

Marco Tartaglia

List of Publications by Citations

Source: <https://exaly.com/author-pdf/1310592/marco-tartaglia-publications-by-citations.pdf>

Version: 2024-04-09

This document has been generated based on the publications and citations recorded by exaly.com. For the latest version of this publication list, visit the link given above.

The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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. <i>Nature Genetics</i> , 2001, 29, 465-8	36.3	1312
327	Somatic mutations in PTPN11 in juvenile myelomonocytic leukemia, myelodysplastic syndromes and acute myeloid leukemia. <i>Nature Genetics</i> , 2003, 34, 148-50	36.3	819
326	Patient-specific induced pluripotent stem-cell-derived models of LEOPARD syndrome. <i>Nature</i> , 2010, 465, 808-12	50.4	573
325	PTPN11 mutations in Noonan syndrome: molecular spectrum, genotype-phenotype correlation, and phenotypic heterogeneity. <i>American Journal of Human Genetics</i> , 2002, 70, 1555-63	11	567
324	Gain-of-function RAF1 mutations cause Noonan and LEOPARD syndromes with hypertrophic cardiomyopathy. <i>Nature Genetics</i> , 2007, 39, 1007-12	36.3	523
323	Noonan syndrome. <i>Lancet, The</i> , 2013, 381, 333-42	40	459
322	Gain-of-function SOS1 mutations cause a distinctive form of Noonan syndrome. <i>Nature Genetics</i> , 2007, 39, 75-9	36.3	440
321	Diversity and functional consequences of germline and somatic PTPN11 mutations in human disease. <i>American Journal of Human Genetics</i> , 2006, 78, 279-90	11	292
320	Mutation of SHOC2 promotes aberrant protein N-myristoylation and causes Noonan-like syndrome with loose anagen hair. <i>Nature Genetics</i> , 2009, 41, 1022-6	36.3	291
319	Somatically acquired JAK1 mutations in adult acute lymphoblastic leukemia. <i>Journal of Experimental Medicine</i> , 2008, 205, 751-8	16.6	285
318	Noonan syndrome and related disorders: genetics and pathogenesis. <i>Annual Review of Genomics and Human Genetics</i> , 2005, 6, 45-68	9.7	246
317	Noonan syndrome and clinically related disorders. <i>Best Practice and Research in Clinical Endocrinology and Metabolism</i> , 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. <i>Blood</i> , 2004, 104, 307-13	2.2	237
315	A restricted spectrum of NRAS mutations causes Noonan syndrome. <i>Nature Genetics</i> , 2010, 42, 27-9	36.3	232
314	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. <i>American Journal of Human Genetics</i> , 2017, 101, 664-685	11	214
313	Germline BRAF mutations in Noonan, LEOPARD, and cardiofaciocutaneous syndromes: molecular diversity and associated phenotypic spectrum. <i>Human Mutation</i> , 2009, 30, 695-702	4.7	213
312	The mutational spectrum of PTPN11 in juvenile myelomonocytic leukemia and Noonan syndrome/myeloproliferative disease. <i>Blood</i> , 2005, 106, 2183-5	2.2	187

311	Germline missense mutations affecting KRAS Isoform B are associated with a severe Noonan syndrome phenotype. <i>American Journal of Human Genetics</i> , 2006 , 79, 129-35	11	183
310	Heterozygous germline mutations in the CBL tumor-suppressor gene cause a Noonan syndrome-like phenotype. <i>American Journal of Human Genetics</i> , 2010 , 87, 250-7	11	179
309	Noonan syndrome-associated SHP2/PTPN11 mutants cause EGF-dependent prolonged GAB1 binding and sustained ERK2/MAPK1 activation. <i>Human Mutation</i> , 2004 , 23, 267-77	4.7	163
308	Noonan syndrome: clinical aspects and molecular pathogenesis. <i>Molecular Syndromology</i> , 2010 , 1, 2-26	1.5	155
307	Noonan syndrome and related disorders: dysregulated RAS-mitogen activated protein kinase signal transduction. <i>Human Molecular Genetics</i> , 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. <i>Annals of the New York Academy of Sciences</i> , 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. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2006 , 91, 1428-33	5.6	132
304	ACE2 gene variants may underlie interindividual variability and susceptibility to COVID-19 in the Italian population. <i>European Journal of Human Genetics</i> , 2020 , 28, 1602-1614	5.3	132
303	Mutations in KCNH1 and ATP6V1B2 cause Zimmermann-Laband syndrome. <i>Nature Genetics</i> , 2015 , 47, 661-7	36.3	128
302	NF1 gene mutations represent the major molecular event underlying neurofibromatosis-Noonan syndrome. <i>American Journal of Human Genetics</i> , 2005 , 77, 1092-101	11	115
301	A Specific Mutational Signature Associated with DNA 8-Oxoguanine Persistence in MUTYH-defective Colorectal Cancer. <i>EBioMedicine</i> , 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. <i>Journal of Food Protection</i> , 1998 , 61, 513-8	2.5	108
299	Germ-line and somatic PTPN11 mutations in human disease. <i>European Journal of Medical Genetics</i> , 2005 , 48, 81-96	2.6	105
298	Diversity, parental germline origin, and phenotypic spectrum of de novo HRAS missense changes in Costello syndrome. <i>Human Mutation</i> , 2007 , 28, 265-72	4.7	104
297	Activating mutations in RRAS underlie a phenotype within the RASopathy spectrum and contribute to leukaemogenesis. <i>Human Molecular Genetics</i> , 2014 , 23, 4315-27	5.6	95
296	p.Arg1809Cys substitution in neurofibromin is associated with a distinctive NF1 phenotype without neurofibromas. <i>European Journal of Human Genetics</i> , 2015 , 23, 1068-71	5.3	89
295	Decreased proliferation and altered differentiation in osteoblasts from genetically and clinically distinct craniosynostotic disorders. <i>American Journal of Pathology</i> , 1999 , 154, 1465-77	5.8	86
294	Functional Dysregulation of CDC42 Causes Diverse Developmental Phenotypes. <i>American Journal of Human Genetics</i> , 2018 , 102, 309-320	11	85

