Christophe Beroud

List of Publications by Year
in descending order

130	12,751	22153	24258
papers	citations	59 h-index	110 g-index

138	138	138	19168
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Diagnostic interest of whole-body MRI in early- and late-onset LAMA2 muscular dystrophies: a large international cohort. Journal of Neurology, 2022, 269, 2414-2429.	3.6	5
2	The RDâ€Connect Genomeâ€Phenome Analysis Platform: Accelerating diagnosis, research, and gene discovery for rare diseases. Human Mutation, 2022, , .	2.5	18
3	Convergence of patient- and physician-reported outcomes in the French National Registry of Facioscapulohumeral Dystrophy. Orphanet Journal of Rare Diseases, 2022, 17, 96.	2.7	2
4	Highlighting the Dystonic Phenotype Related to <scp>GNAO1</scp>. Movement Disorders, 2022, 37, 1547-1554.	3.9	25
5	The lncRNA 44s2 Study Applicability to the Design of 45-55 Exon Skipping Therapeutic Strategy for DMD. Biomedicines, 2021, 9, 219.	3.2	4
6	Whole-body muscle MRI characteristics of LAMA2-related congenital muscular dystrophy children: An emerging pattern. Neuromuscular Disorders, 2021, 31, 814-823.	0.6	4
7	Standardisation of pathogenicity classification for somatic alterations in solid tumours and haematologic malignancies. European Journal of Cancer, 2021, 159, 1-15.	2.8	7
8	X-linked muscular dystrophy in a Labrador Retriever strain: phenotypic and molecular characterisation. Skeletal Muscle, 2020, 10, 23.	4.2	12
9	Pathogenic FBN1 Genetic Variation and Aortic Dissection in Patients With MarfanÂSyndrome. Journal of the American College of Cardiology, 2020, 75, 843-853.	2.8	38
10	The ELIXIR Human Copy Number Variations Community: building bioinformatics infrastructure for research. F1000Research, 2020, 9, 1229.	1.6	5
11	UMD-MEN1 Database: An Overview of the 370 MEN1 Variants Present in 1676 Patients From the French Population. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 753-764.	3.6	39
12	Whole Exome Sequencing Reveals a Large Genetic Heterogeneity and Revisits the Causes of Hypertrophic Cardiomyopathy. Circulation Genomic and Precision Medicine, 2019, 12, e002500.	3.6	9
13	Clinical Significance of Aortic Root Modification Associated With Bicuspid Aortic Valve in Marfan Syndrome. Circulation: Cardiovascular Imaging, 2019, 12, e008129.	2.6	15
14	Proposition of adjustments to the ACMGâ€AMP framework for the interpretation of MEN1 missense variants. Human Mutation, 2019, 40, 661-674.	2.5	21
15	Single-cell genetic analysis validates cytopathological identification of circulating cancer cells in patients with clear cell renal cell carcinoma. Oncotarget, 2018, 9, 20058-20074.	1.8	20
16	The French National Registry of patients with Facioscapulohumeral muscular dystrophy. Orphanet Journal of Rare Diseases, 2018, 13, 218.	2.7	11
17	Muscular MRI-based algorithm to differentiate inherited myopathies presenting with spinal rigidity. European Radiology, 2018, 28, 5293-5303.	4.5	28
18	Loss of Calmodulin- and Radial-Spoke-Associated Complex Protein CFAP251 Leads to Immotile Spermatozoa Lacking Mitochondria and Infertility in Men. American Journal of Human Genetics, 2018, 103, 413-420.	6.2	74

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19	VarAFT: a variant annotation and filtration system for human next generation sequencing data. Nucleic Acids Research, 2018, 46, W545-W553.	14.5	136
20	Clinical Outcomes in Duchenne Muscular Dystrophy: A Study of 5345 Patients from the TREAT-NMD DMD Global Database. Journal of Neuromuscular Diseases, 2017, 4, 293-306.	2.6	125
