

# Giulio Piluso

## List of Publications by Year in descending order

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Version: 2024-02-01

96  
papers

3,462  
citations

159525

30  
h-index

149623

56  
g-index

97  
all docs

97  
docs citations

97  
times ranked

4229  
citing authors

#	ARTICLE	IF	CITATIONS
1	Therapeutic homology-independent targeted integration in retina and liver. <i>Nature Communications</i> , 2022, 13, 1963.	5.8	14
2	Genotype-Phenotype Correlations in Neurofibromatosis Type 1: Identification of Novel and Recurrent NF1 Gene Variants and Correlations with Neurocognitive Phenotype. <i>Genes</i> , 2022, 13, 1130.	1.0	10
3	Linked-Read Whole Genome Sequencing Solves a Double DMD Gene Rearrangement. <i>Genes</i> , 2021, 12, 133.	1.0	8
4	Nephroplex: a kidney-focused NGS panel highlights the challenges of PKD1 sequencing and identifies a founder BBS4 mutation. <i>Journal of Nephrology</i> , 2021, 34, 1855-1874.	0.9	6
5	Missense mutations in small muscle protein X-linked (SMPX) cause distal myopathy with protein inclusions. <i>Acta Neuropathologica</i> , 2021, 142, 375-393.	3.9	6
6	Solving unsolved rare neurological diseases – a Solve-RD viewpoint. <i>European Journal of Human Genetics</i> , 2021, 29, 1332-1336.	1.4	4
7	A novel MEIS2 mutation explains the complex phenotype in a boy with a typical NF1 microdeletion syndrome. <i>European Journal of Medical Genetics</i> , 2021, 64, 104190.	0.7	3
8	Exome reanalysis and proteomic profiling identified TRIP4 as a novel cause of cerebellar hypoplasia and spinal muscular atrophy (PCH1). <i>European Journal of Human Genetics</i> , 2021, 29, 1348-1353.	1.4	10
9	Poikiloderma With Neutropenia and Mastocytosis: A Case Report and a Review of Dermatological Signs. <i>Frontiers in Medicine</i> , 2021, 8, 680363.	1.2	3
10	Solve-RD: systematic pan-European data sharing and collaborative analysis to solve rare diseases. <i>European Journal of Human Genetics</i> , 2021, 29, 1325-1331.	1.4	49
11	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. <i>European Journal of Human Genetics</i> , 2021, 29, 1337-1347.	1.4	34
12	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.	1.0	6
13	Enhancing cyst-like lesions of the white matter in tuberous sclerosis complex: a novel neuroradiological finding. <i>Neuroradiology</i> , 2021, 63, 971-974.	1.1	5
14	Intermittent macrothrombocytopenia in a novel patient with Takenouchi-Kosaki syndrome and review of literature. <i>European Journal of Medical Genetics</i> , 2021, 64, 104358.	0.7	3
15	Expanding the Neuroradiological Phenotype of 18q Deletion Syndrome. <i>Indian Pediatrics</i> , 2021, 58, 1187-1188.	0.2	0
16	Expanding the Neuroradiological Phenotype of 18q Deletion Syndrome.. <i>Indian Pediatrics</i> , 2021, 58, 1187-1188.	0.2	0
17	Clinical spectrum of individuals with pathogenic NF1 missense variants affecting p.Met1149, p.Arg1276, and p.Lys1423: genotype-phenotype study in neurofibromatosis type 1. <i>Human Mutation</i> , 2020, 41, 299-315.	1.1	80
18	NEW GENES IN NEUROMUSCULAR DISEASES. <i>Neuromuscular Disorders</i> , 2020, 30, S46.	0.3	1