293	SHP-2 and myeloid malignancies. <i>Current Opinion in Hematology</i> , 2004 , 11, 44-50	3.3	84
292	SOS1 mutations in Noonan syndrome: molecular spectrum, structural insights on pathogenic effects, and genotype-phenotype correlations. <i>Human Mutation</i> , 2011 , 32, 760-72	4.7	82
291	Genotypic and phenotypic characterization of Noonan syndrome: new data and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 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. <i>Neuro-Oncology</i> , 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. <i>Neuro-Oncology</i> , 2021 , 23, i36-i36	1	78
288	RAS signaling dysregulation in human embryonal Rhabdomyosarcoma. <i>Genes Chromosomes and Cancer</i> , 2009 , 48, 975-82	5	77
287	Mutations in PAX2 associate with adult-onset FSGS. <i>Journal of the American Society of Nephrology: JASN</i> , 2014 , 25, 1942-53	12.7	76
286	Mutation of the receptor tyrosine phosphatase PTPRC (CD45) in T-cell acute lymphoblastic leukemia. <i>Blood</i> , 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. <i>American Journal of Human Genetics</i> , 2015 , 96, 816-25	11	75
284	A novel disorder involving dyshematopoiesis, inflammation, and HLH due to aberrant CDC42 function. <i>Journal of Experimental Medicine</i> , 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 oncogene (BRAF(V599Ins)). <i>Clinical Endocrinology</i> , 2006 , 64, 105-9	3.4	70
282	Phosphatase-defective LEOPARD syndrome mutations in PTPN11 gene have gain-of-function effects during Drosophila development. <i>Human Molecular Genetics</i> , 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. <i>Journal of Clinical Investigation</i> , 2011 , 121, 3479-91	15.9	68
280	Cardiomyopathies in Noonan syndrome and the other RASopathies. <i>Progress in Pediatric Cardiology</i> , 2015 , 39, 13-19	0.4	67
279	Multiple giant cell lesions in patients with Noonan syndrome and cardio-facio-cutaneous syndrome. <i>European Journal of Human Genetics</i> , 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. <i>Journal of Food Protection</i> , 2003 , 66, 103-9	2.5	65
277	A restricted spectrum of mutations in the SMAD4 tumor-suppressor gene underlies Myhre syndrome. <i>American Journal of Human Genetics</i> , 2012 , 90, 161-9	11	64
276	Somatic PTPN11 mutations in childhood acute myeloid leukaemia. <i>British Journal of Haematology</i> , 2005 , 129, 333-9	4.5	64

275	PTPN2 negatively regulates oncogenic JAK1 in T-cell acute lymphoblastic leukemia. <i>Blood</i> , 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. <i>Human Molecular Genetics</i> , 2008 , 17, 2018-29	5.6	62
273	Mutations in ZBTB20 cause Primrose syndrome. <i>Nature Genetics</i> , 2014 , 46, 815-7	36.3	61
272	Cognitive profile of disorders associated with dysregulation of the RAS/MAPK signaling cascade. <i>American Journal of Medical Genetics, Part A</i> , 2009 , 149A, 140-6	2.5	61
271	Paternal germline origin and sex-ratio distortion in transmission of PTPN11 mutations in Noonan syndrome. <i>American Journal of Human Genetics</i> , 2004 , 75, 492-7	11	61
270	Organoids as a new model for improving regenerative medicine and cancer personalized therapy in renal diseases. <i>Cell Death and Disease</i> , 2019 , 10, 201	9.8	61
269	RASopathies: Clinical Diagnosis in the First Year of Life. <i>Molecular Syndromology</i> , 2011 , 1, 282-289	1.5	59
268	Noncanonical GLI1 signaling promotes stemness features and in vivo growth in lung adenocarcinoma. <i>Oncogene</i> , 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. <i>Journal of Biological Chemistry</i> , 2009 , 284, 6773-81	5.4	58
266	Spectrum of MEK1 and MEK2 gene mutations in cardio-facio-cutaneous syndrome and genotype-phenotype correlations. <i>European Journal of Human Genetics</i> , 2009 , 17, 733-40	5.3	57
265	Transgenic Drosophila models of Noonan syndrome causing PTPN11 gain-of-function mutations. <i>Human Molecular Genetics</i> , 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. <i>American Journal of Human Genetics</i> , 2016 , 99, 962-973 ¹¹	55	
263	RAF1 mutations in childhood-onset dilated cardiomyopathy. <i>Nature Genetics</i> , 2014 , 46, 635-639	36.3	54
262	Modeling medulloblastoma in vivo and with human cerebellar organoids. <i>Nature Communications</i> , 2020 , 11, 583	17.4	54
261	Phenotypic and genotypic characterisation of Noonan-like/multiple giant cell lesion syndrome. <i>Journal of Medical Genetics</i> , 2005 , 42, e11	5.8	53
260	Myeloid Dysregulation in a Human Induced Pluripotent Stem Cell Model of PTPN11-Associated Juvenile Myelomonocytic Leukemia. <i>Cell Reports</i> , 2015 , 13, 504-515	10.6	52
259	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. <i>American Journal of Human Genetics</i> , 2020 , 106, 356-370	11	51
258	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. <i>Human Mutation</i> , 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. <i>Oncogene</i> , 2006 , 25, 4235-40	9.2	49
256	Acquired PTPN11 mutations occur rarely in adult patients with myelodysplastic syndromes and chronic myelomonocytic leukemia. <i>Leukemia Research</i> , 2005 , 29, 459-62	2.7	49
255	Heterozygous germline mutations in A2ML1 are associated with a disorder clinically related to Noonan syndrome. <i>European Journal of Human Genetics</i> , 2015 , 23, 317-24	5.3	48
254	Cardiac defects, morbidity and mortality in patients affected by RASopathies. CARNET study results. <i>International Journal of Cardiology</i> , 2017 , 245, 92-98	3.2	48
253	CHK1-targeted therapy to deplete DNA replication-stressed, p53-deficient, hyperdiploid colorectal cancer stem cells. <i>Gut</i> , 2018 , 67, 903-917	19.2	45
252	Phenotypic analysis of individuals with Costello syndrome due to HRAS p.G13C. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 706-16	2.5	45
251	Jackson-Weiss syndrome: identification of two novel FGFR2 missense mutations shared with Crouzon and Pfeiffer craniosynostotic disorders. <i>Human Genetics</i> , 1997 , 101, 47-50	6.3	45
250	Mutations in KCNK4 that Affect Gating Cause a Recognizable Neurodevelopmental Syndrome. <i>American Journal of Human Genetics</i> , 2018 , 103, 621-630	11	45
249	LYRM7 mutations cause a multifocal cavitating leukoencephalopathy with distinct MRI appearance. <i>Brain</i> , 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. <i>Cell Reports</i> , 2020 , 30, 2963-2977.e6	10.6	43
247	Behavioral profile in RASopathies. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 934-42	2.5	42
246	Childhood onset tubular aggregate myopathy associated with de novo STIM1 mutations. <i>Journal of Neurology</i> , 2014 , 261, 870-6	5.5	41
245	Craniosynostosis in patients with Noonan syndrome caused by germline KRAS mutations. <i>American Journal of Medical Genetics, Part A</i> , 2009 , 149A, 1036-40	2.5	40
244	Trp290Cys mutation in exon IIIa of the fibroblast growth factor receptor 2 (FGFR2) gene is associated with Pfeiffer syndrome. <i>Human Genetics</i> , 1997 , 99, 602-6	6.3	40
243	Activating PTPN11 mutations play a minor role in pediatric and adult solid tumors. <i>Cancer Genetics and Cytogenetics</i> , 2006 , 166, 124-9		40
242	Absence of PTPN11 mutations in 28 cases of cardiofaciocutaneous (CFC) syndrome. <i>Human Genetics</i> , 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. <i>Human Mutation</i> , 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. <i>Human Molecular Genetics</i> , 2019 , 28, 1007-1022	5.6	38

