated with prenatalâ€onset hypertrophic cardiomyopathy. Human Mutation, 2019, 40, 1046-1056.	1.1	18
192	Bi-allelic LoF NRROS Variants Impairing Active TGF-Î²1 Delivery Cause a Severe Infantile-Onset Neurodegenerative Condition with Intracranial Calcification. American Journal of Human Genetics, 2020, 106, 559-569.	2.6	18
193	Primrose syndrome: Characterization of the phenotype in 42 patients. Clinical Genetics, 2020, 97, 890-901.	1.0	18
194	Fibroblast growth factor receptor mutational screening in newborns affected by metopic synostosis. Child's Nervous System, 1999, 15, 389-394.	0.6	17
195	PTPN11 mutations in childhood acute lymphoblastic leukemia occur as a secondary event associated with high hyperdiploidy. Leukemia, 2010, 24, 232-235.	3.3	17
196	SHOC2 subcellular shuttling requires the KEKE motif-rich region andN-terminal leucine-rich repeat domain and impacts on ERK signalling. Human Molecular Genetics, 2016, 25, 3824-3835.	1.4	17
197	De novo p.T362R mutation in MORC2 causes early onset cerebellar ataxia, axonal polyneuropathy and nocturnal hypoventilation. Brain, 2017, 140, e34-e34.	3.7	17
198	Neurotransmitter trafficking defect in a patient with clathrin (CLTC) variation presenting with intellectual disability and early-onset parkinsonism. Parkinsonism and Related Disorders, 2019, 61, 207-210.	1.1	17

#	Article	IF	Citations
199	Infantile-Onset Syndromic Cerebellar Ataxia and CACNA1G Mutations. Pediatric Neurology, 2020, 104, 40-45.	1.0	17
200	Structural Determinants of Phosphopeptide Binding to the N-Terminal Src Homology 2 Domain of the SHP2 Phosphatase. Journal of Chemical Information and Modeling, 2020, 60, 3157-3171.	2.5	17
201	Atypical cardiac defects in patients with RASopathies: Updated data on CARNET study. Birth Defects Research, 2020, 112, 725-731.	0.8	17
202	Variants of SOS2 are a rare cause of Noonan syndrome with particular predisposition for lymphatic complications. European Journal of Human Genetics, 2021, 29, 51-60.	1.4	17
203	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.	1.1	17
204	Rare and de novo coding variants in chromodomain genes in Chiari I malformation. American Journal of Human Genetics, 2021, 108, 100-114.	2.6	17
205	Targeting Oncogenic Src Homology 2 Domain-Containing Phosphatase 2 (SHP2) by Inhibiting Its Proteinâ€“Protein Interactions. Journal of Medicinal Chemistry, 2021, 64, 15973-15990.	2.9	17
206	Mutations at the C-terminus of CDC42 cause distinct hematopoietic and autoinflammatory disorders. Journal of Allergy and Clinical Immunology, 2022, 150, 223-228.	1.5	17
207	A Competitive PCR-Based Method to Measure Human Fibroblast Growth Factor Receptor 1â€“4 (FGFR1â€“4) Gene Expression. DNA and Cell Biology, 2001, 20, 367-379.	0.9	16
208	Congenital heart defects in Noonan syndrome and RIT1 mutation. Genetics in Medicine, 2016, 18, 1320.	1.1	16
209	TARP syndrome: Long-term survival, anatomic patterns of congenital heart defects, differential diagnosis and pathogenetic considerations. European Journal of Medical Genetics, 2019, 62, 103534.	0.7	16
210	Co-occurring WARS2 and CHRNA6 mutations in a child with a severe form of infantile parkinsonism. Parkinsonism and Related Disorders, 2020, 72, 75-79.	1.1	16
211	A genotype-first approach to exploring Mendelian cardiovascular traits with clear external manifestations. Genetics in Medicine, 2021, 23, 94-102.	1.1	16
212	Natural history and life-threatening complications in Myhre syndrome and review of the literature. European Journal of Pediatrics, 2016, 175, 1307-1315.	1.3	15
