

# Emanuele Agolini

## List of Publications by Year in descending order

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72  
papers

1,292  
citations

471509

17  
h-index

414414

32  
g-index

76  
all docs

76  
docs citations

76  
times ranked

2670  
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in ANKRD11 Cause KBG Syndrome, Characterized by Intellectual Disability, Skeletal Malformations, and Macrodonia. <i>American Journal of Human Genetics</i> , 2011, 89, 289-294.	6.2	205
2	p.Arg1809Cys substitution in neurofibromin is associated with a distinctive NF1 phenotype without neurofibromas. <i>European Journal of Human Genetics</i> , 2015, 23, 1068-1071.	2.8	113
3	Mutations in PVRL4, Encoding Cell Adhesion Molecule Nectin-4, Cause Ectodermal Dysplasia-Syndactyly Syndrome. <i>American Journal of Human Genetics</i> , 2010, 87, 265-273.	6.2	98
4	COVID-19 and Genetic Variants of Protein Involved in the SARS-CoV-2 Entry into the Host Cells. <i>Genes</i> , 2020, 11, 1010.	2.4	88
5	Novel congenital disorder of <i>O</i> -linked glycosylation caused by GALNT2 loss of function. <i>Brain</i> , 2020, 143, 1114-1126.	7.6	46
6	Specific combinations of biallelic <i>POLR3A</i> variants cause Wiedemann-Rautenstrauch syndrome. <i>Journal of Medical Genetics</i> , 2018, 55, 837-846.	3.2	44
7	De Barsy Syndrome: A genetically heterogeneous autosomal recessive cutis laxa syndrome related to P5CS and PYCR1 dysfunction. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 927-931.	1.2	37
8	BRAF V600E Inhibitor (Vemurafenib) for BRAF V600E Mutated Low Grade Gliomas. <i>Frontiers in Oncology</i> , 2018, 8, 526.	2.8	37
9	Nectin-4 Mutations Causing Ectodermal Dysplasia with Syndactyly Perturb the Rac1 Pathway and the Kinetics of Adherens Junction Formation. <i>Journal of Investigative Dermatology</i> , 2014, 134, 2146-2153.	0.7	33
10	Inhibition of HECT E3 ligases as potential therapy for COVID-19. <i>Cell Death and Disease</i> , 2021, 12, 310.	6.3	33
11	Episignatures Stratifying Helsmoortel-Van Der Aa Syndrome Show Modest Correlation with Phenotype. <i>American Journal of Human Genetics</i> , 2020, 107, 555-563.	6.2	32
12	Expanding the clinical and molecular spectrum of <i>PRMT7</i> mutations: 3 additional patients and review. <i>Clinical Genetics</i> , 2018, 93, 675-681.	2.0	28
13	Congenital emphysematous lung disease associated with a novel Filamin A mutation. Case report and literature review. <i>BMC Pediatrics</i> , 2019, 19, 86.	1.7	27
14	<i>COL1A2</i> -related overlap disorder: A novel connective tissue disorder incorporating the osteogenesis imperfecta/Ehlers-Danlos syndrome overlap. <i>Clinical Genetics</i> , 2020, 97, 396-406.	2.0	27
15	KBG syndrome: Common and uncommon clinical features based on 31 new patients. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1073-1083.	1.2	27
16	Pathogenic Variants in GPC4 Cause Keipert Syndrome. <i>American Journal of Human Genetics</i> , 2019, 104, 914-924.	6.2	23
17	HLA-haploidentical TCR $\alpha$ <sup>hi</sup> /CD19 <sup>+</sup> -depleted stem cell transplantation in children and young adults with Fanconi anemia. <i>Blood Advances</i> , 2021, 5, 1333-1339.	5.2	22
18	Low-Grade Gliomas in Patients with Noonan Syndrome: Case-Based Review of the Literature. <i>Diagnostics</i> , 2020, 10, 582.	2.6	21

