Marco Tartaglia

List of Publications by Year in Descending Order

328 papers	15,161 citations	60 h-index	115 g-index
374 ext. papers	18,228 ext. citations	6.1 avg, IF	6 L-index

#	Paper	IF	Citations
328	Toward the in vitro understanding of iPSC nucleoskeletal and cytoskeletal biology, and their relevance for organoid development **2022**, 137-150		
327	Prevalence of bladder cancer in Costello syndrome: new insights to drive clinical decision-making.. *Clinical Genetics*, **2022**,	4	1
326	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	2.6	1
325	Metabolic profiling of Costello syndrome: Insights from a single-center cohort.. *European Journal of Medical Genetics*, **2022**, 104439	2.6	0
324	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders.. *Human Genetics and Genomics Advances*, **2022**, 3, 100075	0.8	1
323	Hypertrophic Cardiomyopathy in RASopathies: Diagnosis, Clinical Characteristics, Prognostic Implications, and Management. *Heart Failure Clinics*, **2022**, 18, 19-29	3.3	2
322	The seventh international RASopathies symposium: Pathways to a cure-expanding knowledge, enhancing research, and therapeutic discovery.. *American Journal of Medical Genetics, Part A*, **2022**,	2.5	2
321	Complex Presentation of Hao-Fountain Syndrome Solved by Exome Sequencing Highlighting Co-Occurring Genomic Variants. *Genes*, **2022**, 13, 889	4.2	0
320	Characterization of bone homeostasis in individuals affected by cardio-facio-cutaneous syndrome. *American Journal of Medical Genetics, Part A*, **2021**, 188, 414	2.5	1
319	RASopathies and hemostatic abnormalities: key role of platelet dysfunction. *Orphanet Journal of Rare Diseases*, **2021**, 16, 499	4.2	0
318	Risk of autoimmune diseases in patients with RASopathies: systematic study of humoral and cellular immunity. *Orphanet Journal of Rare Diseases*, **2021**, 16, 410	4.2	0
317	Genotype-cardiac phenotype correlations in a large single-center cohort of patients affected by RASopathies: Clinical implications and literature review. *American Journal of Medical Genetics, Part A*, **2021**,	2.5	2
316	SPRED2 loss-of-function causes a recessive Noonan syndrome-like phenotype. *American Journal of Human Genetics*, **2021**, 108, 2112-2129	11	2
315	Targeting Oncogenic Src Homology 2 Domain-Containing Phosphatase 2 (SHP2) by Inhibiting Its Protein-Protein Interactions. *Journal of Medicinal Chemistry*, **2021**, 64, 15973-15990	8.3	4
314	Management of cardiac aspects in children with Noonan syndrome - results from a European clinical practice survey among paediatric cardiologists. *European Journal of Medical Genetics*, **2021**, 65, 104372	2.6	2
313	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	2.6	0
312	Neonatal Manifestations of Chronic Granulomatous Disease: MAS/HLH and Necrotizing Pneumonia as Unusual Phenotypes and Review of the Literature. *Journal of Clinical Immunology*, **2021**, 1	5.7	1

311	Etanercept as a successful therapy in autoinflammatory syndrome related to TRNT1 mutations: a case-based review. *Clinical Rheumatology*, **2021**, 40, 4341-4348	3.9	2
310	Clinical and molecular characterization of patients with adenylosuccinate lyase deficiency. *Orphanet Journal of Rare Diseases*, **2021**, 16, 112	4.2	2
309	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	11	12
308	Enlarged spinal nerve roots in RASopathies: Report of two cases. *European Journal of Medical Genetics*, **2021**, 64, 104187	2.6	0
307	Copy number variation analysis implicates novel pathways in patients with oculo-auriculo-vertebral-spectrum and congenital heart defects. *Clinical Genetics*, **2021**, 100, 268-279	4	2
306	Biallelic mutations in RNF220 cause laminopathies featuring leukodystrophy, ataxia and deafness. *Brain*, **2021**, 144, 3020-3035	11.2	1
305	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	5.7	0
304	Dissecting the Role of PCDH19 in Clustering Epilepsy by Exploiting Patient-Specific Models of Neurogenesis. *Journal of Clinical Medicine*, **2021**, 10,	5.1	3
303	Broadening the phenotypic spectrum of Beta3GalT6-associated phenotypes. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3153-3160	2.5	0
302	Biallelic Variants Associated with Intellectual Disability and Neurodevelopmental Disorders Alter TRESK Channel Activity. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	1
301	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	1	78
300	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	5.3	11
299	Embryopathy Following Maternal Biliopancreatic Diversion: Is Bariatric Surgery Really Safe?. *Obesity Surgery*, **2021**, 31, 445-450	3.7	2
298	Melanocytic nevi in RASopathies: insights on dermatological diagnostic handles. *Journal of the European Academy of Dermatology and Venereology*, **2021**, 35, e83-e85	4.6	3
297	A genotype-first approach to exploring Mendelian cardiovascular traits with clear external manifestations. *Genetics in Medicine*, **2021**, 23, 94-102	8.1	4
296	The clinical significance of A2ML1 variants in Noonan syndrome has to be reconsidered. *European Journal of Human Genetics*, **2021**, 29, 524-527	5.3	2
295	De novo DHDDS variants cause a neurodevelopmental and neurodegenerative disorder with myoclonus. *Brain*, **2021**,	11.2	2
294	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	11	8

