Emanuele Agolini

List of Publications by Year
in descending order

72	1,292	471509 17	414414 32
papers	citations	h-index	g-index

76	76	76	2670
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A Novel CHEK2 Variant Identified by Next-Generation Sequencing in an Italian Family with Li-Fraumeni Syndrome: Case Report. SN Comprehensive Clinical Medicine, 2022, 4, 1.	0.6	0
2	De Novo Mutation in KMT2C Manifesting as Kleefstra Syndrome 2: Case Report and Literature Review. Pediatric Reports, 2022, 14, 131-139.	1.3	12
3	The Fight Just Bornâ€"Neonatal Cancer: Rare Occurrence with a Favorable Outcome but Challenging Management. Cancers, 2022, 14, 2244.	3.7	0
4	Expanding the novel <scp> MAPKAPK5 </scp> â€"related developmental disorderâ€™s genotypeâ€phenotype correlation: patient report and 19 months followâ€up. Clinical Genetics, 2022, , .	2.0	2
5	Liquid Biopsy with Detection of NRASQ61K Mutation in Cerebrospinal Fluid: An Alternative Tool for the Diagnosis of Primary Pediatric Leptomeningeal Melanoma. Diagnostics, 2022, 12, 1609.	2.6	2
6	Fetal early motor neuron disruption and prenatal molecular diagnosis in a severe BICD2 â€opathy. American Journal of Medical Genetics, Part A, 2021, 185, 1509-1514.	1.2	1
7	Medulloblastoma Associated with Down Syndrome: From a Rare Event Leading to a Pathogenic Hypothesis. Diagnostics, 2021, 11, 254.	2.6	3
8	HLA-haploidentical TCRÎ±Î²+/CD19+-depleted stem cell transplantation in children and young adults with Fanconi anemia. Blood Advances, 2021, 5, 1333-1339.	5.2	22
9	Inhibition of HECT E3 ligases as potential therapy for COVID-19. Cell Death and Disease, 2021, 12, 310.	6.3	33
10	Biallelic hypomorphic variants in ALDH1A2 cause a novel lethal human multiple congenital anomaly syndrome encompassing diaphragmatic, pulmonary, and cardiovascular defects. Human Mutation, 2021, 42, 506-519.	2.5	12
11	Molecular Characterization of Medulloblastoma in a Patient with Neurofibromatosis Type 1: Case Report and Literature Review. Diagnostics, 2021, 11, 647.	2.6	4
12	Expansion of the clinical and molecular spectrum of an <scp>XPD</scp>â€related disorder linked to biallelic mutations in <scp>ERCC2</scp> gene. Clinical Genetics, 2021, 99, 842-848.	2.0	4
13	ANKRD11 variants: <scp>KBG</scp> syndrome and beyond. Clinical Genetics, 2021, 100, 187-200.	2.0	21
14	<scp>TSPEAR</scp> variants are primarily associated with ectodermal dysplasia and tooth agenesis but not hearing loss: A novel cohort study. American Journal of Medical Genetics, Part A, 2021, 185, 2417-2433.	1.2	10
15	Whole Exome Sequencing Is the Minimal Technological Approach in Probands Born to Consanguineous Couples. Genes, 2021, 12, 962.	2.4	0
16	Clinical presentation and molecular characterization of a novel patient with variant <scp>POC1A</scp>â€related syndrome. Clinical Genetics, 2021, 99, 540-546.	2.0	7
17	Pediatric gastrointestinal stromal tumor: Report of two novel patients harboring germline variants in SDHB and SDHC genes. Cancer Genetics, 2020, 241, 61-65.	0.4	4
18	Expanding the clinical and molecular spectrum of lethal congenital contracture syndrome 8 associated with biallelic variants of ADCY6. Clinical Genetics, 2020, 97, 649-654.	2.0	4

