Carmen Orellana

List of Publications by Year
in descending order

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58 papers	1,230 citations	394390 19 h-index	434170 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. American Journal of Human Genetics, 2015, 97, 922-932.	6.2	101
2	High diagnostic yield of syndromic intellectual disability by targeted next-generation sequencing. Journal of Medical Genetics, 2017, 54, 87-92.	3.2	93
3	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 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. American Journal of Human Genetics, 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. American Journal of Human Genetics, 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. Journal of Medical Genetics, 2008, 45, 432-437.	3.2	47
7	Refining the phenotype associated with GNB1 mutations: Clinical data on 18 newly identified patients and review of the literature. American Journal of Medical Genetics, Part A, 2018, 176, 2259-2275.	1.2	47
8	Reciprocal deletion and duplication at 2q23.1 indicates a role for MBD5 in autism spectrum disorder. European Journal of Human Genetics, 2014, 22, 57-63.	2.8	42
9	The Doublecortin Gene, A New Molecular Marker to Detect Minimal Residual Disease in Neuroblastoma. Diagnostic Molecular Pathology, 2005, 14, 53-57.	2.1	41
10	Hypermethylation of apoptotic genes as independent prognostic factor in neuroblastoma disease. Molecular Carcinogenesis, 2011, 50, 153-162.	2.7	39
11	Infectious and Immunologic Phenotype of MECP2 Duplication Syndrome. Journal of Clinical Immunology, 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. Pediatric Research, 2015, 78, 533-539.	2.3	35
13	Evaluation of MLPA for the detection of cryptic subtelomeric rearrangements. Translational Research, 2006, 147, 295-300.	2.3	33
14	Multiâ€system involvement in a severe variant of fibrodysplasia ossificans progressiva (ACVR1) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 2265-2271.	1.2	33
15	De novo mutations in genes of mediator complex causing syndromic intellectual disability: mediatorpathy or transcriptomopathy?. Pediatric Research, 2016, 80, 809-815.	2.3	27
16	Expression of aquaporins early in human pregnancy. Early Human Development, 2012, 88, 589-594.	1.8	25
17	Pediatric Brain Tumors: Loss of Heterozygosity at 17p and TP53 Gene Mutations. Cancer Genetics and Cytogenetics, 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. Cytogenetic and Genome Research, 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. Journal of Cancer Research and Clinical Oncology, 2010, 136, 1415-1421.	2.5	20
20	Hidden etiology of cerebral palsy: genetic and clinical heterogeneity and efficient diagnosis by next-generation sequencing. Pediatric Research, 2021, 90, 284-288.	2.3	20
21	Minimal disease detection in peripheral blood and bone marrow from patients with non-metastatic neuroblastoma. Journal of Cancer Research and Clinical Oncology, 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). Journal of Investigative Dermatology, 1999, 113, 285-286.	0.7	18
23	Novel UBE3A mutations causing Angelman syndrome: Different parental origin for single nucleotide changes and multiple nucleotide deletions or insertions. American Journal of Medical Genetics, Part A, 2009, 149A, 343-348.	1.2	18
24	Enrichment of ultraconserved elements among genomic imbalances causing mental delay and congenital anomalies. BMC Medical Genomics, 2010, 3, 54.	1.5	18
25	Phenotype profiling of patients with intellectual disability and copy number variations. European Journal of Paediatric Neurology, 2014, 18, 558-566.	1.6	18
26	Molecular characterization of Spanish patients with MECP2 duplication syndrome. Clinical Genetics, 2020, 97, 610-620.	2.0	16
27	MAGE-A1 expression is associated with good prognosis in neuroblastoma tumors. Journal of Cancer Research and Clinical Oncology, 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. American Journal of Human Genetics, 2021, 108, 929-941.	6.2	15
29	There Is No Evidence That the SDHB Gene Is Involved in Neuroblastoma Development. Oncology Research, 2005, 15, 393-398.	1.5	15
30	Submicroscopic Duplication of the Wolf-Hirschhorn Critical Region with a 4p Terminal Deletion. Cytogenetic and Genome Research, 2009, 125, 103-108.	1.1	14
31	Mutation screening of AURKB and SYCP3 in patients with reproductive problems. Molecular Human Reproduction, 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. American Journal of Medical Genetics, Part A, 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. American Journal of Medical Genetics, Part A, 2015, 167, 1614-1620.	1.2	12
34	Subtelomeric analysis of pediatric astrocytoma: subchromosomal instability is a distinctive feature of pleomorphic xanthoastrocytoma. Journal of Neuro-Oncology, 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. Cancer Genetics and Cytogenetics, 1998, 105, 103-108.	1.0	10
36	Robust, Easy, and Dose-Sensitive Methylation Test for the Diagnosis of Praderâ€“Willi and Angelman Syndromes. Genetic Testing and Molecular Biomarkers, 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 ATRX gene. American Journal of Medical Genetics, Part A, 2014, 164, 918-923.	1.2	10
38	Haploinsufficiency of the MYT1L gene causes intellectual disability frequently associated with behavioral disorder. Genetics in Medicine, 2015, 17, 683-684.	2.4	10
39	Chimeric Genes in Deletions and Duplications Associated with Intellectual Disability. International Journal of Genomics, 2017, 2017, 1-11.	1.6	10
40	A Novel Mutation of MAGEL2 in a Patient with Schaaf-Yang Syndrome and Hypopituitarism. International Journal of Endocrinology and Metabolism, 2018, In Press, e67329.	1.0	9
41	Localization of non-specific X-linked mental retardation gene (MRX73) to Xp22.2. American Journal of Medical Genetics Part A, 2001, 102, 200-204.	2.4	8
42	The Genetic Landscape of Mitochondrial Diseases in Spain: A Nationwide Call. Genes, 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. Cytogenetic and Genome Research, 2011, 133, 78-83.	1.1	7
44	Large deletion in the Factor <scp>VIII</scp> gene (<scp>F</scp>8) involving segmental duplications in int22h shows no haematological phenotype in female carriers, but may be embryonic lethal in males. British Journal of Haematology, 2012, 158, 138-140.	2.5	7
45	A novel missense mutation in the NSDHL gene identified in a Lithuanian family by targeted nextâ€generation sequencing causes CK syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1342-1348.	1.2	6
46	Localization of MRX82: 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. Journal of Assisted Reproduction and Genetics, 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. European Journal of Medical Genetics, 2010, 53, 76-79.	1.3	5
49	Tyrosinemia type 1 and Angelman syndrome due to paternal uniparental isodisomy 15. Journal of Inherited Metabolic Disease, 2009, 32, 349-353.	3.6	4
50	Hypomethylation of the KCNQ1OT1 imprinting center of chromosome 11 associated to Sotos-like features. Journal of Human Genetics, 2012, 57, 153-156.	2.3	4
51	Generation of a disease-specific iPS cell line derived from a patient with Charcot-Marie-Tooth type 2K lacking functional GDAP1 gene. Stem Cell Research, 2017, 18, 1-4.	0.7	4
52	Recombinant X chromosome in a prenatal diagnosis. Cytogenetic and Genome Research, 2006, 112, 337-340.	1.1	3
53	Clinical findings and molecular characterization of six subtelomeric imbalances. Clinical Genetics, 2007, 71, 474-479.	2.0	3
54	In Pursuit of New Imprinting Syndromes by Epimutation Screening in Idiopathic Neurodevelopmental Disorder Patients. BioMed Research International, 2015, 2015, 1-8.	1.9	3

#	Article	IF	Citations
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