293	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	4.2	7
292	Melanotic Neuroectodermal Tumor of Infancy (MNTI) and Pineal Anlage Tumor (PAT) Harbor A Medulloblastoma Signature by DNA Methylation Profiling. *Cancers*, **2021**, 13,	6.6	4
291	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	8.1	5
290	Expanding the clinical phenotype of the ultra-rare Skraban-Deardorff syndrome: Two novel individuals with WDR26 loss-of-function variants and a literature review. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1712-1720	2.5	1
289	Cardiac Defects and Genetic Syndromes: Old Uncertainties and New Insights. *Genes*, **2021**, 12,	4.2	5
288	Clinical variability of neurofibromatosis 1: A modifying role of cooccurring PTPN11 variants and atypical brain MRI findings. *Clinical Genetics*, **2021**, 100, 563-572	4	2
287	Epilepsy and Mutations: Phenotypes, Natural History and Genotype-Phenotype Correlations. *Genes*, **2021**, 12,	4.2	1
286	Childhood-onset dystonia-causing KMT2B variants result in a distinctive genomic hypermethylation profile. *Clinical Epigenetics*, **2021**, 13, 157	7.7	0
285	A Rare Case of Brachyolmia with Amelogenesis Imperfecta Caused by a New Pathogenic Splicing Variant in. *Genes*, **2021**, 12,	4.2	0
284	DICER1-associated malignancies mimicking germ cell neoplasms: Report of two cases and review of the literature. *Pathology Research and Practice*, **2021**, 225, 153553	3.4	3
283	Hyperactive HRAS dysregulates energetic metabolism in fibroblasts from patients with Costello syndrome via enhanced production of reactive oxidizing species. *Human Molecular Genetics*, **2021**,	5.6	2
282	Rare and de novo coding variants in chromodomain genes in Chiari I malformation. *American Journal of Human Genetics*, **2021**, 108, 100-114	11	3
281	Compound heterozygosity for PTPN11 variants in a subject with Noonan syndrome provides insights into the mechanism of SHP2-related disorders. *Clinical Genetics*, **2021**, 99, 457-461	4	2
280	Induced Pluripotent Stem Cells (iPSCs) and Gene Therapy: A New Era for the Treatment of Neurological Diseases.. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	3
279	Bone tissue homeostasis and risk of fractures in Costello syndrome: A 4-year follow-up study.. *American Journal of Medical Genetics, Part A*, **2021**,	2.5	1
278	Refinement of the clinical and mutational spectrum of UBE2A deficiency syndrome. *Clinical Genetics*, **2020**, 98, 172-178	4	2
277	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. *Genetics in Medicine*, **2020**, 22, 1338-1347	8.1	9
276	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	5.3	10

275	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	6.1	6
274	Developmental and epileptic encephalopathy due to SZT2 genomic variants: Emerging features of a syndromic condition. *Epilepsy and Behavior*, **2020**, 108, 107097	3.2	2
273	Very mild isolated intellectual disability caused by adenylosuccinate lyase deficiency: a new phenotype. *Molecular Genetics and Metabolism Reports*, **2020**, 23, 100592	1.8	3
272	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	3.9	6
271	Atypical cardiac defects in patients with RASopathies: Updated data on CARNET study. *Birth Defects Research*, **2020**, 112, 725-731	2.9	6
270	Synonymous GATA2 mutations result in selective loss of mutated RNA and are common in patients with GATA2 deficiency. *Leukemia*, **2020**, 34, 2673-2687	10.7	18
269	The Interplay between CD27 and CD27 B Cells Ensures the Flexibility, Stability, and Resilience of Human B Cell Memory. *Cell Reports*, **2020**, 30, 2963-2977.e6	10.6	43
268	Bi-allelic LoF NRROS Variants Impairing Active TGF-1 Delivery Cause a Severe Infantile-Onset Neurodegenerative Condition with Intracranial Calcification. *American Journal of Human Genetics*, **2020**, 106, 559-569	11	7
267	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	2.6	3
266	DNA Methylation Profiling for Diagnosing Undifferentiated Sarcoma with Capicua Transcriptional Receptor () Alterations. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	10
265	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	4.1	6
264	Cant syndrome versus Zimmermann-Laband syndrome: Report of nine individuals with ABCC9 variants. *European Journal of Medical Genetics*, **2020**, 63, 103996	2.6	2
263	Defining the phenotype of FHF1 developmental and epileptic encephalopathy. *Epilepsia*, **2020**, 61, e71-e78	6.4	2
262	Pathogenic PTPN11 variants involving the poly-glutamine Gln -Gln -Gln stretch highlight the relevance of helix B in SHP2 functional regulation. *Human Mutation*, **2020**, 41, 1171-1182	4.7	2
261	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. *American Journal of Human Genetics*, **2020**, 106, 356-370	11	51
260	Defining language disorders in children and adolescents with Noonan Syndrome. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1069	2.3	1
259	Co-occurring WARS2 and CHRNA6 mutations in a child with a severe form of infantile parkinsonism. *Parkinsonism and Related Disorders*, **2020**, 72, 75-79	3.6	9
258	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	11	16

257	Primrose syndrome: Characterization of the phenotype in 42 patients. *Clinical Genetics*, **2020**, 97, 890-901	4	5
256	MODL-23. DNA METHYLATION AND COPY NUMBER VARIATION PROFILE FOR CHARACTERIZATION OF PEDIATRIC BRAIN TUMOR PRIMARY CELL LINES. *Neuro-Oncology*, **2020**, 22, iii415-iii415	1	78
255	De Novo VPS4A Mutations Cause Multisystem Disease with Abnormal Neurodevelopment. *American Journal of Human Genetics*, **2020**, 107, 1129-1148	11	11
254	Modeling medulloblastoma in vivo and with human cerebellar organoids. *Nature Communications*, **2020**, 11, 583	17.4	54
253	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. *Nature Communications*, **2020**, 11, 595	17.4	18
252	Skeletal abnormalities are common features in Aym□Gripp syndrome. *Clinical Genetics*, **2020**, 97, 362-369	4	4
251	Frameshift mutations at the C-terminus of HIST1H1E result in a specific DNA hypomethylation signature. *Clinical Epigenetics*, **2020**, 12, 7	7.7	23
250	Infantile-Onset Syndromic Cerebellar Ataxia and CACNA1G Mutations. *Pediatric Neurology*, **2020**, 104, 40-45	2.9	12
249	A Recurrent Pathogenic Variant of 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	4.5	2
248	Antioxidant Amelioration of Riboflavin Transporter Deficiency in Motoneurons Derived from Patient-Specific Induced Pluripotent Stem Cells. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	2
247	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	5.3	132
246	A Recurrent Gain-of-Function Mutation in CLCN6, Encoding the ClC-6 Cl/H-Exchanger, Causes Early-Onset Neurodegeneration. *American Journal of Human Genetics*, **2020**, 107, 1062-1077	11	7
245	Histone H3.3 beyond cancer: Germline mutations in cause a previously unidentified neurodegenerative disorder in 46 patients. *Science Advances*, **2020**, 6,	14.3	12
244	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. *American Journal of Human Genetics*, **2020**, 107, 499-513	11	25
243	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	3.6	1
242	Mitochondrial and Peroxisomal Alterations Contribute to Energy Dysmetabolism in Riboflavin Transporter Deficiency. *Oxidative Medicine and Cellular Longevity*, **2020**, 2020, 6821247	6.7	6
241	Common atrium/atrioventricular canal defect and postaxial polydactyly: A mild clinical subtype of Ellis-van Creveld syndrome caused by hypomorphic mutations in the EVC gene. *Human Mutation*, **2020**, 41, 2087-2093	4.7	2
240	Low-Grade Gliomas in Patients with Noonan Syndrome: Case-Based Review of the Literature. *Diagnostics*, **2020**, 10,	3.8	3

