Pietro Chiurazzi

List of Publications by Year in descending order

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76 papers	3,318 citations	201674 27 h-index	155660 55 g-index
80 all docs	80 docs citations	80 times ranked	5156 citing authors

#	Article	IF	Citations
1	Mechanisms of the FMR1 Repeat Instability: How Does the CGG Sequence Expand?. International Journal of Molecular Sciences, 2022, 23, 5425.	4.1	7
2	PacMAGI: A pipeline including accurate indel detection for the analysis of PacBio sequencing data applied to RPE65. Gene, 2022, 832, 146554.	2.2	4
3	Mother and Daughter Carrying of the Same Pathogenic Variant in FGFR2 with Discordant Phenotype. Genes, 2022, 13, 1161.	2.4	0
4	Deep brain stimulation in Fragile X syndrome with tardive dystonia. Neurological Sciences, 2021, 42, 2987-2989.	1.9	2
5	USH2A-Related Retinitis Pigmentosa: Staging of Disease Severity and Morpho-Functional Studies. Diagnostics, 2021, 11, 213.	2.6	5
6	Neuroacanthocytosis Syndromes in an Italian Cohort: Clinical Spectrum, High Genetic Variability and Muscle Involvement. Genes, 2021, 12, 344.	2.4	6
7	DNA Methylation, Mechanisms of FMR1 Inactivation and Therapeutic Perspectives for Fragile X Syndrome. Biomolecules, 2021, 11, 296.	4.0	19
8	Inherited Retinal Diseases Due to RPE65 Variants: From Genetic Diagnostic Management to Therapy. International Journal of Molecular Sciences, 2021, 22, 7207.	4.1	19
9	Co-Occurrence of Fragile X Syndrome with a Second Genetic Condition: Three Independent Cases of Double Diagnosis. Genes, 2021, 12, 1909.	2.4	4
10	A next generation sequencing gene panel for use in the diagnosis of anorexia nervosa. Eating and Weight Disorders, 2021, , 1.	2.5	9
11	Variant Selection and Interpretation: An Example of Modified VarSome Classifier of ACMG Guidelines in the Diagnostic Setting. Genes, 2021, 12, 1885.	2.4	4
12	Validating clinical characteristics of primary failure of eruption (PFE) associated with PTH1R variants. Progress in Orthodontics, 2021, 22, 43.	3.5	10
13	Methylated premutation of the FMR1 gene in three sisters: correlating CGG expansion and epigenetic inactivation. European Journal of Human Genetics, 2020, 28, 567-575.	2.8	6
14	Two rare PROX1 variants in patients with lymphedema. Molecular Genetics & Genomic Medicine, 2020, 8, e1424.	1.2	4
15	NLRP12 gene mutations and auto-inflammatory diseases: ever-changing evidence. Rheumatology, 2020, 59, 3129-3136.	1.9	11
16	Aldo-Keto Reductase 1C1 (AKR1C1) as the First Mutated Gene in a Family with Nonsyndromic Primary Lipedema. International Journal of Molecular Sciences, 2020, 21, 6264.	4.1	27
17	Genetic contributions to the etiology of anorexia nervosa: New perspectives in molecular diagnosis and treatment. Molecular Genetics & Genomic Medicine, 2020, 8, e1244.	1.2	21
18	Myotonic dystrophy type 1 cosegregating with autosomal dominant polycystic kidney disease type 2. Neurological Sciences, 2020, 41, 3761-3763.	1.9	0