239	TBCE Mutations Cause Early-Onset Progressive Encephalopathy with Distal Spinal Muscular Atrophy. <i>American Journal of Human Genetics</i> , 2016 , 99, 974-983	11	37
238	RAS signaling pathway mutations and hypertrophic cardiomyopathy: getting into and out of the thick of it. <i>Journal of Clinical Investigation</i> , 2011 , 121, 844-7	15.9	36
237	Cancer Stem Cell-Based Models of Colorectal Cancer Reveal Molecular Determinants of Therapy Resistance. <i>Stem Cells Translational Medicine</i> , 2016 , 5, 511-23	6.9	35
236	Prenatal features of Noonan syndrome: prevalence and prognostic value. <i>Prenatal Diagnosis</i> , 2011 , 31, 949-54	3.2	35
235	Loss of function of the E3 ubiquitin-protein ligase UBE3B causes Kaufman oculocerebrofacial syndrome. <i>Journal of Medical Genetics</i> , 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. <i>Human Molecular Genetics</i> , 2010 , 19, 790-802	5.6	33
233	Structural, Functional, and Clinical Characterization of a Novel PTPN11 Mutation Cluster Underlying Noonan Syndrome. <i>Human Mutation</i> , 2017 , 38, 451-459	4.7	32
232	Cooperating JAK1 and JAK3 mutants increase resistance to JAK inhibitors. <i>Blood</i> , 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. <i>Cell Death and Disease</i> , 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. <i>Clinical Genetics</i> , 2003 , 63, 423-6 ⁴		31
229	Specific combinations of biallelic variants cause Wiedemann-Rautenstrauch syndrome. <i>Journal of Medical Genetics</i> , 2018 , 55, 837-846	5.8	31
228	Aberrant Function of the C-Terminal Tail of HIST1H1E Accelerates Cellular Senescence and Causes Premature Aging. <i>American Journal of Human Genetics</i> , 2019 , 105, 493-508	11	30
227	Further delineation of an entity caused by CREBBP and EP300 mutations but not resembling Rubinstein-Taybi syndrome. <i>American Journal of Medical Genetics, Part A</i> , 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. <i>Journal of Biological Chemistry</i> , 2015 , 290, 29022-34	5.4	29
225	Recessive Inactivating Mutations in TBCK, Encoding a Rab GTPase-Activating Protein, Cause Severe Infantile Syndromic Encephalopathy. <i>American Journal of Human Genetics</i> , 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. <i>Neurogenetics</i> , 2018 , 19, 111-121	3	28
223	Not only dominant, not only optic atrophy: expanding the clinical spectrum associated with OPA1 mutations. <i>Orphanet Journal of Rare Diseases</i> , 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. <i>Human Molecular Genetics</i> , 2014 , 23, 3607-17	5.6	28

221	CREBBP mutations in individuals without Rubinstein-Taybi syndrome phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 2681-93	2.5	28
220	Novel mutations in IBA57 are associated with leukodystrophy and variable clinical phenotypes. <i>Journal of Neurology</i> , 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. <i>Clinical Genetics</i> , 2017 , 91, 86-91	4	26
218	Clinical, biochemical and molecular characterization of prosaposin deficiency. <i>Clinical Genetics</i> , 2016 , 90, 220-9	4	26
217	Congenital heart disease and genetic syndromes: new insights into molecular mechanisms. <i>Expert Review of Molecular Diagnostics</i> , 2017 , 17, 861-870	3.8	26
216	Biallelic mutations in early-onset, variably progressive neurodegeneration. <i>Neurology</i> , 2018 , 91, e319-e380	26	
215	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. <i>American Journal of Human Genetics</i> , 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. <i>Neuropathology and Applied Neurobiology</i> , 2018 , 44, 687-706	5.2	24
213	Clinical Presentation and Natural History of Hypertrophic Cardiomyopathy in RASopathies. <i>Heart Failure Clinics</i> , 2018 , 14, 225-235	3.3	24
212	Understanding Growth Failure in Costello Syndrome: Increased Resting Energy Expenditure. <i>Journal of Pediatrics</i> , 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. <i>Human Molecular Genetics</i> , 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. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 2389-93	2.5	24
209	Genotype and phenotype spectrum of NRAS germline variants. <i>European Journal of Human Genetics</i> , 2017 , 25, 823-831	5.3	23
208	Wiedemann-Rautenstrauch syndrome: A phenotype analysis. <i>American Journal of Medical Genetics, Part A</i> , 2017 , 173, 1763-1772	2.5	23
207	Activating Mutations of RRAS2 Are a Rare Cause of Noonan Syndrome. <i>American Journal of Human Genetics</i> , 2019 , 104, 1223-1232	11	23
206	A new bioavailable fenretinide formulation with antiproliferative, antimetabolic, and cytotoxic effects on solid tumors. <i>Cell Death and Disease</i> , 2019 , 10, 529	9.8	23
205	Novel SMAD4 mutation causing Myhre syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 1835-40	2.5	23
204	Enhanced human brain associative plasticity in Costello syndrome. <i>Journal of Physiology</i> , 2010 , 588, 3445-356	23	