239	Functional analysis of variants and their proximal interactomes implicates impaired kinase activity and chromatin maintenance defects in their pathogenesis. *Journal of Medical Genetics*, **2020**,	5.8	2
238	Mitochondrial Abnormalities in Induced Pluripotent Stem Cells-Derived Motor Neurons from Patients with Riboflavin Transporter Deficiency. *Antioxidants*, **2020**, 9,	7.1	2
237	Activating MRAS mutations cause Noonan syndrome associated with hypertrophic cardiomyopathy. *Human Molecular Genetics*, **2020**, 29, 1772-1783	5.6	14
236	Upfront treatment with mTOR inhibitor everolimus in pediatric low-grade gliomas: A single-center experience. *International Journal of Cancer*, **2020**, 148, 2522	7.5	5
235	The activating p.Ser466Arg change in STAT1 causes a peculiar phenotype with features of interferonopathies. *Clinical Genetics*, **2019**, 96, 585-589	4	3
234	Prevalence, Type, and Molecular Spectrum of Mutations in Patients with Neurofibromatosis Type 1 and Congenital Heart Disease. *Genes*, **2019**, 10,	4.2	8
233	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	11	30
232	Expanding the clinical spectrum associated with PACS2 mutations. *Clinical Genetics*, **2019**, 95, 525-531	4	10
231	Role of DNA Methylation Profile in Diagnosing Astroblastoma: A Case Report and Literature Review. *Frontiers in Genetics*, **2019**, 10, 391	4.5	14
230	POGZ-related epilepsy: Case report and review of the literature. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1631-1636	2.5	10
229	Activating Mutations of RRAS2 Are a Rare Cause of Noonan Syndrome. *American Journal of Human Genetics*, **2019**, 104, 1223-1232	11	23
228	Clinical and functional characterization of a novel RASopathy-causing SHOC2 mutation associated with prenatal-onset hypertrophic cardiomyopathy. *Human Mutation*, **2019**, 40, 1046-1056	4.7	6
227	Anti-Hypothalamus and Anti-Pituitary Auto-antibodies in ROHHAD Syndrome: Additional Evidence Supporting an Autoimmune Etiopathogenesis. *Hormone Research in Paediatrics*, **2019**, 92, 124-132	3.3	15
226	Cost-effectiveness of exome sequencing: an Italian pilot study on undiagnosed patients. *New Genetics and Society*, **2019**, 38, 249-263	1.9	2
225	NBAS pathogenic variants: Defining the associated clinical and facial phenotype and genotype-phenotype correlations. *Human Mutation*, **2019**, 40, 721-728	4.7	15
224	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	2.5	11
223	Copy number variants in autism spectrum disorders. *Progress in Neuro-Psychopharmacology and Biological Psychiatry*, **2019**, 92, 421-427	5.5	16
222	Biallelic Variants in the Nuclear Pore Complex Protein NUP93 Are Associated with Non-progressive Congenital Ataxia. *Cerebellum*, **2019**, 18, 422-432	4.3	7

221	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	11	16
220	Pediatric patients with RASopathy-associated hypertrophic cardiomyopathy: the multifaceted consequences of PTPN11 mutations. *Orphanet Journal of Rare Diseases*, **2019**, 14, 163	4.2	3
219	De Novo Variants Disturbing the Transactivation Capacity of POU3F3 Cause a Characteristic Neurodevelopmental Disorder. *American Journal of Human Genetics*, **2019**, 105, 403-412	11	17
218	A new bioavailable fenretinide formulation with antiproliferative, antimetabolic, and cytotoxic effects on solid tumors. *Cell Death and Disease*, **2019**, 10, 529	9.8	23
217	A novel disorder involving dyshematopoiesis, inflammation, and HLH due to aberrant CDC42 function. *Journal of Experimental Medicine*, **2019**, 216, 2778-2799	16.6	71
216	Obsessive Compulsive Symptoms and Psychopathological Profile in Children and Adolescents with KBG syndrome. *Brain Sciences*, **2019**, 9,	3.4	1
215	Isoform-specific NF1 mRNA levels correlate with disease severity in Neurofibromatosis type 1. *Orphanet Journal of Rare Diseases*, **2019**, 14, 261	4.2	11
214	Organoids as a new model for improving regenerative medicine and cancer personalized therapy in renal diseases. *Cell Death and Disease*, **2019**, 10, 201	9.8	61
213	No metagenomic evidence of tumorigenic viruses in cancers from a selected cohort of immunosuppressed subjects. *Scientific Reports*, **2019**, 9, 19815	4.9	
212	TARP syndrome: Long-term survival, anatomic patterns of congenital heart defects, differential diagnosis and pathogenetic considerations. *European Journal of Medical Genetics*, **2019**, 62, 103534	2.6	6
211	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	5.6	38
210	Phenotypic Features of Epidermolysis Bullosa Simplex due to KLHL24 Mutations in 3 Italian Cases. *Acta Dermato-Venereologica*, **2019**, 99, 238-239	2.2	4
209	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	3.6	9
208	Atrioventricular canal defect and genetic syndromes: The unifying role of sonic hedgehog. *Clinical Genetics*, **2019**, 95, 268-276	4	16
207	Further delineation of an entity caused by CREBBP and EP300 mutations but not resembling Rubinstein-Taybi syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 862-876	2.5	29
206	Colorectal cancer spheroid biobanks: multi-level approaches to drug sensitivity studies. *Cell Biology and Toxicology*, **2018**, 34, 459-469	7.4	12
205	The miR-139-5p regulates proliferation of supratentorial paediatric low-grade gliomas by targeting the PI3K/AKT/mTORC1 signalling. *Neuropathology and Applied Neurobiology*, **2018**, 44, 687-706	5.2	24
204	Functional Dysregulation of CDC42 Causes Diverse Developmental Phenotypes. *American Journal of Human Genetics*, **2018**, 102, 309-320	11	85

