Sergi Beltran

List of Publications by Year in Descending Order

78 papers	5,088 citations	30 h-index	71 g-index
92 ext. papers	6,977 ext. citations	9 avg, IF	7.07 L-index

#	Paper	IF	Citations
78	Diagnosis of Genetic White Matter Disorders by Singleton Whole-Exome and Genome Sequencing Using Interactome-Driven Prioritization.. *Neurology*, **2022**,	6.5	2
77	Systematic Collaborative Reanalysis of Genomic Data Improves Diagnostic Yield in Neurologic Rare Diseases.. *Journal of Molecular Diagnostics*, **2022**, 24, 529-542	5.1	1
76	GA4GH: International policies and standards for data sharing across genomic research and healthcare.. *Cell Genomics*, **2021**, 1, 100029-100029		20
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	5.3	0
74	Solving unsolved rare neurological diseases-a Solve-RD viewpoint. *European Journal of Human Genetics*, **2021**, 29, 1332-1336	5.3	0
73	Solve-RD: systematic pan-European data sharing and collaborative analysis to solve rare diseases. *European Journal of Human Genetics*, **2021**, 29, 1325-1331	5.3	10
72	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. *European Journal of Human Genetics*, **2021**, 29, 1337-1347	5.3	4
71	Genetic lesions in MYC and STAT3 drive oncogenic transcription factor overexpression in plasmablastic lymphoma. *Haematologica*, **2021**, 106, 1120-1128	6.6	12
70	ELIXIR-EXCELERATE: establishing Europe's data infrastructure for the life science research of the future. *EMBO Journal*, **2021**, 40, e107409	13	11
69	Autosomal recessive variants in alter the tubulin ring complex leading to neurodevelopmental disease. *IScience*, **2021**, 24, 101948	6.1	1
68	Successful treatment of intractable epilepsy with ketogenic diet therapy in twins with ALG3-CDG. *Brain and Development*, **2020**, 42, 539-545	2.2	6
67	Improved Diagnosis of Rare Disease Patients through Systematic Detection of Runs of Homozygosity. *Journal of Molecular Diagnostics*, **2020**, 22, 1205-1215	5.1	8
66	Diagnostic value of bone marrow core biopsy patterns in lymphoplasmacytic lymphoma/Waldenström macroglobulinaemia and description of its mutational profiles by targeted NGS. *Journal of Clinical Pathology*, **2020**, 73, 571-577	3.9	1
65	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	2.6	5
64	Pan-cancer analysis of whole genomes. *Nature*, **2020**, 578, 82-93	50.4	840
63	Exome sequencing identifies mutations in three cases diagnosed with Retinitis Pigmentosa and hearing impairment. *Molecular Vision*, **2020**, 26, 216-225	2.3	2
62	A call for global action for rare diseases in Africa. *Nature Genetics*, **2020**, 52, 21-26	36.3	11

61	Framework for quality assessment of whole genome cancer sequences. *Nature Communications*, **2020**, 11, 5040	17.4	1
60	Behr syndrome and hypertrophic cardiomyopathy in a family with a novel UCHL1 deletion. *Journal of Neurology*, **2020**, 267, 3643-3649	5.5	3
59	-related autosomal recessive encephalopathy in 2 Turkish children. *Neurology: Genetics*, **2020**, 6, e392	3.8	4
58	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	4.2	8
57	The ethylene receptors CpETR1A and CpETR2B cooperate in the control of sex determination in Cucurbita pepo. *Journal of Experimental Botany*, **2020**, 71, 154-167	7	15
56	Leveraging European infrastructures to access 1 million human genomes by 2022. *Nature Reviews Genetics*, **2019**, 20, 693-701	30.1	36
55	Clonal dynamics monitoring during clinical evolution in chronic lymphocytic leukaemia. *Scientific Reports*, **2019**, 9, 975	4.9	4
54	Chromosome 12p Amplification in Triple-Negative/Mutated Breast Cancer Associates with Emergence of Docetaxel Resistance and Carboplatin Sensitivity. *Cancer Research*, **2019**, 79, 4258-4270	10.1	6
53	Increasing phenotypic annotation improves the diagnostic rate of exome sequencing in a rare neuromuscular disorder. *Human Mutation*, **2019**, 40, 1797-1812	4.7	9
52	Mutations in TIMM50 cause severe mitochondrial dysfunction by targeting key aspects of mitochondrial physiology. *Human Mutation*, **2019**, 40, 1700-1712	4.7	12
51	Integrated Analysis of Germline and Tumor DNA Identifies New Candidate Genes Involved in Familial Colorectal Cancer. *Cancers*, **2019**, 11,	6.6	8
50	and hijack immunoglobulin light-chain enhancers in cyclin D1 mantle cell lymphoma. *Blood*, **2019**, 133, 940-951	2.2	48
49	Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. *Nucleic Acids Research*, **2019**, 47, D1018-D1027	20.1	333
48	Mutations in Vps15 perturb neuronal migration in mice and are associated with neurodevelopmental disease in humans. *Nature Neuroscience*, **2018**, 21, 207-217	25.5	24
47	Rare germline copy number variants in colorectal cancer predisposition characterized by exome sequencing analysis. *Journal of Genetics and Genomics*, **2018**, 45, 41-45	4	11
46	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	2.5	17
45	Phenomic and Genomic Characterization of a Mutant Platform in. *Frontiers in Plant Science*, **2018**, 9, 1049	6.2	11
44	Heterozygous mutation causes familial ataxia with cognitive affective syndrome (SCA48). *Neurology*, **2018**, 91, e1988-e1998	6.5	59