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19	Compound heterozygosity for an expanded (GAA) and a (GAAGGA) repeat at FXN locus: from a diagnostic pitfall to potential clues to the pathogenesis of Friedreich ataxia. Neurogenetics, 2020, 21, 279-287.	1.4	2
20	Reversion to Normal of FMR1 Expanded Alleles: A Rare Event in Two Independent Fragile X Syndrome Families. Genes, 2020, 11, 248.	2.4	7
21	Altered mitochondrial function in cells carrying a premutation or unmethylated full mutation of the FMR1 gene. Human Genetics, 2020, 139, 227-245.	3.8	16
22	Insights into Genetic Susceptibility to Melanoma by Gene Panel Testing: Potential Pathogenic Variants in ACD, ATM, BAP1, and POT1. Cancers, 2020, 12, 1007.	3.7	19
23	Natural small molecules as inhibitors of coronavirus lipid-dependent attachment to host cells: a possible strategy for reducing SARS-COV-2 infectivity?. Acta Biomedica, 2020, 91, 161-164.	0.3	89
24	Genetic analysis of intellectual disability and autism. Acta Biomedica, 2020, 91, e2020003.	0.3	18
25	Bioinformatic analysis indicates that SARS-CoV-2 is unrelated to known artificial coronaviruses. European Review for Medical and Pharmacological Sciences, 2020, 24, 4558-4564.	0.7	14
26	Italian SARS-CoV-2 patients in intensive care: towards an identikit for subjects at risk?. European Review for Medical and Pharmacological Sciences, 2020, 24, 9698-9704.	0.7	9
27	A novel nonsense PTH1R variant shows incomplete penetrance of primary failure of eruption: a case report. BMC Oral Health, 2019, 19, 249.	2.3	5
28	Combined use of medically-assisted reproductive techniques: a new bioethical issue. Acta Biomedica, 2019, 90, 58-61.	0.3	2
29	Genetics of lipedema: new perspectives on genetic research and molecular diagnoses. European Review for Medical and Pharmacological Sciences, 2019, 23, 5581-5594.	0.7	36
30	PipeMAGI: an integrated and validated workflow for analysis of NGS data for clinical diagnostics. European Review for Medical and Pharmacological Sciences, 2019, 23, 6753-6765.	0.7	17
31	Reactivation of the FMR1 Gene. , 2017, , 341-360.		0
32	Advances in understanding â€“ genetic basis of intellectual disability. F1000Research, 2016, 5, 599.	1.6	95
33	CGG Repeat-Induced FMR1 Silencing Depends on the Expansion Size in Human iPSCs and Neurons Carrying Unmethylated Full Mutations. Stem Cell Reports, 2016, 7, 1059-1071.	4.8	25
34	Rho Kinase Inhibition Is Essential During In Vitro Neurogenesis and Promotes Phenotypic Rescue of Human Induced Pluripotent Stem Cell-Derived Neurons With Oligophrenin-1 Loss of Function. Stem Cells Translational Medicine, 2016, 5, 860-869.	3.3	17
35	Defining the role of the CGGBP1 protein in FMR1 gene expression. European Journal of Human Genetics, 2016, 24, 697-703.	2.8	6
36	ZC4H2, an XLID gene, is required for the generation of a specific subset of CNS interneurons. Human Molecular Genetics, 2015, 24, 4848-4861.	2.9	48

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37	Molecular Analysis, Pathogenic Mechanisms, and Readthrough Therapy on a Large Cohort of<scp>K</scp>abuki Syndrome Patients. Human Mutation, 2014, 35, 841-850.	2.5	87
38	Epigenetics, fragile X syndrome and transcriptional therapy. American Journal of Medical Genetics, Part A, 2013, 161, 2797-2808.	1.2	31
39	Role of CTCF Protein in Regulating FMR1 Locus Transcription. PLoS Genetics, 2013, 9, e1003601.	3.5	38
40	Targeted high-throughput sequencing for diagnosis of genetically heterogeneous diseases: efficient mutation detection in Bardet-Biedl and AlstrÃ¶m Syndromes. Journal of Medical Genetics, 2012, 49, 502-512.	3.2	104
41	Mutations in KANSL1 cause the 17q21.31 microdeletion syndrome phenotype. Nature Genetics, 2012, 44, 636-638.	21.4	148
42	Mental retardation: Is naming the real issue?. American Journal of Medical Genetics, Part A, 2011, 155, 974-975.	1.2	10
43	The Pittâ€Hopkins syndrome: Report of 16 new patients and clinical diagnostic criteria. American Journal of Medical Genetics, Part A, 2011, 155, 1536-1545.	1.2	55
44	Insertion of 16 amino acids in the BAR domain of the oligophrenin 1 protein causes mental retardation and cerebellar hypoplasia in an Italian family. Human Mutation, 2011, 32, E2294-E2307.	2.5	20
45	Unexpected finding of a paternal premutation of the fragile X FMR1 gene in a female fetus of a premutation carrier mother. American Journal of Medical Genetics, Part A, 2010, 152A, 409-412.	1.2	0
46	A doubleâ€blind, parallel, multicenter comparison of <scp>L</scp>â€acetylcarnitine with placebo on the attention deficit hyperactivity disorder in fragile X syndrome boys. American Journal of Medical Genetics, Part A, 2008, 146A, 803-812.	1.2	91
47	Epigenetic analysis reveals a euchromatic configuration in the FMR1 unmethylated full mutations. European Journal of Human Genetics, 2008, 16, 1487-1498.	2.8	87
48	A unique case of reversion to normal size of a maternal premutation FMR1 allele in a normal boy. European Journal of Human Genetics, 2008, 16, 209-214.	2.8	24
49	XLMR genes: update 2007. European Journal of Human Genetics, 2008, 16, 422-434.	2.8	155
50	Modest reactivation of the mutant FMR1 gene by valproic acid is accompanied by histone modifications but not DNA demethylation. Pharmacogenetics and Genomics, 2008, 18, 738-741.	1.5	39
51	Ovarian dysfunction and FMR1 alleles in a large Italian family with POF and FRAXA disorders: case report. BMC Medical Genetics, 2007, 8, 18.	2.1	14
52	MRX87 family with Aristaless Xdup24bp mutation and implication for polyAlanine expansions. BMC Medical Genetics, 2007, 8, 25.	2.1	20
53	A new function for the fragile X mental retardation protein in regulation of PSD-95 mRNA stability. Nature Neuroscience, 2007, 10, 578-587.	14.8	318
54	A truncating mutation in theIL1RAPL1 gene is responsible for X-linked mental retardation in the MRX21 family. American Journal of Medical Genetics, Part A, 2006, 140A, 482-487.	1.2	51