203	Functional evaluation of natural killer cell cytotoxic activity in NFKB2-mutated patients. *Immunology Letters*, **2018**, 194, 40-43	4.1	9
202	Data on cardiac defects, morbidity and mortality in patients affected by RASopathies. CARNET study results. *Data in Brief*, **2018**, 16, 649-654	1.2	5
201	Expanding the histopathological spectrum of CFL2-related myopathies. *Clinical Genetics*, **2018**, 93, 1234-1239	4	7
200	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	3	28
199	Whole exome sequencing in an Italian family with isolated maxillary canine agenesis and canine eruption anomalies. *Archives of Oral Biology*, **2018**, 91, 96-102	2.8	3
198	Neurobehavioral features in individuals with Kabuki syndrome. *Molecular Genetics & Genomic Medicine*, **2018**, 6, 322-331	2.3	17
197	Clinical Presentation and Natural History of Hypertrophic Cardiomyopathy in RASopathies. *Heart Failure Clinics*, **2018**, 14, 225-235	3.3	24
196	Defective kinesin binding of TUBB2A causes progressive spastic ataxia syndrome resembling sacsinopathy. *Human Molecular Genetics*, **2018**, 27, 1892-1904	5.6	18
195	CHK1-targeted therapy to deplete DNA replication-stressed, p53-deficient, hyperdiploid colorectal cancer stem cells. *Gut*, **2018**, 67, 903-917	19.2	45
194	Clinical spectrum of Kabuki-like syndrome caused by HNRNPK haploinsufficiency. *Clinical Genetics*, **2018**, 93, 401-407	4	16
193	Psychopathological features in Noonan syndrome. *European Journal of Paediatric Neurology*, **2018**, 22, 170-177	3.8	13
192	Biallelic mutations in DYNC2LI1 are a rare cause of Ellis-van Creveld syndrome. *Clinical Genetics*, **2018**, 93, 632-639	4	16
191	Expanding the clinical and molecular spectrum of PRMT7 mutations: 3 additional patients and review. *Clinical Genetics*, **2018**, 93, 675-681	4	18
190	Heterozygous missense mutations in NFATC1 are associated with atrioventricular septal defect. *Human Mutation*, **2018**, 39, 1428-1441	4.7	10
189	Clinical and functional characterization of two novel ZBTB20 mutations causing Primrose syndrome. *Human Mutation*, **2018**, 39, 959-964	4.7	9
188	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	3.6	9
187	Mutations in KCNK4 that Affect Gating Cause a Recognizable Neurodevelopmental Syndrome. *American Journal of Human Genetics*, **2018**, 103, 621-630	11	45
186	Assessing the gene-disease association of 19 genes with the RASopathies using the ClinGen gene curation framework. *Human Mutation*, **2018**, 39, 1485-1493	4.7	39

185	Specific combinations of biallelic variants cause Wiedemann-Rautenstrauch syndrome. *Journal of Medical Genetics*, **2018**, 55, 837-846	5.8	31
184	Biallelic mutations in early-onset, variably progressive neurodegeneration. *Neurology*, **2018**, 91, e319-e330	5.5	26
183	A novel mutation in NDUFB11 unveils a new clinical phenotype associated with lactic acidosis and sideroblastic anemia. *Clinical Genetics*, **2017**, 91, 441-447	4	18
182	Mutations in the IRBIT domain of ITPR1 are a frequent cause of autosomal dominant nonprogressive congenital ataxia. *Clinical Genetics*, **2017**, 91, 86-91	4	26
181	Microcephaly, intractable seizures and developmental delay caused by biallelic variants in TBCD: further delineation of a new chaperone-mediated tubulinopathy. *Clinical Genetics*, **2017**, 91, 725-738	4	18
180	Structural, Functional, and Clinical Characterization of a Novel PTPN11 Mutation Cluster Underlying Noonan Syndrome. *Human Mutation*, **2017**, 38, 451-459	4.7	32
179	Aberrant HRAS transcript processing underlies a distinctive phenotype within the RASopathy clinical spectrum. *Human Mutation*, **2017**, 38, 798-804	4.7	11
178	De novo p.T362R mutation in MORC2 causes early onset cerebellar ataxia, axonal polyneuropathy and nocturnal hypoventilation. *Brain*, **2017**, 140, e34	11.2	8
177	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	2.5	8
176	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	2.5	17
175	Genotype and phenotype spectrum of NRAS germline variants. *European Journal of Human Genetics* , **2017**, 25, 823-831	5.3	23
174	Wiedemann-Rautenstrauch syndrome: A phenotype analysis. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1763-1772	2.5	23
173	A Specific Mutational Signature Associated with DNA 8-Oxoguanine Persistence in MUTYH-defective Colorectal Cancer. *EBioMedicine*, **2017**, 20, 39-49	8.8	112
172	Noncanonical GLI1 signaling promotes stemness features and in vivo growth in lung adenocarcinoma. *Oncogene*, **2017**, 36, 4641-4652	9.2	58
171	Visual perception skills: a comparison between patients with Noonan syndrome and 22q11.2 deletion syndrome. *Genes, Brain and Behavior*, **2017**, 16, 627-634	3.6	1
170	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	3.8	6
169	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. *American Journal of Human Genetics*, **2017**, 101, 664-685	11	214
168	Biallelic mutations in the homeodomain of NKX6-2 underlie a severe hypomyelinating leukodystrophy. *Brain*, **2017**, 140, 2550-2556	11.2	15

