Carmen Orellana

List of Publications by Year
in descending order

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		394421	434195
58	1,230	19	31
papers	citations	h-index	g-index

61	61	61	2865
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	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
2	Prevalence of pathogenic copy number variants among children conceived by donor oocyte. Scientific Reports, 2021, 11, 6752.	3.3	1
3	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
4	The Genetic Landscape of Mitochondrial Diseases in Spain: A Nationwide Call. Genes, 2021, 12, 1590.	2.4	8
5	Molecular characterization of Spanish patients with MECP2 duplication syndrome. Clinical Genetics, 2020, 97, 610-620.	2.0	16
6	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
7	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
8	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	2.4	80
9	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
10	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
11	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
12	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
13	High diagnostic yield of syndromic intellectual disability by targeted next-generation sequencing. Journal of Medical Genetics, 2017, 54, 87-92.	3.2	93
14	Chimeric Genes in Deletions and Duplications Associated with Intellectual Disability. International Journal of Genomics, 2017, 2017, 1-11.	1.6	10
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	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
17	In Pursuit of New Imprinting Syndromes by Epimutation Screening in Idiopathic Neurodevelopmental Disorder Patients. BioMed Research International, 2015, 2015, 1-8.	1.9	3
18	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. American Journal of Human Genetics, 2015, 97, 922-932.	6.2	101

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19	Haploinsufficiency of the MYT1L gene causes intellectual disability frequently associated with behavioral disorder. Genetics in Medicine, 2015, 17, 683-684.	2.4	10
20	Infectious and Immunologic Phenotype of MECP2 Duplication Syndrome. Journal of Clinical Immunology, 2015, 35, 168-181.	3.8	35
21	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
22	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
23	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
24	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
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	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
27	Mutation screening of AURKB and SYCP3 in patients with reproductive problems. Molecular Human Reproduction, 2013, 19, 102-108.	2.8	14
28	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
29	Expression of aquaporins early in human pregnancy. Early Human Development, 2012, 88, 589-594.	1.8	25
30	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
31	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
32	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
33	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
34	Hypermethylation of apoptotic genes as independent prognostic factor in neuroblastoma disease. Molecular Carcinogenesis, 2011, 50, 153-162.	2.7	39
35	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
36	Enrichment of ultraconserved elements among genomic imbalances causing mental delay and congenital anomalies. BMC Medical Genomics, 2010, 3, 54.	1.5	18

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37	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
38	Response: rare chromosomal complement of trisomy 21 in a boy conceived only by IVF. Reproductive BioMedicine Online, 2010, 21, 723.	2.4	0
39	Submicroscopic Duplication of the Wolf-Hirschhorn Critical Region with a 4p Terminal Deletion. Cytogenetic and Genome Research, 2009, 125, 103-108.	1.1	14
40	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
41	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
42	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
43	Tyrosinemia type 1 and Angelman syndrome due to paternal uniparental isodisomy 15. Journal of Inherited Metabolic Disease, 2009, 32, 349-353.	3.6	4
44	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
45	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
46	Clinical findings and molecular characterization of six subtelomeric imbalances. Clinical Genetics, 2007, 71, 474-479.	2.0	3
47	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
48	Evaluation of MLPA for the detection of cryptic subtelomeric rearrangements. Translational Research, 2006, 147, 295-300.	2.3	33
49	A subtelomeric translocation apparently implied in multiple abortions. Journal of Assisted Reproduction and Genetics, 2006, 23, 97-101.	2.5	5
50	Recombinant X chromosome in a prenatal diagnosis. Cytogenetic and Genome Research, 2006, 112, 337-340.	1.1	3
51	The Doublecortin Gene, A New Molecular Marker to Detect Minimal Residual Disease in Neuroblastoma. Diagnostic Molecular Pathology, 2005, 14, 53-57.	2.1	41
52	There Is No Evidence That the SDHB Gene Is Involved in Neuroblastoma Development. Oncology Research, 2005, 15, 393-398.	1.5	15
53	Screening for microdeletions of the X-chromosome in non-specific mental retardation. , 2004, 124A, 99-101.		0
54	Localization of MRX82: A new nonsyndromic X-linked mental retardation locus to Xq24-q25 in a Basque family. , 2004, 131A, 174-178.		5

#	Article	IF	Citations
55	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
56	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
57	Pediatric Brain Tumors: Loss of Heterozygosity at 17p and TP53 Gene Mutations. Cancer Genetics and Cytogenetics, 1998, 102, 93-99.	1.0	23
58	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