203	ALL-associated JAK1 mutations confer hypersensitivity to the antiproliferative effect of type I interferon. <i>Blood</i> , 2010 , 115, 3287-95	2.2	23
202	Frameshift mutations at the C-terminus of HIST1H1E result in a specific DNA hypomethylation signature. <i>Clinical Epigenetics</i> , 2020 , 12, 7	7.7	23
201	Molecular Diversity and Associated Phenotypic Spectrum of Germline CBL Mutations. <i>Human Mutation</i> , 2015 , 36, 787-96	4.7	22
200	Mutations in Fibronectin Cause a Subtype of Spondylometaphyseal Dysplasia with "Corner Fractures". <i>American Journal of Human Genetics</i> , 2017 , 101, 815-823	11	22
199	Genomic duplication of PTPN11 is an uncommon cause of Noonan syndrome. <i>American Journal of Medical Genetics, Part A</i> , 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. <i>Proteins: Structure, Function and Bioinformatics</i> , 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. <i>Journal of the Neurological Sciences</i> , 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. <i>Gene</i> , 2017 , 628, 141-145	3.8	20
195	Hydrops fetalis in a preterm newborn heterozygous for the c.4A>G SHOC2 mutation. <i>American Journal of Medical Genetics, Part A</i> , 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. <i>Circulation: Cardiovascular Genetics</i> , 2012 , 5, 317-26	19	
193	Atrioventricular canal defect in patients with RASopathies. <i>European Journal of Human Genetics</i> , 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. <i>Clinical Genetics</i> , 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. <i>Clinical Genetics</i> , 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. <i>Leukemia</i> , 2020 , 34, 2673-2687	10.7	18
189	Defective kinesin binding of TUBB2A causes progressive spastic ataxia syndrome resembling saccinopathy. <i>Human Molecular Genetics</i> , 2018 , 27, 1892-1904	5.6	18
188	Expanding the clinical and molecular spectrum of PRMT7 mutations: 3 additional patients and review. <i>Clinical Genetics</i> , 2018 , 93, 675-681	4	18
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. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 1772-9	2.5	18
186	Long term memory profile of disorders associated with dysregulation of the RAS-MAPK signaling cascade. <i>Behavior Genetics</i> , 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. <i>Journal of Biological Chemistry</i> , 2012 , 287, 27066-77	5.4	18
184	Visual function in Noonan and LEOPARD syndrome. <i>Neuropediatrics</i> , 2008 , 39, 335-40	1.6	18
183	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. <i>Nature Communications</i> , 2020 , 11, 595	17.4	18
182	Expanding the molecular diversity and phenotypic spectrum of glycerol 3-phosphate dehydrogenase 1 deficiency. <i>Journal of Inherited Metabolic Disease</i> , 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. <i>American Journal of Medical Genetics, Part A</i> , 2017 , 173, 1965-1969	2.5	17
180	Neurobehavioral features in individuals with Kabuki syndrome. <i>Molecular Genetics & Genomic Medicine</i> , 2018 , 6, 322-331	2.3	17
179	De Novo Variants Disturbing the Transactivation Capacity of POU3F3 Cause a Characteristic Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 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. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 2786-94	2.5	17
177	PTPN11 mutations in childhood acute lymphoblastic leukemia occur as a secondary event associated with high hyperdiploidy. <i>Leukemia</i> , 2010 , 24, 232-5	10.7	17
176	Copy number variants in autism spectrum disorders. <i>Progress in Neuro-Psychopharmacology and Biological Psychiatry</i> , 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. <i>American Journal of Human Genetics</i> , 2020 , 106, 484-495	11	16
174	Clinical spectrum of Kabuki-like syndrome caused by HNRNPK haploinsufficiency. <i>Clinical Genetics</i> , 2018 , 93, 401-407	4	16
173	Biallelic mutations in DYNC2LI1 are a rare cause of Ellis-van Creveld syndrome. <i>Clinical Genetics</i> , 2018 , 93, 632-639	4	16
172	De Novo Missense Variants in FBXW11 Cause Diverse Developmental Phenotypes Including Brain, Eye, and Digit Anomalies. <i>American Journal of Human Genetics</i> , 2019 , 105, 640-657	11	16
171	Acute lymphoblastic leukaemia in Noonan syndrome. <i>British Journal of Haematology</i> , 2006 , 133, 448-50	4.5	16
170	Atrioventricular canal defect and genetic syndromes: The unifying role of sonic hedgehog. <i>Clinical Genetics</i> , 2019 , 95, 268-276	4	16
169	Biallelic mutations in the homeodomain of NKX6-2 underlie a severe hypomyelinating leukodystrophy. <i>Brain</i> , 2017 , 140, 2550-2556	11.2	15
168	Anti-Hypothalamus and Anti-Pituitary Auto-antibodies in ROHHAD Syndrome: Additional Evidence Supporting an Autoimmune Etiopathogenesis. <i>Hormone Research in Paediatrics</i> , 2019 , 92, 124-132	3.3	15

167	NBAS pathogenic variants: Defining the associated clinical and facial phenotype and genotype-phenotype correlations. <i>Human Mutation</i> , 2019 , 40, 721-728	4.7	15
166	Decreased bone mineral density in Costello syndrome. <i>Molecular Genetics and Metabolism</i> , 2014 , 111, 41-5	3.7	15
165	Phenotypic variability associated with the invariant SHOC2 c.4A>G (p.Ser2Gly) missense mutation. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 3120-5	2.5	15
164	A competitive PCR-based method to measure human fibroblast growth factor receptor 1-4 (FGFR1-4) gene expression. <i>DNA and Cell Biology</i> , 2001 , 20, 367-79	3.6	15
163	Congenital heart defects in Noonan syndrome and RIT1 mutation. <i>Genetics in Medicine</i> , 2016 , 18, 1320	8.1	15
162	Role of DNA Methylation Profile in Diagnosing Astroblastoma: A Case Report and Literature Review. <i>Frontiers in Genetics</i> , 2019 , 10, 391	4.5	14
161	Novel - Fusion Gene in Pediatric Ependymomas Discovered by Clonal Expansion of Stem Cells in Absence of Exogenous Mitogens. <i>Cancer Research</i> , 2017 , 77, 5860-5872	10.1	14
160	Cyclosporine attenuates cardiomyocyte hypertrophy induced by RAF1 mutants in Noonan and LEOPARD syndromes. <i>Journal of Molecular and Cellular Cardiology</i> , 2011 , 51, 4-15	5.8	14
159	Rat nicastrin gene: cDNA isolation, mRNA variants and expression pattern analysis. <i>Molecular Brain Research</i> , 2005 , 136, 12-22		14
158	Differences in the prevalence of PTPN11 mutations in FAB M5 paediatric acute myeloid leukaemia. <i>British Journal of Haematology</i> , 2005 , 130, 801-3	4.5	14
157	Activating MRAS mutations cause Noonan syndrome associated with hypertrophic cardiomyopathy. <i>Human Molecular Genetics</i> , 2020 , 29, 1772-1783	5.6	14
156	Psychopathological features in Noonan syndrome. <i>European Journal of Paediatric Neurology</i> , 2018 , 22, 170-177	3.8	13
155	Fibroblast growth factor receptor mutational screening in newborns affected by metopic synostosis. <i>Childs Nervous System</i> , 1999 , 15, 389-93; discussion 393-4	1.7	13
154	Colorectal cancer spheroid biobanks: multi-level approaches to drug sensitivity studies. <i>Cell Biology and Toxicology</i> , 2018 , 34, 459-469	7.4	12
153	Natural history and life-threatening complications in Myhre syndrome and review of the literature. <i>European Journal of Pediatrics</i> , 2016 , 175, 1307-15	4.1	12
152	Clinical lumping and molecular splitting of LEOPARD and NF1/NF1-Noonan syndromes. <i>American Journal of Medical Genetics, Part A</i> , 2007 , 143A, 1009-11	2.5	12
151	Infantile-Onset Syndromic Cerebellar Ataxia and CACNA1G Mutations. <i>Pediatric Neurology</i> , 2020 , 104, 40-45	2.9	12
150	Histone H3.3 beyond cancer: Germline mutations in cause a previously unidentified neurodegenerative disorder in 46 patients. <i>Science Advances</i> , 2020 , 6,	14.3	12