21	Exome Sequencing Identifies Two Variants of the Alkylglycerol Monooxygenase Gene as a Cause of Relapses in Visceral Leishmaniasis in Children, in Sudan. Journal of Infectious Diseases, 2017, 216, 22-28.	4.0	11
22	New advances in DPYD genotype and risk of severe toxicity under capecitabine. PLoS ONE, 2017, 12, e0175998.	2.5	82
23	Simple Sequence Mutations. , 2017, , 217-230.		0
24	UMDâ€Predictor: A Highâ€Throughput Sequencing Compliant System for Pathogenicity Prediction of any Human cDNA Substitution. Human Mutation, 2016, 37, 439-446.	2.5	104
25	Coverage analysis of lists of genes involved in heterogeneous genetic diseases following benchtop exome sequencing using the ion proton. Journal of Genetics, 2016, 95, 203-208.	0.7	3
26	How to Identify Pathogenic Mutations among All Those Variations: Variant Annotation and Filtration in the Genome Sequencing Era. Human Mutation, 2016, 37, 1272-1282.	2.5	28
27	Novel heterozygous mutation in ANO3 responsible for craniocervical dystonia. Movement Disorders, 2016, 31, 1251-1252.	3.9	24
28	BRCA Share: A Collection of Clinical BRCA Gene Variants. Human Mutation, 2016, 37, 1318-1328.	2.5	57
29	Actionable Genes, Core Databases, and Locus-Specific Databases. Human Mutation, 2016, 37, 1299-1307.	2.5	6
30	WES/WGS Reporting of Mutations from Cardiovascular â€œActionableâ€•Genes in Clinical Practice: A Key Role for UMD Knowledgebases in the Era of Big Databases. Human Mutation, 2016, 37, 1308-1317.	2.5	5
31	High-Throughput Sequencing in the Context of Human Genetic Diseases: Now and Tomorrow. Human Mutation, 2016, 37, 1247-1247.	2.5	0
32	Consideration surrounding incidental findings throughout multigene panel testing in cancer genetics. Clinical Genetics, 2016, 89, 267-268.	2.0	1
33	Improving molecular diagnosis of distal myopathies by targeted next-generation sequencing: TableÂ1. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 340-342.	1.9	20
34	A mutation in the Gardos channel is associated with hereditary xerocytosis. Blood, 2015, 126, 1273-1280.	1.4	97
35	Laminin Î±2 Deficiency-Related Muscular Dystrophy Mimicking Emery-Dreifuss andÂCollagen VI related Diseases. Journal of Neuromuscular Diseases, 2015, 2, 229-240.	2.6	30
36	Comparing targeted exome and whole exome approaches for genetic diagnosis of neuromuscular disorders. Applied & Translational Genomics, 2015, 7, 26-31.	2.1	18

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37	High Prevalence of PRPH2 in Autosomal Dominant Retinitis Pigmentosa in France and Characterization of Biochemical and Clinical Features. American Journal of Ophthalmology, 2015, 159, 302-314.	3.3	29
38	Rare inherited disorders with renal involvementâ€"approach to the patient. Kidney International, 2015, 87, 901-908.	5.2	18
39	The TREAT-NMD DMD Global Database: Analysis of More than 7,000 Duchenne Muscular Dystrophy Mutations. Human Mutation, 2015, 36, 395-402.	2.5	507
40	Genomic variations integrated database forMUTYH-associated adenomatous polyposis. Journal of Medical Genetics, 2015, 52, 25-27.	3.2	2
41	Identification of Splicing Defects Caused by Mutations in the Dysferlin Gene. Human Mutation, 2014, 35, 1532-1541.	2.5	22
42	Clinical heterogeneity and a high proportion of novel mutations in a Chinese cohort of patients with dysferlinopathy. Neurology India, 2014, 62, 635.	0.4	27
43	The UMD-APC Database, a Model of Nation-Wide Knowledge Base: Update with Data from 3,581 Variations. Human Mutation, 2014, 35, 532-536.	2.5	15