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19	<i>ANKRD11</i> variants: KBG syndrome and beyond. <i>Clinical Genetics</i> , 2021, 100, 187-200.	2.0	21
20	Helsmoortel-Van der Aa Syndrome as emerging clinical diagnosis in intellectually disabled children with autistic traits and ocular involvement. <i>European Journal of Paediatric Neurology</i> , 2018, 22, 552-557.	1.6	19
21	Infantile-Onset Syndromic Cerebellar Ataxia and CACNA1G Mutations. <i>Pediatric Neurology</i> , 2020, 104, 40-45.	2.1	17
22	Cancer Predisposition Syndromes and Medulloblastoma in the Molecular Era. <i>Frontiers in Oncology</i> , 2020, 10, 566822.	2.8	17
23	ALG12-CDG: novel glyco-phenotype insights endorse the molecular defect. <i>Glycoconjugate Journal</i> , 2019, 36, 461-472.	2.7	16
24	TUBB3 E410K syndrome: Case report and review of the clinical spectrum of TUBB3 mutations. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1977-1984.	1.2	15
25	A case report on filamin A gene mutation and progressive pulmonary disease in an infant. <i>Medicine (United States)</i> , 2018, 97, e13033.	1.0	12
26	Biallelic mutations in the TOGARAM1 gene cause a novel primary ciliopathy. <i>Journal of Medical Genetics</i> , 2020, 58, jmedgenet-2020-106833.	3.2	12
27	Biallelic hypomorphic variants in ALDH1A2 cause a novel lethal human multiple congenital anomaly syndrome encompassing diaphragmatic, pulmonary, and cardiovascular defects. <i>Human Mutation</i> , 2021, 42, 506-519.	2.5	12
28	De Novo Mutation in KMT2C Manifesting as Kleefstra Syndrome 2: Case Report and Literature Review. <i>Pediatric Reports</i> , 2022, 14, 131-139.	1.3	12
29	Proliferative vasculopathy and hydranencephaly—hydrocephaly syndrome or Fowler syndrome: Report of a family and insight into the disease's mechanism. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2018, 6, 446-451.	1.2	11
30	Exon-Trapping Assay Improves Clinical Interpretation of COL11A1 and COL11A2 Intronic Variants in Stickler Syndrome Type 2 and Otospondylomegapiphyseal Dysplasia. <i>Genes</i> , 2020, 11, 1513.	2.4	11
31	Vemurafenib Treatment of Pleomorphic Xanthoastrocytoma in a Child With Down Syndrome. <i>Frontiers in Oncology</i> , 2019, 9, 277.	2.8	10
32	Novel TNXB Variants in Two Italian Patients with Classical-Like Ehlers-Danlos Syndrome. <i>Genes</i> , 2019, 10, 967.	2.4	10
33	Burkitt lymphoma in a patient with Kabuki syndrome carrying a novel <i>KMT2D</i> mutation. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 113-117.	1.2	10
34	A novel patient with White-Sutton syndrome refines the mutational and clinical repertoire of the <i>POGZ</i> -related phenotype and suggests further observations. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1791-1795.	1.2	10
35	<i>TSPEAR</i> variants are primarily associated with ectodermal dysplasia and tooth agenesis but not hearing loss: A novel cohort study. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2417-2433.	1.2	10
36	Nonsense variant of <i>ATP8B1</i> gene in heterozygosis and benign recurrent intrahepatic cholestasis: A case report and review of literature. <i>World Journal of Hepatology</i> , 2020, 12, 64-71.	2.0	9

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37	Genetic identification and molecular modeling characterization of a novel POU3F4 variant in two Italian deaf brothers. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2020, 129, 109790.	1.0	8
38	A Nationwide Genetic Testing Survey in Italy, Year 2007. <i>Genetic Testing and Molecular Biomarkers</i> , 2010, 14, 17-22.	0.7	7
39	The splice c.1815G>A variant in KIAA0586 results in a phenotype bridging short-rib-polydactyly and oral-facial-digital syndrome. <i>Medicine (United States)</i> , 2020, 99, e19169.	1.0	7
40	Clinical presentation and molecular characterization of a novel patient with variant <sc>POC1A</sc>-related syndrome. <i>Clinical Genetics</i> , 2021, 99, 540-546.	2.0	7
41	Autism spectrum disorder in a patient with a genomic rearrangement that only involves the EPHA5 gene. <i>Psychiatric Genetics</i> , 2019, 29, 86-90.	1.1	5
42	WWP1 germline variants are associated with normocephalic autism spectrum disorder. <i>Cell Death and Disease</i> , 2020, 11, 529.	6.3	5
43	14q12q13.2 microdeletion syndrome: Clinical characterization of a new patient, review of the literature, and further evidence of a candidate region for CNS anomalies. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2020, 8, e1289.	1.2	5
44	Refinement of the clinical and mutational spectrum of <sc>UBE2A</sc> deficiency syndrome. <i>Clinical Genetics</i> , 2020, 98, 172-178.	2.0	5
45	Nectinopathies: an emerging group of ectodermal dysplasia syndromes. <i>Giornale Italiano Di Dermatologia E Venereologia</i> , 2013, 148, 59-64.	0.8	5
46	Pediatric gastrointestinal stromal tumor: Report of two novel patients harboring germline variants in SDHB and SDHC genes. <i>Cancer Genetics</i> , 2020, 241, 61-65.	0.4	4
47	Expanding the clinical and molecular spectrum of lethal congenital contracture syndrome 8 associated with biallelic variants <i>of ADCY6</i>. <i>Clinical Genetics</i> , 2020, 97, 649-654.	2.0	4
48	CUGC for lysinuric protein intolerance (LPI). <i>European Journal of Human Genetics</i> , 2020, 28, 1129-1134.	2.8	4
49	Molecular Characterization of Medulloblastoma in a Patient with Neurofibromatosis Type 1: Case Report and Literature Review. <i>Diagnostics</i> , 2021, 11, 647.	2.6	4
50	Expansion of the clinical and molecular spectrum of an <sc>XPD</sc>-related disorder linked to biallelic mutations in <sc>ERCC2</sc> gene. <i>Clinical Genetics</i> , 2021, 99, 842-848.	2.0	4
51	The p.Arg377Trp variant in <i>ACTL6A</i> underlines a recognizable BAFopathy phenotype. <i>Clinical Genetics</i> , 2020, 97, 672-674.	2.0	4
52	Novel homozygous mutation in exon 5 of <i>WFS1</i> gene in an Apulian family with mild phenotypic expression of Wolfram syndrome. <i>Clinical Genetics</i> , 2014, 86, 197-198.	2.0	3
53	Two novel mutations in exon 3 of PHOX2B gene: think about congenital central hypoventilation syndrome in patients with Hirschsprung disease. <i>Italian Journal of Pediatrics</i> , 2019, 45, 49.	2.6	3
54	Primary muscle involvement in a 15-year-old girl with the recurrent homozygous c.362dupC variant in <i>FKBP14</i>. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 317-321.	1.2	3

