Marco Tartaglia

List of Publications by Citations

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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	Mutations in PTPN11, encoding the protein tyrosine phosphatase SHP-2, cause Noonan syndrome. *Nature Genetics*, **2001**, 29, 465-8	36.3	1312
327	Somatic mutations in PTPN11 in juvenile myelomonocytic leukemia, myelodysplastic syndromes and acute myeloid leukemia. *Nature Genetics*, **2003**, 34, 148-50	36.3	819
326	Patient-specific induced pluripotent stem-cell-derived models of LEOPARD syndrome. *Nature*, **2010** , 465, 808-12	50.4	573
325	PTPN11 mutations in Noonan syndrome: molecular spectrum, genotype-phenotype correlation, and phenotypic heterogeneity. *American Journal of Human Genetics*, **2002**, 70, 1555-63	11	567
324	Gain-of-function RAF1 mutations cause Noonan and LEOPARD syndromes with hypertrophic cardiomyopathy. *Nature Genetics*, **2007**, 39, 1007-12	36.3	523
323	Noonan syndrome. *Lancet, The*, **2013**, 381, 333-42	40	459
322	Gain-of-function SOS1 mutations cause a distinctive form of Noonan syndrome. *Nature Genetics*, **2007**, 39, 75-9	36.3	440
321	Diversity and functional consequences of germline and somatic PTPN11 mutations in human disease. *American Journal of Human Genetics*, **2006**, 78, 279-90	11	292
320	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
319	Somatically acquired JAK1 mutations in adult acute lymphoblastic leukemia. *Journal of Experimental Medicine*, **2008**, 205, 751-8	16.6	285
318	Noonan syndrome and related disorders: genetics and pathogenesis. *Annual Review of Genomics and Human Genetics*, **2005**, 6, 45-68	9.7	246
317	Noonan syndrome and clinically related disorders. *Best Practice and Research in Clinical Endocrinology and Metabolism*, **2011**, 25, 161-79	6.5	240
316	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
315	A restricted spectrum of NRAS mutations causes Noonan syndrome. *Nature Genetics*, **2010**, 42, 27-9	36.3	232
314	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. *American Journal of Human Genetics*, **2017**, 101, 664-685	11	214
313	Germline BRAF mutations in Noonan, LEOPARD, and cardiofaciocutaneous syndromes: molecular diversity and associated phenotypic spectrum. *Human Mutation*, **2009**, 30, 695-702	4.7	213
312	The mutational spectrum of PTPN11 in juvenile myelomonocytic leukemia and Noonan syndrome/myeloproliferative disease. *Blood*, **2005**, 106, 2183-5	2.2	187

311	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
310	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
309	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
308	Noonan syndrome: clinical aspects and molecular pathogenesis. *Molecular Syndromology*, **2010**, 1, 2-26	1.5	155
307	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
306	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
305	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
304	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
303	Mutations in KCNH1 and ATP6V1B2 cause Zimmermann-Laband syndrome. *Nature Genetics*, **2015**, 47, 661-7	36.3	128
302	NF1 gene mutations represent the major molecular event underlying neurofibromatosis-Noonan syndrome. *American Journal of Human Genetics*, **2005**, 77, 1092-101	11	115
301	A Specific Mutational Signature Associated with DNA 8-Oxoguanine Persistence in MUTYH-defective Colorectal Cancer. *EBioMedicine*, **2017**, 20, 39-49	8.8	112
300	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
299	Germ-line and somatic PTPN11 mutations in human disease. *European Journal of Medical Genetics*, **2005**, 48, 81-96	2.6	105
298	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
297	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
296	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
295	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
294	Functional Dysregulation of CDC42 Causes Diverse Developmental Phenotypes. *American Journal of Human Genetics*, **2018**, 102, 309-320	11	85

293	SHP-2 and myeloid malignancies. *Current Opinion in Hematology*, **2004**, 11, 44-50	3.3	84
292	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
291	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
290	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
289	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
288	RAS signaling dysregulation in human embryonal Rhabdomyosarcoma. *Genes Chromosomes and Cancer*, **2009**, 48, 975-82	5	77
287	Mutations in PAX2 associate with adult-onset FSGS. *Journal of the American Society of Nephrology: JASN*, **2014**, 25, 1942-53	12.7	76
286	Mutation of the receptor tyrosine phosphatase PTPRC (CD45) in T-cell acute lymphoblastic leukemia. *Blood*, **2012**, 119, 4476-9	2.2	76
285	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
284	A novel disorder involving dyshematopoiesis, inflammation, and HLH due to aberrant CDC42 function. *Journal of Experimental Medicine*, **2019**, 216, 2778-2799	16.6	71
283	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
282	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
281	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
280	Cardiomyopathies in Noonan syndrome and the other RASopathies. *Progress in Pediatric Cardiology*, **2015**, 39, 13-19	0.4	67
279	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
278	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
277	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
276	Somatic PTPN11 mutations in childhood acute myeloid leukaemia. *British Journal of Haematology*, **2005**, 129, 333-9	4.5	64