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19	COL1â€related overlap disorder: A novel connective tissue disorder incorporating the osteogenesis imperfecta/Ehlersâ€Danlos syndrome overlap. Clinical Genetics, 2020, 97, 396-406.	2.0	27
20	Genetic identification and molecular modeling characterization of a novel POU3F4 variant in two Italian deaf brothers. International Journal of Pediatric Otorhinolaryngology, 2020, 129, 109790.	1.0	8
21	Infantile-Onset Syndromic Cerebellar Ataxia and CACNA1G Mutations. Pediatric Neurology, 2020, 104, 40-45.	2.1	17
22	Biallelic mutations in the TOGARAM1 gene cause a novel primary ciliopathy. Journal of Medical Genetics, 2020, 58, jmedgenet-2020-106833.	3.2	12
23	WWP1 germline variants are associated with normocephalic autism spectrum disorder. Cell Death and Disease, 2020, 11, 529.	6.3	5
24	Microcephalic osteodysplastic primordial dwarfism type II and pachygyria: Morphometric analysis in a 2â€yearâ€old girl. American Journal of Medical Genetics, Part A, 2020, 182, 2372-2376.	1.2	2
25	Cancer Predisposition Syndromes and Medulloblastoma in the Molecular Era. Frontiers in Oncology, 2020, 10, 566822.	2.8	17
26	COVID-19 and Genetic Variants of Protein Involved in the SARS-CoV-2 Entry into the Host Cells. Genes, 2020, 11, 1010.	2.4	88
27	Low-Grade Gliomas in Patients with Noonan Syndrome: Case-Based Review of the Literature. Diagnostics, 2020, 10, 582.	2.6	21
28	Episignatures Stratifying Helsmoortel-Van Der Aa Syndrome Show Modest Correlation with Phenotype. American Journal of Human Genetics, 2020, 107, 555-563.	6.2	32
29	Exon-Trapping Assay Improves Clinical Interpretation of COL11A1 and COL11A2 Intronic Variants in Stickler Syndrome Type 2 and Otospondylomegaepiphyseal Dysplasia. Genes, 2020, 11, 1513.	2.4	11
30	A novel patient with <scp>Whiteâ€“Sutton</scp> syndrome refines the mutational and clinical repertoire of the POGZâ€related phenotype and suggests further observations. American Journal of Medical Genetics, Part A, 2020, 182, 1791-1795.	1.2	10
31	14q12q13.2 microdeletion syndrome: Clinical characterization of a new patient, review of the literature, and further evidence of a candidate region for CNS anomalies. Molecular Genetics & Genomic Medicine, 2020, 8, e1289.	1.2	5
32	Refinement of the clinical and mutational spectrum of <scp>UBE2A</scp> deficiency syndrome. Clinical Genetics, 2020, 98, 172-178.	2.0	5
33	TUBB3 E410K syndrome: Case report and review of the clinical spectrum of TUBB3 mutations. American Journal of Medical Genetics, Part A, 2020, 182, 1977-1984.	1.2	15
34	Further delineation of the neurodevelopmental phenotypic spectrum associated to 14q11.2 microduplication. Neurological Sciences, 2020, 41, 3751-3753.	1.9	3
35	The splice c.1815G>A variant in KIAA0586 results in a phenotype bridging short-rib-polydactyly and oral-facial-digital syndrome. Medicine (United States), 2020, 99, e19169.	1.0	7
36	KBG syndrome: Common and uncommon clinical features based on 31 new patients. American Journal of Medical Genetics, Part A, 2020, 182, 1073-1083.	1.2	27

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37	Phenytoin intoxication associated with omeprazole administration in a child with defective CYP2C9. European Journal of Clinical Pharmacology, 2020, 76, 731-732.	1.9	3
38	CUGC for hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome. European Journal of Human Genetics, 2020, 28, 982-987.	2.8	3
39	CUGC for lysinuric protein intolerance (LPI). European Journal of Human Genetics, 2020, 28, 1129-1134.	2.8	4
40	Novel congenital disorder of O-linked glycosylation caused by GALNT2 loss of function. Brain, 2020, 143, 1114-1126.	7.6	46
41	The p.Arg377Trp variant in ACTL6A underlines a recognizable BAFâ€opathy phenotype. Clinical Genetics, 2020, 97, 672-674.	2.0	4
42	Nonsense variant of ATP8B1 gene in heterozygosis and benign recurrent intrahepatic cholestasis: A case report and review of literature. World Journal of Hepatology, 2020, 12, 64-71.	2.0	9
43	Structural modeling of a novel TERC variant in a patient with aplastic anemia and short telomeres. Annals of Hematology, 2019, 98, 805-807.	1.8	1
44	P.04.15 THE DISCOVERY OF A NOVEL GENETIC MUTATION IN BENIGN RECURRENT INTRAHEPATIC CHOLESTASIS WIDENS THE INSIGHTS INTO DISORDER RELATED-MOLECULAR ANALYSIS. Digestive and Liver Disease, 2019, 51, e178.	0.9	0
45	ALG12-CDG: novel glycophenotype insights endorse the molecular defect. Glycoconjugate Journal, 2019, 36, 461-472.	2.7	16
46	Hermansky-Pudlak Syndrome Subtype 4 (HPS-4): A Novel Mutation in a 44 Years Old Italian Woman with Severe Pulmonary Fibrosis. , 2019, , .		0
47	Two novel mutations in exon 3 of PHOX2B gene: think about congenital central hypoventilation syndrome in patients with Hirschsprung disease. Italian Journal of Pediatrics, 2019, 45, 49.	2.6	3
48	Vemurafenib Treatment of Pleomorphic Xanthoastrocytoma in a Child With Down Syndrome. Frontiers in Oncology, 2019, 9, 277.	2.8	10
49	Pathogenic Variants in GPC4 Cause Keipert Syndrome. American Journal of Human Genetics, 2019, 104, 914-924.	6.2	23
50	Congenital emphysematous lung disease associated with a novel Filamin A mutation. Case report and literature review. BMC Pediatrics, 2019, 19, 86.	1.7	27
51	Autism spectrum disorder in a patient with a genomic rearrangement that only involves the EPHA5 gene. Psychiatric Genetics, 2019, 29, 86-90.	1.1	5
52	Novel TNXB Variants in Two Italian Patients with Classical-Like Ehlers-Danlos Syndrome. Genes, 2019, 10, 967.	2.4	10
53	Primary muscle involvement in a 15â€yearâ€old girl with the recurrent homozygous c.362dupC variant in FKBP14. American Journal of Medical Genetics, Part A, 2019, 179, 317-321.	1.2	3
54	Burkitt lymphoma in a patient with Kabuki syndrome carrying a novel KMT2D mutation. American Journal of Medical Genetics, Part A, 2019, 179, 113-117.	1.2	10