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55	Differential epigenetic modifications in the FMR1 gene of the fragile X syndrome after reactivating pharmacological treatments. European Journal of Human Genetics, 2005, 13, 641-648.	2.8	83
56	Fragile X syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2005, 137C, 32-37.	1.6	60
57	Molecular dissection of the events leading to inactivation of the FMR1 gene. Human Molecular Genetics, 2005, 14, 267-277.	2.9	120
58	Clinical Picture, Evolution and Peculiar Molecular Findings in a Very Large Pedigree with Wolfram Syndrome. Journal of Pediatric Endocrinology and Metabolism, 2005, 18, 1391-7.	0.9	12
59	X-Linked Mental Retardation (XLMR): From Clinical Conditions to Cloned Genes. Critical Reviews in Clinical Laboratory Sciences, 2004, 41, 117-158.	6.1	27
60	Expansion to full mutation of a FMR1 intermediate allele over two generations. European Journal of Human Genetics, 2004, 12, 333-336.	2.8	38
61	Understanding the biological underpinnings of fragile X syndrome. Current Opinion in Pediatrics, 2003, 15, 559-566.	2.0	26
62	Quantitative analysis of DNA demethylation and transcriptional reactivation of the FMR1 gene in fragile X cells treated with 5-azadeoxycytidine. Nucleic Acids Research, 2002, 30, 3278-3285.	14.5	103
63	A novel gene, FAM11A, associated with the FRAXF CpG island is transcriptionally silent in FRAXF full mutation. European Journal of Human Genetics, 2002, 10, 767-772.	2.8	21
64	Pharmacological reactivation of inactive genes: the fragile X experience. Brain Research Bulletin, 2001, 56, 383-387.	3.0	13
65	XLMR genes: update 2000. European Journal of Human Genetics, 2001, 9, 71-81.	2.8	74
66	Effects of Melatonin Treatment in Septic Newborns. Pediatric Research, 2001, 50, 756-760.	2.3	452
67	Genetics of mental retardation. Current Opinion in Pediatrics, 2000, 12, 529-535.	2.0	39
68	Syndromic XLMR genes (MRXS): Update 2000. American Journal of Medical Genetics Part A, 2000, 94, 361-363.	2.4	23
69	X-linked mental retardation. American Journal of Medical Genetics Part A, 2000, 97, 173-173.	2.4	2
70	X-Linked Mental Retardation. Advances in Genetics, 1999, 41, 55-94.	1.8	16
71	SMT3A,a Human Homologue of theS. cerevisiae SMT3Gene, Maps to Chromosome 21qter and Defines a Novel Gene Family. Genomics, 1997, 40, 362-366.	2.9	112
72	XLMRgenes: Update 1996. , 1996, 64, 147-157.		88

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73	No apparent involvement of the FMR1 gene in five patients with phenotypic manifestations of the fragile X syndrome. American Journal of Medical Genetics Part A, 1994, 51, 309-314.	2.4	13
74	Unstable triplets and their mutational mechanism: Size reduction of the CGG repeat vs. germline mosaicism in the fragile X syndrome. American Journal of Medical Genetics Part A, 1994, 51, 517-521.	2.4	27
75	XLMR genes: Update 1994. American Journal of Medical Genetics Part A, 1994, 51, 542-549.	2.4	51
76	XLMR genes: Update 1992. American Journal of Medical Genetics Part A, 1992, 43, 373-382.	2.4	31