213	Heterozygous missense mutations in NFATC1 are associated with atrioventricular septal defect. Human Mutation, 2018, 39, 1428-1441.	1.1	15
214	Isoform-specific NF1 mRNA levels correlate with disease severity in Neurofibromatosis type 1. Orphanet Journal of Rare Diseases, 2019, 14, 261.	1.2	15
215	De novo DHDDS variants cause a neurodevelopmental and neurodegenerative disorder with myoclonus. Brain, 2022, 145, 208-223.	3.7	15
216	Rat nicastrin gene: cDNA isolation, mRNA variants and expression pattern analysis. Molecular Brain Research, 2005, 136, 12-22.	2.5	14

#	Article	IF	Citations
217	GH Therapy and first final height data in Noonanâ€like syndrome with loose anagen hair (Mazzanti) Tj ETQq1 1 0.784314 rgBT /Overlock	0.7	14
218	Noonan syndromeâ€like disorder with loose anagen hair: A second case with neuroblastoma. American Journal of Medical Genetics, Part A, 2015, 167, 1902-1907.	0.7	14
219	AberrantHRAStranscript processing underlies a distinctive phenotype within the RASopathy clinical spectrum. Human Mutation, 2017, 38, 798-804.	1.1	14
220	Exome sequencing in children of women with skewed X-inactivation identifies atypical cases and complex phenotypes. European Journal of Paediatric Neurology, 2017, 21, 475-484.	0.7	14
221	Colorectal cancer spheroid biobanks: multi-level approaches to drug sensitivity studies. Cell Biology and Toxicology, 2018, 34, 459-469.	2.4	14
222	Developmental and epileptic encephalopathy due to SZT2 genomic variants: Emerging features of a syndromic condition. Epilepsy and Behavior, 2020, 108, 107097.	0.9	14
223	Analysis of three RFLPs of the COL1A2 (Type I Collagen) in the Amhara and the Oromo of Ethiopia. Annals of Human Biology, 2002, 29, 432-441.	0.4	13
224	Clinical lumping and molecular splitting of LEOPARD and NF1/NF1-Noonan syndromes. American Journal of Medical Genetics, Part A, 2007, 143A, 1009-1011.	0.7	13
225	Prevalence, Type, and Molecular Spectrum of NF1 Mutations in Patients with Neurofibromatosis Type 1 and Congenital Heart Disease. Genes, 2019, 10, 675.	1.0	13
226	Mitochondrial and Peroxisomal Alterations Contribute to Energy Dysmetabolism in Riboflavin Transporter Deficiency. Oxidative Medicine and Cellular Longevity, 2020, 2020, 1-19.	1.9	13
227	Dissecting the Role of PCDH19 in Clustering Epilepsy by Exploiting Patient-Specific Models of Neurogenesis. Journal of Clinical Medicine, 2021, 10, 2754.	1.0	13
228	Epilepsy and BRAF Mutations: Phenotypes, Natural History and Genotype-Phenotype Correlations. Genes, 2021, 12, 1316.	1.0	13
229	Recurrent de novo missense variants across multiple histone H4 genes underlie a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 750-758.	2.6	13
230	Induced Pluripotent Stem Cells (iPSCs) and Gene Therapy: A New Era for the Treatment of Neurological Diseases. International Journal of Molecular Sciences, 2021, 22, 13674.	1.8	13
231	Dystonia in Costello syndrome. Parkinsonism and Related Disorders, 2012, 18, 798-800.	1.1	12
232	Transcriptional hallmarks of noonan syndrome and noonanâ€like syndrome with loose anagen hair. Human Mutation, 2012, 33, 703-709.	1.1	12
233	A PTPN11 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
234	Functional evaluation of natural killer cell cytotoxic activity in NFKB2-mutated patients. Immunology Letters, 2018, 194, 40-43.	1.1	12

