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	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
2	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
3	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
4	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
5	Incomplete penetrance and phenotypic variability of 6q16 deletions including SIM1. European Journal of Human Genetics, 2015, 23, 1010-1018.	1.4	35
6	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
7	Variable phenotype in 17q12 microdeletions: Clinical and molecular characterization of a new case. Gene, 2014, 538, 373-378.	1.0	28
8	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
9	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
10	De novo microduplication of CHL1 in a patient with non-syndromic developmental phenotypes. Molecular Cytogenetics, 2015, 8, 66.	0.4	19
11	Excess of runs of homozygosity is associated with severe cognitive impairment in intellectual disability. Genetics in Medicine, 2015, 17, 396-399.	1.1	19
12	Genomic inversions and GOLGA core duplicons underlie disease instability at the 15q25 locus. PLoS Genetics, 2019, 15, e1008075.	1.5	17
13	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
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	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
16	Report of a patient and further clinical and molecular characterization of interstitial 4p16.3 microduplication. Molecular Cytogenetics, 2015, 8, 15.	0.4	12
17	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
18	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

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19	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
20	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
21	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
22	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
23	Putative TMPRSS3/GJB2 digenic inheritance of hearing loss detected by targeted resequencing. Molecular and Cellular Probes, 2017, 33, 24-27.	0.9	8
24	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
25	A de novo 11p13 Microduplication in a Patient with Some Features Invoking Silver-Russell Syndrome. Molecular Syndromology, 2014, 5, 11-18.	0.3	7
26	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
27	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
28	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
29	Clinical and molecular characterization of a de novo 19p13.3 microdeletion. Molecular Cytogenetics, 2016, 9, 40.	0.4	6
30	<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
31	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
32	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
33	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
34	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
35	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
36	Novel STAG1 Frameshift Mutation in a Patient Affected by a Syndromic Form of Neurodevelopmental Disorder. Genes, 2021, 12, 1116.	1.0	2

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37	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
38	Pharmacogenomics of Pediatric Cardiac Arrest: Cisplatin Treatment Worsened by a Ryanodine Receptor 2 Gene Mutation. Neurology International, 2022, 12, 80-88.	0.2	1