Jean Muller

List of Publications by Year
in descending order

94 papers	18,313 citations	87888 38 h-index	38395 95 g-index
103 all docs	103 docs citations	103 times ranked	31587 citing authors

#	Article	IF	Citations
1	Enterotypes of the human gut microbiome. Nature, 2011, 473, 174-180.	27.8	5,800
2	The STRING database in 2011: functional interaction networks of proteins, globally integrated and scored. Nucleic Acids Research, 2011, 39, D561-D568.	14.5	3,014
3	STRING 8--a global view on proteins and their functional interactions in 630 organisms. Nucleic Acids Research, 2009, 37, D412-D416.	14.5	2,195
4	The Ecoresponsive Genome of Daphnia pulex. Science, 2011, 331, 555-561.	12.6	1,086
5	Functional and Evolutionary Insights from the Genomes of Three Parasitoid Nasonia Species. Science, 2010, 327, 343-348.	12.6	808
6	eggNOG v3.0: orthologous groups covering 1133 organisms at 41 different taxonomic ranges. Nucleic Acids Research, 2012, 40, D284-D289.	14.5	490
7	eggNOG: automated construction and annotation of orthologous groups of genes. Nucleic Acids Research, 2007, 36, D250-D254.	14.5	428
8	BBS10 encodes a vertebrate-specific chaperonin-like protein and is a major BBS locus. Nature Genetics, 2006, 38, 521-524.	21.4	259
9	AnnotSV: an integrated tool for structural variations annotation. Bioinformatics, 2018, 34, 3572-3574.	4.1	231
10	Efficient strategy for the molecular diagnosis of intellectual disability using targeted high-throughput sequencing. Journal of Medical Genetics, 2014, 51, 724-736.	3.2	229
11	Identification of a Novel BBS Gene (BBS12) Highlights the Major Role of a Vertebrate-Specific Branch of Chaperonin-Related Proteins in Bardet-Biedl Syndrome. American Journal of Human Genetics, 2007, 80, 1-11.	6.2	219
12	eggNOG v2.0: extending the evolutionary genealogy of genes with enhanced non-supervised orthologous groups, species and functional annotations. Nucleic Acids Research, 2010, 38, D190-D195.	14.5	202
13	DPY19L2 Deletion as a Major Cause of Globozoospermia. American Journal of Human Genetics, 2011, 88, 344-350.	6.2	172
14	Exome sequencing of Bardetâ€“Biedl syndrome patient identifies a null mutation in the BBSome subunitBBIP1(BBS18). Journal of Medical Genetics, 2014, 51, 132-136.	3.2	124
15	Orthology prediction methods: A quality assessment using curated protein families. BioEssays, 2011, 33, 769-780.	2.5	121
16	AlstrÃ¶m Syndrome: Mutation Spectrum ofALMS1. Human Mutation, 2015, 36, 660-668.	2.5	117
17	Exome sequencing reveals a nonsense mutation inTEX15causing spermatogenic failure in a Turkish family. Human Molecular Genetics, 2015, 24, 5581-5588.	2.9	111
18	Identification of 28 novel mutations in the Bardetâ€“Biedl syndrome genes: the burden of private mutations in an extensively heterogeneous disease. Human Genetics, 2010, 127, 583-593.	3.8	109

