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	A comprehensive WGS-based pipeline for the identification of new candidate genes in inherited retinal dystrophies. Npj Genomic Medicine, 2022, 7, 17.	3.8	7
2	CSVS, a crowdsourcing database of the Spanish population genetic variability. Nucleic Acids Research, 2021, 49, D1130-D1137.	14.5	34
3	RMRP, RMST, FTX and IPW: novel potential long non-coding RNAs in medullary thyroid cancer. Orphanet Journal of Rare Diseases, 2021, 16, 4.	2.7	10
4	Genome-wide analysis of DNA methylation in Hirschsprung enteric precursor cells: unraveling the epigenetic landscape of enteric nervous system development. Clinical Epigenetics, 2021, 13, 51.	4.1	4
5	What is new about the genetic background of Hirschsprung disease?. Clinical Genetics, 2020, 97, 114-124.	2.0	24
6	Identification of New Potential LncRNA Biomarkers in Hirschsprung Disease. International Journal of Molecular Sciences, 2020, 21, 5534.	4.1	10
7	ChIP-Seq-Based Approach in Mouse Enteric Precursor Cells Reveals New Potential Genes with a Role in Enteric Nervous System Development and Hirschsprung Disease. International Journal of Molecular Sciences, 2020, 21, 9061.	4.1	4
8	A Multi-Strategy Sequencing Workflow in Inherited Retinal Dystrophies: Routine Diagnosis, Addressing Unsolved Cases and Candidate Genes Identification. International Journal of Molecular Sciences, 2020, 21, 9355.	4.1	5
9	Unmasking Retinitis Pigmentosa complex cases by a whole genome sequencing algorithm based on open-access tools: hidden recessive inheritance and potential oligogenic variants. Journal of Translational Medicine, 2020, 18, 73.	4.4	23
10	A complementary study approach unravels novel players in the pathoetiology of Hirschsprung disease. PLoS Genetics, 2020, 16, e1009106.	3.5	7
11	Epigenetic Mechanisms in Hirschsprung Disease. International Journal of Molecular Sciences, 2019, 20, 3123.	4.1	23
12	Influencers on Thyroid Cancer Onset: Molecular Genetic Basis. Genes, 2019, 10, 913.	2.4	28
13	Expanding the clinical and mutational spectrum of germline ABL1 mutations-associated syndrome. Medicine (United States), 2019, 98, e14782.	1.0	8
14	Correlation between SMA type and SMN2 copy number revisited: An analysis of 625 unrelated Spanish patients and a compilation of 2834 reported cases. Neuromuscular Disorders, 2018, 28, 208-215.	0.6	273
15	Searching the second hit in patients with inherited retinal dystrophies and monoallelic variants in ABCA4, USH2A and CEP290 by whole-gene targeted sequencing. Scientific Reports, 2018, 8, 13312.	3.3	17
16	Whole exome sequencing coupled with unbiased functional analysis reveals new Hirschsprung disease genes. Genome Biology, 2017, 18, 48.	8.8	72
17	Unravelling the genetic basis of simplex Retinitis Pigmentosa cases. Scientific Reports, 2017, 7, 41937.	3.3	71
18	A Scoring System to Predict the Severity of Hirschsprung Disease at Diagnosis and Its Correlation With Molecular Genetics. Pediatric and Developmental Pathology, 2017, 20, 28-37.	1.0	2

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19	ESR2 Gene and Medullary Thyroid Carcinoma. Thyroid, 2017, 27, 1456-1457.	4.5	3
20	Overexpression of DNMT3b target genes during Enteric Nervous System development contribute to the onset of Hirschsprung disease. Scientific Reports, 2017, 7, 6221.	3.3	18
21	Multilayer OMIC Data in Medullary Thyroid Carcinoma Identifies the STAT3 Pathway as a Potential Therapeutic Target in RETM918T Tumors. Clinical Cancer Research, 2017, 23, 1334-1345.	7.0	34
22	Ultrasound, Echocardiography, MRI, and Genetic Analysis of a Fetus with Congenital Diaphragmatic Hernia and Partial 11q Trisomy. Case Reports in Obstetrics and Gynecology, 2017, 2017, 1-4.	0.3	3
