Giulio Piluso

List of Publications by Year
in descending order

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	Autosomal recessive limbgirdle muscular dystrophy, LGMD2F, is caused by a mutation in the Î'â€"sarcoglycan gene. Nature Genetics, 1996, 14, 195-198.	9.4	417
2	Identification of the Syrian hamster cardiomyopathy gene. Human Molecular Genetics, 1997, 6, 601-607.	1.4	253
3	Improvement of survival in Duchenne Muscular Dystrophy: retrospective analysis of 835 patients. Acta Myologica, 2012, 31, 121-5.	1.5	221
4	Identification of a novel sarcoglycan gene at 5q33 encoding a sarcolemmal 35 kDa glycoprotein. Human Molecular Genetics, 1996, 5, 1179-1186.	1.4	173
5	Limb girdle muscular dystrophies. Current Opinion in Neurology, 2011, 24, 429-436.	1.8	136
6	Î³1- and Î³2-Syntrophins, Two Novel Dystrophin-binding Proteins Localized in Neuronal Cells. Journal of Biological Chemistry, 2000, 275, 15851-15860.	1.6	117
7	Molecular diagnosis in LGMD2A: Mutation analysis or protein testing?. Human Mutation, 2004, 24, 52-62.	1.1	109
8	Interaction of Vault Particles with Estrogen Receptor in the MCF-7 Breast Cancer Cell. Journal of Cell Biology, 1998, 141, 1301-1310.	2.3	93
9	Extensive scanning of the calpain-3 gene broadens the spectrum of LGMD2A phenotypes. Journal of Medical Genetics, 2005, 42, 686-693.	1.5	92
10	Mutations that impair interaction properties of TRIM32 associated with limb-girdle muscular dystrophy 2H. Human Mutation, 2008, 29, 240-247.	1.1	92
11	The genetic basis of undiagnosed muscular dystrophies and myopathies. Neurology, 2016, 87, 71-76.	1.5	92
12	A Missense Mutation in CASK Causes FG Syndrome in an Italian Family. American Journal of Human Genetics, 2009, 84, 162-177.	2.6	82
13	Clinical spectrum of individuals with pathogenic N F1 missense variants affecting p.Met1149, p.Arg1276, and p.Lys1423: genotypeâ€"phenotype study in neurofibromatosis type 1. Human Mutation, 2020, 41, 299-315.	1.1	80
14	Lack of replication of genetic associations with human longevity. Biogerontology, 2008, 9, 85-92.	2.0	69
15	Next-Generation Sequencing Identifies Transportin 3 as the Causative Gene for LGMD1F. PLoS ONE, 2013, 8, e63536.	1.1	69
16	Muscular dystrophy with marked Dysferlin deficiency is consistently caused by primary dysferlin gene mutations. European Journal of Human Genetics, 2011, 19, 974-980.	1.4	67
17	Molecular and muscle pathology in a series of caveolinopathy patients. Human Mutation, 2005, 25, 82-89.	1.1	64
18	The retinoblastoma-interacting zinc-finger protein RIZ is a downstream effector of estrogen action. Proceedings of the National Academy of Sciences of the United States of America, 2000, 97, 3130-3135.	3.3	56

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19	Solve-RD: systematic pan-European data sharing and collaborative analysis to solve rare diseases. European Journal of Human Genetics, 2021, 29, 1325-1331.	1.4	49
20	SSCP detection of novel mutations in patients with Emery-Dreifuss muscular dystrophy: definition of a small C-terminal region required for emerin function. Human Molecular Genetics, 1995, 4, 2003-2004.	1.4	48
21	Motor Chip: A Comparative Genomic Hybridization Microarray for Copy-Number Mutations in 245 Neuromuscular Disorders. Clinical Chemistry, 2011, 57, 1584-1596.	1.5	48
22	Spectrum of muscular dystrophies associated with sarcolemmal-protein genetic defects. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2015, 1852, 585-593.	1.8	48
