

Salud Borrego

List of Publications by Year in descending order

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86
papers

2,899
citations

186265

28
h-index

189892

50
g-index

86
all docs

86
docs citations

86
times ranked

3440
citing authors

#	ARTICLE	IF	CITATIONS
1	Correlation between SMA type and SMN2 copy number revisited: An analysis of 625 unrelated Spanish patients and a compilation of 2834 reported cases. <i>Neuromuscular Disorders</i> , 2018, 28, 208-215.	0.6	273
2	Differential Contributions of Rare and Common, Coding and Noncoding Ret Mutations to Multifactorial Hirschsprung Disease Liability. <i>American Journal of Human Genetics</i> , 2010, 87, 60-74.	6.2	230
3	EYS, encoding an ortholog of <i>Drosophila</i> spacemaker, is mutated in autosomal recessive retinitis pigmentosa. <i>Nature Genetics</i> , 2008, 40, 1285-1287.	21.4	175
4	Specific polymorphisms in the RET proto-oncogene are over-represented in patients with Hirschsprung disease and may represent loci modifying phenotypic expression. <i>Journal of Medical Genetics</i> , 1999, 36, 771-774.	3.2	142
5	Contribution of rare and common variants determine complex diseases—Hirschsprung disease as a model. <i>Developmental Biology</i> , 2013, 382, 320-329.	2.0	119
6	Functional Loss of Semaphorin 3C and/or Semaphorin 3D and Their Epistatic Interaction with Ret Are Critical to Hirschsprung Disease Liability. <i>American Journal of Human Genetics</i> , 2015, 96, 581-596.	6.2	118
7	A Founding Locus within the RET Proto-Oncogene May Account for a Large Proportion of Apparently Sporadic Hirschsprung Disease and a Subset of Cases of Sporadic Medullary Thyroid Carcinoma. <i>American Journal of Human Genetics</i> , 2003, 72, 88-100.	6.2	100
8	267 Spanish Exomes Reveal Population-Specific Differences in Disease-Related Genetic Variation. <i>Molecular Biology and Evolution</i> , 2016, 33, 1205-1218.	8.9	78
9	Whole exome sequencing coupled with unbiased functional analysis reveals new Hirschsprung disease genes. <i>Genome Biology</i> , 2017, 18, 48.	8.8	72
10	Unravelling the genetic basis of simplex Retinitis Pigmentosa cases. <i>Scientific Reports</i> , 2017, 7, 41937.	3.3	71
11	Mutation spectrum of EYS in Spanish patients with autosomal recessive retinitis pigmentosa. <i>Human Mutation</i> , 2010, 31, E1772-E1800.	2.5	69
12	Molecular Analysis of the RET and GDNF Genes in a Family with Multiple Endocrine Neoplasia Type 2A and Hirschsprung Disease. <i>Journal of Clinical Endocrinology and Metabolism</i> , 1998, 83, 3361-3364.	3.6	61
13	New roles of EDNRB and EDN3 in the pathogenesis of Hirschsprung disease. <i>Genetics in Medicine</i> , 2010, 12, 39-43.	2.4	59
14	Improving the management of Inherited Retinal Dystrophies by targeted sequencing of a population-specific gene panel. <i>Scientific Reports</i> , 2016, 6, 23910.	3.3	51
15	Hirschsprung's disease as a model of complex genetic etiology. <i>Histology and Histopathology</i> , 2013, 28, 1117-36.	0.7	48
16	Involvement of SOX10 in the pathogenesis of Hirschsprung disease: report of a truncating mutation in an isolated patient. <i>Journal of Molecular Medicine</i> , 2010, 88, 507-514.	3.9	40
17	Copy-Number Variations in <i>EYS</i> : A Significant Event in the Appearance of arRP. , 2011, 52, 5625.		40
18	Trans-ethnic meta-analysis of genome-wide association studies for Hirschsprung disease. <i>Human Molecular Genetics</i> , 2016, 25, ddw333.	2.9	38