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19	PipeAlign: a new toolkit for protein family analysis. Nucleic Acids Research, 2003, 31, 3829-3832.	14.5	108
20	Targeted high-throughput sequencing for diagnosis of genetically heterogeneous diseases: efficient mutation detection in Bardet-Biedl and AlstrÃ¶m Syndromes. Journal of Medical Genetics, 2012, 49, 502-512.	3.2	104
21	Globozoospermia is mainly due to DPY19L2 deletion via non-allelic homologous recombination involving two recombination hotspots. Human Molecular Genetics, 2012, 21, 3695-3702.	2.9	100
22	Sequence and Comparative Genomic Analysis of Actin-related Proteins. Molecular Biology of the Cell, 2005, 16, 5736-5748.	2.1	99
23	Homozygosity Mapping and Candidate Prioritization Identify Mutations, Missed by Whole-Exome Sequencing, in SMOC2, Causing Major Dental Developmental Defects. American Journal of Human Genetics, 2011, 89, 773-781.	6.2	88
24	Next generation sequencing for molecular diagnosis of neuromuscular diseases. Acta Neuropathologica, 2012, 124, 273-283.	7.7	80
25	VaRank: a simple and powerful tool for ranking genetic variants. PeerJ, 2015, 3, e796.	2.0	80
26	20 ans aprÃ¨s: a second mutation in MAOA identified by targeted high-throughput sequencing in a family with altered behavior and cognition. European Journal of Human Genetics, 2014, 22, 776-783.	2.8	75
27	Muâ€opioid receptor activation induces transcriptional plasticity in the central extended amygdala. European Journal of Neuroscience, 2008, 27, 2973-2984.	2.6	74
28	Mutations in SDCCAG8/NPHP10 Cause Bardet-Biedl Syndrome and Are Associated with Penetrant Renal Disease and Absent Polydactyly. Molecular Syndromology, 2010, 1, 273-281.	0.8	73
29	Highly Sensitive Diagnosis of 43 Monogenic Forms of Diabetes or Obesity Through One-Step PCR-Based Enrichment in Combination With Next-Generation Sequencing. Diabetes Care, 2014, 37, 460-467.	8.6	69
30	Identification of a novel mutation confirms the implication of IFT172 (BBS20) in Bardetâ€“Biedl syndrome. Journal of Human Genetics, 2016, 61, 447-450.	2.3	64
31	Defective Membrane Remodeling in Neuromuscular Diseases: Insights from Animal Models. PLoS Genetics, 2012, 8, e1002595.	3.5	62
32	Osteosclerotic bone dysplasia in siblings with a Fam20C mutation. Clinical Genetics, 2011, 80, 177-183.	2.0	61
33	Autosomal Recessive Cerebellar Ataxia Type 3 Due toANO10Mutations. JAMA Neurology, 2014, 71, 1305.	9.0	57
34	Pitfalls of homozygosity mapping: an extended consanguineous Bardetâ€“Biedl syndrome family with two mutant genes (BBS2, BBS10), three mutations, but no triallelism. European Journal of Human Genetics, 2006, 14, 1195-1203.	2.8	56
35	AQUA: automated quality improvement for multiple sequence alignments. Bioinformatics, 2010, 26, 263-265.	4.1	53
36	The Molecular Architecture of Native BBSome Obtained by an Integrated Structural Approach. Structure, 2019, 27, 1384-1394.e4.	3.3	51

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37	Rare De Novo Missense Variants in RNA Helicase DDX6 Cause Intellectual Disability and Dysmorphic Features and Lead to P-Body Defects and RNA Dysregulation. American Journal of Human Genetics, 2019, 105, 509-525.	6.2	50
38	Intragenic FMR1 disease-causing variants: a significant mutational mechanism leading to Fragile-X syndrome. European Journal of Human Genetics, 2017, 25, 423-431.	2.8	48
39	Transcriptome analysis identifies genes with enriched expression in the mouse central extended amygdala. Neuroscience, 2008, 156, 950-965.	2.3	47
40	Proteasome subunit PSMC3 variants cause neurosensory syndrome combining deafness and cataract due to proteotoxic stress. EMBO Molecular Medicine, 2020, 12, e11861.	6.9	43
41	Mutations in the latent TGF-beta binding protein 3 (LTBP3) gene cause brachyolmia with amelogenesis imperfecta. Human Molecular Genetics, 2015, 24, 3038-3049.	2.9	40
42	Genetic evaluation of patients with non-syndromic male infertility. Journal of Assisted Reproduction and Genetics, 2018, 35, 1939-1951.	2.5	39
43	The phenotypic and molecular genetic spectrum of AlstrÃ¶m syndrome in 44 Turkish kindreds and a literature review of AlstrÃ¶m syndrome in Turkey. Journal of Human Genetics, 2015, 60, 1-9.	2.3	38
44	A no-stop mutation in MAGEB4 is a possible cause of rare X-linked azoospermia and oligozoospermia in a consanguineous Turkish family. Journal of Assisted Reproduction and Genetics, 2017, 34, 683-694.	2.5	38
45	AnnotSV and knotAnnotSV: a web server for human structural variations annotations, ranking and analysis. Nucleic Acids Research, 2021, 49, W21-W28.	14.5	38
46	Targeted next generation sequencing application in cardiac channelopathies: Analysis of a cohort of autopsy-negative sudden unexplained deaths. Forensic Science International, 2015, 254, 5-11.	2.2	35
47	Mesoaxial polydactyly is a major feature in Bardetâ€“Biedl syndrome patients with <scp>LZTFL1</scp> (<scp>BBS17</scp>) mutations. Clinical Genetics, 2014, 85, 476-481.	2.0	34
48	Olfaction evaluation and correlation with brain atrophy in Bardetâ€Biedl syndrome. Clinical Genetics, 2014, 86, 521-529.	2.0	32
49	Uncommon nucleotide excision repair phenotypes revealed by targeted high-throughput sequencing. Orphanet Journal of Rare Diseases, 2016, 11, 26.	2.7	32
50	The Data Use Ontology to streamline responsible access to human biomedical datasets. Cell Genomics, 2021, 1, 100028.	6.5	31
51	Identification and Characterization of Known Biallelic Mutations in the IFT27 (BBS19) Gene in a Novel Family With Bardet-Biedl Syndrome. Frontiers in Genetics, 2019, 10, 21.	2.3	30
52	Bardetâ€Biedl syndrome: Antenatal presentation of fortyâ€five fetuses with biallelic pathogenic variants in known Bardetâ€Biedl syndrome genes. Clinical Genetics, 2019, 95, 384-397.	2.0	30
53	Clinical and genetic characterization of Bardetâ€“Biedl syndrome in Tunisia: defining a strategy for molecular diagnosis. Clinical Genetics, 2014, 85, 172-177.	2.0	28
54	A maximum likelihood approximation method for Dirichlet's parameter estimation. Computational Statistics and Data Analysis, 2008, 52, 1315-1322.	1.2	26

