Sergi Beltran

List of Publications by Year
in descending order

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84 papers	8,231 citations	126708 33 h-index	58464 82 g-index
92 all docs	92 docs citations	92 times ranked	20000 citing authors

#	Article	IF	Citations
1	Pan-cancer analysis of whole genomes. Nature, 2020, 578, 82-93.	13.7	1,966
2	Transcriptome and genome sequencing uncovers functional variation in humans. Nature, 2013, 501, 506-511.	13.7	1,857
3	Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. Nucleic Acids Research, 2019, 47, D1018-D1027.	6.5	539
4	The Matchmaker Exchange: A Platform for Rare Disease Gene Discovery. Human Mutation, 2015, 36, 915-921.	1.1	390
5	A comprehensive assessment of somatic mutation detection in cancer using whole-genome sequencing. Nature Communications, 2015, 6, 10001.	5.8	266
6	Effect of predicted protein-truncating genetic variants on the human transcriptome. Science, 2015, 348, 666-669.	6.0	252
7	Reproducibility of high-throughput mRNA and small RNA sequencing across laboratories. Nature Biotechnology, 2013, 31, 1015-1022.	9.4	251
8	Recurrent inactivation of STAG2 in bladder cancer is not associated with aneuploidy. Nature Genetics, 2013, 45, 1464-1469.	9.4	224
9	Runs of homozygosity reveal signatures of positive selection for reproduction traits in breed and non-breed horses. BMC Genomics, 2015, 16, 764.	1.2	125
10	Whole-exome sequencing in splenic marginal zone lymphoma reveals mutations in genes involved in marginal zone differentiation. Leukemia, 2014, 28, 1334-1340.	3.3	115
11	Transcriptomics of In Vitro Immune-Stimulated Hemocytes from the Manila Clam Ruditapes philippinarum Using High-Throughput Sequencing. PLoS ONE, 2012, 7, e35009.	1.1	106
12	Genome-wide analysis reveals that Smad3 and JMJD3 HDM co-activate the neural developmental program. Development (Cambridge), 2012, 139, 2681-2691.	1.2	100
13	High-Throughput Sequence Analysis of Turbot (Scophthalmus maximus) Transcriptome Using 454-Pyrosequencing for the Discovery of Antiviral Immune Genes. PLoS ONE, 2012, 7, e35369.	1.1	100
14	GA4GH: International policies and standards for data sharing across genomic research and healthcare. Cell Genomics, 2021, 1, 100029.	3.0	94
15	Whole-exome sequencing identifies rare pathogenic variants in new predisposition genes for familial colorectal cancer. Genetics in Medicine, 2015, 17, 131-142.	1.1	82
16	Heterozygous STUB1 mutation causes familial ataxia with cognitive affective syndrome (SCA48). Neurology, 2018, 91, e1988-e1998.	1.5	81
17	Shared Oncogenic Pathways Implicated in Both Virus-Positive and UV-Induced Merkel Cell Carcinomas. Journal of Investigative Dermatology, 2017, 137, 197-206.	0.3	78
18	CCND2 and CCND3 hijack immunoglobulin light-chain enhancers in cyclin D1â^' mantle cell lymphoma. Blood, 2019, 133, 940-951.	0.6	77

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19	Two Novel Mutations in theBCKDK(Branched-Chain Keto-Acid Dehydrogenase Kinase) Gene Are Responsible for a Neurobehavioral Deficit in Two Pediatric Unrelated Patients. Human Mutation, 2014, 35, 470-477.	1.1	70
20	Leveraging European infrastructures to access 1 million human genomes by 2022. Nature Reviews Genetics, 2019, 20, 693-701.	7.7	69
21	Deep RNA Sequencing of the Skeletal Muscle Transcriptome in Swimming Fish. PLoS ONE, 2013, 8, e53171.	1.1	62
22	The Fanconi anemia DNA damage repair pathway in the spotlight for germline predisposition to colorectal cancer. European Journal of Human Genetics, 2016, 24, 1501-1505.	1.4	59
23	Thermal evolution of gene expression profiles in Drosophila subobscura. BMC Evolutionary Biology, 2007, 7, 42.	3.2	58
24	Gene expression following induction of regeneration in Drosophila wing imaginal discs. Expression profile of regenerating wing discs. BMC Developmental Biology, 2010, 10, 94.	2.1	56
25	Exome Sequencing Reveals AMER1 as a Frequently Mutated Gene in Colorectal Cancer. Clinical Cancer Research, 2015, 21, 4709-4718.	3.2	52
26	Splenic diffuse red pulp small B-cell lymphoma displays increased expression of cyclin D3 and recurrent CCND3 mutations. Blood, 2017, 129, 1042-1045.	0.6	52
27	Solve-RD: systematic pan-European data sharing and collaborative analysis to solve rare diseases. European Journal of Human Genetics, 2021, 29, 1325-1331.	1.4	49
28	RNA polymerase II progression through H3K27me3-enriched gene bodies requires JMJD3 histone demethylase. Molecular Biology of the Cell, 2013, 24, 351-360.	0.9	48
29	Recommendations for whole genome sequencing in diagnostics for rare diseases. European Journal of Human Genetics, 2022, 30, 1017-1021.	1.4	48
30	From Wetâ€Lab to Variations: Concordance and Speed of Bioinformatics Pipelines for Whole Genome and Whole Exome Sequencing. Human Mutation, 2016, 37, 1263-1271.	1.1	47
31	The GA4GH Phenopacket schema defines a computable representation of clinical data. Nature Biotechnology, 2022, 40, 817-820.	9.4	38
32	Transcriptomic Characterization of the Larval Stage in Gilthead Seabream (Sparus aurata) by 454 Pyrosequencing. Marine Biotechnology, 2012, 14, 423-435.	1.1	37
33	The ethylene receptors CpETR1A and CpETR2B cooperate in the control of sex determination in Cucurbita pepo. Journal of Experimental Botany, 2020, 71, 154-167.	2.4	37
34	Genetic lesions in MYC and STAT3 drive oncogenic transcription factor overexpression in plasmablastic lymphoma. Haematologica, 2021, 106, 1120-1128.	1.7	37
35	Benchmarking of Whole Exome Sequencing and Ad Hoc Designed Panels for Genetic Testing of Hereditary Cancer. Scientific Reports, 2017, 7, 37984.	1.6	35
36	Mutations inTRAPPC11are associated with a congenital disorder of glycosylation. Human Mutation, 2017, 38, 148-151.	1.1	34