23	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. BioMed Research International, 2017, 2017, 1-7.	1.9	4
24	Dnmt3b knock-down in enteric precursors reveals a possible mechanism by which this de novo methyltransferase is involved in the enteric nervous system development and the onset of Hirschsprung disease. Oncotarget, 2017, 8, 106443-106453.	1.8	6
25	The NER-related gene GTF2H5 predicts survival in high-grade serous ovarian cancer patients. Journal of Gynecologic Oncology, 2016, 27, e7.	2.2	30
26	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. American Journal of Medical Genetics, Part A, 2016, 170, 1268-1273.	1.2	8
27	Improving the management of Inherited Retinal Dystrophies by targeted sequencing of a population-specific gene panel. Scientific Reports, 2016, 6, 23910.	3.3	51
28	Identification of different mechanisms leading to PAX6 down-regulation as potential events contributing to the onset of Hirschsprung disease. Scientific Reports, 2016, 6, 21160.	3.3	8
29	Coâ€segregation of a homozygous <scp>SMN</scp>1 deletion and a heterozygous <scp>PMP</scp>22 duplication in a patient. Clinical Case Reports (discontinued), 2016, 4, 879-884.	0.5	3
30	Trans-ethnic meta-analysis of genome-wide association studies for Hirschsprung disease. Human Molecular Genetics, 2016, 25, ddw333.	2.9	38
31	G534E Variant in HABP2 and Nonmedullary Thyroid Cancer. Thyroid, 2016, 26, 987-988.	4.5	17
32	267 Spanish Exomes Reveal Population-Specific Differences in Disease-Related Genetic Variation. Molecular Biology and Evolution, 2016, 33, 1205-1218.	8.9	78
33	Exome sequencing reveals a high genetic heterogeneity on familial Hirschsprung disease. Scientific Reports, 2015, 5, 16473.	3.3	29
34	Next-generation-based targeted sequencing as an efficient tool for the study of the genetic background in Hirschsprung patients. BMC Medical Genetics, 2015, 16, 89.	2.1	14
35	Identification of epistatic interactions through genome-wide association studies in sporadic medullary and juvenile papillary thyroid carcinomas. BMC Medical Genomics, 2015, 8, 83.	1.5	15
36	Experience of Preimplantation Genetic Diagnosis for Hemophilia at the University Hospital Virgen Del RocÃo in Spain: Technical and Clinical Overview. BioMed Research International, 2015, 2015, 1-8.	1.9	10

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37	Clinical and Technical Overview of Preimplantation Genetic Diagnosis for Fragile X Syndrome: Experience at the University Hospital Virgen del Rocio in Spain. BioMed Research International, 2015, 2015, 1-6.	1.9	4
38	Functional Loss of Semaphorin 3C and/or Semaphorin 3D and Their Epistatic Interaction with Ret Are Critical to Hirschsprung Disease Liability. American Journal of Human Genetics, 2015, 96, 581-596.	6.2	118
39	Deletion at 6q24.2â€“26 predicts longer survival of highâ€grade serous epithelial ovarian cancer patients. Molecular Oncology, 2015, 9, 422-436.	4.6	17
40	Reâ€evaluation casts doubt on the pathogenicity of homozygous USH2A p.C759F. American Journal of Medical Genetics, Part A, 2015, 167, 1597-1600.	1.2	21
41	Exome Sequencing Reveals Novel and Recurrent Mutations with Clinical Significance in Inherited Retinal Dystrophies. PLoS ONE, 2014, 9, e116176.	2.5	16
42	Involvement of DNMT3B in the pathogenesis of Hirschsprung disease and its possible role as a regulator of neurogenesis in the human enteric nervous system. Genetics in Medicine, 2014, 16, 703-710.	2.4	29