167	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	3.2	7
166	Congenital heart disease and genetic syndromes: new insights into molecular mechanisms. *Expert Review of Molecular Diagnostics*, **2017**, 17, 861-870	3.8	26
165	Somatic mosaicism represents an underestimated event underlying collagen 6-related disorders. *European Journal of Paediatric Neurology*, **2017**, 21, 873-883	3.8	4
164	Cardiac defects, morbidity and mortality in patients affected by RASopathies. CARNET study results. *International Journal of Cardiology*, **2017**, 245, 92-98	3.2	48
163	A syndromic extreme insulin resistance caused by biallelic mutations in exon 10. *European Journal of Endocrinology*, **2017**, 177, K21-K27	6.5	6
162	Novel - Fusion Gene in Pediatric Ependymomas Discovered by Clonal Expansion of Stem Cells in Absence of Exogenous Mitogens. *Cancer Research*, **2017**, 77, 5860-5872	10.1	14
161	Congenital myopathy with protein aggregates and nemaline bodies related to CFL2 mutations. *Neuromuscular Disorders*, **2017**, 27, S186	2.9	3
160	Mutations in Fibronectin Cause a Subtype of Spondylometaphyseal Dysplasia with "Corner Fractures". *American Journal of Human Genetics*, **2017**, 101, 815-823	11	22
159	Identification of novel and hotspot mutations in the channel domain of ITPR1 in two patients with Gillespie syndrome. *Gene*, **2017**, 628, 141-145	3.8	20
158	The phenotypic and molecular spectrum of PEHO syndrome and PEHO-like disorders. *Brain*, **2017**, 140, e49	11.2	11
157	Not only dominant, not only optic atrophy: expanding the clinical spectrum associated with OPA1 mutations. *Orphanet Journal of Rare Diseases*, **2017**, 12, 89	4.2	28
156	Novel mutations in IBA57 are associated with leukodystrophy and variable clinical phenotypes. *Journal of Neurology*, **2017**, 264, 102-111	5.5	27
155	HIPK2-T566 autophosphorylation diversely contributes to UV- and doxorubicin-induced HIPK2 activation. *Oncotarget*, **2017**, 8, 16744-16754	3.3	5
154	SHOC2 subcellular shuttling requires the KEKE motif-rich region and N-terminal leucine-rich repeat domain and impacts on ERK signalling. *Human Molecular Genetics*, **2016**, 25, 3824-3835	5.6	11
153	Natural history and life-threatening complications in Myhre syndrome and review of the literature. *European Journal of Pediatrics*, **2016**, 175, 1307-15	4.1	12
152	Cancer Stem Cell-Based Models of Colorectal Cancer Reveal Molecular Determinants of Therapy Resistance. *Stem Cells Translational Medicine*, **2016**, 5, 511-23	6.9	35
151	Whole exome sequencing is necessary to clarify ID/DD cases with de novo copy number variants of uncertain significance: Two proof-of-concept examples. *American Journal of Medical Genetics, Part A*, **2016**, 170, 1772-9	2.5	18
150	Clinical, biochemical and molecular characterization of prosaposin deficiency. *Clinical Genetics*, **2016**, 90, 220-9	4	26

149	LYRM7 mutations cause a multifocal cavitating leukoencephalopathy with distinct MRI appearance. *Brain*, **2016**, 139, 782-94	11.2	44
148	Understanding Growth Failure in Costello Syndrome: Increased Resting Energy Expenditure. *Journal of Pediatrics*, **2016**, 170, 322-4	3.6	24
147	Differential Effects of HRAS Mutation on LTP-Like Activity Induced by Different Protocols of Repetitive Transcranial Magnetic Stimulation. *Brain Stimulation*, **2016**, 9, 33-8	5.1	9
146	Congenital immunodeficiency in an individual with Wiedemann-Steiner syndrome due to a novel missense mutation in KMT2A. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2389-93	2.5	24
145	Expanding the molecular diversity and phenotypic spectrum of glycerol 3-phosphate dehydrogenase 1 deficiency. *Journal of Inherited Metabolic Disease*, **2016**, 39, 689-695	5.4	18
144	CREBBP mutations in individuals without Rubinstein-Taybi syndrome phenotype. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2681-93	2.5	28
143	Recessive Inactivating Mutations in TBCK, Encoding a Rab GTPase-Activating Protein, Cause Severe Infantile Syndromic Encephalopathy. *American Journal of Human Genetics*, **2016**, 98, 772-81	11	29
142	Congenital heart defects in Noonan syndrome and RIT1 mutation. *Genetics in Medicine*, **2016**, 18, 1320	8.1	15
141	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	11	55
140	TBCE Mutations Cause Early-Onset Progressive Encephalopathy with Distal Spinal Muscular Atrophy. *American Journal of Human Genetics*, **2016**, 99, 974-983	11	37
139	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	3.2	21
138	Cardiomyopathies in Noonan syndrome and the other RASopathies. *Progress in Pediatric Cardiology*, **2015**, 39, 13-19	0.4	67
137	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, 816-25	11	75
136	Molecular Diversity and Associated Phenotypic Spectrum of Germline CBL Mutations. *Human Mutation*, **2015**, 36, 787-96	4.7	22
135	Mutations in KCNH1 and ATP6V1B2 cause Zimmermann-Laband syndrome. *Nature Genetics*, **2015**, 47, 661-7	36.3	128
134	Myeloid Dysregulation in a Human Induced Pluripotent Stem Cell Model of PTPN11-Associated Juvenile Myelomonocytic Leukemia. *Cell Reports*, **2015**, 13, 504-515	10.6	52
133	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	9.8	31
132	p.Arg1809Cys substitution in neurofibromin is associated with a distinctive NF1 phenotype without neurofibromas. *European Journal of Human Genetics*, **2015**, 23, 1068-71	5.3	89

131	Heterozygous germline mutations in A2ML1 are associated with a disorder clinically related to Noonan syndrome. *European Journal of Human Genetics*, **2015**, 23, 317-24	5.3	48
130	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. *Human Mutation*, **2015**, 36, 1080-7	4.7	51
129	Response to long-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**, 167A, 2786-94	2.5	17
128	Noonan syndrome-like disorder with loose anagen hair: a second case with neuroblastoma. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 1902-7	2.5	7
127	Behavioral phenotype in Costello syndrome with atypical mutation: a case report. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2015**, 168B, 66-71	3.5	5
126	BCM-95 and (2-hydroxypropyl)-Ecyclodextrin reverse autophagy dysfunction and deplete stored lipids in Sap C-deficient fibroblasts. *Human Molecular Genetics*, **2015**, 24, 4198-211	5.6	7
125	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, 29022-34	5.4	29
124	Childhood onset tubular aggregate myopathy associated with de novo STIM1 mutations. *Journal of Neurology*, **2014**, 261, 870-6	5.5	41
123	Behavioral profile in RASopathies. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 934-42	2.5	42
122	Hydrops fetalis in a preterm newborn heterozygous for the c.4A>G SHOC2 mutation. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 1015-20	2.5	19
121	RAF1 mutations in childhood-onset dilated cardiomyopathy. *Nature Genetics*, **2014**, 46, 635-639	36.3	54
120	Gaucher disease due to saposin C deficiency is an inherited lysosomal disease caused by rapidly degraded mutant proteins. *Human Molecular Genetics*, **2014**, 23, 5814-26	5.6	24
119	Mutations in ZBTB20 cause Primrose syndrome. *Nature Genetics*, **2014**, 46, 815-7	36.3	61
118	Novel SMAD4 mutation causing Myhre syndrome. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 1835-40	2.5	23
117	Decreased bone mineral density in Costello syndrome. *Molecular Genetics and Metabolism*, **2014**, 111, 41-5	3.7	15
116	Cooperating JAK1 and JAK3 mutants increase resistance to JAK inhibitors. *Blood*, **2014**, 124, 3924-31	2.2	32
115	Phenotypic variability associated with the invariant SHOC2 c.4A>G (p.Ser2Gly) missense mutation. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 3120-5	2.5	15
114	Mutations in PAX2 associate with adult-onset FSGS. *Journal of the American Society of Nephrology: JASN*, **2014**, 25, 1942-53	12.7	76