275	PTPN2 negatively regulates oncogenic JAK1 in T-cell acute lymphoblastic leukemia. *Blood*, **2011**, 117, 7090-8	2.2	63
274	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
273	Mutations in ZBTB20 cause Primrose syndrome. *Nature Genetics*, **2014**, 46, 815-7	36.3	61
272	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
271	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
270	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
269	RASopathies: Clinical Diagnosis in the First Year of Life. *Molecular Syndromology*, **2011**, 1, 282-289	1.5	59
268	Noncanonical GLI1 signaling promotes stemness features and in vivo growth in lung adenocarcinoma. *Oncogene*, **2017**, 36, 4641-4652	9.2	58
267	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
266	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
265	Transgenic Drosophila models of Noonan syndrome causing PTPN11 gain-of-function mutations. *Human Molecular Genetics*, **2006**, 15, 543-53	5.6	57
264	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
263	RAF1 mutations in childhood-onset dilated cardiomyopathy. *Nature Genetics*, **2014**, 46, 635-639	36.3	54
262	Modeling medulloblastoma in vivo and with human cerebellar organoids. *Nature Communications*, **2020**, 11, 583	17.4	54
261	Phenotypic and genotypic characterisation of Noonan-like/multiple giant cell lesion syndrome. *Journal of Medical Genetics*, **2005**, 42, e11	5.8	53
260	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
259	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
258	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. *Human Mutation*, **2015**, 36, 1080-7	4.7	51

257	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
256	Acquired PTPN11 mutations occur rarely in adult patients with myelodysplastic syndromes and chronic myelomonocytic leukemia. *Leukemia Research*, **2005**, 29, 459-62	2.7	49
255	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
254	Cardiac defects, morbidity and mortality in patients affected by RASopathies. CARNET study results. *International Journal of Cardiology*, **2017**, 245, 92-98	3.2	48
253	CHK1-targeted therapy to deplete DNA replication-stressed, p53-deficient, hyperdiploid colorectal cancer stem cells. *Gut*, **2018**, 67, 903-917	19.2	45
252	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
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250	Mutations in KCNK4 that Affect Gating Cause a Recognizable Neurodevelopmental Syndrome. *American Journal of Human Genetics*, **2018**, 103, 621-630	11	45
249	LYRM7 mutations cause a multifocal cavitating leukoencephalopathy with distinct MRI appearance. *Brain*, **2016**, 139, 782-94	11.2	44
248	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
247	Behavioral profile in RASopathies. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 934-42	2.5	42
246	Childhood onset tubular aggregate myopathy associated with de novo STIM1 mutations. *Journal of Neurology*, **2014**, 261, 870-6	5.5	41
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243	Activating PTPN11 mutations play a minor role in pediatric and adult solid tumors. *Cancer Genetics and Cytogenetics*, **2006**, 166, 124-9		40
242	Absence of PTPN11 mutations in 28 cases of cardiofaciocutaneous (CFC) syndrome. *Human Genetics*, **2002**, 111, 421-7	6.3	39
241	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
240	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

239	TBCE Mutations Cause Early-Onset Progressive Encephalopathy with Distal Spinal Muscular Atrophy. *American Journal of Human Genetics*, **2016**, 99, 974-983	11	37
238	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
237	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
236	Prenatal features of Noonan syndrome: prevalence and prognostic value. *Prenatal Diagnosis*, **2011**, 31, 949-54	3.2	35
235	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
234	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
233	Structural, Functional, and Clinical Characterization of a Novel PTPN11 Mutation Cluster Underlying Noonan Syndrome. *Human Mutation*, **2017**, 38, 451-459	4.7	32
232	Cooperating JAK1 and JAK3 mutants increase resistance to JAK inhibitors. *Blood*, **2014**, 124, 3924-31	2.2	32
231	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
230	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
229	Specific combinations of biallelic variants cause Wiedemann-Rautenstrauch syndrome. *Journal of Medical Genetics*, **2018**, 55, 837-846	5.8	31
228	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
227	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
226	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
225	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
224	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
223	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
222	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