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19	Association Study of 69 Genes in the Ret Pathway Identifies Low-penetrance Loci in Sporadic Medullary Thyroid Carcinoma. <i>Cancer Research</i> , 2007, 67, 9561-9567.	0.9	36
20	Mutation Screening of Multiple Genes in Spanish Patients with Autosomal Recessive Retinitis Pigmentosa by Targeted Resequencing. <i>PLoS ONE</i> , 2011, 6, e27894.	2.5	36
21	Comprehensive Analysis of NRG1 Common and Rare Variants in Hirschsprung Patients. <i>PLoS ONE</i> , 2012, 7, e36524.	2.5	36
22	Novel mutations at RET ligand genes preventing receptor activation are associated to Hirschsprung's disease. <i>Journal of Molecular Medicine</i> , 2011, 89, 471-480.	3.9	35
23	Mutational Spectrum of Semaphorin 3A and Semaphorin 3D Genes in Spanish Hirschsprung patients. <i>PLoS ONE</i> , 2013, 8, e54800.	2.5	34
24	Multilayer OMIC Data in Medullary Thyroid Carcinoma Identifies the STAT3 Pathway as a Potential Therapeutic Target in <i>RET</i> M918T Tumors. <i>Clinical Cancer Research</i> , 2017, 23, 1334-1345.	7.0	34
25	CSVS, a crowdsourcing database of the Spanish population genetic variability. <i>Nucleic Acids Research</i> , 2021, 49, D1130-D1137.	14.5	34
26	An Impairment of Long Distance SOX10 Regulatory Elements Underlies Isolated Hirschsprung Disease. <i>Human Mutation</i> , 2014, 35, 303-307.	2.5	33
27	Contributions of PHOX2B in the Pathogenesis of Hirschsprung Disease. <i>PLoS ONE</i> , 2013, 8, e54043.	2.5	30
28	The NER-related gene <i>GTF2H5</i> predicts survival in high-grade serous ovarian cancer patients. <i>Journal of Gynecologic Oncology</i> , 2016, 27, e7.	2.2	30
29	A complex additive model of inheritance for Hirschsprung disease is supported by both RET mutations and predisposing RET haplotypes. <i>Genetics in Medicine</i> , 2006, 8, 704-710.	2.4	29
30	Involvement of DNMT3B in the pathogenesis of Hirschsprung disease and its possible role as a regulator of neurogenesis in the human enteric nervous system. <i>Genetics in Medicine</i> , 2014, 16, 703-710.	2.4	29
31	Exome sequencing reveals a high genetic heterogeneity on familial Hirschsprung disease. <i>Scientific Reports</i> , 2015, 5, 16473.	3.3	29
32	A map of human microRNA variation uncovers unexpectedly high levels of variability. <i>Genome Medicine</i> , 2012, 4, 62.	8.2	28
33	Influencers on Thyroid Cancer Onset: Molecular Genetic Basis. <i>Genes</i> , 2019, 10, 913.	2.4	28
34	Analysis of <i>RET</i> Polymorphisms and Haplotypes in the Context of Sporadic Medullary Thyroid Carcinoma. <i>Thyroid</i> , 2006, 16, 411-417.	4.5	25
35	NTF-3, a gene involved in the enteric nervous system development, as a candidate gene for Hirschsprung disease. <i>Journal of Pediatric Surgery</i> , 2008, 43, 1308-1311.	1.6	24
36	What is new about the genetic background of Hirschsprung disease?. <i>Clinical Genetics</i> , 2020, 97, 114-124.	2.0	24