44	Mapping the differences in care for 5,000 Spinal Muscular Atrophy patients, a survey of 24 national registries in North America, Australasia and Europe. Journal of Neurology, 2014, 261, 152-163.	3.6	76
45	RD-Connect: An Integrated Platform Connecting Databases, Registries, Biobanks and Clinical Bioinformatics for Rare Disease Research. Journal of General Internal Medicine, 2014, 29, 780-787.	2.6	159
46	Diagnostic approach to the congenital muscular dystrophies. Neuromuscular Disorders, 2014, 24, 289-311.	0.6	275
47	Dispelling myths about rare disease registry system development. Source Code for Biology and Medicine, 2013, 8, 21.	1.7	49
48	UMD-MLH1/MSH2/MSH6 databases: description and analysis of genetic variations in French Lynch syndrome families. Database: the Journal of Biological Databases and Curation, 2013, 2013, bat036-bat036.	3.0	15
49	The TREAT-NMD Duchenne Muscular Dystrophy Registries: Conception, Design, and Utilization by Industry and Academia. Human Mutation, 2013, 34, 1449-1457.	2.5	94
50	Description and analysis of genetic variants in French hereditary breast and ovarian cancer families recorded in the UMD-BRCA1/BRCA2 databases. Nucleic Acids Research, 2012, 40, D992-D1002.	14.5	84
51	Aortic Event Rate in the Marfan Population. Circulation, 2012, 125, 226-232.	1.6	165
52	VarioML framework for comprehensive variation data representation and exchange. BMC Bioinformatics, 2012, 13, 254.	2.6	17
53	Surgical management of patients with Marfan syndrome: Evolution throughout the years. Archives of Cardiovascular Diseases, 2012, 105, 84-90.	1.6	5
54	In-Frame Mutations in Exon 1 of SKI Cause Dominant Shprintzen-Goldberg Syndrome. American Journal of Human Genetics, 2012, 91, 950-957.	6.2	95

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55	Bioinformatics and Mutations Leading to Exon Skipping. Methods in Molecular Biology, 2012, 867, 17-35.	0.9	12
56	Mutation spectrum in the large GTPase dynamin 2, and genotype-phenotype correlation in autosomal dominant centronuclear myopathy. Human Mutation, 2012, 33, 949-959.	2.5	115
57	Motor and respiratory heterogeneity in Duchenne patients: Implication for clinical trials. European Journal of Paediatric Neurology, 2012, 16, 149-160.	1.6	112
58	The new Ghent criteria for Marfan syndrome: what do they change?. Clinical Genetics, 2012, 81, 433-442.	2.0	90
59	UMD-DYSF, a novel locus specific database for the compilation and interactive analysis of mutations in the dysferlin gene. Human Mutation, 2012, 33, E2317-E2331.	2.5	35
60	Hydrophobic pulses predict transmembrane helix irregularities and channel transmembrane units. BMC Bioinformatics, 2011, 12, 135.	2.6	4
61	Polymorphisms of MAMLD1 gene in hypospadias. Journal of Pediatric Urology, 2011, 7, 585-591.	1.1	33
62	DYT6 dystonia: Review of the literature and creation of the UMD locus-specific database (LSDB) for mutations in the THAP1 gene. Human Mutation, 2011, 32, 1213-1224.	2.5	126
63	Missense mutations of conserved glycine residues in fibrillin-1 highlight a potential subtype of cb-EGF-like domains. Human Mutation, 2010, 31, E1021-E1042.	2.5	9
64	MUT-TP53 2.0: a novel versatile matrix for statistical analysis of TP53 mutations in human cancera. Human Mutation, 2010, 31, 1020-1025.	2.5	26
65	UMD-CFTR: A database dedicated to CF and CFTR-related disorders. Human Mutation, 2010, 31, 1011-1019.	2.5	23