43	Splenic diffuse red pulp small B-cell lymphoma displays increased expression of cyclin D3 and recurrent CCND3 mutations. *Blood*, **2017**, 129, 1042-1045	2.2	31
42	Benchmarking of Whole Exome Sequencing and Ad Hoc Designed Panels for Genetic Testing of Hereditary Cancer. *Scientific Reports*, **2017**, 7, 37984	4.9	31
41	Mutations in TRAPPC11 are associated with a congenital disorder of glycosylation. *Human Mutation* , **2017**, 38, 148-151	4.7	26
40	Shared Oncogenic Pathways Implicated in Both Virus-Positive and UV-Induced Merkel Cell Carcinomas. *Journal of Investigative Dermatology*, **2017**, 137, 197-206	4.3	55
39	Whole exome sequencing as a diagnostic tool for patients with ciliopathy-like phenotypes. *PLoS ONE*, **2017**, 12, e0183081	3.7	4
38	Growth phase-dependent control of R27 conjugation is mediated by the interplay between the plasmid-encoded regulatory circuit TrhR/TrhY-HtdA and the cAMP regulon. *Environmental Microbiology*, **2016**, 18, 5277-5287	5.2	5
37	Identification of protein-damaging mutations in 10 swine taste receptors and 191 appetite-reward genes. *BMC Genomics*, **2016**, 17, 685	4.5	4
36	Whole exome sequencing analysis reveals TRPV3 as a risk factor for cardioembolic stroke. *Thrombosis and Haemostasis*, **2016**, 116, 1165-1171	7	5
35	Somatic Embryonic FGFR2 Mutations in Keratinocytic Epidermal Nevi. *Journal of Investigative Dermatology*, **2016**, 136, 1718-1721	4.3	15
34	The Fanconi anemia DNA damage repair pathway in the spotlight for germline predisposition to colorectal cancer. *European Journal of Human Genetics*, **2016**, 24, 1501-5	5.3	35
33	From Wet-Lab to Variations: Concordance and Speed of Bioinformatics Pipelines for Whole Genome and Whole Exome Sequencing. *Human Mutation*, **2016**, 37, 1263-1271	4.7	33
32	Exome Sequencing Reveals AMER1 as a Frequently Mutated Gene in Colorectal Cancer. *Clinical Cancer Research*, **2015**, 21, 4709-18	12.9	35
31	Human genomics. Effect of predicted protein-truncating genetic variants on the human transcriptome. *Science*, **2015**, 348, 666-9	33.3	170
30	The Matchmaker Exchange: a platform for rare disease gene discovery. *Human Mutation*, **2015**, 36, 915-21	4.7	280
29	Whole-exome sequencing identifies rare pathogenic variants in new predisposition genes for familial colorectal cancer. *Genetics in Medicine*, **2015**, 17, 131-42	8.1	71
28	Runs of homozygosity reveal signatures of positive selection for reproduction traits in breed and non-breed horses. *BMC Genomics*, **2015**, 16, 764	4.5	77
27	Colorectal adenomas contain multiple somatic mutations that do not coincide with synchronous adenocarcinoma specimens. *PLoS ONE*, **2015**, 10, e0119946	3.7	7
26	Genomic characterization of mutant laboratory mouse strains by exome sequencing and annotation lift-over. *BMC Genomics*, **2015**, 16, 351	4.5	

