Claudio Graziano

List of Publications by Year
in descending order

80	1,787	304602	315616
papers	citations	22 h-index	38 g-index

85	85	85	4183
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Refining the mutational spectrum and geneâ€“phenotype correlates in pontocerebellar hypoplasia: results of a multicentric study. Journal of Medical Genetics, 2022, 59, 399-409.	1.5	13
2	Recurrent NF1 gene variants and their genotype/phenotype correlations in patients with Neurofibromatosis type I. Genes Chromosomes and Cancer, 2022, 61, 10-21.	1.5	6
3	Adult phenotype in Koolen-de Vries/KANSL1 haploinsufficiency syndrome. Journal of Medical Genetics, 2022, 59, 189-195.	1.5	6
4	Increased intracranial arterial tortuosity is associated with worse cardiovascular outcome in patients with Loeys-Dietz syndrome. Journal of Clinical Neuroscience, 2022, 96, 38-42.	0.8	1
5	Consolidation of the clinical and genetic definition of aSOX4-related neurodevelopmental syndrome. Journal of Medical Genetics, 2022, 59, 1058-1068.	1.5	10
6	There Is More Than Meets the Eye: Identification of Dual Molecular Diagnosis in Patients Affected by Hearing Loss. Biomedicines, 2022, 10, 12.	1.4	2
7	HDAC9structural variants disruptingTWIST1transcriptional regulation lead to craniofacial and limb malformations. Genome Research, 2022, 32, 1242-1253.	2.4	5
8	Clinical spectrum and followâ€up in six individuals with Lambâ€“Shaffer syndrome (<scp>SOX5</scp>). American Journal of Medical Genetics, Part A, 2021, 185, 608-613.	0.7	6
9	A New Homozygous CACNB2 Mutation has Functional Relevance and Supports a Role for Calcium Channels in Autism Spectrum Disorder. Journal of Autism and Developmental Disorders, 2021, 51, 377-381.	1.7	5
10	Preferences of Italian patients for return of secondary findings from clinical genome/exome sequencing. Journal of Genetic Counseling, 2021, 30, 665-675.	0.9	2
11	A Novel Phenotype of Junctional Epidermolysis Bullosa with Transient Skin Fragility and Predominant Ocular Involvement Responsive to Human Amniotic Membrane Eyedrops. Genes, 2021, 12, 716.	1.0	5
12	Coronary Artery Aneurysms in Patients With Marfan Syndrome: Frequent, Progressive, and Relevant. Canadian Journal of Cardiology, 2021, 37, 1225-1231.	0.8	7
13	Expanding the Neurological Phenotype of Ring Chromosome 10 Syndrome: A Case Report and Review of the Literature. Genes, 2021, 12, 1513.	1.0	2
14	Deciphering the pathogenesis of the COL4â€related hematuric nephritis: A genotype/phenotype study. Molecular Genetics & Genomic Medicine, 2021, 9, e1576.	0.6	2
15	An international cohort study of autosomal dominant tubulointerstitial kidney disease due to mutations identifies distinct clinical subtypes. Kidney International, 2020, 98, 1589-1604.	2.6	27
16	Lights and Shadows in the Genetics of Syndromic and Non-Syndromic Hearing Loss in the Italian Population. Genes, 2020, 11, 1237.	1.0	13
17	Intracranial Arterial Tortuosity in Marfan Syndrome and Loeys-Dietz Syndrome: Tortuosity Index Evaluation Is Useful in the Differential Diagnosis. American Journal of Neuroradiology, 2020, 41, 1916-1922.	1.2	12
18	P0085MAYO AND PRO-PKD SCORE CONCORDANCE FOR PROGRESSION OF RENAL FALIURE EVALUATION IN ADPKD PATIENTS. Nephrology Dialysis Transplantation, 2020, 35, .	0.4	0

