Pietro Palumbo

List of Publications by Year
in descending order

38 papers	602 citations	687220 13 h-index	677027 22 g-index
38 all docs	38 docs citations	38 times ranked	1517 citing authors

#	Article	IF	Citations
1	Pharmacogenomics of Pediatric Cardiac Arrest: Cisplatin Treatment Worsened by a Ryanodine Receptor 2 Gene Mutation. Neurology International, 2022, 12, 80-88.	0.2	1
2	GDF5 mutation case report and a systematic review of molecular and clinical spectrum: Expanding current knowledge on genotype-phenotype correlations. Bone, 2021, 144, 115803.	1.4	7
3	Whole Exome Sequencing Reveals a Novel AUTS2 In-Frame Deletion in a Boy with Global Developmental Delay, Absent Speech, Dysmorphic Features, and Cerebral Anomalies. Genes, 2021, 12, 229.	1.0	8
4	A Novel Genetic Variant in the WFS1 Gene in a Patient with Partial Uniparental Mero-Isodisomy of Chromosome 4. International Journal of Molecular Sciences, 2021, 22, 8082.	1.8	1
5	Novel STAG1 Frameshift Mutation in a Patient Affected by a Syndromic Form of Neurodevelopmental Disorder. Genes, 2021, 12, 1116.	1.0	2
6	The recurrent SETBP1 c.2608Gâ€‰>â€‰A, p.(Gly870Ser) variant in a patient with Schinzel-Giedion syndrome: an illustrative case of the utility of whole exome sequencing in a critically ill neonate. Italian Journal of Pediatrics, 2020, 46, 74.	1.0	6
7	A Private 16q24.2q24.3 Microduplication in a Boy with Intellectual Disability, Speech Delay and Mild Dysmorphic Features. Genes, 2020, 11, 707.	1.0	10
8	Compound Phenotype Due to Recessive Variants in LARP7 and OTOG Genes Disclosed by an Integrated Approach of SNP-Array and Whole Exome Sequencing. Genes, 2020, 11, 379.	1.0	3
9	Long QT syndrome in chromosome 7q35q36.3 deletion involving KCNH2 gene: Warning for chlorpheniramine prescription. Molecular Genetics & Genomic Medicine, 2019, 7, e855.	0.6	4
10	Genomic inversions and GOLGA core duplicons underlie disease instability at the 15q25 locus. PLoS Genetics, 2019, 15, e1008075.	1.5	17
11	Sudden death in mild hypertrophic cardiomyopathy with compound DSG2/DSC2/MYH6 mutations: Revisiting phenotype after genetic assessment in a master runner athlete. Journal of Electrocardiology, 2019, 53, 95-99.	0.4	10
12	Refinement of the critical 7p22.1 deletion region: Haploinsufficiency of ACTB is the cause of the 7p22.1 microdeletion-related developmental disorders. European Journal of Medical Genetics, 2018, 61, 248-252.	0.7	10
13	Sudden cardiac death in J wave syndrome with short QT associated to a novel mutation in Nav 1.8 coding gene SCN10A: First case report for a possible pharmacogenomic role. Journal of Electrocardiology, 2018, 51, 809-813.	0.4	10
14	Clinical and molecular characterization of an emerging chromosome 22q13.31 microdeletion syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 391-398.	0.7	15
15	Putative TMPRSS3/GJB2 digenic inheritance of hearing loss detected by targeted resequencing. Molecular and Cellular Probes, 2017, 33, 24-27.	0.9	8
16	Clinical and molecular characterization of a second family with the 12q14 microdeletion syndrome and review of the literature. American Journal of Medical Genetics, Part A, 2017, 173, 1922-1930.	0.7	7
17	The epilepsy phenotype in adult patients with intellectual disability and pathogenic copy number variants. Seizure: the Journal of the British Epilepsy Association, 2017, 53, 86-93.	0.9	4
18	Developmental Coordination Disorder in a Patient with Mental Disability and a Mild Phenotype Carrying Terminal 6q26-qter Deletion. Frontiers in Genetics, 2017, 8, 206.	1.1	7