221	CREBBP mutations in individuals without Rubinstein-Taybi syndrome phenotype. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2681-93	2.5	28
220	Novel mutations in IBA57 are associated with leukodystrophy and variable clinical phenotypes. *Journal of Neurology*, **2017**, 264, 102-111	5.5	27
219	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
218	Clinical, biochemical and molecular characterization of prosaposin deficiency. *Clinical Genetics*, **2016** , 90, 220-9	4	26
217	Congenital heart disease and genetic syndromes: new insights into molecular mechanisms. *Expert Review of Molecular Diagnostics*, **2017**, 17, 861-870	3.8	26
216	Biallelic mutations in early-onset, variably progressive neurodegeneration. *Neurology*, **2018**, 91, e319-e330	5.5	26
215	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. *American Journal of Human Genetics*, **2020**, 107, 499-513	11	25
214	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
213	Clinical Presentation and Natural History of Hypertrophic Cardiomyopathy in RASopathies. *Heart Failure Clinics*, **2018**, 14, 225-235	3.3	24
212	Understanding Growth Failure in Costello Syndrome: Increased Resting Energy Expenditure. *Journal of Pediatrics*, **2016**, 170, 322-4	3.6	24
211	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
210	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
209	Genotype and phenotype spectrum of NRAS germline variants. *European Journal of Human Genetics* , **2017**, 25, 823-831	5.3	23
208	Wiedemann-Rautenstrauch syndrome: A phenotype analysis. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1763-1772	2.5	23
207	Activating Mutations of RRAS2 Are a Rare Cause of Noonan Syndrome. *American Journal of Human Genetics*, **2019**, 104, 1223-1232	11	23
206	A new bioavailable fenretinide formulation with antiproliferative, antimetabolic, and cytotoxic effects on solid tumors. *Cell Death and Disease*, **2019**, 10, 529	9.8	23
205	Novel SMAD4 mutation causing Myhre syndrome. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 1835-40	2.5	23
204	Enhanced human brain associative plasticity in Costello syndrome. *Journal of Physiology*, **2010**, 588, 3445-56	5.9	23

203	ALL-associated JAK1 mutations confer hypersensitivity to the antiproliferative effect of type I interferon. *Blood*, **2010**, 115, 3287-95	2.2	23
202	Frameshift mutations at the C-terminus of HIST1H1E result in a specific DNA hypomethylation signature. *Clinical Epigenetics*, **2020**, 12, 7	7.7	23
201	Molecular Diversity and Associated Phenotypic Spectrum of Germline CBL Mutations. *Human Mutation*, **2015**, 36, 787-96	4.7	22
200	Mutations in Fibronectin Cause a Subtype of Spondylometaphyseal Dysplasia with "Corner Fractures". *American Journal of Human Genetics*, **2017**, 101, 815-823	11	22
199	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
198	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
197	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
196	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
195	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
194	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
193	Atrioventricular canal defect in patients with RASopathies. *European Journal of Human Genetics*, **2013**, 21, 200-4	5.3	19
192	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
191	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
190	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
189	Defective kinesin binding of TUBB2A causes progressive spastic ataxia syndrome resembling sacsinopathy. *Human Molecular Genetics*, **2018**, 27, 1892-1904	5.6	18
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187	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
186	Long term memory profile of disorders associated with dysregulation of the RAS-MAPK signaling cascade. *Behavior Genetics*, **2011**, 41, 423-9	3.2	18

185	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
184	Visual function in Noonan and LEOPARD syndrome. *Neuropediatrics*, **2008**, 39, 335-40	1.6	18
183	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. *Nature Communications*, **2020**, 11, 595	17.4	18
182	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
181	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
180	Neurobehavioral features in individuals with Kabuki syndrome. *Molecular Genetics & Genomic Medicine*, **2018**, 6, 322-331	2.3	17
179	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
178	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
177	PTPN11 mutations in childhood acute lymphoblastic leukemia occur as a secondary event associated with high hyperdiploidy. *Leukemia*, **2010**, 24, 232-5	10.7	17
176	Copy number variants in autism spectrum disorders. *Progress in Neuro-Psychopharmacology and Biological Psychiatry*, **2019**, 92, 421-427	5.5	16
175	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
174	Clinical spectrum of Kabuki-like syndrome caused by HNRNPK haploinsufficiency. *Clinical Genetics*, **2018**, 93, 401-407	4	16
173	Biallelic mutations in DYNC2LI1 are a rare cause of Ellis-van Creveld syndrome. *Clinical Genetics*, **2018**, 93, 632-639	4	16
172	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
171	Acute lymphoblastic leukaemia in Noonan syndrome. *British Journal of Haematology*, **2006**, 133, 448-50	4.5	16
170	Atrioventricular canal defect and genetic syndromes: The unifying role of sonic hedgehog. *Clinical Genetics*, **2019**, 95, 268-276	4	16
169	Biallelic mutations in the homeodomain of NKX6-2 underlie a severe hypomyelinating leukodystrophy. *Brain*, **2017**, 140, 2550-2556	11.2	15
168	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

167	NBAS pathogenic variants: Defining the associated clinical and facial phenotype and genotype-phenotype correlations. *Human Mutation*, **2019**, 40, 721-728	4.7	15
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