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19	Kidney Transplant in Fabry Disease: A Revision of the Literature. Medicina (Lithuania), 2020, 56, 284.	0.8	9
20	Gene Panel Analysis in a Large Cohort of Patients With Autosomal Dominant Polycystic Kidney Disease Allows the Identification of 80 Potentially Causative Novel Variants and the Characterization of a Complex Genetic Architecture in a Subset of Families. Frontiers in Genetics, 2020, 11, 464.	1.1	26
21	Autozygosity-driven genetic diagnosis in consanguineous families from Italy and the Greater Middle East. Human Genetics, 2020, 139, 1429-1441.	1.8	8
22	High prevalence of arterial dissection in patients with Loeysâ€“Dietz syndrome and cerebral aneurysm. Vascular Medicine, 2020, 25, 218-220.	0.8	10
23	Aortic arch geometry predicts outcome in patients with Loeysâ€“Dietz syndrome independent of the causative gene. American Journal of Medical Genetics, Part A, 2020, 182, 1673-1680.	0.7	4
24	Next generation sequencing study in a cohort of Italian patients with syndromic hearing loss. Hearing Research, 2019, 381, 107769.	0.9	7
25	A new MEFV gene mutation in an Iranian patient with familial Mediterranean fever. Reumatismo, 2019, 71, 85-87.	0.4	1
26	Novel Mutations and Unreported Clinical Features in KBG Syndrome. Molecular Syndromology, 2019, 10, 130-138.	0.3	23
27	Sleep in Mowat-Wilson Syndrome: a clinical and video-polysomnographic study. Sleep Medicine, 2019, 61, 44-51.	0.8	4
28	Challenges in the clinical interpretation of small de novo copy number variants in neurodevelopmental disorders. Gene, 2019, 706, 162-171.	1.0	9
29	<i>HDAC8</i> Loss of Function and <i>SHOX</i> Haploinsufficiency: Two Independent Genetic Defects Responsible for a Complex Phenotype. Cytogenetic and Genome Research, 2019, 157, 135-140.	0.6	3
30	De Novo SOX4 Variants Cause a Neurodevelopmental Disease Associated with Mild Dysmorphism. American Journal of Human Genetics, 2019, 104, 246-259.	2.6	40
31	Genomic Studies in a Large Cohort of Hearing Impaired Italian Patients Revealed Several New Alleles, a Rare Case of Uniparental Disomy (UPD) and the Importance to Search for Copy Number Variations. Frontiers in Genetics, 2018, 9, 681.	1.1	25
32	Next Generation Molecular Diagnosis of Hereditary Spastic Paraplegias: An Italian Cross-Sectional Study. Frontiers in Neurology, 2018, 9, 981.	1.1	64
33	A mosaic intragenic microduplication of LAMA1 and a constitutional 18p11.32 microduplication in a patient with keratosis pilaris and intellectual disability. American Journal of Medical Genetics, Part A, 2018, 176, 2395-2403.	0.7	9
34	Insights into Mutation Effect in Three Poikiloderma with Neutropenia Patients by Transcript Analysis and Disease Evolution of Reported Patients with the Same Pathogenic Variants. Journal of Clinical Immunology, 2018, 38, 494-502.	2.0	6
35	Two novel <scp>PRNP</scp> truncating mutations broaden the spectrum of prion amyloidosis. Annals of Clinical and Translational Neurology, 2018, 5, 777-783.	1.7	15
36	Bi-allelic mutations in TRAPPC2L result in a neurodevelopmental disorder and have an impact on RAB11 in fibroblasts. Journal of Medical Genetics, 2018, 55, 753-764.	1.5	39