149	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. <i>American Journal of Human Genetics</i> , 2021 , 108, 502-516	11	12
148	Aberrant HRAS transcript processing underlies a distinctive phenotype within the RASopathy clinical spectrum. <i>Human Mutation</i> , 2017 , 38, 798-804	4.7	11
147	Pain in individuals with RASopathies: Prevalence and clinical characterization in a sample of 80 affected patients. <i>American Journal of Medical Genetics, Part A</i> , 2019 , 179, 940-947	2.5	11
146	SHOC2 subcellular shuttling requires the KEKE motif-rich region and N-terminal leucine-rich repeat domain and impacts on ERK signalling. <i>Human Molecular Genetics</i> , 2016 , 25, 3824-3835	5.6	11
145	Isoform-specific NF1 mRNA levels correlate with disease severity in Neurofibromatosis type 1. <i>Orphanet Journal of Rare Diseases</i> , 2019 , 14, 261	4.2	11
144	The phenotypic and molecular spectrum of PEHO syndrome and PEHO-like disorders. <i>Brain</i> , 2017 , 140, e49	11.2	11
143	EcoRI, Rsal, and MspI RFLPs of the COL1A2 gene (type I collagen) in the Cayapa, a Native American population of Ecuador. <i>Human Biology</i> , 1994 , 66, 979-89	1.2	11
142	Genetic characterization of the Cayapa Indians of Ecuador and their genetic relationships to other Native American populations. <i>Human Biology</i> , 1994 , 66, 299-322	1.2	11
141	De Novo VPS4A Mutations Cause Multisystem Disease with Abnormal Neurodevelopment. <i>American Journal of Human Genetics</i> , 2020 , 107, 1129-1148	11	11
140	Variants of SOS2 are a rare cause of Noonan syndrome with particular predisposition for lymphatic complications. <i>European Journal of Human Genetics</i> , 2021 , 29, 51-60	5.3	11
139	Expanding the clinical spectrum associated with PACS2 mutations. <i>Clinical Genetics</i> , 2019 , 95, 525-531	4	10
138	POGZ-related epilepsy: Case report and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2019 , 179, 1631-1636	2.5	10
137	A second cohort of CHD3 patients expands the molecular mechanisms known to cause Snijders Blok-Campeau syndrome. <i>European Journal of Human Genetics</i> , 2020 , 28, 1422-1431	5.3	10
136	DNA Methylation Profiling for Diagnosing Undifferentiated Sarcoma with Capicua Transcriptional Receptor () Alterations. <i>International Journal of Molecular Sciences</i> , 2020 , 21,	6.3	10
135	Heterozygous missense mutations in NFATC1 are associated with atrioventricular septal defect. <i>Human Mutation</i> , 2018 , 39, 1428-1441	4.7	10
134	Dystonia in Costello syndrome. <i>Parkinsonism and Related Disorders</i> , 2012 , 18, 798-800	3.6	10
133	Analysis of three RFLPs of the COL1A2 (Type I Collagen) in the Amhara and the Oromo of Ethiopia. <i>Annals of Human Biology</i> , 2002 , 29, 432-41	1.7	10
132	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. <i>Genetics in Medicine</i> , 2020 , 22, 1338-1347	8.1	9

131	Co-occurring WARS2 and CHRNA6 mutations in a child with a severe form of infantile parkinsonism. <i>Parkinsonism and Related Disorders</i> , 2020 , 72, 75-79	3.6	9
130	Functional evaluation of natural killer cell cytotoxic activity in NFKB2-mutated patients. <i>Immunology Letters</i> , 2018 , 194, 40-43	4.1	9
129	Differential Effects of HRAS Mutation on LTP-Like Activity Induced by Different Protocols of Repetitive Transcranial Magnetic Stimulation. <i>Brain Stimulation</i> , 2016 , 9, 33-8	5.1	9
128	Clinical and functional characterization of two novel ZBTB20 mutations causing Primrose syndrome. <i>Human Mutation</i> , 2018 , 39, 959-964	4.7	9
127	A PTPN11 allele encoding a catalytically impaired SHP2 protein in a patient with a Noonan syndrome phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 2351-5	2.5	9
126	Increased sleep spindle activity in patients with Costello syndrome (HRAS gene mutation). <i>Journal of Clinical Neurophysiology</i> , 2011 , 28, 314-8	2.2	9
125	Reactive oxygen species and epidermal growth factor are antagonistic cues controlling SHP-2 dimerization. <i>Molecular and Cellular Biology</i> , 2012 , 32, 1998-2009	4.8	9
124	Induction of both CD8+ and CD4+ T-cell-mediated responses in colorectal cancer patients by colon antigen-1. <i>Clinical Cancer Research</i> , 2008 , 14, 7292-303	12.9	9
123	VarGenius executes cohort-level DNA-seq variant calling and annotation and allows to manage the resulting data through a PostgreSQL database. <i>BMC Bioinformatics</i> , 2018 , 19, 477	3.6	9
122	Neurotransmitter trafficking defect in a patient with clathrin (CLTC) variation presenting with intellectual disability and early-onset parkinsonism. <i>Parkinsonism and Related Disorders</i> , 2019 , 61, 207-210	3.6	9
121	De novo p.T362R mutation in MORC2 causes early onset cerebellar ataxia, axonal polyneuropathy and nocturnal hypoventilation. <i>Brain</i> , 2017 , 140, e34	11.2	8
120	Alterations in metabolic patterns have a key role in diagnosis and progression of primrose syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2017 , 173, 1896-1902	2.5	8
119	Prevalence, Type, and Molecular Spectrum of Mutations in Patients with Neurofibromatosis Type 1 and Congenital Heart Disease. <i>Genes</i> , 2019 , 10,	4.2	8
118	GH Therapy and first final height data in Noonan-like syndrome with loose anagen hair (Mazzanti syndrome). <i>American Journal of Medical Genetics, Part A</i> , 2013 , 161A, 2756-61	2.5	8
117	Hyperthrophic cardiomyopathy and the PTPN11 gene. <i>American Journal of Medical Genetics, Part A</i> , 2005 , 136, 93-4	2.5	8
116	SCUBE3 loss-of-function causes a recognizable recessive developmental disorder due to defective bone morphogenetic protein signaling. <i>American Journal of Human Genetics</i> , 2021 , 108, 115-133	11	8
115	Biallelic Variants in the Nuclear Pore Complex Protein NUP93 Are Associated with Non-progressive Congenital Ataxia. <i>Cerebellum</i> , 2019 , 18, 422-432	4.3	7
114	Bi-allelic LoF NRROS Variants Impairing Active TGF- β Delivery Cause a Severe Infantile-Onset Neurodegenerative Condition with Intracranial Calcification. <i>American Journal of Human Genetics</i> , 2020 , 106, 559-569	11	7