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37	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. European Journal of Human Genetics, 2021, 29, 1337-1347.	1.4	34
38	New genes emerging for colorectal cancer predisposition. World Journal of Gastroenterology, 2014, 20, 1961.	1.4	34
39	Functional dissection of the ash2 and ash1 transcriptomes provides insights into the transcriptional basis of wing phenotypes and reveals conserved protein interactions. Genome Biology, 2007, 8, R67.	13.9	31
40	Tandem RNA Chimeras Contribute to Transcriptome Diversity in Human Population and Are Associated with Intronic Genetic Variants. PLoS ONE, 2014, 9, e104567.	1.1	31
41	A call for global action for rare diseases in Africa. Nature Genetics, 2020, 52, 21-26.	9.4	31
42	Mutations in Vps15 perturb neuronal migration in mice and are associated with neurodevelopmental disease in humans. Nature Neuroscience, 2018, 21, 207-217.	7.1	30
43	Recessive variants of MuSK are associated with late onset CMS and predominant limb girdle weakness. American Journal of Medical Genetics, Part A, 2018, 176, 1594-1601.	0.7	25
44	Next generation sequencing gives an insight into the characteristics of highly selected breeds versus non-breed horses in the course of domestication. BMC Genomics, 2014, 15, 562.	1.2	24
45	Phenomic and Genomic Characterization of a Mutant Platform in Cucurbita pepo. Frontiers in Plant Science, 2018, 9, 1049.	1.7	23
46	Increasing phenotypic annotation improves the diagnostic rate of exome sequencing in a rare neuromuscular disorder. Human Mutation, 2019, 40, 1797-1812.	1.1	22
47	Transcriptional network controlled by the trithorax-group gene ash2 in Drosophila melanogaster. Proceedings of the National Academy of Sciences of the United States of America, 2003, 100, 3293-3298.	3.3	21
48	A guide to writing systematic reviews of rare disease treatments to generate FAIR-compliant datasets: building a Treatabolome. Orphanet Journal of Rare Diseases, 2020, 15, 206.	1.2	21
49	ELIXIRâ€EXCELERATE: establishing Europe's data infrastructure for the life science research of the future. EMBO Journal, 2021, 40, e107409.	3.5	18
50	The RDâ€Connect Genomeâ€Phenome Analysis Platform: Accelerating diagnosis, research, and gene discovery for rare diseases. Human Mutation, 2022, , .	1.1	18
51	Somatic Embryonic FGFR2 Mutations inÂKeratinocytic Epidermal Nevi. Journal of Investigative Dermatology, 2016, 136, 1718-1721.	0.3	17
52	Chromosome 12p Amplification in Triple-Negative/BRCA1-Mutated Breast Cancer Associates with Emergence of Docetaxel Resistance and Carboplatin Sensitivity. Cancer Research, 2019, 79, 4258-4270.	0.4	17
53	Conserved chromosomal clustering of genes governed by chromatin regulators in Drosophila. Genome Biology, 2008, 9, R134.	13.9	16
54	Mutations in TIMM50 cause severe mitochondrial dysfunction by targeting key aspects of mitochondrial physiology. Human Mutation, 2019, 40, 1700-1712.	1.1	16

