

Carmen Orellana

List of Publications by Year in descending order

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Version: 2024-02-01

58
papers

1,230
citations

394421

19
h-index

434195

31
g-index

61
all docs

61
docs citations

61
times ranked

2865
citing authors

#	ARTICLE	IF	CITATIONS
1	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. <i>American Journal of Human Genetics</i> , 2015, 97, 922-932.	6.2	101
2	High diagnostic yield of syndromic intellectual disability by targeted next-generation sequencing. <i>Journal of Medical Genetics</i> , 2017, 54, 87-92.	3.2	93
3	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffin-Siris syndrome. <i>Genetics in Medicine</i> , 2019, 21, 1295-1307.	2.4	80
4	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. <i>American Journal of Human Genetics</i> , 2019, 104, 164-178.	6.2	59
5	De Novo Variants in the F-Box Protein FBXO11 in 20 Individuals with a Variable Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2018, 103, 305-316.	6.2	48
6	Detection of known and novel genomic rearrangements by array based comparative genomic hybridisation: deletion of ZNF533 and duplication of CHARGE syndrome genes. <i>Journal of Medical Genetics</i> , 2008, 45, 432-437.	3.2	47
7	Refining the phenotype associated with <i>GNB1</i> mutations: Clinical data on 18 newly identified patients and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2259-2275.	1.2	47
8	Reciprocal deletion and duplication at 2q23.1 indicates a role for MBD5 in autism spectrum disorder. <i>European Journal of Human Genetics</i> , 2014, 22, 57-63.	2.8	42
9	The Doublecortin Gene, A New Molecular Marker to Detect Minimal Residual Disease in Neuroblastoma. <i>Diagnostic Molecular Pathology</i> , 2005, 14, 53-57.	2.1	41
10	Hypermethylation of apoptotic genes as independent prognostic factor in neuroblastoma disease. <i>Molecular Carcinogenesis</i> , 2011, 50, 153-162.	2.7	39
11	Infectious and Immunologic Phenotype of MECP2 Duplication Syndrome. <i>Journal of Clinical Immunology</i> , 2015, 35, 168-181.	3.8	35
12	Novel mutations of NFIX gene causing Marshall-Smith syndrome or Sotos-like syndrome: one gene, two phenotypes. <i>Pediatric Research</i> , 2015, 78, 533-539.	2.3	35
13	Evaluation of MLPA for the detection of cryptic subtelomeric rearrangements. <i>Translational Research</i> , 2006, 147, 295-300.	2.3	33
14	Multi-system involvement in a severe variant of fibrodysplasia ossificans progressiva (<i>ACVR1</i>) Tj ETQq0 0 0 rgBT /Overlock 10 T 2265-2271.	1.2	33
15	De novo mutations in genes of mediator complex causing syndromic intellectual disability: mediatoropathy or transcriptomopathy?. <i>Pediatric Research</i> , 2016, 80, 809-815.	2.3	27
16	Expression of aquaporins early in human pregnancy. <i>Early Human Development</i> , 2012, 88, 589-594.	1.8	25
17	Pediatric Brain Tumors: Loss of Heterozygosity at 17p and TP53 Gene Mutations. <i>Cancer Genetics and Cytogenetics</i> , 1998, 102, 93-99.	1.0	23
18	De novo Interstitial Triplication of <i>MECP2</i> in a Girl with Neurodevelopmental Disorder and Random X Chromosome Inactivation. <i>Cytogenetic and Genome Research</i> , 2011, 135, 93-101.	1.1	22