43	Atypical Association of Angelman Syndrome and Klinefelter Syndrome in a Boy with 47,XXY Karyotype and Deletion 15q11.2-q13. Case Reports in Genetics, 2014, 2014, 1-4.	0.2	2
44	Experience of Preimplantation Genetic Diagnosis with HLA Matching at the University Hospital Virgen del RocÃo in Spain: Technical and Clinical Overview. BioMed Research International, 2014, 2014, 1-8.	1.9	18
45	An Impairment of Long Distance SOX10 Regulatory Elements Underlies Isolated Hirschsprung Disease. Human Mutation, 2014, 35, 303-307.	2.5	33
46	Deciphering intrafamilial phenotypic variability by exome sequencing in a Bardetâ€“Biedl family. Molecular Genetics & Genomic Medicine, 2014, 2, 124-133.	1.2	13
47	Novel RP1 mutations and a recurrent BBS1variant explain the co-existence of two distinct retinal phenotypes in the same pedigree. BMC Genetics, 2014, 15, 143.	2.7	18
48	Somatic and germâ€line mosaicism of deletion 15q11.2â€“q13 in a mother of dyzigotic twins with Angelman syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 370-376.	1.2	8
49	Waardenburg syndrome type 4: Report of two new cases caused by SOX 10 mutations in Spain. American Journal of Medical Genetics, Part A, 2014, 164, 542-547.	1.2	18
50	Contribution of rare and common variants determine complex diseasesâ€”Hirschsprung disease as a model. Developmental Biology, 2013, 382, 320-329.	2.0	119
51	Pathways systematically associated to Hirschsprungâ€™s disease. Orphanet Journal of Rare Diseases, 2013, 8, 187.	2.7	17
52	Novel One-Step Multiplex PCR-Based Method for HLA Typing and Preimplantational Genetic Diagnosis of -Thalassemia. BioMed Research International, 2013, 2013, 1-9.	1.9	9
53	Contributions of PHOX2B in the Pathogenesis of Hirschsprung Disease. PLoS ONE, 2013, 8, e54043.	2.5	30
54	Mutational Spectrum of Semaphorin 3A and Semaphorin 3D Genes in Spanish Hirschsprung patients. PLoS ONE, 2013, 8, e54800.	2.5	34

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55	Hirschsprung's disease as a model of complex genetic etiology. Histology and Histopathology, 2013, 28, 1117-36.	0.7	48
56	Whole-exome sequencing identifies novel compound heterozygous mutations in USH2A in Spanish patients with autosomal recessive retinitis pigmentosa. Molecular Vision, 2013, 19, 2187-95.	1.1	17
57	Male and female differential reproductive rate could explain parental transmission asymmetry of mutation origin in Hirschsprung disease. European Journal of Human Genetics, 2012, 20, 917-920.	2.8	8
58	Four new loci associations discovered by pathway-based and network analyses of the genome-wide variability profile of Hirschsprungâ€™s disease. Orphanet Journal of Rare Diseases, 2012, 7, 103.	2.7	7
59	A map of human microRNA variation uncovers unexpectedly high levels of variability. Genome Medicine, 2012, 4, 62.	8.2	28
60	Comprehensive Analysis of NRG1 Common and Rare Variants in Hirschsprung Patients. PLoS ONE, 2012, 7, e36524.	2.5	36
61	Association of Xâ€linked hydrocephalus and Hirschsprung disease: Report of a new patient with a mutation in the L1CAM gene. American Journal of Medical Genetics, Part A, 2012, 158A, 816-820.	1.2	14
62	Expression of PROKR1 and PROKR2 in Human Enteric Neural Precursor Cells and Identification of Sequence Variants Suggest a Role in HSCR. PLoS ONE, 2011, 6, e23475.	2.5	22
63	Mutation Screening of Multiple Genes in Spanish Patients with Autosomal Recessive Retinitis Pigmentosa by Targeted Resequencing. PLoS ONE, 2011, 6, e27894.	2.5	36
64	Novel mutations at RET ligand genes preventing receptor activation are associated to Hirschsprungâ€™s disease. Journal of Molecular Medicine, 2011, 89, 471-480.	3.9	35