113	A mild form of adenylosuccinate lyase deficiency in absence of typical brain MRI features diagnosed by whole exome sequencing. <i>Italian Journal of Pediatrics</i> , 2017 , 43, 65	3.2	7
112	Expanding the histopathological spectrum of CFL2-related myopathies. <i>Clinical Genetics</i> , 2018 , 93, 1234-1239	7	
111	Transcriptional hallmarks of Noonan syndrome and Noonan-like syndrome with loose anagen hair. <i>Human Mutation</i> , 2012 , 33, 703-9	4.7	7
110	Noonan syndrome-like disorder with loose anagen hair: a second case with neuroblastoma. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 1902-7	2.5	7
109	BCM-95 and (2-hydroxypropyl)-β-cyclodextrin reverse autophagy dysfunction and deplete stored lipids in Sap C-deficient fibroblasts. <i>Human Molecular Genetics</i> , 2015 , 24, 4198-211	5.6	7
108	Geroderma osteodysplastica maps to a 4 Mb locus on chromosome 1q24. <i>American Journal of Medical Genetics, Part A</i> , 2008 , 146A, 3034-7	2.5	7
107	Rapid communication: nucleotide sequence of porcine and ovine tRNA(Lys) and ATPase8 mitochondrial genes. <i>Journal of Animal Science</i> , 1998 , 76, 2207-8	0.7	7
106	A Recurrent Gain-of-Function Mutation in CLCN6, Encoding the CLC-6 Cl/H-Exchanger, Causes Early-Onset Neurodegeneration. <i>American Journal of Human Genetics</i> , 2020 , 107, 1062-1077	11	7
105	Musculo-skeletal phenotype of Costello syndrome and cardio-facio-cutaneous syndrome: insights on the functional assessment status. <i>Orphanet Journal of Rare Diseases</i> , 2021 , 16, 43	4.2	7
104	Exome sequencing in children of women with skewed X-inactivation identifies atypical cases and complex phenotypes. <i>European Journal of Paediatric Neurology</i> , 2017 , 21, 475-484	3.8	6
103	Clinical and functional characterization of a novel RASopathy-causing SHOC2 mutation associated with prenatal-onset hypertrophic cardiomyopathy. <i>Human Mutation</i> , 2019 , 40, 1046-1056	4.7	6
102	Structural Determinants of Phosphopeptide Binding to the N-Terminal Src Homology 2 Domain of the SHP2 Phosphatase. <i>Journal of Chemical Information and Modeling</i> , 2020 , 60, 3157-3171	6.1	6
101	Ethnobotany of dye plants in Southern Italy, Mediterranean Basin: floristic catalog and two centuries of analysis of traditional botanical knowledge heritage. <i>Journal of Ethnobiology and Ethnomedicine</i> , 2020 , 16, 31	3.9	6
100	Atypical cardiac defects in patients with RASopathies: Updated data on CARNET study. <i>Birth Defects Research</i> , 2020 , 112, 725-731	2.9	6
99	Biallelic TRNT1 variants in a child with B cell immunodeficiency, periodic fever and developmental delay without sideroblastic anemia (SIFD variant). <i>Immunology Letters</i> , 2020 , 225, 64-65	4.1	6
98	A syndromic extreme insulin resistance caused by biallelic mutations in exon 10. <i>European Journal of Endocrinology</i> , 2017 , 177, K21-K27	6.5	6
97	Early fetal death associated with compound heterozygosity for Noonan syndrome-causative PTPN11 mutations. <i>American Journal of Medical Genetics, Part A</i> , 2007 , 143A, 1249-52	2.5	6
96	Rapid communication: nucleotide sequence of chamois, alpine ibex, and red deer tRNA(Lys) and ATPase8 mitochondrial genes. <i>Journal of Animal Science</i> , 1999 , 77, 3398-9	0.7	6

95	Mitochondrial and Peroxisomal Alterations Contribute to Energy Dysmetabolism in Riboflavin Transporter Deficiency. <i>Oxidative Medicine and Cellular Longevity</i> , 2020 , 2020, 6821247	6.7	6
94	TARP syndrome: Long-term survival, anatomic patterns of congenital heart defects, differential diagnosis and pathogenetic considerations. <i>European Journal of Medical Genetics</i> , 2019 , 62, 103534	2.6	6
93	Primrose syndrome: Characterization of the phenotype in 42 patients. <i>Clinical Genetics</i> , 2020 , 97, 890-904		5
92	Data on cardiac defects, morbidity and mortality in patients affected by RASopathies. CARNET study results. <i>Data in Brief</i> , 2018 , 16, 649-654	1.2	5
91	Behavioral phenotype in Costello syndrome with atypical mutation: a case report. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2015 , 168B, 66-71	3.5	5
90	Loss of CBL E3-ligase activity in B-lineage childhood acute lymphoblastic leukaemia. <i>British Journal of Haematology</i> , 2012 , 159, 115-9	4.5	5
89	HIPK2-T566 autophosphorylation diversely contributes to UV- and doxorubicin-induced HIPK2 activation. <i>Oncotarget</i> , 2017 , 8, 16744-16754	3.3	5
88	When to test fetuses for RASopathies? Proposition from a systematic analysis of 352 multicenter cases and a postnatal cohort. <i>Genetics in Medicine</i> , 2021 , 23, 1116-1124	8.1	5
87	Cardiac Defects and Genetic Syndromes: Old Uncertainties and New Insights. <i>Genes</i> , 2021 , 12,	4.2	5
86	Upfront treatment with mTOR inhibitor everolimus in pediatric low-grade gliomas: A single-center experience. <i>International Journal of Cancer</i> , 2020 , 148, 2522	7.5	5
85	Somatic mosaicism represents an underestimated event underlying collagen 6-related disorders. <i>European Journal of Paediatric Neurology</i> , 2017 , 21, 873-883	3.8	4
84	Worldwide distribution of phosphoglucomutase 1 (PGM1) polymorphism detected by isoelectric focusing: A review. <i>International Journal of Anthropology</i> , 1994 , 9, 81-112		4
83	Red-cell enzyme polymorphisms in the Reggio Calabria province (Italy). <i>Human Heredity</i> , 1990 , 40, 308-10.	1	4
82	Targeting Oncogenic Src Homology 2 Domain-Containing Phosphatase 2 (SHP2) by Inhibiting Its Protein-Protein Interactions. <i>Journal of Medicinal Chemistry</i> , 2021 , 64, 15973-15990	8.3	4
81	Skeletal abnormalities are common features in Aymé-Gripp syndrome. <i>Clinical Genetics</i> , 2020 , 97, 362-369	4	
80	Phenotypic Features of Epidermolysis Bullosa Simplex due to KLHL24 Mutations in 3 Italian Cases. <i>Acta Dermato-Venereologica</i> , 2019 , 99, 238-239	2.2	4
79	A genotype-first approach to exploring Mendelian cardiovascular traits with clear external manifestations. <i>Genetics in Medicine</i> , 2021 , 23, 94-102	8.1	4
78	Melanotic Neuroectodermal Tumor of Infancy (MNTI) and Pineal Anlage Tumor (PAT) Harbor A Medulloblastoma Signature by DNA Methylation Profiling. <i>Cancers</i> , 2021 , 13,	6.6	4