113	Activating mutations in RRAS underlie a phenotype within the RASopathy spectrum and contribute to leukaemogenesis. *Human Molecular Genetics*, **2014**, 23, 4315-27	5.6	95
112	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, 3607-17	5.6	28
111	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**, 164A, 2351-5	2.5	9
110	Noonan syndrome. *Lancet, The*, **2013**, 381, 333-42	40	459
109	Atrioventricular canal defect in patients with RASopathies. *European Journal of Human Genetics*, **2013**, 21, 200-4	5.3	19
108	Loss of function of the E3 ubiquitin-protein ligase UBE3B causes Kaufman oculocerebrofacial syndrome. *Journal of Medical Genetics*, **2013**, 50, 493-9	5.8	33
107	GH Therapy and first final height data in Noonan-like syndrome with loose anagen hair (Mazzanti syndrome). *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2756-61	2.5	8
106	Progressive extreme heterotopic calcification. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 1706-13	2.5	2
105	Transcriptional hallmarks of Noonan syndrome and Noonan-like syndrome with loose anagen hair. *Human Mutation*, **2012**, 33, 703-9	4.7	7
104	Loss of CBL E3-ligase activity in B-lineage childhood acute lymphoblastic leukaemia. *British Journal of Haematology*, **2012**, 159, 115-9	4.5	5
103	Dystonia in Costello syndrome. *Parkinsonism and Related Disorders*, **2012**, 18, 798-800	3.6	10
102	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, 317-26		19
101	Mutation of the receptor tyrosine phosphatase PTPRC (CD45) in T-cell acute lymphoblastic leukemia. *Blood*, **2012**, 119, 4476-9	2.2	76
100	A restricted spectrum of mutations in the SMAD4 tumor-suppressor gene underlies Myhre syndrome. *American Journal of Human Genetics*, **2012**, 90, 161-9	11	64
99	Reactive oxygen species and epidermal growth factor are antagonistic cues controlling SHP-2 dimerization. *Molecular and Cellular Biology*, **2012**, 32, 1998-2009	4.8	9
98	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, 27066-77	5.4	18
97	Noonan syndrome and clinically related disorders. *Best Practice and Research in Clinical Endocrinology and Metabolism*, **2011**, 25, 161-79	6.5	240
96	Cyclosporine attenuates cardiomyocyte hypertrophy induced by RAF1 mutants in Noonan and LEOPARD syndromes. *Journal of Molecular and Cellular Cardiology*, **2011**, 51, 4-15	5.8	14

95	Efficient one-step chromatographic purification and functional characterization of recombinant human Saposin C. *Protein Expression and Purification*, **2011**, 78, 209-15	2	2
94	Increased sleep spindle activity in patients with Costello syndrome (HRAS gene mutation). *Journal of Clinical Neurophysiology*, **2011**, 28, 314-8	2.2	9
93	PTPN2 negatively regulates oncogenic JAK1 in T-cell acute lymphoblastic leukemia. *Blood*, **2011**, 117, 7090-8	2.2	63
92	Long term memory profile of disorders associated with dysregulation of the RAS-MAPK signaling cascade. *Behavior Genetics*, **2011**, 41, 423-9	3.2	18
91	Prenatal features of Noonan syndrome: prevalence and prognostic value. *Prenatal Diagnosis*, **2011**, 31, 949-54	3.2	35
90	Phenotypic analysis of individuals with Costello syndrome due to HRAS p.G13C. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 706-16	2.5	45
89	SOS1 mutations in Noonan syndrome: molecular spectrum, structural insights on pathogenic effects, and genotype-phenotype correlations. *Human Mutation*, **2011**, 32, 760-72	4.7	82
88	RASopathies: Clinical Diagnosis in the First Year of Life. *Molecular Syndromology*, **2011**, 1, 282-289	1.5	59
87	Germline PTPN11 mutation affecting exon 8 in a case of syndromic juvenile myelomonocytic leukemia. *Leukemia Research*, **2011**, 35, e13-4	2.7	
86	Disruption of the histone acetyltransferase MYST4 leads to a Noonan syndrome-like phenotype and hyperactivated MAPK signaling in humans and mice. *Journal of Clinical Investigation*, **2011**, 121, 3479-91	15.9	68
85	RAS signaling pathway mutations and hypertrophic cardiomyopathy: getting into and out of the thick of it. *Journal of Clinical Investigation*, **2011**, 121, 844-7	15.9	36
84	PTPN11 mutations in childhood acute lymphoblastic leukemia occur as a secondary event associated with high hyperdiploidy. *Leukemia*, **2010**, 24, 232-5	10.7	17
83	Patient-specific induced pluripotent stem-cell-derived models of LEOPARD syndrome. *Nature*, **2010**, 465, 808-12	50.4	573
82	Enhanced human brain associative plasticity in Costello syndrome. *Journal of Physiology*, **2010**, 588, 3445-56	5.5	23
81	A restricted spectrum of NRAS mutations causes Noonan syndrome. *Nature Genetics*, **2010**, 42, 27-9	36.3	232
80	Disorders of dysregulated signal traffic through the RAS-MAPK pathway: phenotypic spectrum and molecular mechanisms. *Annals of the New York Academy of Sciences*, **2010**, 1214, 99-121	6.5	132
79	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	5.6	33
78	Noonan syndrome: clinical aspects and molecular pathogenesis. *Molecular Syndromology*, **2010**, 1, 2-26	1.5	155