66	Cardiovascular manifestations in men and women carrying a FBN1 mutation. European Heart Journal, 2010, 31, 2223-2229.	2.2	133
67	Consensus Statement on Standard of Care for Congenital Muscular Dystrophies. Journal of Child Neurology, 2010, 25, 1559-1581.	1.4	200
68	Locus Reference Genomic sequences: an improved basis for describing human DNA variants. Genome Medicine, 2010, 2, 24.	8.2	100
69	Comparison of Clinical Presentations and Outcomes Between Patients With TGFBR2 and FBN1 Mutations in Marfan Syndrome and Related Disorders. Circulation, 2009, 120, 2541-2549.	1.6	203
70	Clinical and Molecular Study of 320 Children With Marfan Syndrome and Related Type I Fibrillinopathies in a Series of 1009 Probands With Pathogenic FBN1 Mutations. Pediatrics, 2009, 123, 391-398.	2.1	146
71	Pathogenic FBN1 mutations in 146 adults not meeting clinical diagnostic criteria for Marfan syndrome: Further delineation of type 1 fibrillinopathies and focus on patients with an isolated major criterion. American Journal of Medical Genetics, Part A, 2009, 149A, 854-860.	1.2	40
72	TheFBN2gene: new mutations, locus-specific database (Universal Mutation DatabaseFBN2), and genotype-phenotype correlations. Human Mutation, 2009, 30, 181-190.	2.5	49

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73	Analysis of theDYSFmutational spectrum in a large cohort of patients. Human Mutation, 2009, 30, E345-E375.	2.5	97
74	UMD-predictor, a new prediction tool for nucleotide substitution pathogenicity-application to four genes:FBN1,FBN2,TGFBR1, andTGFBR2. Human Mutation, 2009, 30, 952-959.	2.5	80
75	Genotype-phenotype analysis in 2,405 patients with a dystrophinopathy using the UMD-DMD database: a model of nationwide knowledgebase. Human Mutation, 2009, 30, 934-945.	2.5	309
76	Clinical and mutation-type analysis from an international series of 198 probands with a pathogenic FBN1 exons 24â€“32 mutation. European Journal of Human Genetics, 2009, 17, 491-501.	2.8	66
77	Human Splicing Finder: an online bioinformatics tool to predict splicing signals. Nucleic Acids Research, 2009, 37, e67-e67.	14.5	2,206
78	A new locus-specific database (LSDB) for mutations in theTGFBR2gene: UMD-TGFBR2. Human Mutation, 2008, 29, 33-38.	2.5	27
79	UMD-USHbases: a comprehensive set of databases to record and analyse pathogenic mutations and unclassified variants in seven Usher syndrome causing genes. Human Mutation, 2008, 29, E76-E87.	2.5	38
80	Contribution of molecular analyses in diagnosing Marfan syndrome and type I fibrillinopathies: an international study of 1009 probands. Journal of Medical Genetics, 2008, 45, 384-390.	3.2	83
81	Effect of Mutation Type and Location on Clinical Outcome in 1,013 Probands with Marfan Syndrome or Related Phenotypes and FBN1 Mutations: An International Study. American Journal of Human Genetics, 2007, 81, 454-466.	6.2	485
82	Protein- and mRNA-based phenotype-genotype correlations in DMD/BMD with point mutations and molecular basis for BMD with nonsense and frameshift mutations in the DMD gene. Human Mutation, 2007, 28, 183-195.	2.5	107
83	Multiexon skipping leading to an artificial DMD protein lacking amino acids from exons 45 through 55 could rescue up to 63% of patients with Duchenne muscular dystrophy. Human Mutation, 2007, 28, 196-202.	2.5	178
84	Large genomic rearrangements in the CFTRgene contribute to CBAVD. BMC Medical Genetics, 2007, 8, 22.	2.1	42
85	Trichloroethylene exposure and somatic mutations of the VHL gene in patients with Renal Cell Carcinoma. Journal of Occupational Medicine and Toxicology, 2007, 2, 13.	2.2	23