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55	Novel exostosinâ€2 missense variants in a family with autosomal recessive exostosinâ€2â€related syndrome: further evidences on the phenotype. <i>Clinical Genetics</i> , 2019, 95, 165-171.	2.0	3
56	Further delineation of the neurodevelopmental phenotypic spectrum associated to 14q11.2 microduplication. <i>Neurological Sciences</i> , 2020, 41, 3751-3753.	1.9	3
57	Phenytoin intoxication associated with omeprazole administration in a child with defective CYP2C9. <i>European Journal of Clinical Pharmacology</i> , 2020, 76, 731-732.	1.9	3
58	CUGC for hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome. <i>European Journal of Human Genetics</i> , 2020, 28, 982-987.	2.8	3
59	Medulloblastoma Associated with Down Syndrome: From a Rare Event Leading to a Pathogenic Hypothesis. <i>Diagnostics</i> , 2021, 11, 254.	2.6	3
60	Facial comedonal acne in orofaciodigital syndrome type 1 caused by a novel frameshift variant in <i><scp>OFD</scp> 1</i>. <i>Clinical and Experimental Dermatology</i> , 2019, 44, 706-708.	1.3	2
61	Microcephalic osteodysplastic primordial dwarfism type II and pachygyria: Morphometric analysis in a 2â€yearâ€old girl. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2372-2376.	1.2	2
62	Expanding the novel <i><scp>MAPKAPK5</scp></i> â€related developmental disorderâ€™s genotypeâ€phenotype correlation: patient report and 19 months followâ€up. <i>Clinical Genetics</i> , 2022, , .	2.0	2
63	Posterior fossa ependymoma in neurodevelopmental syndrome caused by a de novo germline pathogenic <i>Polr2a</i> variant. <i>American Journal of Medical Genetics, Part A</i> , 0, , .	1.2	2
64	Liquid Biopsy with Detection of NRASQ61K Mutation in Cerebrospinal Fluid: An Alternative Tool for the Diagnosis of Primary Pediatric Leptomeningeal Melanoma. <i>Diagnostics</i> , 2022, 12, 1609.	2.6	2
65	Structural modeling of a novel TERC variant in a patient with aplastic anemia and short telomeres. <i>Annals of Hematology</i> , 2019, 98, 805-807.	1.8	1
66	Fetal early motor neuron disruption and prenatal molecular diagnosis in a severe BICD2 â€opathy. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1509-1514.	1.2	1
67	Pilot Study of Warfarin Genotype Polymorphisms in Pediatric Patients with Ventricular Assist Devices. <i>Journal of Heart and Lung Transplantation</i> , 2017, 36, S281.	0.6	0
68	P.04.15 THE DISCOVERY OF A NOVEL GENETIC MUTATION IN BENIGN RECURRENT INTRAHEPATIC CHOLESTASIS WIDENS THE INSIGHTS INTO DISORDER RELATED-MOLECULAR ANALYSIS. <i>Digestive and Liver Disease</i> , 2019, 51, e178.	0.9	0
69	Hermansky-Pudlak Syndrome Subtype 4 (HPS-4): A Novel Mutation in a 44 Years Old Italian Woman with Severe Pulmonary Fibrosis. , 2019, , .		0
70	Whole Exome Sequencing Is the Minimal Technological Approach in Proband Born to Consanguineous Couples. <i>Genes</i> , 2021, 12, 962.	2.4	0
71	A Novel CHEK2 Variant Identified by Next-Generation Sequencing in an Italian Family with Li-Fraumeni Syndrome: Case Report. <i>SN Comprehensive Clinical Medicine</i> , 2022, 4, 1.	0.6	0
72	The Fight Just Bornâ€™ Neonatal Cancer: Rare Occurrence with a Favorable Outcome but Challenging Management. <i>Cancers</i> , 2022, 14, 2244.	3.7	0