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19	A novel RAB39B mutation and concurrent de novo NF1 mutation in a boy with neurofibromatosis type 1, intellectual disability, and autism: a case report. BMC Neurology, 2020, 20, 327.	0.8	21
20	The position of nonsense mutations can predict the phenotype severity: A survey on the DMD gene. PLoS ONE, 2020, 15, e0237803.	1.1	25
21	Identification and Characterization of Splicing Defects by Single-Molecule Real-Time Sequencing Technology (PacBio). Journal of Neuromuscular Diseases, 2020, 7, 477-481.	1.1	7
22	Clinical and Genetic Findings in Children with Neurofibromatosis Type 1, Legius Syndrome, and Other Related Neurocutaneous Disorders. Genes, 2019, 10, 580.	1.0	25
23	O.27Ultra-exome: a new tool to solve the unsolved NMD. Neuromuscular Disorders, 2019, 29, S123-S124.	0.3	0
24	Report on a child with neurofibromatosis type 2 and unilateral moyamoya: further evidence of cerebral vasculopathy in NF2. Neurological Sciences, 2019, 40, 1475-1476.	0.9	3
25	Assessment of de novo copy-number variations in Italian patients with schizophrenia: Detection of putative mutations involving regulatory enhancer elements. World Journal of Biological Psychiatry, 2019, 20, 126-136.	1.3	12
26	Targeted gene panel screening is an effective tool to identify undiagnosed late onset Pompe disease. Neuromuscular Disorders, 2018, 28, 586-591.	0.3	24
27	<i>UBE2A</i> deficiency in two siblings: A novel splicing variant inherited from a maternal germline mosaicism. American Journal of Medical Genetics, Part A, 2018, 176, 722-726.	0.7	9
28	Multiple spinal nerve enlargement and <i>SOS1</i> mutation: Further evidence of overlap between neurofibromatosis type 1 and Noonan phenotype. Clinical Genetics, 2018, 93, 138-143.	1.0	17
29	A Novel 12q13.2-q13.3 Microdeletion Syndrome With Combined Features of Diamond Blackfan Anemia, Pierre Robin Sequence and Klippel Feil Deformity. Frontiers in Genetics, 2018, 9, 549.	1.1	7
30	Copy Number Variants Account for a Tiny Fraction of Undiagnosed Myopathic Patients. Genes, 2018, 9, 524.	1.0	7
31	Whole exome sequencing identifies MRV1 as a susceptibility gene for moyamoya syndrome in neurofibromatosis type 1. PLoS ONE, 2018, 13, e0200446.	1.1	24
32	Seizures in children with neurofibromatosis type 1: is neurofibromatosis type 1 enough?. Italian Journal of Pediatrics, 2018, 44, 41.	1.0	27
33	Moyamoya syndrome in children with neurofibromatosis type 1: Italian-French experience. American Journal of Medical Genetics, Part A, 2017, 173, 1521-1530.	0.7	36
34	A novel SHANK3 interstitial microdeletion in a family with intellectual disability and brain MRI abnormalities resembling Unidentified Bright Objects. European Journal of Paediatric Neurology, 2017, 21, 902-906.	0.7	5
35	Expanding the phenotype of <i>RTTN</i> variations: a new family with primary microcephaly, severe growth failure, brain malformations and dermatitis. Clinical Genetics, 2016, 90, 445-450.	1.0	21
36	Identification of an intragenic deletion in the SGCB gene through a re-evaluation of negative next generation sequencing results. Neuromuscular Disorders, 2016, 26, 367-369.	0.3	12

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37	The genetic basis of undiagnosed muscular dystrophies and myopathies. <i>Neurology</i> , 2016, 87, 71-76.	1.5	92
38	Novel findings associated with MTM1 suggest a higher number of female symptomatic carriers. <i>Neuromuscular Disorders</i> , 2016, 26, 292-299.	0.3	25
39	A novel diagnostic method to detect truncated neurofibromin in neurofibromatosis 1. <i>Journal of Neurochemistry</i> , 2015, 135, 1123-1128.	2.1	13
40	Spectrum of muscular dystrophies associated with sarcolemmal-protein genetic defects. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2015, 1852, 585-593.	1.8	48
41	Arg1809 substitution in neurofibromin: further evidence of a genotype-phenotype correlation in neurofibromatosis type 1. <i>European Journal of Human Genetics</i> , 2015, 23, 1460-1461.	1.4	35
42	Novel mutations in LMNA A/C gene and associated phenotypes. <i>Acta Myologica</i> , 2015, 34, 116-9.	1.5	10
43	Giant thrombosed intracavernous carotid artery aneurysm presenting as Tolosa&ndash;Hunt syndrome in a patient harboring a new pathogenic neurofibromatosis type 1 mutation: a case report and review of the literature. <i>Neuropsychiatric Disease and Treatment</i> , 2014, 10, 135.	1.0	10
44	LEOPARD syndrome: clinical dilemmas in differential diagnosis of RASopathies. <i>BMC Medical Genetics</i> , 2014, 15, 44.	2.1	33
45	C.O.7. <i>Neuromuscular Disorders</i> , 2014, 24, 851.	0.3	0
46	Use of a Lower Dosage Liver-Detargeted AAV Vector to Prevent Hamster Muscular Dystrophy. <i>Human Gene Therapy</i> , 2013, 24, 424-430.	1.4	7
47	O.17 Mutation spectrum of limb-girdle muscular dystrophies by New Generation Sequencing approaches. <i>Neuromuscular Disorders</i> , 2013, 23, 849-850.	0.3	0
48	Identification and molecular characterization of a novel 55â€b deletion recurrent in southern Italy: the Italian <sup>G</sup>(<sup>A</sup>)&sup>3</sup>(&sup>A</sup>)&sup>2</sup> thalassemia. <i>European Journal of Haematology</i> , 2013, 90, 214-219.	1.1	8
49	Familial trisomy 6p in mother and daughter. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 1675-1681.	0.7	3
50	Next-Generation Sequencing Identifies Transportin 3 as the Causative Gene for LGMD1F. <i>PLoS ONE</i> , 2013, 8, e63536.	1.1	69
51	D.O.3 Next generation sequencing applications are ready for genetic diagnosis of muscular dystrophies. <i>Neuromuscular Disorders</i> , 2012, 22, 806.	0.3	0
52	Enhancer Chip: Detecting Human Copy Number Variations in Regulatory Elements. <i>PLoS ONE</i> , 2012, 7, e52264.	1.1	8
53	Identification of a functional estrogen-responsive enhancer element in the promoter 2 of <i>PRDM2</i> gene in breast cancer cell lines. <i>Journal of Cellular Physiology</i> , 2012, 227, 964-975.	2.0	22
54	Improvement of survival in Duchenne Muscular Dystrophy: retrospective analysis of 835 patients. <i>Acta Myologica</i> , 2012, 31, 121-5.	1.5	221