#	Article	IF	Citations
235	Co-occurrence of mutations in KIF7 and KIAA0556 in Joubert syndrome with ocular coloboma, pituitary malformation and growth hormone deficiency: a case report and literature review. BMC Pediatrics, 2020, 20, 120.	0.7	12
236	Melanotic Neuroectodermal Tumor of Infancy (MNTI) and Pineal Anlage Tumor (PAT) Harbor A Medulloblastoma Signature by DNA Methylation Profiling. Cancers, 2021, 13, 706.	1.7	12
237	Clinical and molecular characterization of patients with adenylosuccinate lyase deficiency. Orphanet Journal of Rare Diseases, 2021, 16, 112.	1.2	12
238	Cardiac Defects and Genetic Syndromes: Old Uncertainties and New Insights. Genes, 2021, 12, 1047.	1.0	12
239	DICER1-associated malignancies mimicking germ cell neoplasms: Report of two cases and review of the literature. Pathology Research and Practice, 2021, 225, 153553.	1.0	12
240	BCM-95 and (2-hydroxypropyl)-Î²-cyclodextrin reverse autophagy dysfunction and deplete stored lipids in Sap C-deficient fibroblasts. Human Molecular Genetics, 2015, 24, 4198-4211.	1.4	11
241	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
242	Alterations in metabolic patterns have a key role in diagnosis and progression of primrose syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 1896-1902.	0.7	11
243	Expanding the histopathological spectrum of CFL2â€related myopathies. Clinical Genetics, 2018, 93, 1234-1239.	1.0	11
244	Clinical and functional characterization of two novel ZBTB20 mutations causing Primrose syndrome. Human Mutation, 2018, 39, 959-964.	1.1	11
245	Mitochondrial Abnormalities in Induced Pluripotent Stem Cells-Derived Motor Neurons from Patients with Riboflavin Transporter Deficiency. Antioxidants, 2020, 9, 1252.	2.2	11
246	Defining the phenotype of FHF1 developmental and epileptic encephalopathy. Epilepsia, 2020, 61, e71-e78.	2.6	11
247	Biallelic mutations in RNF220 cause laminopathies featuring leukodystrophy, ataxia and deafness. Brain, 2021, 144, 3020-3035.	3.7	11
248	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
249	Management of cardiac aspects in children with Noonan syndrome â€“ results from a European clinical practice survey among paediatric cardiologists. European Journal of Medical Genetics, 2021, 65, 104372.	0.7	11
250	Neonatal Manifestations of Chronic Granulomatous Disease: MAS/HLH and Necrotizing Pneumonia as Unusual Phenotypes and Review of the Literature. Journal of Clinical Immunology, 2022, 42, 299-311.	2.0	11
251	EcoRI, RsaI, and MspI RFLPs of the COL1A2 gene (type I collagen) in the Cayapa, a Native American population of Ecuador. Human Biology, 1994, 66, 979-89.	0.4	11
252	Genetic characterization of the Cayapa Indians of Ecuador and their genetic relationships to other Native American populations. Human Biology, 1994, 66, 299-322.	0.4	11

#	Article	IF	Citations
253	Incremental net benefit of whole genome sequencing for newborns and children with suspected genetic disorders: Systematic review and meta-analysis of cost-effectiveness evidence. Health Policy, 2022, 126, 337-345.	1.4	11
254	Induction of Both CD8+ and CD4+ T-Cellâ€“Mediated Responses in Colorectal Cancer Patients by Colon Antigen-1. Clinical Cancer Research, 2008, 14, 7292-7303.	3.2	10
255	Increased Sleep Spindle Activity in Patients With Costello Syndrome (HRAS Gene Mutation). Journal of Clinical Neurophysiology, 2011, 28, 314-318.	0.9	10
256	The activating p.Ser466Arg change in STAT1 causes a peculiar phenotype with features of interferonopathies. Clinical Genetics, 2019, 96, 585-589.	1.0	10