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19	Epigenetic alterations in disseminated neuroblastoma tumour cells: influence of TMS1 gene hypermethylation in relapse risk in NB patients. <i>Journal of Cancer Research and Clinical Oncology</i> , 2010, 136, 1415-1421.	2.5	20
20	Hidden etiology of cerebral palsy: genetic and clinical heterogeneity and efficient diagnosis by next-generation sequencing. <i>Pediatric Research</i> , 2021, 90, 284-288.	2.3	20
21	Minimal disease detection in peripheral blood and bone marrow from patients with non-metastatic neuroblastoma. <i>Journal of Cancer Research and Clinical Oncology</i> , 2011, 137, 1263-1272.	2.5	19
22	X-Linked Anhidrotic (Hypohidrotic) Ectodermal Dysplasia Caused by a Novel Mutation in EDA1 Gene: 406T>G (Leu55Arg). <i>Journal of Investigative Dermatology</i> , 1999, 113, 285-286.	0.7	18
23	Novel <i>UBE3A</i> mutations causing Angelman syndrome: Different parental origin for single nucleotide changes and multiple nucleotide deletions or insertions. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 343-348.	1.2	18
24	Enrichment of ultraconserved elements among genomic imbalances causing mental delay and congenital anomalies. <i>BMC Medical Genomics</i> , 2010, 3, 54.	1.5	18
25	Phenotype profiling of patients with intellectual disability and copy number variations. <i>European Journal of Paediatric Neurology</i> , 2014, 18, 558-566.	1.6	18
26	Molecular characterization of Spanish patients with <i>MECP2</i> duplication syndrome. <i>Clinical Genetics</i> , 2020, 97, 610-620.	2.0	16
27	MAGE-A1 expression is associated with good prognosis in neuroblastoma tumors. <i>Journal of Cancer Research and Clinical Oncology</i> , 2009, 135, 523-531.	2.5	15
28	Haploinsufficiency of the Sin3/HDAC corepressor complex member SIN3B causes a syndromic intellectual disability/autism spectrum disorder. <i>American Journal of Human Genetics</i> , 2021, 108, 929-941.	6.2	15
29	There Is No Evidence That the <i>SDHB</i> Gene Is Involved in Neuroblastoma Development. <i>Oncology Research</i> , 2005, 15, 393-398.	1.5	15
30	Submicroscopic Duplication of the Wolf-Hirschhorn Critical Region with a 4p Terminal Deletion. <i>Cytogenetic and Genome Research</i> , 2009, 125, 103-108.	1.1	14
31	Mutation screening of AURKB and SYCP3 in patients with reproductive problems. <i>Molecular Human Reproduction</i> , 2013, 19, 102-108.	2.8	14
32	Duplication of 14q11.2 associates with short stature and mild mental retardation: A putative relation with quantitative trait loci. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 382-384.	1.2	12
33	Pure duplication of 19p13.3 in three members of a family with intellectual disability and literature review. Definition of a new microduplication syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1614-1620.	1.2	12
34	Subtelomeric analysis of pediatric astrocytoma: subchromosomal instability is a distinctive feature of pleomorphic xanthoastrocytoma. <i>Journal of Neuro-Oncology</i> , 2009, 93, 175-182.	2.9	11
35	A Novel TP53 Germ-Line Mutation Identified in a Girl with a Primitive Neuroectodermal Tumor and Her Father. <i>Cancer Genetics and Cytogenetics</i> , 1998, 105, 103-108.	1.0	10
36	Robust, Easy, and Dose-Sensitive Methylation Test for the Diagnosis of Prader-Willi and Angelman Syndromes. <i>Genetic Testing and Molecular Biomarkers</i> , 2006, 10, 174-177.	1.7	10