#	Article	IF	Citations
19	Clinical and molecular characterization of a de novo 19p13.3 microdeletion. Molecular Cytogenetics, 2016, 9, 40.	0.4	6
20	<i>PARK2</i> Microduplication: Clinical and Molecular Characterization of a Further Case and Review of the Literature. Molecular Syndromology, 2016, 7, 282-286.	0.3	6
21	De novo microduplication of CHL1 in a patient with non-syndromic developmental phenotypes. Molecular Cytogenetics, 2015, 8, 66.	0.4	19
22	Maternal uniparental isodisomy (iUPD) of chromosome 4 in a subject with mild intellectual disability and speech delay. American Journal of Medical Genetics, Part A, 2015, 167, 2219-2222.	0.7	5
23	Paternal uniparental disomy chromosome 14â€like syndrome due a maternal de novo 160â€‰kb deletion at the 14q32.2 region not encompassing the IGâ€•and the MEG3â€DMRs: Patient report and genotypeâ€“phenotype correlation. American Journal of Medical Genetics, Part A, 2015, 167, 3130-3138.	0.7	15
24	Excess of runs of homozygosity is associated with severe cognitive impairment in intellectual disability. Genetics in Medicine, 2015, 17, 396-399.	1.1	19
25	Microdeletion of 12q24.31: Report of a girl with intellectual disability, stereotypies, seizures and facial dysmorphisms. American Journal of Medical Genetics, Part A, 2015, 167, 438-444.	0.7	32
26	Report of a patient and further clinical and molecular characterization of interstitial 4p16.3 microduplication. Molecular Cytogenetics, 2015, 8, 15.	0.4	12
27	Target sequencing approach intended to discover new mutations in non-syndromic intellectual disability. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2015, 781, 32-36.	0.4	10
28	Incomplete penetrance and phenotypic variability of 6q16 deletions including SIM1. European Journal of Human Genetics, 2015, 23, 1010-1018.	1.4	35
29	Testis development in the absence of SRY: chromosomal rearrangements at SOX9 and SOX3. European Journal of Human Genetics, 2015, 23, 1025-1032.	1.4	59
30	A de novo 11p13 Microduplication in a Patient with Some Features Invoking Silver-Russell Syndrome. Molecular Syndromology, 2014, 5, 11-18.	0.3	7
31	TBR1 is the candidate gene for intellectual disability in patients with a 2q24.2 interstitial deletion. American Journal of Medical Genetics, Part A, 2014, 164, 828-833.	0.7	52
32	A novel CISD2 intragenic deletion, optic neuropathy and platelet aggregation defect in Wolfram syndrome type 2. BMC Medical Genetics, 2014, 15, 88.	2.1	59
33	Variable phenotype in 17q12 microdeletions: Clinical and molecular characterization of a new case. Gene, 2014, 538, 373-378.	1.0	28
34	20 novel point mutations and one large deletion in EXT1 and EXT2 genes: Report of diagnostic screening in a large Italian cohort of patients affected by hereditary multiple exostosis. Gene, 2013, 515, 339-348.	1.0	24
35	8q12.1q12.3 de novo microdeletion involving the CHD7 gene in a patient without the major features of CHARGE syndrome: Case report and critical review of the literature. Gene, 2013, 513, 209-213.	1.0	8
36	3p14.1 de novo microdeletion involving the FOXP1 gene in an adult patient with autism, severe speech delay and deficit of motor coordination. Gene, 2013, 516, 107-113.	1.0	38

#	Article	IF	Citations
37	An emerging phenotype of interstitial 15q25.2 microdeletions: Clinical report and review. American Journal of Medical Genetics, Part A, 2012, 158A, 3182-3189.	0.7	14
38	A novel deletion in 2q24.1q24.2 in a girl with mental retardation and generalized hypotonia: a case report. Molecular Cytogenetics, 2012, 5, 1.	0.4	24