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55	Next generation sequencing (NGS) strategies for the genetic testing of myopathies. <i>Acta Myologica</i> , 2012, 31, 196-200.	1.5	27
56	P2.27 Full exome resequencing by next generation sequencing (NGS) combined with chip analysis for the genetic testing of unclassified myopathic patients. <i>Neuromuscular Disorders</i> , 2011, 21, 668.	0.3	0
57	Worsening of Cardiomyopathy Using Deflazacort in an Animal Model Rescued by Gene Therapy. <i>PLoS ONE</i> , 2011, 6, e24729.	1.1	19
58	Limb girdle muscular dystrophies. <i>Current Opinion in Neurology</i> , 2011, 24, 429-436.	1.8	136
59	Novel missense mutations and unexpected multiple changes of RYR1 gene in 75 malignant hyperthermia families. <i>Clinical Genetics</i> , 2011, 79, 438-447.	1.0	34
60	Muscular dystrophy with marked Dysferlin deficiency is consistently caused by primary dysferlin gene mutations. <i>European Journal of Human Genetics</i> , 2011, 19, 974-980.	1.4	67
61	Motor Chip: A Comparative Genomic Hybridization Microarray for Copy-Number Mutations in 245 Neuromuscular Disorders. <i>Clinical Chemistry</i> , 2011, 57, 1584-1596.	1.5	48
62	Combined deficiency of alpha and epsilon sarcoglycan disrupts the cardiac dystrophin complex. <i>Human Molecular Genetics</i> , 2011, 20, 4644-4654.	1.4	35
63	Reliable resequencing of the human dystrophin locus by universal long polymerase chain reaction and massive pyrosequencing. <i>Analytical Biochemistry</i> , 2010, 406, 176-184.	1.1	15
64	P3.14 New AAVs for the muscle gene therapy in sarcoglycan deficient animals. <i>Neuromuscular Disorders</i> , 2010, 20, 645.	0.3	0
65	An Italian case of hereditary myopathy with early respiratory failure (HMERF) not associated with the titin kinase domain R279W mutation. <i>Neuromuscular Disorders</i> , 2010, 20, 730-734.	0.3	15
66	One Hundred Twenty-One Dystrophin Point Mutations Detected from Stored DNA Samples by Combinatorial Denaturing High-Performance Liquid Chromatography. <i>Journal of Molecular Diagnostics</i> , 2010, 12, 65-73.	1.2	17
67	Mendelian bases of myopathies, cardiomyopathies, and neuromyopathies. <i>Acta Myologica</i> , 2010, 29, 1-20.	1.5	7
68	A Missense Mutation in CASK Causes FC Syndrome in an Italian Family. <i>American Journal of Human Genetics</i> , 2009, 84, 162-177.	2.6	82
69	Lack of replication of genetic associations with human longevity. <i>Biogerontology</i> , 2008, 9, 85-92.	2.0	69
70	Mutations that impair interaction properties of TRIM32 associated with limb-girdle muscular dystrophy 2H. <i>Human Mutation</i> , 2008, 29, 240-247.	1.1	92
71	G.P.10.01 Dysferlinopathies in Southern Italy. <i>Neuromuscular Disorders</i> , 2008, 18, 790.	0.3	0
72	G.P.10.11 T-CAP, the gene responsible for LGMD2G, may interact with dysferlin. <i>Neuromuscular Disorders</i> , 2008, 18, 793.	0.3	0