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37	A new PLA2G6 mutation in a family with infantile neuroaxonal dystrophy. Journal of the Neurological Sciences, 2017, 381, 209-212.	0.3	9
38	Novel Mutations in Neurogenic Chronic Intestinal Pseudo-Obstruction Identified by High-Throughput Sequencing. Gastroenterology, 2017, 152, S129.	0.6	0
39	A de novo PUF60 mutation in a child with a syndromic form of coloboma and persistent fetal vasculature. Ophthalmic Genetics, 2017, 38, 590-592.	0.5	16
40	Patterns of Novel Alleles and Genotype/Phenotype Correlations Resulting from the Analysis of 108 Previously Undetected Mutations in Patients Affected by Neurofibromatosis Type I. International Journal of Molecular Sciences, 2017, 18, 2071.	1.8	11
41	Unravelling the Complexity of Inherited Retinal Dystrophies Molecular Testing: Added Value of Targeted Next-Generation Sequencing. BioMed Research International, 2016, 2016, 1-14.	0.9	47
42	New patients with Temple syndrome caused by 14q32 deletion: Genotypeâ€phenotype correlations and risk of thyroid cancer. American Journal of Medical Genetics, Part A, 2016, 170, 162-169.	0.7	15
43	Recurrent De Novo and Biallelic Variation of ATAD3A , Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. American Journal of Human Genetics, 2016, 99, 831-845.	2.6	146
44	Cytogenetic and molecular characterization of a recombinant X chromosome in a family with a severe neurologic phenotype and macular degeneration. Molecular Cytogenetics, 2015, 8, 58.	0.4	4
45	Syndromic intellectual disability: A new phenotype caused by an aromatic amino acid decarboxylase gene (DDC) variant. Gene, 2015, 559, 144-148.	1.0	27
46	Usher syndrome: An effective sequencing approach to establish a genetic and clinical diagnosis. Hearing Research, 2015, 320, 18-23.	0.9	26
47	Validation of CFTR intronic variants identified during cystic fibrosis population screening by a minigene splicing assay. Clinical Chemistry and Laboratory Medicine, 2015, 53, 1719-23.	1.4	12
48	HCFC1 loss-of-function mutations disrupt neuronal and neural progenitor cells of the developing brain. Human Molecular Genetics, 2015, 24, 3335-3347.	1.4	47
49	Mutations in RAD21 Disrupt Regulation of APOB in Patients With Chronic Intestinal Pseudo-Obstruction. Gastroenterology, 2015, 148, 771-782.e11.	0.6	71
50	Unbalanced Translocations Involving Chromosome Region 10q25.3q26.3 in Patients with Intellectual Disability and Complex Phenotypes. Cytogenetic and Genome Research, 2014, 144, 169-177.	0.6	1
51	Single gene microdeletions and microduplication of 3p26.3 in three unrelated families: CNTN6 as a new candidate gene for intellectual disability. Molecular Cytogenetics, 2014, 7, 97.	0.4	51
52	A mutation in PAK3 with a dual molecular effect deregulates the RAS/MAPK pathway and drives an X-linked syndromic phenotype. Human Molecular Genetics, 2014, 23, 3607-3617.	1.4	33
53	Maternally inherited genetic variants of CADPS 2 are present in Autism Spectrum Disorders and Intellectual Disability patients. EMBO Molecular Medicine, 2014, 6, 795-809.	3.3	42
54	9q31.1q31.3 deletion in two patients with similar clinical features: A newly recognized microdeletion syndrome?. American Journal of Medical Genetics, Part A, 2014, 164, 685-690.	0.7	9