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55	Facial comedonal acne in orofaciodigital syndrome type 1 caused by a novel frameshift variant in <scp>OFD</scp> 1 . Clinical and Experimental Dermatology, 2019, 44, 706-708.	1.3	2
56	Novel exostosinâ€2 missense variants in a family with autosomal recessive exostosinâ€2â€related syndrome: further evidences on the phenotype. Clinical Genetics, 2019, 95, 165-171.	2.0	3
57	Proliferative vasculopathy and hydranencephalyâ€“hydrocephaly syndrome or Fowler syndrome: Report of a family and insight into the disease's mechanism. Molecular Genetics & Genomic Medicine, 2018, 6, 446-451.	1.2	11
58	Helsmoortel-Van der Aa Syndrome as emerging clinical diagnosis in intellectually disabled children with autistic traits and ocular involvement. European Journal of Paediatric Neurology, 2018, 22, 552-557.	1.6	19
59	Expanding the clinical and molecular spectrum of PRMT7 mutations: 3 additional patients and review. Clinical Genetics, 2018, 93, 675-681.	2.0	28
60	A case report on filamin A gene mutation and progressive pulmonary disease in an infant. Medicine (United States), 2018, 97, e13033.	1.0	12
61	BRAF V600E Inhibitor (Vemurafenib) for BRAF V600E Mutated Low Grade Gliomas. Frontiers in Oncology, 2018, 8, 526.	2.8	37
62	Specific combinations of biallelic POLR3A variants cause Wiedemann-Rautenstrauch syndrome. Journal of Medical Genetics, 2018, 55, 837-846.	3.2	44
63	Pilot Study of Warfarin Genotype Polymorphisms in Pediatric Patients with Ventricular Assist Devices. Journal of Heart and Lung Transplantation, 2017, 36, S281.	0.6	0
64	p.Arg1809Cys substitution in neurofibromin is associated with a distinctive NF1 phenotype without neurofibromas. European Journal of Human Genetics, 2015, 23, 1068-1071.	2.8	113
65	Nectin-4 Mutations Causing Ectodermal Dysplasia with Syndactyly Perturb the Rac1 Pathway and the Kinetics of Adherens Junction Formation. Journal of Investigative Dermatology, 2014, 134, 2146-2153.	0.7	33
66	Novel homozygous mutation in exon 5 of <scp>WFS1</scp> gene in an Apulian family with mild phenotypic expression of Wolfram syndrome. Clinical Genetics, 2014, 86, 197-198.	2.0	3
67	Nectinopathies: an emerging group of ectodermal dysplasia syndromes. Giornale Italiano Di Dermatologia E Venereologia, 2013, 148, 59-64.	0.8	5
68	De Barsy Syndrome: A genetically heterogeneous autosomal recessive cutis laxa syndrome related to P5CS and PYCR1 dysfunction. American Journal of Medical Genetics, Part A, 2012, 158A, 927-931.	1.2	37
69	Mutations in ANKRD11 Cause KBG Syndrome, Characterized by Intellectual Disability, Skeletal Malformations, and Macrodontia. American Journal of Human Genetics, 2011, 89, 289-294.	6.2	205
70	Mutations in PVRL4, Encoding Cell Adhesion Molecule Nectin-4, Cause Ectodermal Dysplasia-Syndactyly Syndrome. American Journal of Human Genetics, 2010, 87, 265-273.	6.2	98
71	A Nationwide Genetic Testing Survey in Italy, Year 2007. Genetic Testing and Molecular Biomarkers, 2010, 14, 17-22.	0.7	7
72	Posterior fossa ependymoma in neurodevelopmental syndrome caused by a de novo germline pathogenic Polr2a variant. American Journal of Medical Genetics, Part A, 0, , .	1.2	2