23	Scanning for Mutations of the Ryanodine Receptor (RYR1) Gene by Denaturing HPLC: Detection of Three Novel Malignant Hyperthermia Alleles. Clinical Chemistry, 2003, 49, 761-768.	1.5	37
24	Moyamoya syndrome in children with neurofibromatosis type 1: Italianâ€“French experience. American Journal of Medical Genetics, Part A, 2017, 173, 1521-1530.	0.7	36
25	The retinoblastoma-interacting zinc-finger protein RIZ is a downstream effector of estrogen action. Proceedings of the National Academy of Sciences of the United States of America, 2000, 97, 3130-3135.	3.3	36
26	Combined deficiency of alpha and epsilon sarcoglycan disrupts the cardiac dystrophin complex. Human Molecular Genetics, 2011, 20, 4644-4654.	1.4	35
27	Arg1809 substitution in neurofibromin: further evidence of a genotypeâ€“phenotype correlation in neurofibromatosis type 1. European Journal of Human Genetics, 2015, 23, 1460-1461.	1.4	35
28	Novel missense mutations and unexpected multiple changes of RYR1 gene in 75 malignant hyperthermia families. Clinical Genetics, 2011, 79, 438-447.	1.0	34
29	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. European Journal of Human Genetics, 2021, 29, 1337-1347.	1.4	34
30	LEOPARD syndrome: clinical dilemmas in differential diagnosis of RASopathies. BMC Medical Genetics, 2014, 15, 44.	2.1	33
31	Identification of a DNA Binding Protein Cooperating with Estrogen Receptor as RIZ (Retinoblastoma) Tj ETQq1 1 0.784314 rgBT /Over 983-989.	1.0	31
32	Prevalence of the 550delA mutation in calpainopathy (LGMD 2A) in Croatia. American Journal of Medical Genetics Part A, 2004, 125A, 152-156.	2.4	31
33	Genetic heterogeneity of FG syndrome: a fourth locus (FGS4) maps to Xp11.4-p11.3 in an Italian family. Human Genetics, 2003, 112, 124-130.	1.8	30
34	Log-PCR: A New Tool for Immediate and Cost-Effective Diagnosis of up to 85% of Dystrophin Gene Mutations. Clinical Chemistry, 2008, 54, 973-981.	1.5	27
35	Seizures in children with neurofibromatosis type 1: is neurofibromatosis type 1 enough?. Italian Journal of Pediatrics, 2018, 44, 41.	1.0	27
36	Next generation sequencing (NGS) strategies for the genetic testing of myopathies. Acta Myologica, 2012, 31, 196-200.	1.5	27

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37	The fourth component of the sarcoglycan complex. FEBS Letters, 1997, 403, 143-148.	1.3	26
38	Novel findings associated with MTM1 suggest a higher number of female symptomatic carriers. Neuromuscular Disorders, 2016, 26, 292-299.	0.3	25
39	Clinical and Genetic Findings in Children with Neurofibromatosis Type 1, Legius Syndrome, and Other Related Neurocutaneous Disorders. Genes, 2019, 10, 580.	1.0	25
40	The position of nonsense mutations can predict the phenotype severity: A survey on the DMD gene. PLoS ONE, 2020, 15, e0237803.	1.1	25
41	Identification and characterization of a novel member of the dystrobrevin gene family. FEBS Letters, 1998, 425, 7-13.	1.3	24
42	Targeted gene panel screening is an effective tool to identify undiagnosed late onset Pompe disease. Neuromuscular Disorders, 2018, 28, 586-591.	0.3	24
43	Whole exome sequencing identifies MRVI1 as a susceptibility gene for moyamoya syndrome in neurofibromatosis type 1. PLoS ONE, 2018, 13, e0200446.	1.1	24
44	Identification of a functional estrogenâ€responsive enhancer element in the promoter 2 of PRDM2 gene in breast cancer cell lines. Journal of Cellular Physiology, 2012, 227, 964-975.	2.0	22
45	Expanding the phenotype of <scp>RTTN</scp> variations: a new family with primary microcephaly, severe growth failure, brain malformations and dermatitis. Clinical Genetics, 2016, 90, 445-450.	1.0	21