86	Negative genetic neonatal screening for cystic fibrosis caused by compound heterozygosity for two large CFTR rearrangements. Clinical Genetics, 2007, 72, 374-377.	2.0	11
87	Transcriptional explorations of CAPN3 identify novel splicing mutations, a largeâ€sized genomic deletion and evidence for messenger RNA decay. Clinical Genetics, 2007, 72, 582-592.	2.0	25
88	Locus-specific mutation databases: pitfalls and good practice based on the p53 experience. Nature Reviews Cancer, 2006, 6, 83-90.	28.4	134
89	The UMD TP53 database and website: update and revisions. Human Mutation, 2006, 27, 14-20.	2.5	125
90	Meta-analysis of the p53 Mutation Database for Mutant p53 Biological Activity Reveals a Methodologic Bias in Mutation Detection. Clinical Cancer Research, 2006, 12, 62-69.	7.0	67

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91	UMD (Universal Mutation Database): 2005 update. Human Mutation, 2005, 26, 184-191.	2.5	101
92	Dysferlin mutations in LGMD2B, Miyoshi myopathy, and atypical dysferlinopathies. Human Mutation, 2005, 26, 165-165.	2.5	104
93	Introducing the online version of the gene table for neuromuscular disease (nuclear genes only). Neuromuscular Disorders, 2005, 15, 88.	0.6	2
94	Impact of cytomorphological detection of circulating tumor cells in patients with liver cancer. Hepatology, 2004, 39, 792-797.	7.3	184
95	Genotype-phenotype correlation in von Hippel-Lindau families with renal lesions. Human Mutation, 2004, 24, 215-224.	2.5	81
96	High incidence of renal tumours in vitamins A and E synthesis workers: A new cause of occupational cancer?. International Journal of Cancer, 2004, 108, 942-944.	5.1	14
97	Dystrophinopathy caused by mid-intronic substitutions activating cryptic exons in the DMD gene. Neuromuscular Disorders, 2004, 14, 10-18.	0.6	46
98	The UMD-p53 database: New mutations and analysis tools. Human Mutation, 2003, 21, 176-181.	2.5	136
99	Significance of TP53 mutations in human cancer: A critical analysis of mutations at CpG dinucleotides. Human Mutation, 2003, 21, 192-200.	2.5	94
100	Update of the UMD-FBN1mutation database and creation of anFBN1polymorphism database. Human Mutation, 2003, 22, 199-208.	2.5	299
101	Prenatal diagnosis of spinal muscular atrophy by genetic analysis of circulating fetal cells. Lancet, The, 2003, 361, 1013-1014.	13.7	45
102	108th ENMC International Workshop, 3rd Workshop of the MYO-CLUSTER project: EUROMEN, 7th International Emery-Dreifuss Muscular Dystrophy (EDMD) Workshop, 13â€“15 September 2002, Naarden, The Netherlands. Neuromuscular Disorders, 2003, 13, 508-515.	0.6	78
103	Low mitochondrial respiratory chain content correlates with tumor aggressiveness in renal cell carcinoma. Carcinogenesis, 2002, 23, 759-768.	2.8	298
104	Enrichment, Immunomorphological, and Genetic Characterization of Fetal Cells Circulating in Maternal Blood. American Journal of Pathology, 2002, 160, 51-58.	3.8	78
105	Diagnosis of pheochromocytoma and laparoscopic adrenalectomy in two anephric patients with von hippel-lindau disease. American Journal of Kidney Diseases, 2002, 39, e6.1-e6.4.	1.9	4
106	Germline mutation profile ofMEN1in multiple endocrine neoplasia type 1: search for correlation between phenotype and the functional domains of the MEN1 protein. Human Mutation, 2002, 20, 35-47.	2.5	137
107	The UMD-LDLR database: additions to the software and 490 new entries to the database. Human Mutation, 2002, 20, 81-87.	2.5	105