77	The activating p.Ser466Arg change in STAT1 causes a peculiar phenotype with features of interferonopathies. <i>Clinical Genetics</i> , 2019 , 96, 585-589	4	3
76	Very mild isolated intellectual disability caused by adenylosuccinate lyase deficiency: a new phenotype. <i>Molecular Genetics and Metabolism Reports</i> , 2020 , 23, 100592	1.8	3
75	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. <i>BMC Pediatrics</i> , 2020 , 20, 120	2.6	3
74	Whole exome sequencing in an Italian family with isolated maxillary canine agenesis and canine eruption anomalies. <i>Archives of Oral Biology</i> , 2018 , 91, 96-102	2.8	3
73	Pediatric patients with RASopathy-associated hypertrophic cardiomyopathy: the multifaceted consequences of PTPN11 mutations. <i>Orphanet Journal of Rare Diseases</i> , 2019 , 14, 163	4.2	3
72	Congenital myopathy with protein aggregates and nemaline bodies related to CFL2 mutations. <i>Neuromuscular Disorders</i> , 2017 , 27, S186	2.9	3
71	Polymorphism at position 882 of the fibroblast growth factor receptor 3 (FGFR3) gene detected by SSCP analysis. <i>Molecular and Cellular Probes</i> , 1998 , 12, 335-7	3.3	3
70	Low-Grade Gliomas in Patients with Noonan Syndrome: Case-Based Review of the Literature. <i>Diagnostics</i> , 2020 , 10,	3.8	3
69	Dissecting the Role of PCDH19 in Clustering Epilepsy by Exploiting Patient-Specific Models of Neurogenesis. <i>Journal of Clinical Medicine</i> , 2021 , 10,	5.1	3
68	Melanocytic nevi in RASopathies: insights on dermatological diagnostic handles. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2021 , 35, e83-e85	4.6	3
67	DICER1-associated malignancies mimicking germ cell neoplasms: Report of two cases and review of the literature. <i>Pathology Research and Practice</i> , 2021 , 225, 153553	3.4	3
66	Rare and de novo coding variants in chromodomain genes in Chiari I malformation. <i>American Journal of Human Genetics</i> , 2021 , 108, 100-114	11	3
65	Induced Pluripotent Stem Cells (iPSCs) and Gene Therapy: A New Era for the Treatment of Neurological Diseases.. <i>International Journal of Molecular Sciences</i> , 2021 , 22,	6.3	3
64	Cost-effectiveness of exome sequencing: an Italian pilot study on undiagnosed patients. <i>New Genetics and Society</i> , 2019 , 38, 249-263	1.9	2
63	Refinement of the clinical and mutational spectrum of UBE2A deficiency syndrome. <i>Clinical Genetics</i> , 2020 , 98, 172-178	4	2
62	Developmental and epileptic encephalopathy due to SQT2 genomic variants: Emerging features of a syndromic condition. <i>Epilepsy and Behavior</i> , 2020 , 108, 107097	3.2	2
61	Cantù syndrome versus Zimmermann-Laband syndrome: Report of nine individuals with ABCC9 variants. <i>European Journal of Medical Genetics</i> , 2020 , 63, 103996	2.6	2
60	Defining the phenotype of FHF1 developmental and epileptic encephalopathy. <i>Epilepsia</i> , 2020 , 61, e71-e78	2	2

59	Pathogenic PTPN11 variants involving the poly-glutamine Gln -Gln -Gln stretch highlight the relevance of helix B in SHP2@ functional regulation. <i>Human Mutation</i> , 2020 , 41, 1171-1182	4.7	2
58	Progressive extreme heterotopic calcification. <i>American Journal of Medical Genetics, Part A</i> , 2013 , 161A, 1706-13	2.5	2
57	Efficient one-step chromatographic purification and functional characterization of recombinant human Saposin C. <i>Protein Expression and Purification</i> , 2011 , 78, 209-15	2	2
56	Genetic heterogeneity among the Hindus and their relationships with the other "Caucasoid" populations: new data on Punjab-Haryana and Rajasthan Indian states. <i>American Journal of Physical Anthropology</i> , 1995 , 98, 257-73	2.5	2
55	Genotype-cardiac phenotype correlations in a large single-center cohort of patients affected by RASopathies: Clinical implications and literature review. <i>American Journal of Medical Genetics, Part A</i> , 2021 ,	2.5	2
54	Hypertrophic Cardiomyopathy in RASopathies: Diagnosis, Clinical Characteristics, Prognostic Implications, and Management. <i>Heart Failure Clinics</i> , 2022 , 18, 19-29	3.3	2
53	SPRED2 loss-of-function causes a recessive Noonan syndrome-like phenotype. <i>American Journal of Human Genetics</i> , 2021 , 108, 2112-2129	11	2
52	Noonan Syndrome		2
51	Management of cardiac aspects in children with Noonan syndrome - results from a European clinical practice survey among paediatric cardiologists. <i>European Journal of Medical Genetics</i> , 2021 , 65, 104372	2.6	2
50	A Recurrent Pathogenic Variant of Underlies Autosomal Recessive Congenital Muscular Dystrophy With Cataracts and Intellectual Disability: Evidence for a Founder Effect in Southern Italy. <i>Frontiers in Genetics</i> , 2020 , 11, 565868	4.5	2
49	Antioxidant Amelioration of Riboflavin Transporter Deficiency in Motoneurons Derived from Patient-Specific Induced Pluripotent Stem Cells. <i>International Journal of Molecular Sciences</i> , 2020 , 21,	6.3	2
48	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. <i>Human Mutation</i> , 2020 , 41, 2087-2093	4.7	2
47	Functional analysis of variants and their proximal interactomes implicates impaired kinase activity and chromatin maintenance defects in their pathogenesis. <i>Journal of Medical Genetics</i> , 2020 ,	5.8	2
46	Mitochondrial Abnormalities in Induced Pluripotent Stem Cells-Derived Motor Neurons from Patients with Riboflavin Transporter Deficiency. <i>Antioxidants</i> , 2020 , 9,	7.1	2
45	Etanercept as a successful therapy in autoinflammatory syndrome related to TRNT1 mutations: a case-based review. <i>Clinical Rheumatology</i> , 2021 , 40, 4341-4348	3.9	2
44	Clinical and molecular characterization of patients with adenylosuccinate lyase deficiency. <i>Orphanet Journal of Rare Diseases</i> , 2021 , 16, 112	4.2	2
43	Copy number variation analysis implicates novel pathways in patients with oculo-auriculo-vertebral-spectrum and congenital heart defects. <i>Clinical Genetics</i> , 2021 , 100, 268-279	4	2
42	Embryopathy Following Maternal Biliopancreatic Diversion: Is Bariatric Surgery Really Safe?. <i>Obesity Surgery</i> , 2021 , 31, 445-450	3.7	2