46	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
47	Worsening of Cardiomyopathy Using Deflazacort in an Animal Model Rescued by Gene Therapy. PLoS ONE, 2011, 6, e24729.	1.1	19
48	One Hundred Twenty-One Dystrophin Point Mutations Detected from Stored DNA Samples by Combinatorial Denaturing High-Performance Liquid Chromatography. Journal of Molecular Diagnostics, 2010, 12, 65-73.	1.2	17
49	Multiple spinal nerve enlargement and <scp>SOS1</scp> mutation: Further evidence of overlap between neurofibromatosis type 1 and Noonan phenotype. Clinical Genetics, 2018, 93, 138-143.	1.0	17
50	Reliable resequencing of the human dystrophin locus by universal long polymerase chain reaction and massive pyrosequencing. Analytical Biochemistry, 2010, 406, 176-184.	1.1	15
51	An Italian case of hereditary myopathy with early respiratory failure (HMERF) not associated with the titin kinase domain R279W mutation. Neuromuscular Disorders, 2010, 20, 730-734.	0.3	15
52	Therapeutic homology-independent targeted integration in retina and liver. Nature Communications, 2022, 13, 1963.	5.8	14
53	A novel diagnostic method to detect truncated neurofibromin in neurofibromatosis 1. Journal of Neurochemistry, 2015, 135, 1123-1128.	2.1	13
54	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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55	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
56	Giant thrombosed intracavernous carotid artery aneurysm presenting as Tolosa–Hunt syndrome in a patient harboring a new pathogenic neurofibromatosis type 1 mutation: a case report and review of the literature. Neuropsychiatric Disease and Treatment, 2014, 10, 135.	1.0	10
57	Exome reanalysis and proteomic profiling identified TRIP4 as a novel cause of cerebellar hypoplasia and spinal muscular atrophy (PCH1). European Journal of Human Genetics, 2021, 29, 1348-1353.	1.4	10
58	Novel mutations in LMNA A/C gene and associated phenotypes. Acta Myologica, 2015, 34, 116-9.	1.5	10
59	Genotype-Phenotype Correlations in Neurofibromatosis Type 1: Identification of Novel and Recurrent NF1 Gene Variants and Correlations with Neurocognitive Phenotype. Genes, 2022, 13, 1130.	1.0	10
60	UBE2A 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
61	Enhancer Chip: Detecting Human Copy Number Variations in Regulatory Elements. PLoS ONE, 2012, 7, e52264.	1.1	8
62	Identification and molecular characterization of a novel 55â€kb deletion recurrent in southern Italy: the Italian ^GÎ³(^AÎ³Î²)Â°â€thalassemia. European Journal of Haematology, 2013, 90, 214-219.	1.1	8
63	Linked-Read Whole Genome Sequencing Solves a Double DMD Gene Rearrangement. Genes, 2021, 12, 133.	1.0	8
64	Use of a Lower Dosage Liver-Detargeted AAV Vector to Prevent Hamster Muscular Dystrophy. Human Gene Therapy, 2013, 24, 424-430.	1.4	7
65	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
66	Copy Number Variants Account for a Tiny Fraction of Undiagnosed Myopathic Patients. Genes, 2018, 9, 524.	1.0	7
67	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
68	Mendelian bases of myopathies, cardiomyopathies, and neuromyopathies. Acta Myologica, 2010, 29, 1-20.	1.5	7
69	Nephroplex: a kidney-focused NGS panel highlights the challenges of PKD1 sequencing and identifies a founder BBS4 mutation. Journal of Nephrology, 2021, 34, 1855-1874.	0.9	6
70	Missense mutations in small muscle protein X-linked (SMPX) cause distal myopathy with protein inclusions. Acta Neuropathologica, 2021, 142, 375-393.	3.9	6