257	Biallelic Variants in the Nuclear Pore Complex Protein NUP93 Are Associated with Non-progressive Congenital Ataxia. Cerebellum, 2019, 18, 422-432.	1.4	10
258	Skeletal abnormalities are common features in AymÃ©â€Gripp syndrome. Clinical Genetics, 2020, 97, 362-369.	1.0	10
259	Biallelic TRNT1 variants in a child with B cell immunodeficiency, periodic fever and developmental delay without sideroblastic anemia (SIFD variant). Immunology Letters, 2020, 225, 64-65.	1.1	10
260	The seventh international <scp>RASopathies</scp> symposium: Pathways to a cureâ€”expanding knowledge, enhancing research, and therapeutic discovery. American Journal of Medical Genetics, Part A, 2022, 188, 1915-1927.	0.7	10
261	Geroderma osteodysplastica maps to a 4 Mb locus on chromosome 1q24. American Journal of Medical Genetics, Part A, 2008, 146A, 3034-3037.	0.7	9
262	Reactive Oxygen Species and Epidermal Growth Factor Are Antagonistic Cues Controlling SHP-2 Dimerization. Molecular and Cellular Biology, 2012, 32, 1998-2009.	1.1	9
263	A mild form of adenylosuccinate lyase deficiency in absence of typical brain MRI features diagnosed by whole exome sequencing. Italian Journal of Pediatrics, 2017, 43, 65.	1.0	9
264	Functional analysis of TLK2 variants and their proximal interactomes implicates impaired kinase activity and chromatin maintenance defects in their pathogenesis. Journal of Medical Genetics, 2022, 59, 170-179.	1.5	9
265	Ethnobotany of dye plants in Southern Italy, Mediterranean Basin: floristic catalog and two centuries of analysis of traditional botanical knowledge heritage. Journal of Ethnobiology and Ethnomedicine, 2020, 16, 31.	1.1	9
266	Copy number variation analysis implicates novel pathways in patients with oculoâ€auriculoâ€vertebralâ€spectrum and congenital heart defects. Clinical Genetics, 2021, 100, 268-279.	1.0	9
267	Red-Cell Enzyme Polymorphisms in the Reggio Calabria Province (Italy). Human Heredity, 1990, 40, 308-310.	0.4	8
268	Hyperthrophic cardiomyopathy and thePTPN11 gene. American Journal of Medical Genetics, Part A, 2005, 136A, 93-94.	0.7	8
269	Early fetal death associated with compound heterozygosity for Noonan syndrome-causativePTPN11 mutations. American Journal of Medical Genetics, Part A, 2007, 143A, 1249-1252.	0.7	8
270	A syndromic extreme insulin resistance caused by biallelic POC1A mutations in exon 10. European Journal of Endocrinology, 2017, 177, K21-K27.	1.9	8

#	Article	IF	Citations
271	Whole exome sequencing in an Italian family with isolated maxillary canine agenesis and canine eruption anomalies. Archives of Oral Biology, 2018, 91, 96-102.	0.8	8
272	A Recurrent Pathogenic Variant of INPP5K Underlies Autosomal Recessive Congenital Muscular Dystrophy With Cataracts and Intellectual Disability: Evidence for a Founder Effect in Southern Italy. Frontiers in Genetics, 2020, 11, 565868.	1.1	8
273	Antioxidant Amelioration of Riboflavin Transporter Deficiency in Motoneurons Derived from Patient-Specific Induced Pluripotent Stem Cells. International Journal of Molecular Sciences, 2020, 21, 7402.	1.8	8
274	Clinical Utility of a Unique Genome-Wide DNA Methylation Signature for KMT2A-Related Syndrome. International Journal of Molecular Sciences, 2022, 23, 1815.	1.8	8
275	Rapid communication: nucleotide sequence of porcine and ovine tRNA(Lys) and ATPase8 mitochondrial genes.. Journal of Animal Science, 1998, 76, 2207.	0.2	7