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37	Epigenetic Mechanisms in Hirschsprung Disease. <i>International Journal of Molecular Sciences</i> , 2019, 20, 3123.	4.1	23
38	Unmasking Retinitis Pigmentosa complex cases by a whole genome sequencing algorithm based on open-access tools: hidden recessive inheritance and potential oligogenic variants. <i>Journal of Translational Medicine</i> , 2020, 18, 73.	4.4	23
39	Expression of PROKR1 and PROKR2 in Human Enteric Neural Precursor Cells and Identification of Sequence Variants Suggest a Role in HSCR. <i>PLoS ONE</i> , 2011, 6, e23475.	2.5	22
40	Re-evaluation casts doubt on the pathogenicity of homozygous <i>USH2A</i> p.C759F. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1597-1600.	1.2	21
41	Novel association of severe neonatal encephalopathy and Hirschsprung disease in a male with a duplication at the Xq28 region. <i>BMC Medical Genetics</i> , 2010, 11, 137.	2.1	20
42	Experience of Preimplantation Genetic Diagnosis with HLA Matching at the University Hospital Virgen del Roc�o in Spain: Technical and Clinical Overview. <i>BioMed Research International</i> , 2014, 2014, 1-8.	1.9	18
43	Novel RP1 mutations and a recurrent BBS1 variant explain the co-existence of two distinct retinal phenotypes in the same pedigree. <i>BMC Genetics</i> , 2014, 15, 143.	2.7	18
44	Waardenburg syndrome type 4: Report of two new cases caused by <i>SOX10</i> mutations in Spain. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 542-547.	1.2	18
45	Overexpression of DNMT3b target genes during Enteric Nervous System development contribute to the onset of Hirschsprung disease. <i>Scientific Reports</i> , 2017, 7, 6221.	3.3	18
46	Pathways systematically associated to Hirschsprung's disease. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 187.	2.7	17
47	Deletion at 6q24.2�26 predicts longer survival of high-grade serous epithelial ovarian cancer patients. <i>Molecular Oncology</i> , 2015, 9, 422-436.	4.6	17
48	G534E Variant in <i>HABP2</i> and Nonmedullary Thyroid Cancer. <i>Thyroid</i> , 2016, 26, 987-988.	4.5	17
49	Searching the second hit in patients with inherited retinal dystrophies and monoallelic variants in ABCA4, USH2A and CEP290 by whole-gene targeted sequencing. <i>Scientific Reports</i> , 2018, 8, 13312.	3.3	17
50	Whole-exome sequencing identifies novel compound heterozygous mutations in USH2A in Spanish patients with autosomal recessive retinitis pigmentosa. <i>Molecular Vision</i> , 2013, 19, 2187-95.	1.1	17
51	Prevalence of 2314delG mutation in Spanish patients with Usher syndrome type II (USH2). <i>Ophthalmic Genetics</i> , 2000, 21, 123-128.	1.2	16
52	Exome Sequencing Reveals Novel and Recurrent Mutations with Clinical Significance in Inherited Retinal Dystrophies. <i>PLoS ONE</i> , 2014, 9, e116176.	2.5	16
53	Identification of epistatic interactions through genome-wide association studies in sporadic medullary and juvenile papillary thyroid carcinomas. <i>BMC Medical Genomics</i> , 2015, 8, 83.	1.5	15
54	Evaluation of the role of RET polymorphisms/haplotypes as modifier loci for MEN 2, and analysis of the correlation with the type of RET mutation in a series of Spanish patients. <i>International Journal of Molecular Medicine</i> , 2006, 17, 575-81.	4.0	15

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55	Comprehensive analysis of RET common and rare variants in a series of Spanish Hirschsprung patients confirms a synergistic effect of both kinds of events. <i>BMC Medical Genetics</i> , 2011, 12, 138.	2.1	14
56	Association of X-linked hydrocephalus and Hirschsprung disease: Report of a new patient with a mutation in the <i>L1CAM</i> gene. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 816-820.	1.2	14
57	Next-generation-based targeted sequencing as an efficient tool for the study of the genetic background in Hirschsprung patients. <i>BMC Medical Genetics</i> , 2015, 16, 89.	2.1	14
58	Deciphering intrafamilial phenotypic variability by exome sequencing in a Bardet-Biedl family. <i>Molecular Genetics & Genomic Medicine</i> , 2014, 2, 124-133.	1.2	13
59	One-step multiplex polymerase chain reaction for preimplantation genetic diagnosis of Huntington disease. <i>Fertility and Sterility</i> , 2010, 93, 2411-2412.	1.0	12
60	Experience of Preimplantation Genetic Diagnosis for Hemophilia at the University Hospital Virgen Del Rocío in Spain: Technical and Clinical Overview. <i>BioMed Research International</i> , 2015, 2015, 1-8.	1.9	10
61	Identification of New Potential LncRNA Biomarkers in Hirschsprung Disease. <i>International Journal of Molecular Sciences</i> , 2020, 21, 5534.	4.1	10
62	RMRP, RMST, FTX and IPW: novel potential long non-coding RNAs in medullary thyroid cancer. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 4.	2.7	10
63	The RETC620S mutation causes multiple endocrine neoplasia type 2A (MEN2A) but not Hirschsprung disease (HSCR) in a family cosegregating both phenotypes. <i>Human Mutation</i> , 2003, 22, 412-415.	2.5	9
64	Novel One-Step Multiplex PCR-Based Method for HLA Typing and Preimplantational Genetic Diagnosis of -Thalassemia. <i>BioMed Research International</i> , 2013, 2013, 1-9.	1.9	9
65	Male and female differential reproductive rate could explain parental transmission asymmetry of mutation origin in Hirschsprung disease. <i>European Journal of Human Genetics</i> , 2012, 20, 917-920.	2.8	8
66	Somatic and germline mosaicism of deletion 15q11.2-q13 in a mother of dizygotic twins with Angelman syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 370-376.	1.2	8
67	Interstitial 10p deletion derived from a maternal ins(16;10)(q22;p13p15.2): Report of the first familial case of 10p monosomy affecting to two familial members of different generations. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1268-1273.	1.2	8
68	Identification of different mechanisms leading to PAX6 down-regulation as potential events contributing to the onset of Hirschsprung disease. <i>Scientific Reports</i> , 2016, 6, 21160.	3.3	8
69	Expanding the clinical and mutational spectrum of germline ABL1 mutations-associated syndrome. <i>Medicine (United States)</i> , 2019, 98, e14782.	1.0	8
70	A novel study of Copy Number Variations in Hirschsprung disease using the Multiple Ligation-dependent Probe Amplification (MLPA) technique. <i>BMC Medical Genetics</i> , 2009, 10, 119.	2.1	7
71	Four new loci associations discovered by pathway-based and network analyses of the genome-wide variability profile of Hirschsprung's disease. <i>Orphanet Journal of Rare Diseases</i> , 2012, 7, 103.	2.7	7
72	A complementary study approach unravels novel players in the pathoetiology of Hirschsprung disease. <i>PLoS Genetics</i> , 2020, 16, e1009106.	3.5	7