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37	Duplication at Xq13.3â€“q21.1 with syndromic intellectual disability, a probable role for the <i>ATRXL</i> gene. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 918-923.	1.2	10
38	Haploinsufficiency of the <i>MYT1L</i> gene causes intellectual disability frequently associated with behavioral disorder. <i>Genetics in Medicine</i> , 2015, 17, 683-684.	2.4	10
39	Chimeric Genes in Deletions and Duplications Associated with Intellectual Disability. <i>International Journal of Genomics</i> , 2017, 2017, 1-11.	1.6	10
40	A Novel Mutation of <i>MAGEL2</i> in a Patient with Schaaf-Yang Syndrome and Hypopituitarism. <i>International Journal of Endocrinology and Metabolism</i> , 2018, In Press, e67329.	1.0	9
41	Localization of non-specific X-linked mental retardation gene (<i>MRX73</i>) to Xp22.2. <i>American Journal of Medical Genetics Part A</i> , 2001, 102, 200-204.	2.4	8
42	The Genetic Landscape of Mitochondrial Diseases in Spain: A Nationwide Call. <i>Genes</i> , 2021, 12, 1590.	2.4	8
43	Partial Duplication of 18q Including a Distal Critical Region for Edwards Syndrome in a Patient with Normal Phenotype and Oligoasthenospermia: Case Report. <i>Cytogenetic and Genome Research</i> , 2011, 133, 78-83.	1.1	7
44	Large deletion in the Factor <i>VIII</i> gene (<i>F8</i>) involving segmental duplications in int22h shows no haematological phenotype in female carriers, but may be embryonic lethal in males. <i>British Journal of Haematology</i> , 2012, 158, 138-140.	2.5	7
45	A novel missense mutation in the <i>NSDHL</i> gene identified in a Lithuanian family by targeted nextâ€“generation sequencing causes CK syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1342-1348.	1.2	6
46	Localization of <i>MRX82</i> : A new nonsyndromic X-linked mental retardation locus to Xq24-q25 in a Basque family. , 2004, 131A, 174-178.		5
47	A subtelomeric translocation apparently implied in multiple abortions. <i>Journal of Assisted Reproduction and Genetics</i> , 2006, 23, 97-101.	2.5	5
48	Prenatal study of common submicroscopic â€œgenomic disordersâ€“using MLPA with subtelomeric/microdeletion syndrome probe mixes, among gestations with ultrasound abnormalities in the first trimester. <i>European Journal of Medical Genetics</i> , 2010, 53, 76-79.	1.3	5
49	Tyrosinemia type 1 and Angelman syndrome due to paternal uniparental isodisomy 15. <i>Journal of Inherited Metabolic Disease</i> , 2009, 32, 349-353.	3.6	4
50	Hypomethylation of the <i>KCNQ1OT1</i> imprinting center of chromosome 11 associated to Sotos-like features. <i>Journal of Human Genetics</i> , 2012, 57, 153-156.	2.3	4
51	Generation of a disease-specific iPSC cell line derived from a patient with Charcot-Marie-Tooth type 2K lacking functional <i>GDAP1</i> gene. <i>Stem Cell Research</i> , 2017, 18, 1-4.	0.7	4
52	Recombinant X chromosome in a prenatal diagnosis. <i>Cytogenetic and Genome Research</i> , 2006, 112, 337-340.	1.1	3
53	Clinical findings and molecular characterization of six subtelomeric imbalances. <i>Clinical Genetics</i> , 2007, 71, 474-479.	2.0	3
54	In Pursuit of New Imprinting Syndromes by Epimutation Screening in Idiopathic Neurodevelopmental Disorder Patients. <i>BioMed Research International</i> , 2015, 2015, 1-8.	1.9	3

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55	Prevalence of pathogenic copy number variants among children conceived by donor oocyte. Scientific Reports, 2021, 11, 6752.	3.3	1
56	Screening for microdeletions of the X-chromosome in non-specific mental retardation. , 2004, 124A, 99-101.		0
57	Response: rare chromosomal complement of trisomy 21 in a boy conceived only by IVF. Reproductive BioMedicine Online, 2010, 21, 723.	2.4	0
58	Mixed Phenotype of Langerâ€™Giedion's and Cornelia de Lange's Syndromes in an 8q23.3-q24.1 Microdeletion without TRPS1 Deletion. Journal of Pediatric Genetics, 2020, 09, 053-057.	0.7	0