276	Obsessive Compulsive Symptoms and Psychopathological Profile in Children and Adolescents with KBG Syndrome. Brain Sciences, 2019, 9, 313.	1.1	7
277	Cost-effectiveness of exome sequencing: an Italian pilot study on undiagnosed patients. New Genetics and Society, 2019, 38, 249-263.	0.7	7
278	Phenotypic Features of Epidermolysis Bullosa Simplex due to KLHL24 Mutations in 3 Italian Cases. Acta Dermato-Venereologica, 2019, 99, 238-239.	0.6	7
279	Common atrium/atrioventricular canal defect and postaxial polydactyly: A mild clinical subtype of Ellisâ€van Creveld syndrome caused by hypomorphic mutations in the EVC gene. Human Mutation, 2020, 41, 2087-2093.	1.1	7
280	CantÃº syndrome versus Zimmermann-Laband syndrome: Report of nine individuals with ABCC9 variants. European Journal of Medical Genetics, 2020, 63, 103996.	0.7	7
281	The clinical significance of A2ML1 variants in Noonan syndrome has to be reconsidered. European Journal of Human Genetics, 2021, 29, 524-527.	1.4	7
282	Characterization of bone homeostasis in individuals affected by cardioâ€facioâ€cutaneous syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 414-421.	0.7	7
283	Expanding the molecular spectrum of pathogenicSHOC2variants underlying Mazzanti syndrome. Human Molecular Genetics, 2022, 31, 2766-2778.	1.4	7
284	Complex Presentation of Hao-Fountain Syndrome Solved by Exome Sequencing Highlighting Co-Occurring Genomic Variants. Genes, 2022, 13, 889.	1.0	7
285	Multidisciplinary Management of Costello Syndrome: Current Perspectives. Journal of Multidisciplinary Healthcare, 0, Volume 15, 1277-1296.	1.1	7
286	Rapid communication: nucleotide sequence of chamois, alpine ibex, and red deer tRNA(Lys) and ATPase8 mitochondrial genes.. Journal of Animal Science, 1999, 77, 3398.	0.2	6
287	Loss of <scp>CBL</scp> E3â€ligase activity in Bâ€lineage childhood acute lymphoblastic leukaemia. British Journal of Haematology, 2012, 159, 115-119.	1.2	6
288	Data on cardiac defects, morbidity and mortality in patients affected by RASopathies. CARNET study results. Data in Brief, 2018, 16, 649-654.	0.5	6

#	Article	IF	Citations
289	7q11.23 Microduplication Syndrome: Clinical and Neurobehavioral Profiling. Brain Sciences, 2020, 10, 839.	1.1	6
290	Melanocytic nevi in RASopathies: insights on dermatological diagnostic handles. Journal of the European Academy of Dermatology and Venereology, 2021, 35, e83-e85.	1.3	6
291	Expanding the clinical phenotype of the ultraâ€rare <scp>Skrabanâ€Deardorff</scp> syndrome: Two novel individuals with <scp>WDR26</scp> lossâ€ofâ€function variants and a literature review. American Journal of Medical Genetics, Part A, 2021, 185, 1712-1720.	0.7	6
292	Etanercept as a successful therapy in autoinflammatory syndrome related to TRNT1 mutations: a case-based review. Clinical Rheumatology, 2021, 40, 4341-4348.	1.0	6
293	Clinical variability of neurofibromatosis 1: A modifying role of cooccurring <scp>PTPN11</scp> variants and atypical brain <scp>MRI</scp> findings. Clinical Genetics, 2021, 100, 563-572.	1.0	6
294	Hyperactive HRAS dysregulates energetic metabolism in fibroblasts from patients with Costello syndrome via enhanced production of reactive oxidizing species. Human Molecular Genetics, 2022, 31, 561-575.	1.4	6
295	HIPK2-T566 autophosphorylation diversely contributes to UV- and doxorubicin-induced HIPK2 activation. Oncotarget, 2017, 8, 16744-16754.	0.8	6
296	Management of growth failure and other endocrine aspects in patients with Noonan syndrome across Europe: A sub-analysis of a European clinical practice survey. European Journal of Medical Genetics, 2022, 65, 104404.	0.7	6