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73	A comprehensive WGS-based pipeline for the identification of new candidate genes in inherited retinal dystrophies. <i>Npj Genomic Medicine</i> , 2022, 7, 17.	3.8	7
74	Novel MLPA procedure using self-designed probes allows comprehensive analysis for CNVs of the genes involved in Hirschsprung disease. <i>BMC Medical Genetics</i> , 2010, 11, 71.	2.1	6
75	<i>Dnmt3b</i> knock-down in enteric precursors reveals a possible mechanism by which this <i>de novo</i> methyltransferase is involved in the enteric nervous system development and the onset of Hirschsprung disease. <i>Oncotarget</i> , 2017, 8, 106443-106453.	1.8	6
76	A Multi-Strategy Sequencing Workflow in Inherited Retinal Dystrophies: Routine Diagnosis, Addressing Unsolved Cases and Candidate Genes Identification. <i>International Journal of Molecular Sciences</i> , 2020, 21, 9355.	4.1	5
77	Cys 634 mutations in the RET proto-oncogene in Spanish families affected by MEN 2A. <i>Human Mutation</i> , 1998, 11, S72-S73.	2.5	4
78	Clinical and Technical Overview of Preimplantation Genetic Diagnosis for Fragile X Syndrome: Experience at the University Hospital Virgen del Rocio in Spain. <i>BioMed Research International</i> , 2015, 2015, 1-6.	1.9	4
79	Preimplantation Genetic Diagnosis for Myotonic Dystrophy Type 1 and Analysis of the Effect of the Disease on the Reproductive Outcome of the Affected Female Patients. <i>BioMed Research International</i> , 2017, 2017, 1-7.	1.9	4
80	ChIP-Seq-Based Approach in Mouse Enteric Precursor Cells Reveals New Potential Genes with a Role in Enteric Nervous System Development and Hirschsprung Disease. <i>International Journal of Molecular Sciences</i> , 2020, 21, 9061.	4.1	4
81	Genome-wide analysis of DNA methylation in Hirschsprung enteric precursor cells: unraveling the epigenetic landscape of enteric nervous system development. <i>Clinical Epigenetics</i> , 2021, 13, 51.	4.1	4
82	Co-segregation of a homozygous <i>SMN1</i> deletion and a heterozygous <i>PMP22</i> duplication in a patient. <i>Clinical Case Reports (discontinued)</i> , 2016, 4, 879-884.	0.5	3
83	<i>ESR2</i> Gene and Medullary Thyroid Carcinoma. <i>Thyroid</i> , 2017, 27, 1456-1457.	4.5	3
84	Ultrasound, Echocardiography, MRI, and Genetic Analysis of a Fetus with Congenital Diaphragmatic Hernia and Partial 11q Trisomy. <i>Case Reports in Obstetrics and Gynecology</i> , 2017, 2017, 1-4.	0.3	3
85	Atypical Association of Angelman Syndrome and Klinefelter Syndrome in a Boy with 47,XXY Karyotype and Deletion 15q11.2-q13. <i>Case Reports in Genetics</i> , 2014, 2014, 1-4.	0.2	2
86	A Scoring System to Predict the Severity of Hirschsprung Disease at Diagnosis and Its Correlation With Molecular Genetics. <i>Pediatric and Developmental Pathology</i> , 2017, 20, 28-37.	1.0	2