71	Clinical variability of neurofibromatosis 1: A modifying role of cooccurring <scp>PTPN11</scp> variants and atypical brain <scp>MRI</scp> findings. Clinical Genetics, 2021, 100, 563-572.	1.0	6
72	Candidate-gene testing for orphan limb-girdle muscular dystrophies. Acta Myologica, 2008, 27, 90-7.	1.5	6

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73	Enhancing cyst-like lesions of the white matter in tuberous sclerosis complex: a novel neuroradiological finding. Neuroradiology, 2021, 63, 971-974.	1.1	5
74	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
75	Solving unsolved rare neurological diseasesâ€"a Solve-RD viewpoint. European Journal of Human Genetics, 2021, 29, 1332-1336.	1.4	4
76	Familial trisomy 6p in mother and daughter. American Journal of Medical Genetics, Part A, 2013, 161, 1675-1681.	0.7	3
77	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
78	A novel MEIS2 mutation explains the complex phenotype in a boy with a typical NF1 microdeletion syndrome. European Journal of Medical Genetics, 2021, 64, 104190.	0.7	3
79	Poikiloderma With Neutropenia and Mastocytosis: A Case Report and a Review of Dermatological Signs. Frontiers in Medicine, 2021, 8, 680363.	1.2	3
80	Intermittent macrothrombocytopenia in a novel patient with Takenouchi-Kosaki syndrome and review of literature. European Journal of Medical Genetics, 2021, 64, 104358.	0.7	3
81	NEW GENES IN NEUROMUSCULAR DISEASES. Neuromuscular Disorders, 2020, 30, S46.	0.3	1
82	G.P.4.03 Mutations in the lamin A/C gene: An emergent cause of fatal arrhythmias in congenital muscular dystrophies. Neuromuscular Disorders, 2006, 16, 675-676.	0.3	0
83	P.P.6 02 Cardiac and respiratory involvement in autosomal recessive limb-girdle muscular dystrophies. Neuromuscular Disorders, 2006, 16, 694.	0.3	0
84	T.O. 6 Systemic delta-sarcoglycan gene transfer into cardiomyopathic BIO14.6 hamsters by AAV. Neuromuscular Disorders, 2006, 16, 724.	0.3	0
85	G.P.8.06 Limb-girdle muscular dystrophies: DNA test following protein test or not?. Neuromuscular Disorders, 2007, 17, 811-812.	0.3	0
86	G.P.10.01 Dysferlinopathies in Southern Italy. Neuromuscular Disorders, 2008, 18, 790.	0.3	0
87	G.P.10.11 T-CAP, the gene responsible for LGMD2G, may interact with dysferlin. Neuromuscular Disorders, 2008, 18, 793.	0.3	0
88	G.P.14.06 LGMD2H patients of non Hutterite origin with mutations in TRIM32 gene. Neuromuscular Disorders, 2008, 18, 817.	0.3	0
89	P3.14 New AAVs for the muscle gene therapy in sarcoglycan deficient animals. Neuromuscular Disorders, 2010, 20, 645.	0.3	0
90	P2.27 Full exome resequencing by next generation sequencing (NGS) combined with chip analysis for the genetic testing of unclassified myopathic patients. Neuromuscular Disorders, 2011, 21, 668.	0.3	0

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91	D.O.3 Next generation sequencing applications are ready for genetic diagnosis of muscular dystrophies. Neuromuscular Disorders, 2012, 22, 806.	0.3	0
92	O.17 Mutation spectrum of limb-girdle muscular dystrophies by New Generation Sequencing approaches. Neuromuscular Disorders, 2013, 23, 849-850.	0.3	0
93	G.O.7. Neuromuscular Disorders, 2014, 24, 851.	0.3	0
94	O.27Ultra-exome: a new tool to solve the unsolved NMD. Neuromuscular Disorders, 2019, 29, S123-S124.	0.3	0
95	Expanding the Neuroradiological Phenotype of 18q Deletion Syndrome. Indian Pediatrics, 2021, 58, 1187-1188.	0.2	0
96	Expanding the Neuroradiological Phenotype of 18q Deletion Syndrome.. Indian Pediatrics, 2021, 58, 1187-1188.	0.2	0