297	Biallelic variants in <scp>ZNF142</scp> lead to a syndromic neurodevelopmental disorder. Clinical Genetics, 2022, 102, 98-109.	1.0	6
298	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
299	Pediatric patients with RASopathy-associated hypertrophic cardiomyopathy: the multifaceted consequences of PTPN11 mutations. Orphanet Journal of Rare Diseases, 2019, 14, 163.	1.2	5
300	Refinement of the clinical and mutational spectrum of <scp>UBE2A</scp> deficiency syndrome. Clinical Genetics, 2020, 98, 172-178.	1.0	5
301	Altered cytoskeletal arrangement in induced pluripotent stem cells and motor neurons from patients with riboflavin transporter deficiency. DMM Disease Models and Mechanisms, 2021, 14, .	1.2	5
302	Adducted Thumb and Peripheral Polyneuropathy: Diagnostic Supports in Suspecting Whiteâ€“Sutton Syndrome: Case Report and Review of the Literature. Genes, 2021, 12, 950.	1.0	5
303	RASopathies and hemostatic abnormalities: key role of platelet dysfunction. Orphanet Journal of Rare Diseases, 2021, 16, 499.	1.2	5
304	Metabolic profiling of Costello syndrome: Insights from a single-center cohort. European Journal of Medical Genetics, 2022, 65, 104439.	0.7	5
305	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.	0.7	5
306	New Insights into the Neurodegeneration Mechanisms Underlying Riboflavin Transporter Deficiency (RTD): Involvement of Energy Dysmetabolism and Cytoskeletal Derangement. Biomedicines, 2022, 10, 1329.	1.4	5

#	Article	IF	Citations
307	The MAP3K7 gene: Further delineation of clinical characteristics and genotype/phenotype correlations. Human Mutation, 2022, 43, 1377-1395.	1.1	5
308	Worldwide distribution of phosphoglucomutase 1 (PGM1) polymorphism detected by isoelectric focusing: A review. International Journal of Anthropology, 1994, 9, 81-112.	0.1	4
309	Polymorphism at position 882 of the fibroblast growth factor receptor 3 (FGFR3) gene detected by SSCP analysis. Molecular and Cellular Probes, 1998, 12, 335-337.	0.9	4
310	Somatic mosaicism represents an underestimated event underlying collagen 6-related disorders. European Journal of Paediatric Neurology, 2017, 21, 873-883.	0.7	4
311	Very mild isolated intellectual disability caused by adenylosuccinate lyase deficiency: a new phenotype. Molecular Genetics and Metabolism Reports, 2020, 23, 100592.	0.4	4
312	Defining language disorders in children and adolescents with Noonan Syndrome. Molecular Genetics & Genomic Medicine, 2020, 8, e1069.	0.6	4
313	â€œAtypicalâ€•Krabbe disease in two siblings harboring biallelic GALC mutations including a deep intronic variant. European Journal of Human Genetics, 2022, , .	1.4	4
314	Potassium Channel KCNH1 Activating Variants Cause Altered Functional and Morphological Ciliogenesis. Molecular Neurobiology, 2022, 59, 4825-4838.	1.9	4
315	Progressive extreme heterotopic calcification. American Journal of Medical Genetics, Part A, 2013, 161, 1706-1713.	0.7	3
316	Visual perception skills: a comparison between patients with <scp>N</scp>oonan syndrome and 22q11.2 deletion syndrome. Genes, Brain and Behavior, 2017, 16, 627-634.	1.1	3
317	Congenital myopathy with protein aggregates and nemaline bodies related to CFL2 mutations. Neuromuscular Disorders, 2017, 27, S186.	0.3	3
318	No metagenomic evidence of tumorigenic viruses in cancers from a selected cohort of immunosuppressed subjects. Scientific Reports, 2019, 9, 19815.	1.6	3