41	The clinical significance of A2ML1 variants in Noonan syndrome has to be reconsidered. <i>European Journal of Human Genetics</i> , 2021 , 29, 524-527	5.3	2
40	De novo DHDDS variants cause a neurodevelopmental and neurodegenerative disorder with myoclonus. <i>Brain</i> , 2021 ,	11.2	2
39	Clinical variability of neurofibromatosis 1: A modifying role of cooccurring PTPN11 variants and atypical brain MRI findings. <i>Clinical Genetics</i> , 2021 , 100, 563-572	4	2
38	Hyperactive HRAS dysregulates energetic metabolism in fibroblasts from patients with Costello syndrome via enhanced production of reactive oxidizing species. <i>Human Molecular Genetics</i> , 2021 ,	5.6	2
37	Compound heterozygosity for PTPN11 variants in a subject with Noonan syndrome provides insights into the mechanism of SHP2-related disorders. <i>Clinical Genetics</i> , 2021 , 99, 457-461	4	2
36	The seventh international RASopathies symposium: Pathways to a cure-expanding knowledge, enhancing research, and therapeutic discovery.. <i>American Journal of Medical Genetics, Part A</i> , 2022 ,	2.5	2
35	Visual perception skills: a comparison between patients with Noonan syndrome and 22q11.2 deletion syndrome. <i>Genes, Brain and Behavior</i> , 2017 , 16, 627-634	3.6	1
34	Defining language disorders in children and adolescents with Noonan Syndrome. <i>Molecular Genetics & Genomic Medicine</i> , 2020 , 8, e1069	2.3	1
33	Obsessive Compulsive Symptoms and Psychopathological Profile in Children and Adolescents with KBG syndrome. <i>Brain Sciences</i> , 2019 , 9,	3.4	1
32	Prevalence of bladder cancer in Costello syndrome: new insights to drive clinical decision-making.. <i>Clinical Genetics</i> , 2022 ,	4	1
31	Management of growth failure and other endocrine aspects in patients with Noonan syndrome across Europe: A sub-analysis of a European clinical practice survey.. <i>European Journal of Medical Genetics</i> , 2022 , 65, 104404	2.6	1
30	A survey of six genetic markers on the populations of Punjab and Rajasthan (India). <i>Gene Geography: A Computerized Bulletin on Human Gene Frequencies</i> , 1991 , 5, 113-21		1
29	PTPN11 Mutational Spectrum in Juvenile Myelomonocytic Leukemia and Noonan Syndrome.. <i>Blood</i> , 2004 , 104, 3417-3417	2.2	1
28	Characterization of bone homeostasis in individuals affected by cardio-facio-cutaneous syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 188, 414	2.5	1
27	Novel diagnostic DNA methylation epigenotypes expand and refine the epigenetic landscapes of Mendelian disorders.. <i>Human Genetics and Genomics Advances</i> , 2022 , 3, 100075	0.8	1
26	Neonatal Manifestations of Chronic Granulomatous Disease: MAS/HLH and Necrotizing Pneumonia as Unusual Phenotypes and Review of the Literature. <i>Journal of Clinical Immunology</i> , 2021 , 1	5.7	1
25	Further insight into the neurobehavioral pattern of children carrying the 2p16.3 heterozygous deletion involving NRXN1: Report of five new cases. <i>Genes, Brain and Behavior</i> , 2020 , 19, e12687	3.6	1
24	Biallelic mutations in RNF220 cause laminopathies featuring leukodystrophy, ataxia and deafness. <i>Brain</i> , 2021 , 144, 3020-3035	11.2	1

23	Biallelic Variants Associated with Intellectual Disability and Neurodevelopmental Disorders Alter TRESK Channel Activity. <i>International Journal of Molecular Sciences</i> , 2021 , 22,	6.3	1
22	Expanding the clinical phenotype of the ultra-rare Skraban-Deardorff syndrome: Two novel individuals with WDR26 loss-of-function variants and a literature review. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 1712-1720	2.5	1
21	Epilepsy and Mutations: Phenotypes, Natural History and Genotype-Phenotype Correlations. <i>Genes</i> , 2021 , 12,	4.2	1
20	Bone tissue homeostasis and risk of fractures in Costello syndrome: A 4-year follow-up study.. <i>American Journal of Medical Genetics, Part A</i> , 2021 ,	2.5	1
19	Metabolic profiling of Costello syndrome: Insights from a single-center cohort.. <i>European Journal of Medical Genetics</i> , 2022 , 104439	2.6	0
18	RASopathies and hemostatic abnormalities: key role of platelet dysfunction. <i>Orphanet Journal of Rare Diseases</i> , 2021 , 16, 499	4.2	0
17	Risk of autoimmune diseases in patients with RASopathies: systematic study of humoral and cellular immunity. <i>Orphanet Journal of Rare Diseases</i> , 2021 , 16, 410	4.2	0
16	European Medical Education Initiative on Noonan syndrome: A clinical practice survey assessing the diagnosis and clinical management of individuals with Noonan syndrome across Europe. <i>European Journal of Medical Genetics</i> , 2021 , 65, 104371	2.6	0
15	Enlarged spinal nerve roots in RASopathies: Report of two cases. <i>European Journal of Medical Genetics</i> , 2021 , 64, 104187	2.6	0
14	Functional Genomics for Undiagnosed Patients: The Impact of Small GTPases Signaling Dysregulation at Pan-Embryo Developmental Scale. <i>Frontiers in Cell and Developmental Biology</i> , 2021 , 9, 642235	5.7	0
13	Broadening the phenotypic spectrum of Beta3GalT6-associated phenotypes. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 3153-3160	2.5	0
12	Childhood-onset dystonia-causing KMT2B variants result in a distinctive genomic hypermethylation profile. <i>Clinical Epigenetics</i> , 2021 , 13, 157	7.7	0
11	A Rare Case of Brachyolmia with Amelogenesis Imperfecta Caused by a New Pathogenic Splicing Variant in. <i>Genes</i> , 2021 , 12,	4.2	0
10	Complex Presentation of Hao-Fountain Syndrome Solved by Exome Sequencing Highlighting Co-Occurring Genomic Variants. <i>Genes</i> , 2022 , 13, 889	4.2	0
9	Multidisciplinary Management of Costello Syndrome: Current Perspectives. <i>Journal of Multidisciplinary Healthcare</i> , Volume 15, 1277-1296	2.8	0
8	Noonan Syndrome and Other RAS/MAPK Pathway Syndromes 122-130		
7	Germline PTPN11 mutation affecting exon 8 in a case of syndromic juvenile myelomonocytic leukemia. <i>Leukemia Research</i> , 2011 , 35, e13-4	2.7	
6	Noonan Syndrome and PTPN11 Mutations 2007 , 263-272		

- 5 Toward the in vitro understanding of iPSC nucleoskeletal and cytoskeletal biology, and their relevance for organoid development **2022**, 137-150
- 4 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
- 3 Linkage disequilibrium at the human phosphoglucomutase 1 locus. *Human Biology*, **1994**, 66, 669-81 1.2
- 2 PTPN11 and RAS Gene Mutation Pattern Identifies an Unique Feature of Upregulated RAS Function in Infant ALL.. *Blood*, **2004**, 104, 996-996 2.2
- 1 No metagenomic evidence of tumorigenic viruses in cancers from a selected cohort of immunosuppressed subjects. *Scientific Reports*, **2019**, 9, 19815 4.9