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73	G.P.14.06 LGMD2H patients of non Hutterite origin with mutations in TRIM32 gene. <i>Neuromuscular Disorders</i> , 2008, 18, 817.	0.3	0
74	Log-PCR: A New Tool for Immediate and Cost-Effective Diagnosis of up to 85% of Dystrophin Gene Mutations. <i>Clinical Chemistry</i> , 2008, 54, 973-981.	1.5	27
75	Candidate-gene testing for orphan limb-girdle muscular dystrophies. <i>Acta Myologica</i> , 2008, 27, 90-7.	1.5	6
76	G.P.8.06 Limb-girdle muscular dystrophies: DNA test following protein test or not?. <i>Neuromuscular Disorders</i> , 2007, 17, 811-812.	0.3	0
77	G.P.4.03 Mutations in the lamin A/C gene: An emergent cause of fatal arrhythmias in congenital muscular dystrophies. <i>Neuromuscular Disorders</i> , 2006, 16, 675-676.	0.3	0
78	P.P.6 02 Cardiac and respiratory involvement in autosomal recessive limb-girdle muscular dystrophies. <i>Neuromuscular Disorders</i> , 2006, 16, 694.	0.3	0
79	T.O. 6 Systemic delta-sarcoglycan gene transfer into cardiomyopathic BIO14.6 hamsters by AAV. <i>Neuromuscular Disorders</i> , 2006, 16, 724.	0.3	0
80	Extensive scanning of the calpain-3 gene broadens the spectrum of LGMD2A phenotypes. <i>Journal of Medical Genetics</i> , 2005, 42, 686-693.	1.5	92
81	Molecular and muscle pathology in a series of caveolinopathy patients. <i>Human Mutation</i> , 2005, 25, 82-89.	1.1	64
82	Prevalence of the 550delA mutation in calpainopathy (LGMD 2A) in Croatia. <i>American Journal of Medical Genetics Part A</i> , 2004, 125A, 152-156.	2.4	31
83	Molecular diagnosis in LGMD2A: Mutation analysis or protein testing?. <i>Human Mutation</i> , 2004, 24, 52-62.	1.1	109
84	Genetic heterogeneity of FG syndrome: a fourth locus (FGS4) maps to Xp11.4-p11.3 in an Italian family. <i>Human Genetics</i> , 2003, 112, 124-130.	1.8	30
85	Scanning for Mutations of the Ryanodine Receptor (RYR1) Gene by Denaturing HPLC: Detection of Three Novel Malignant Hyperthermia Alleles. <i>Clinical Chemistry</i> , 2003, 49, 761-768.	1.5	37
86	$\beta$ 1- and $\beta$ 2-Syntrophins, Two Novel Dystrophin-binding Proteins Localized in Neuronal Cells. <i>Journal of Biological Chemistry</i> , 2000, 275, 15851-15860.	1.6	117
87	The retinoblastoma-interacting zinc-finger protein RIZ is a downstream effector of estrogen action. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2000, 97, 3130-3135.	3.3	56
88	The retinoblastoma-interacting zinc-finger protein RIZ is a downstream effector of estrogen action. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2000, 97, 3130-3135.	3.3	36
89	Identification of a DNA Binding Protein Cooperating with Estrogen Receptor as RIZ (Retinoblastoma) Tj ETQq1 1 0.784314 rgBT /Overl 983-989.	1.0	31
90	Identification and characterization of a novel member of the dystrobrevin gene family. <i>FEBS Letters</i> , 1998, 425, 7-13.	1.3	24

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91	Interaction of Vault Particles with Estrogen Receptor in the MCF-7 Breast Cancer Cell. <i>Journal of Cell Biology</i> , 1998, 141, 1301-1310.	2.3	93
92	Identification of the Syrian hamster cardiomyopathy gene. <i>Human Molecular Genetics</i> , 1997, 6, 601-607.	1.4	253
93	The fourth component of the sarcoglycan complex. <i>FEBS Letters</i> , 1997, 403, 143-148.	1.3	26
94	Autosomal recessive limb-girdle muscular dystrophy, LGMD2F, is caused by a mutation in the "sarcoglycan gene. <i>Nature Genetics</i> , 1996, 14, 195-198.	9.4	417
95	Identification of a novel sarcoglycan gene at 5q33 encoding a sarcolemmal 35 kDa glycoprotein. <i>Human Molecular Genetics</i> , 1996, 5, 1179-1186.	1.4	173
96	SSCP detection of novel mutations in patients with Emery-Dreifuss muscular dystrophy: definition of a small C-terminal region required for emerin function. <i>Human Molecular Genetics</i> , 1995, 4, 2003-2004.	1.4	48