319	Further insight into the neurobehavioral pattern of children carrying the 2p16.3 heterozygous deletion involving NRXN1 : Report of five new cases. Genes, Brain and Behavior, 2020, 19, e12687.	1.1	3
320	Pathogenic PTPN11 variants involving the polyâ€glutamine Gln ²⁵⁵ â€Gln ²⁵⁶ â€Gln ²⁵⁷ stretch highlight the relevance of helix B in SHP2's functional regulation. Human Mutation, 2020, 41, 1171-1182.	1.1	3
321	Manic and Depressive Symptoms in Children Diagnosed with Noonan Syndrome. Brain Sciences, 2021, 11, 233.	1.1	3
322	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
323	Broadening the phenotypic spectrum of Beta3GalT6 â€associated phenotypes. American Journal of Medical Genetics, Part A, 2021, 185, 3153-3160.	0.7	3
324	Co-Occurring Heterozygous CNOT3 and SMAD6 Truncating Variants: Unusual Presentation and Refinement of the IDDSADF Phenotype. Genes, 2021, 12, 1009.	1.0	3

#	Article	IF	Citations
325	KCNK18 Biallelic Variants Associated with Intellectual Disability and Neurodevelopmental Disorders Alter TRESK Channel Activity. International Journal of Molecular Sciences, 2021, 22, 6064.	1.8	3
326	European Medical Education Initiative on Noonan syndrome: A clinical practice survey assessing the diagnosis and clinical management of individuals with Noonan syndrome across Europe. European Journal of Medical Genetics, 2021, 65, 104371.	0.7	3
327	Prevalence of bladder cancer in Costello syndrome: New insights to drive clinical decisionâ€making. Clinical Genetics, 2022, 101, 454-458.	1.0	3
328	Genetic heterogeneity among the Hindus and their relationships with the other â€œCaucasoidâ€• populations: New data on Punjab-Haryana and Rajasthan Indian States. American Journal of Physical Anthropology, 1995, 98, 257-273.	2.1	2
329	Efficient one-step chromatographic purification and functional characterization of recombinant human Saposin C. Protein Expression and Purification, 2011, 78, 209-215.	0.6	2
330	Embryopathy Following Maternal Biliopancreatic Diversion: Is Bariatric Surgery Really Safe?. Obesity Surgery, 2021, 31, 445-450.	1.1	2
331	Cognitive and Adaptive Characterization of Children and Adolescents with KBG Syndrome: An Explorative Study. Journal of Clinical Medicine, 2021, 10, 1523.	1.0	2
332	Enlarged spinal nerve roots in RASopathies: Report of two cases. European Journal of Medical Genetics, 2021, 64, 104187.	0.7	2
333	Posterior Lissencephaly Associated with Subcortical Band Heterotopia Due to a Variation in the CEP85L Gene: A Case Report and Refining of the Phenotypic Spectrum. Genes, 2021, 12, 1208.	1.0	2
334	A Rare Case of Brachyolmia with Amelogenesis Imperfecta Caused by a New Pathogenic Splicing Variant in LTBP3. Genes, 2021, 12, 1406.	1.0	2
335	Recognition Memory in Noonan Syndrome. Brain Sciences, 2021, 11, 169.	1.1	2
336	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
337	<scp>SHP2</scp>'s gainâ€ofâ€function in <scp>Werner</scp> syndrome causes childhood disease onset likely resulting from negative genetic interaction. Clinical Genetics, 2022, 102, 12-21.	1.0	2
338	Posterior fossa ependymoma in neurodevelopmental syndrome caused by a de novo germline pathogenic Polr2a variant. American Journal of Medical Genetics, Part A, 0, , .	0.7	2
339	Brain Abnormalities in Patients with Germline Variants in H3F3: Novel Imaging Findings and Neurologic Symptoms Beyond Somatic Variants and Brain Tumors. American Journal of Neuroradiology, 2022, 43, 1048-1053.	1.2	2