77	ALL-associated JAK1 mutations confer hypersensitivity to the antiproliferative effect of type I interferon. *Blood*, **2010**, 115, 3287-95	2.2	23
76	Heterozygous germline mutations in the CBL tumor-suppressor gene cause a Noonan syndrome-like phenotype. *American Journal of Human Genetics*, **2010**, 87, 250-7	11	179
75	Acute lymphoblastic leukemia-associated JAK1 mutants activate the Janus kinase/STAT pathway via interleukin-9 receptor alpha homodimers. *Journal of Biological Chemistry*, **2009**, 284, 6773-81	5.4	58
74	Phosphatase-defective LEOPARD syndrome mutations in PTPN11 gene have gain-of-function effects during Drosophila development. *Human Molecular Genetics*, **2009**, 18, 193-201	5.6	68
73	Cognitive profile of disorders associated with dysregulation of the RAS/MAPK signaling cascade. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 140-6	2.5	61
72	Craniosynostosis in patients with Noonan syndrome caused by germline KRAS mutations. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 1036-40	2.5	40
71	Genomic duplication of PTPN11 is an uncommon cause of Noonan syndrome. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 2122-8	2.5	22
70	Germline BRAF mutations in Noonan, LEOPARD, and cardiofaciocutaneous syndromes: molecular diversity and associated phenotypic spectrum. *Human Mutation*, **2009**, 30, 695-702	4.7	213
69	RAS signaling dysregulation in human embryonal Rhabdomyosarcoma. *Genes Chromosomes and Cancer*, **2009**, 48, 975-82	5	77
68	Multiple giant cell lesions in patients with Noonan syndrome and cardio-facio-cutaneous syndrome. *European Journal of Human Genetics*, **2009**, 17, 420-5	5.3	65
67	Spectrum of MEK1 and MEK2 gene mutations in cardio-facio-cutaneous syndrome and genotype-phenotype correlations. *European Journal of Human Genetics*, **2009**, 17, 733-40	5.3	57
66	Mutation of SHOC2 promotes aberrant protein N-myristoylation and causes Noonan-like syndrome with loose anagen hair. *Nature Genetics*, **2009**, 41, 1022-6	36.3	291
65	Somatically acquired JAK1 mutations in adult acute lymphoblastic leukemia. *Journal of Experimental Medicine*, **2008**, 205, 751-8	16.6	285
64	Induction of both CD8+ and CD4+ T-cell-mediated responses in colorectal cancer patients by colon antigen-1. *Clinical Cancer Research*, **2008**, 14, 7292-303	12.9	9
63	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, 2018-29	5.6	62
62	Visual function in Noonan and LEOPARD syndrome. *Neuropediatrics*, **2008**, 39, 335-40	1.6	18
61	Geroderma osteodysplastica maps to a 4 Mb locus on chromosome 1q24. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 3034-7	2.5	7
60	Clinical lumping and molecular splitting of LEOPARD and NF1/NF1-Noonan syndromes. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 1009-11	2.5	12

59	Early fetal death associated with compound heterozygosity for Noonan syndrome-causative PTPN11 mutations. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 1249-52	2.5	6
58	Diversity, parental germline origin, and phenotypic spectrum of de novo HRAS missense changes in Costello syndrome. *Human Mutation*, **2007**, 28, 265-72	4.7	104
57	Gain-of-function SOS1 mutations cause a distinctive form of Noonan syndrome. *Nature Genetics*, **2007**, 39, 75-9	36.3	440
56	Gain-of-function RAF1 mutations cause Noonan and LEOPARD syndromes with hypertrophic cardiomyopathy. *Nature Genetics*, **2007**, 39, 1007-12	36.3	523
55	Noonan Syndrome and PTPN11 Mutations **2007**, 263-272		
54	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*, **2007**, 66, 963-74	4.2	22
53	Activating PTPN11 mutations play a minor role in pediatric and adult solid tumors. *Cancer Genetics and Cytogenetics*, **2006**, 166, 124-9		40
52	Noonan syndrome and related disorders: dysregulated RAS-mitogen activated protein kinase signal transduction. *Human Molecular Genetics*, **2006**, 15 Spec No 2, R220-6	5.6	153
51	Transgenic Drosophila models of Noonan syndrome causing PTPN11 gain-of-function mutations. *Human Molecular Genetics*, **2006**, 15, 543-53	5.6	57
50	Missense mutation in the transcription factor NKX2-5: a novel molecular event in the pathogenesis of thyroid dysgenesis. *Journal of Clinical Endocrinology and Metabolism*, **2006**, 91, 1428-33	5.6	132
49	Diversity and functional consequences of germline and somatic PTPN11 mutations in human disease. *American Journal of Human Genetics*, **2006**, 78, 279-90	11	292
48	Germline missense mutations affecting KRAS Isoform B are associated with a severe Noonan syndrome phenotype. *American Journal of Human Genetics*, **2006**, 79, 129-35	11	183
47	Acute lymphoblastic leukaemia in Noonan syndrome. *British Journal of Haematology*, **2006**, 133, 448-50	4.5	16
46	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 (BRAF(V599lns)). *Clinical Endocrinology*, **2006**, 64, 105-9	3.4	70
45	Biochemical and molecular characterization of the novel BRAF(V599Ins) mutation detected in a classic papillary thyroid carcinoma. *Oncogene*, **2006**, 25, 4235-40	9.2	49
44	NF1 gene mutations represent the major molecular event underlying neurofibromatosis-Noonan syndrome. *American Journal of Human Genetics*, **2005**, 77, 1092-101	11	115
43	The mutational spectrum of PTPN11 in juvenile myelomonocytic leukemia and Noonan syndrome/myeloproliferative disease. *Blood*, **2005**, 106, 2183-5	2.2	187
42	Rat nicastrin gene: cDNA isolation, mRNA variants and expression pattern analysis. *Molecular Brain Research*, **2005**, 136, 12-22		14