65	Comprehensive analysis of RET common and rare variants in a series of Spanish Hirschsprung patients confirms a synergistic effect of both kinds of events. BMC Medical Genetics, 2011, 12, 138.	2.1	14
66	Copy-Number Variations inEYS:A Significant Event in the Appearance of arRP. , 2011, 52, 5625.		40
67	Differential Contributions of Rare and Common, Coding and Noncoding Ret Mutations to Multifactorial Hirschsprung Disease Liability. American Journal of Human Genetics, 2010, 87, 60-74.	6.2	230
68	Involvement of SOX10 in the pathogenesis of Hirschsprung disease: report of a truncating mutation in an isolated patient. Journal of Molecular Medicine, 2010, 88, 507-514.	3.9	40
69	Mutation spectrum of EYS in Spanish patients with autosomal recessive retinitis pigmentosa. Human Mutation, 2010, 31, E1772-E1800.	2.5	69
70	Novel association of severe neonatal encephalopathy and Hirschsprung disease in a male with a duplication at the Xq28 region. BMC Medical Genetics, 2010, 11, 137.	2.1	20
71	Novel MLPA procedure using self-designed probes allows comprehensive analysis for CNVs of the genes involved in Hirschsprung disease. BMC Medical Genetics, 2010, 11, 71.	2.1	6
72	New roles of EDNRB and EDN3 in the pathogenesis of Hirschsprung disease. Genetics in Medicine, 2010, 12, 39-43.	2.4	59

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73	One-step multiplex polymerase chain reaction for preimplantation genetic diagnosis of Huntington disease. Fertility and Sterility, 2010, 93, 2411-2412.	1.0	12
74	A novel study of Copy Number Variations in Hirschsprung disease using the Multiple Ligation-dependent Probe Amplification (MLPA) technique. BMC Medical Genetics, 2009, 10, 119.	2.1	7
75	EYS, encoding an ortholog of Drosophila spacemaker, is mutated in autosomal recessive retinitis pigmentosa. Nature Genetics, 2008, 40, 1285-1287.	21.4	175
76	NTF-3, a gene involved in the enteric nervous system development, as a candidate gene for Hirschsprung disease. Journal of Pediatric Surgery, 2008, 43, 1308-1311.	1.6	24
77	Association Study of 69 Genes in the Ret Pathway Identifies Low-penetrance Loci in Sporadic Medullary Thyroid Carcinoma. Cancer Research, 2007, 67, 9561-9567.	0.9	36
78	Analysis of RET Polymorphisms and Haplotypes in the Context of Sporadic Medullary Thyroid Carcinoma. Thyroid, 2006, 16, 411-417.	4.5	25
79	A complex additive model of inheritance for Hirschsprung disease is supported by both RET mutations and predisposing RET haplotypes. Genetics in Medicine, 2006, 8, 704-710.	2.4	29
80	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. International Journal of Molecular Medicine, 2006, 17, 575-81.	4.0	15
81	TheRETC620S mutation causes multiple endocrine neoplasia type 2A (MEN2A) but not Hirschsprung disease (HSCR) in a family cosegregating both phenotypes. Human Mutation, 2003, 22, 412-415.	2.5	9
82	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. American Journal of Human Genetics, 2003, 72, 88-100.	6.2	100
83	Prevalence of 2314delG mutation in Spanish patients with Usher syndrome type II (USH2). Ophthalmic Genetics, 2000, 21, 123-128.	1.2	16
84	Specific polymorphisms in the RETproto-oncogene are over-represented in patients with Hirschsprung disease and may represent loci modifying phenotypic expression. Journal of Medical Genetics, 1999, 36, 771-774.	3.2	142
85	Cys 634 mutations in the RET proto-oncogene in Spanish families affected by MEN 2A. Human Mutation, 1998, 11, S72-S73.	2.5	4
86	Molecular Analysis of theretandGDNFGenes in a Family with Multiple Endocrine Neoplasia Type 2A and Hirschsprung Disease1. Journal of Clinical Endocrinology and Metabolism, 1998, 83, 3361-3364.	3.6	61