108	Retinal hemangioblastoma in von Hippel-Lindau disease: a clinical and molecular study. Investigative Ophthalmology and Visual Science, 2002, 43, 3067-74.	3.3	91

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109	Association of GSTT1 non-null and NAT1 slow/rapid genotypes with von Hippel-Lindau tumour suppressor gene transversions in sporadic renal cell carcinoma. Pharmacogenetics and Genomics, 2001, 11, 521-535.	5.7	26
110	Assessing TP53 status in human tumours to evaluate clinical outcome. Nature Reviews Cancer, 2001, 1, 233-239.	28.4	587
111	p53 Website and analysis of p53 gene mutations in human cancer: Forging a link between epidemiology and carcinogenesis. Human Mutation, 2000, 15, 105-113.	2.5	231
112	UMD (Universal Mutation Database): A generic software to build and analyze locus-specific databases. Human Mutation, 2000, 15, 86-94.	2.5	184
113	Central nervous system hemangioblastomas, endolymphatic sac tumors, and von Hippel-Lindau disease. Neurosurgical Review, 2000, 23, 1-22.	2.4	147
114	Mutations of the VHL gene in sporadic renal cell carcinoma: Definition of a risk factor for VHL patients to develop an RCC. Human Mutation, 1999, 13, 464-475.	2.5	126
115	APC gene: database of germline and somatic mutations in human tumors and cell lines. Nucleic Acids Research, 1998, 26, 269-270.	14.5	119
116	Marfan Database (third edition): new mutations and new routines for the software. Nucleic Acids Research, 1998, 26, 229-233.	14.5	97
117	LDLR Database (second edition): new additions to the database and the software, and results of the first molecular analysis. Nucleic Acids Research, 1998, 26, 248-252.	14.5	77
118	p53 gene mutation: software and database. Nucleic Acids Research, 1998, 26, 200-204.	14.5	132
119	Software and database for the analysis of mutations in the VHL gene. Nucleic Acids Research, 1998, 26, 256-258.	14.5	119
120	Software and database for the analysis of mutations in the human WT1 gene. Nucleic Acids Research, 1998, 26, 271-274.	14.5	40
121	Marfan Database (second edition): software and database for the analysis of mutations in the human FBN1 gene. Nucleic Acids Research, 1997, 25, 147-150.	14.5	32
122	p53 and APC gene mutations: software and databases. Nucleic Acids Research, 1997, 25, 138-138.	14.5	17
123	Software and database for the analysis of mutations in the human LDL receptor gene. Nucleic Acids Research, 1997, 25, 172-180.	14.5	50
124	APC gene: database of germline and somatic mutations in human tumors and cell lines. Nucleic Acids Research, 1996, 24, 121-124.	14.5	286
125	Report of the Sixth International Workshop on Human Chromosome 3 Mapping 1995. Cytogenetic and Genome Research, 1996, 72, 255-270.	1.1	17
126	Correlations of allelic imbalance of chromosome 14 with adverse prognostic parameters in 148 renal cell carcinomas. , 1996, 17, 215-224.		41

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127	Software and database for the analysis of mutations in the human FBN1 gene. Nucleic Acids Research, 1996, 24, 137-140.	14.5	41
128	p53 gene mutation: software and database. Nucleic Acids Research, 1996, 24, 147-150.	14.5	82
129	Correlations of allelic imbalance of chromosome 14 with adverse prognostic parameters in 148 renal cell carcinomas. Genes Chromosomes and Cancer, 1996, 17, 215-224.	2.8	2
130	Constitutional and somatic deletions of two different regions of maternal chromosome 11 in Wilms tumor. Genomics, 1990, 7, 434-438.	2.9	25