41	Germ-line and somatic PTPN11 mutations in human disease. *European Journal of Medical Genetics*, **2005**, 48, 81-96	2.6	105
40	Noonan syndrome and related disorders: genetics and pathogenesis. *Annual Review of Genomics and Human Genetics*, **2005**, 6, 45-68	9.7	246
39	Acquired PTPN11 mutations occur rarely in adult patients with myelodysplastic syndromes and chronic myelomonocytic leukemia. *Leukemia Research*, **2005**, 29, 459-62	2.7	49
38	Somatic PTPN11 mutations in childhood acute myeloid leukaemia. *British Journal of Haematology*, **2005**, 129, 333-9	4.5	64
37	Differences in the prevalence of PTPN11 mutations in FAB M5 paediatric acute myeloid leukaemia. *British Journal of Haematology*, **2005**, 130, 801-3	4.5	14
36	Genotypic and phenotypic characterization of Noonan syndrome: new data and review of the literature. *American Journal of Medical Genetics, Part A*, **2005**, 134A, 165-70	2.5	80
35	Hyperthrophic cardiomyopathy and the PTPN11 gene. *American Journal of Medical Genetics, Part A*, **2005**, 136, 93-4	2.5	8
34	Phenotypic and genotypic characterisation of Noonan-like/multiple giant cell lesion syndrome. *Journal of Medical Genetics*, **2005**, 42, e11	5.8	53
33	Noonan syndrome-associated SHP2/PTPN11 mutants cause EGF-dependent prolonged GAB1 binding and sustained ERK2/MAPK1 activation. *Human Mutation*, **2004**, 23, 267-77	4.7	163
32	Paternal germline origin and sex-ratio distortion in transmission of PTPN11 mutations in Noonan syndrome. *American Journal of Human Genetics*, **2004**, 75, 492-7	11	61
31	SHP-2 and myeloid malignancies. *Current Opinion in Hematology*, **2004**, 11, 44-50	3.3	84
30	Genetic evidence for lineage-related and differentiation stage-related contribution of somatic PTPN11 mutations to leukemogenesis in childhood acute leukemia. *Blood*, **2004**, 104, 307-13	2.2	237
29	PTPN11 Mutational Spectrum in Juvenile Myelomonocytic Leukemia and Noonan Syndrome.. *Blood*, **2004**, 104, 3417-3417	2.2	1
28	PTPN11 and RAS Gene Mutation Pattern Identifies an Unique Feature of Upregulated RAS Function in Infant ALL.. *Blood*, **2004**, 104, 996-996	2.2	
27	A competitive polymerase chain reaction-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, 103-9	2.5	65
26	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, 423-6	4	31
25	Somatic mutations in PTPN11 in juvenile myelomonocytic leukemia, myelodysplastic syndromes and acute myeloid leukemia. *Nature Genetics*, **2003**, 34, 148-50	36.3	819
24	Absence of PTPN11 mutations in 28 cases of cardiofaciocutaneous (CFC) syndrome. *Human Genetics*, **2002**, 111, 421-7	6.3	39

23	Analysis of three RFLPs of the COL1A2 (Type I Collagen) in the Amhara and the Oromo of Ethiopia. *Annals of Human Biology*, **2002**, 29, 432-41	1.7	10
22	PTPN11 mutations in Noonan syndrome: molecular spectrum, genotype-phenotype correlation, and phenotypic heterogeneity. *American Journal of Human Genetics*, **2002**, 70, 1555-63	11	567
21	Mutations in PTPN11, encoding the protein tyrosine phosphatase SHP-2, cause Noonan syndrome. *Nature Genetics*, **2001**, 29, 465-8	36.3	1312
20	A competitive PCR-based method to measure human fibroblast growth factor receptor 1-4 (FGFR1-4) gene expression. *DNA and Cell Biology*, **2001**, 20, 367-79	3.6	15
19	Rapid communication: nucleotide sequence of chamois, alpine ibex, and red deer tRNA(Lys) and ATPase8 mitochondrial genes. *Journal of Animal Science*, **1999**, 77, 3398-9	0.7	6
18	Fibroblast growth factor receptor mutational screening in newborns affected by metopic synostosis. *Childls Nervous System*, **1999**, 15, 389-93; discussion 393-4	1.7	13
17	Decreased proliferation and altered differentiation in osteoblasts from genetically and clinically distinct craniosynostotic disorders. *American Journal of Pathology*, **1999**, 154, 1465-77	5.8	86
16	Polymorphism at position 882 of the fibroblast growth factor receptor 3 (FGFR3) gene detected by SSCP analysis. *Molecular and Cellular Probes*, **1998**, 12, 335-7	3.3	3
15	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, 513-8	2.5	108
14	Rapid communication: nucleotide sequence of porcine and ovine tRNA(Lys) and ATPase8 mitochondrial genes. *Journal of Animal Science*, **1998**, 76, 2207-8	0.7	7
13	Trp290Cys mutation in exon IIIa of the fibroblast growth factor receptor 2 (FGFR2) gene is associated with Pfeiffer syndrome. *Human Genetics*, **1997**, 99, 602-6	6.3	40
12	Jackson-Weiss syndrome: identification of two novel FGFR2 missense mutations shared with Crouzon and Pfeiffer craniosynostotic disorders. *Human Genetics*, **1997**, 101, 47-50	6.3	45
11	Genetic heterogeneity among the Hindus and their relationships with the other "Caucasoid" populations: new data on Punjab-Haryana and Rajasthan Indian states. *American Journal of Physical Anthropology*, **1995**, 98, 257-73	2.5	2
10	Worldwide distribution of phosphoglucomutase 1 (PGM1) polymorphism detected by isoelectric focusing: A review. *International Journal of Anthropology*, **1994**, 9, 81-112		4
9	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	1.2	11
8	Linkage disequilibrium at the human phosphoglucomutase 1 locus. *Human Biology*, **1994**, 66, 669-81	1.2	
7	Genetic characterization of the Cayapa Indians of Ecuador and their genetic relationships to other Native American populations. *Human Biology*, **1994**, 66, 299-322	1.2	11
6	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		1

5	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		
4	Red-cell enzyme polymorphisms in the Reggio Calabria province (Italy). *Human Heredity*, **1990**, 40, 308-10	1.1	4
3	Noonan Syndrome and Other RAS/MAPK Pathway Syndromes122-130		
2	Noonan Syndrome		2
1	Multidisciplinary Management of Costello Syndrome: Current Perspectives. *Journal of Multidisciplinary Healthcare*,Volume 15, 1277-1296	2.8	0