25	A comprehensive assessment of somatic mutation detection in cancer using whole-genome sequencing. *Nature Communications*, **2015**, 6, 10001	17.4	199
24	Mutational Status of Splenic Diffuse Red Pulp Small B-Cell Lymphoma Revealed By Whole Exome Sequencing. *Blood*, **2015**, 126, 1448-1448	2.2	
23	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	4.5	18
22	Whole-exome sequencing in splenic marginal zone lymphoma reveals mutations in genes involved in marginal zone differentiation. *Leukemia*, **2014**, 28, 1334-40	10.7	90
21	Tandem RNA chimeras contribute to transcriptome diversity in human population and are associated with intronic genetic variants. *PLoS ONE*, **2014**, 9, e104567	3.7	27
20	Two novel mutations in the BCKDK (branched-chain keto-acid dehydrogenase kinase) gene are responsible for a neurobehavioral deficit in two pediatric unrelated patients. *Human Mutation*, **2014**, 35, 470-7	4.7	49
19	New genes emerging for colorectal cancer predisposition. *World Journal of Gastroenterology*, **2014**, 20, 1961-71	5.6	31
18	Recurrent inactivation of STAG2 in bladder cancer is not associated with aneuploidy. *Nature Genetics*, **2013**, 45, 1464-9	36.3	186
17	Reproducibility of high-throughput mRNA and small RNA sequencing across laboratories. *Nature Biotechnology*, **2013**, 31, 1015-22	44.5	187
16	Transcriptome and genome sequencing uncovers functional variation in humans. *Nature*, **2013**, 501, 506-11	50.4	1323
15	RNA polymerase II progression through H3K27me3-enriched gene bodies requires JMJD3 histone demethylase. *Molecular Biology of the Cell*, **2013**, 24, 351-60	3.5	36
14	Deep RNA sequencing of the skeletal muscle transcriptome in swimming fish. *PLoS ONE*, **2013**, 8, e53171	3.7	48
13	Genome-wide analysis reveals that Smad3 and JMJD3 HDM co-activate the neural developmental program. *Development (Cambridge)*, **2012**, 139, 2681-91	6.6	77
12	Transcriptomics of in vitro immune-stimulated hemocytes from the Manila clam Ruditapes philippinarum using high-throughput sequencing. *PLoS ONE*, **2012**, 7, e35009	3.7	88
11	High-throughput sequence analysis of turbot (Scophthalmus maximus) transcriptome using 454-pyrosequencing for the discovery of antiviral immune genes. *PLoS ONE*, **2012**, 7, e35369	3.7	90
10	Transcriptomic characterization of the larval stage in gilthead seabream (Sparus aurata) by 454 pyrosequencing. *Marine Biotechnology*, **2012**, 14, 423-35	3.4	33
9	Mutational Status of Splenic Marginal Zone Lymphoma Revealed by Whole Exome Sequencing.. *Blood*, **2012**, 120, 2698-2698	2.2	
8	Characterization of Subclonal Changes Along Progression in Multiple Myeloma.. *Blood*, **2012**, 120, 2924-2924	22	0

7	Drosophila melanogaster SAP18 protein is required for environmental stress responses. *FEBS Letters*, **2011**, 585, 275-80	3.8	6
6	Gene expression following induction of regeneration in Drosophila wing imaginal discs. Expression profile of regenerating wing discs. *BMC Developmental Biology*, **2010**, 10, 94	3.1	42
5	Conserved chromosomal clustering of genes governed by chromatin regulators in Drosophila. *Genome Biology*, **2008**, 9, R134	18.3	15
4	Thermal evolution of gene expression profiles in Drosophila subobscura. *BMC Evolutionary Biology*, **2007**, 7, 42	3	50
3	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	18.3	29
2	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, 3293-8	11.5	19
1	Framework for quality assessment of whole genome, cancer sequences		6