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55	Array CGH analysis of a cohort of Russian patients with intellectual disability. Gene, 2014, 536, 145-150.	1.0	40
56	Novel INF2 mutations in an Italian cohort of patients with focal segmental glomerulosclerosis, renal failure and Charcot-Marie-Tooth neuropathy. Nephrology Dialysis Transplantation, 2014, 29, iv80-iv86.	0.4	28
57	Su2019 Functional Characterization of a Novel RAD21 Mutation in Familial Chronic Intestinal Pseudo-Obstruction (CIPO). Gastroenterology, 2014, 146, S-524.	0.6	0
58	Maternally inherited genetic variants of CADPS 2 are present in Autism Spectrum Disorders and Intellectual Disability patients. EMBO Molecular Medicine, 2014, 6, 1639-1639.	3.3	9
59	DifferentTGM1mutation spectra in Italian and Portuguese patients with autosomal recessive congenital ichthyosis: evidence of founder effects in Portugal. British Journal of Dermatology, 2013, 168, 1364-1367.	1.4	3
60	EEC- and ADULT-AssociatedTP63Mutations Exhibit Functional Heterogeneity Toward P63 Responsive Sequences. Human Mutation, 2013, 34, 894-904.	1.1	19
61	Functional Analysis of Missense Mutations ofOAT, Causing Gyrate Atrophy of Choroid and Retina. Human Mutation, 2013, 34, 229-236.	1.1	23
62	An additional family with association of hereditary thrombocytosis and transverse limb deficiency: Confirmation of a rare clinical spectrum. American Journal of Medical Genetics, Part A, 2012, 158A, 3211-3213.	0.7	2
63	Genetics of human enteric neuropathies. Progress in Neurobiology, 2012, 96, 176-189.	2.8	36
64	Intracranial calcification in early infantile Krabbe disease: nothing new under the sun. Developmental Medicine and Child Neurology, 2012, 54, 376-379.	1.1	14
65	Test genetici e consenso informato. Salute E Societa, 2012, , 68-95.	0.0	0
66	FA2H-related disorders: a novel c.270+3A>T splice-site mutation leads to a complex neurodegenerative phenotype. Developmental Medicine and Child Neurology, 2011, 53, 958-961.	1.1	39
67	Two distinct thyroid tumours in a patient with Cowden syndrome carrying both a 10q23 and a mitochondrial DNA germline deletion. Journal of Medical Genetics, 2011, 48, 779-782.	1.5	14
68	Spinocerebellar ataxia type 12 identified in two Italian families may mimic sporadic ataxia. Movement Disorders, 2010, 25, 1269-1273.	2.2	25
69	Mutations and polymorphisms of the skeletal muscle ÃŽÂ±-actin gene (ACTA1). Human Mutation, 2009, 30, 1267-1277.	1.1	198
70	Functional polymorphisms of the microsomal epoxide hydrolase gene: A reappraisal on a early-onset lung cancer patients series. Lung Cancer, 2009, 63, 187-193.	0.9	22
71	Association of hereditary thrombocythemia and distal limb defects with a thrombopoietin gene mutation. Blood, 2009, 114, 1655-1657.	0.6	21
72	A de novo nonsense mutation ofPAX6 gene in a patient with aniridia, ataxia, and mental retardation. American Journal of Medical Genetics, Part A, 2007, 143A, 1802-1805.	0.7	28

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73	Fatal hypertrophic cardiomyopathy and nemaline myopathy associated with ACTA1 K336E mutation. Neuromuscular Disorders, 2006, 16, 548-552.	0.3	83
74	Psychological consequences of prenatal diagnosis in a case of familial Angelman Syndrome. Prenatal Diagnosis, 2006, 26, 1156-1159.	1.1	2
75	MYO7A mutation screening in Usher syndrome type I patients from diverse origins. Journal of Medical Genetics, 2006, 44, e71-e71.	1.5	53
76	De novoÎ±-actin mutations in monozygotic twins. Clinical Genetics, 2005, 68, 91-92.	1.0	3
77	Sequence diversity within the HA-1 gene as detected by melting temperature assay without oligonucleotide probes. BMC Medical Genetics, 2005, 6, 36.	2.1	5
78	PRV-1 , erythroid colonies and platelet Mpl are unrelated to thrombosis in essential thrombocythaemia. British Journal of Haematology, 2004, 127, 214-219.	1.2	21
79	Alpha-actin gene mutations and polymorphisms in Italian patients with nemaline myopathy. International Journal of Molecular Medicine, 2004, 13, 805-9.	1.8	15
80	Clinical and genetic heterogeneity in autosomal recessive nemaline myopathy. Neuromuscular Disorders, 1999, 9, 564-572.	0.3	84