340	PLEC gene mutations cause familial disto-proximal myopathy and long QT syndrome mimicking mitochondrial disease. Neuromuscular Disorders, 2017, 27, S150-S151.	0.3	1
341	FRI0540â€…A NOVEL AUTOINFLAMMATORY DISEASE CHARACTERIZED BY NEONATAL-ONSET CYTOPENIA WITH AUTOINFLAMMATION, RASH, AND HEMOPHAGOCYTOSIS (NOCARH) DUE TO ABERRANT CDC42 FUNCTION. , 2019, , .		1
342	PTPN11 Mutational Spectrum in Juvenile Myelomonocytic Leukemia and Noonan Syndrome.. Blood, 2004, 104, 3417-3417.	0.6	1

#	Article	IF	Citations
343	A survey of six genetic markers on the populations of Punjab and Rajasthan (India). Gene Geography: A Computerized Bulletin on Human Gene Frequencies, 1991, 5, 113-21.	0.1	1
344	Modeling PCDH19-CE: From 2D Stem Cell Model to 3D Brain Organoids. International Journal of Molecular Sciences, 2022, 23, 3506.	1.8	1
345	MiRLog and dbmiR: Prioritization and functional annotation tools to study human microRNA sequence variants. Human Mutation, 2022, , .	1.1	1
346	Characterization of Cognitive, Language and Adaptive Profiles of Children and Adolescents with Malan Syndrome. Journal of Clinical Medicine, 2022, 11, 4078.	1.0	1
347	RASopathies and sigmoid-shaped ventricular septum morphology: evidence of a previously unappreciated cardiac phenotype. Pediatric Research, 2023, 93, 752-754.	1.1	1
348	Germline PTPN11 mutation affecting exon 8 in a case of syndromic juvenile myelomonocytic leukemia. Leukemia Research, 2011, 35, e13-e14.	0.4	0
349	Distal spinal muscular atrophy and ataxia with cerebellar atrophy in two unrelated patients; a new phenotypic variant of HRD and recessive KCS syndrome related to TBCE. Neuromuscular Disorders, 2015, 25, S222.	0.3	0
350	Noonan Syndrome and Other RAS/MAPK Pathway Syndromes. , 0, , 122-130.		0
351	Identification of novel SEC61G-EGFR fusions in pediatric ependymoma. European Journal of Cancer, 2016, 69, S51.	1.3	0
352	Front Cover, Volume 40, Issue 6. Human Mutation, 2019, 40, i.	1.1	0
353	FRI0539â€…WNT6 MUTATION CAUSES AN EARLY ONSET GRANULOMATOSUS INTESTINAL DISEASE WITH RECURRENT HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS (HLH). , 2019, , .		0
354	TMOD-05. GENOME-WIDE DNA METHYLATION PROFILE: A POWERFUL STRATEGY TO RECAPITULATE HETEROGENEITY OF PEDIATRIC BRAIN TUMORS IN PRIMARY CELL LINES. Neuro-Oncology, 2021, 23, i36-i36.	0.6	0
355	PTPN11 and RAS Gene Mutation Pattern Identifies an Unique Feature of Upregulated RAS Function in Infant ALL.. Blood, 2004, 104, 996-996.	0.6	0
356	Abstract 2912: Protein pathway activation mapping of leukemia-associated JAK1 mutants. , 2011, , .		0
357	Measuring the Confluence of iPSCs using an Automated Imaging System.. Journal of Visualized Experiments, 2020, , .	0.2	0
358	MODL-23. DNA METHYLATION AND COPY NUMBER VARIATION PROFILE FOR CHARACTERIZATION OF PEDIATRIC BRAIN TUMOR PRIMARY CELL LINES. Neuro-Oncology, 2020, 22, iii415-iii415.	0.6	0
359	Elucidating the clinical spectrum and molecular basis of HYAL2 deficiency. Genetics in Medicine, 2022, 24, 631-644.	1.1	0
360	Toward the inÂvitro understanding of iPSC nucleoskeletal and cytoskeletal biology, and their relevance for organoid development. , 2022, , 137-150.		0

#	Article	IF	Citations
361	ESD, GLO1, PGD, PGM1 and PGM2 gene frequencies in the Salerno Province (Italy). Gene Geography: A Computerized Bulletin on Human Gene Frequencies, 1991, 5, 103-6.	0.1	0
362	Linkage disequilibrium at the human phosphoglucomutase 1 locus. Human Biology, 1994, 66, 669-81.	0.4	0