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55	Integrated Analysis of Germline and Tumor DNA Identifies New Candidate Genes Involved in Familial Colorectal Cancer. Cancers, 2019, 11, 362.	1.7	16
56	Improved Diagnosis of Rare Disease Patients through Systematic Detection of Runs of Homozygosity. Journal of Molecular Diagnostics, 2020, 22, 1205-1215.	1.2	14
57	Diagnosis of Genetic White Matter Disorders by Singleton Whole-Exome and Genome Sequencing Using Interactome-Driven Prioritization. Neurology, 2022, , 10.1212/WNL.0000000000013278.	1.5	13
58	Colorectal Adenomas Contain Multiple Somatic Mutations That Do Not Coincide with Synchronous Adenocarcinoma Specimens. PLoS ONE, 2015, 10, e0119946.	1.1	11
59	Rare germline copy number variants in colorectal cancer predisposition characterized by exome sequencing analysis. Journal of Genetics and Genomics, 2018, 45, 41-45.	1.7	11
60	A single nucleotide deletion resulting in a frameshift in exon 4 of TAB2 is associated with a polyvalular syndrome. European Journal of Medical Genetics, 2020, 63, 103854.	0.7	10
61	Drosophila melanogaster SAP18 protein is required for environmental stress responses. FEBS Letters, 2011, 585, 275-280.	1.3	9
62	COL4A1 -related autosomal recessive encephalopathy in 2 Turkish children. Neurology: Genetics, 2020, 6, e392.	0.9	9
63	Successful treatment of intractable epilepsy with ketogenic diet therapy in twins with ALG3-CDG. Brain and Development, 2020, 42, 539-545.	0.6	9
64	Growth phaseâ€dependent control of R27 conjugation is mediated by the interplay between the plasmidâ€encoded regulatory circuit TrhR/TrhYâ€HtdA and the cAMP regulon. Environmental Microbiology, 2016, 18, 5277-5287.	1.8	8
65	Clonal dynamics monitoring during clinical evolution in chronic lymphocytic leukaemia. Scientific Reports, 2019, 9, 975.	1.6	8
66	Behr syndrome and hypertrophic cardiomyopathy in a family with a novel UCHL1 deletion. Journal of Neurology, 2020, 267, 3643-3649.	1.8	8
67	Solving the unsolved rare diseases in Europe. European Journal of Human Genetics, 2021, 29, 1319-1320.	1.4	8
68	Whole exome sequencing as a diagnostic tool for patients with ciliopathy-like phenotypes. PLoS ONE, 2017, 12, e0183081.	1.1	8
69	Diagnostic value of bone marrow core biopsy patterns in lymphoplasmacytic lymphoma/WaldenstrÃ¶m macroglobulinaemia and description of its mutational profiles by targeted NGS. Journal of Clinical Pathology, 2020, 73, 571-577.	1.0	7
70	Whole exome sequencing analysis reveals TRPV3 as a risk factor for cardioembolic stroke/subtitle. Thrombosis and Haemostasis, 2016, 116, 1165-1771.	1.8	6
71	Autosomal recessive variants in TUBGCP2 alter the Î³-tubulin ring complex leading to neurodevelopmental disease. IScience, 2021, 24, 101948.	1.9	6
72	Systematic Collaborative Reanalysis of Genomic Data Improves Diagnostic Yield in Neurologic Rare Diseases. Journal of Molecular Diagnostics, 2022, 24, 529-542.	1.2	6

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73	Identification of protein-damaging mutations in 10 swine taste receptors and 191 appetite-reward genes. BMC Genomics, 2016, 17, 685.	1.2	5
74	Framework for quality assessment of whole genome cancer sequences. Nature Communications, 2020, 11, 5040.	5.8	5
75	Fine-scale population structure in five rural populations from the Spanish Eastern Pyrenees using high-coverage whole-genome sequence data. European Journal of Human Genetics, 2021, 29, 1557-1565.	1.4	4
76	Solving unsolved rare neurological diseasesâ€"a Solve-RD viewpoint. European Journal of Human Genetics, 2021, 29, 1332-1336.	1.4	4
77	Exome sequencing identifies mutations in three cases diagnosed with Retinitis Pigmentosa and hearing impairment. Molecular Vision, 2020, 26, 216-225.	1.1	2
78	Characterization of Subclonal Changes Along Progression in Multiple Myeloma.. Blood, 2012, 120, 2924-2924.	0.6	1
79	438: Comprehensive mutational landscape of human stable colorectal tumors in stage II. European Journal of Cancer, 2014, 50, S105-S106.	1.3	0
80	Genomic characterization of mutant laboratory mouse strains by exome sequencing and annotation lift-over. BMC Genomics, 2015, 16, 351.	1.2	0
81	RD-Connect: Data sharing and analysis for rare disease research within the integrated platform and through GA4GH beacon and matchmaker exchange. Neuromuscular Disorders, 2016, 26, S160-S161.	0.3	0
82	MITOCHONDRIAL DISEASES (Posters). Neuromuscular Disorders, 2018, 28, S85-S86.	0.3	0
83	Mutational Status of Splenic Marginal Zone Lymphoma Revealed by Whole Exome Sequencing.. Blood, 2012, 120, 2698-2698.	0.6	0
84	Mutational Status of Splenic Diffuse Red Pulp Small B-Cell Lymphoma Revealed By Whole Exome Sequencing. Blood, 2015, 126, 1448-1448.	0.6	0