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55	Gene Expression Is Altered in the Lateral Hypothalamus upon Activation of the mu Opioid Receptor. Annals of the New York Academy of Sciences, 2008, 1129, 175-184.	3.8	26
56	KD4v: comprehensible knowledge discovery system for missense variant. Nucleic Acids Research, 2012, 40, W71-W75.	14.5	26
57	Predominantly Cone-System Dysfunction as Rare Form of Retinal Degeneration in Patients With Molecularly Confirmed Bardet-Biedl Syndrome. American Journal of Ophthalmology, 2015, 160, 364-372.e1.	3.3	26
58	Expanding the spectrum of PEX10-related peroxisomal biogenesis disorders: slowly progressive recessive ataxia. Journal of Neurology, 2016, 263, 1552-1558.	3.6	26
59	Molecular and clinical study of a cohort of 110 Algerian patients with autosomal recessive ataxia. BMC Medical Genetics, 2015, 16, 36.	2.1	25
60	A Nitrile Hydratase in the Eukaryote Monosiga brevicollis. PLoS ONE, 2008, 3, e3976.	2.5	24
61	MSV3d: database of human MisSense variants mapped to 3D protein structure. Database: the Journal of Biological Databases and Curation, 2012, 2012, bas018-bas018.	3.0	24
62	Identifying Single Copy Orthologs in Metazoa. PLoS Computational Biology, 2011, 7, e1002269.	3.2	23
63	Time-resolved analysis of transcriptional events during SNAI1-triggered epithelial to mesenchymal transition. Biochemical and Biophysical Research Communications, 2009, 385, 485-491.	2.1	22
64	Evidence for secondary-variant genetic burden and non-random distribution across biological modules in a recessive ciliopathy. Nature Genetics, 2020, 52, 1145-1150.	21.4	22
65	Whole-genome sequencing in patients with ciliopathies uncovers a novel recurrent tandem duplication in IFT140. Human Mutation, 2018, 39, 983-992.	2.5	21
66	A <scp>BBS1 SVA</scp> F retrotransposon insertion is a frequent cause of <scp>Bardetâ€Biedl</scp> syndrome. Clinical Genetics, 2021, 99, 318-324.	2.0	21
67	Homozygous Splice Site Mutation in ZP1 Causes Familial Oocyte Maturation Defect. Genes, 2020, 11, 382.	2.4	19
68	ICDS database: interrupted CoDing sequences in prokaryotic genomes. Nucleic Acids Research, 2006, 34, D338-D343.	14.5	18
69	A Novel Mutation in the <i>ROGDI</i> Gene in a Patient with KohlschÃ¼tter-TÃ¶nz Syndrome. Molecular Syndromology, 2014, 5, 293-298.	0.8	18
70	Validation of a clinical practice-based algorithm for the diagnosis of autosomal recessive cerebellar ataxias based on NGS identified cases. Journal of Neurology, 2016, 263, 1314-1322.	3.6	15
71	Genes for spinocerebellar ataxia with blindness and deafness (SCABD/SCAR3, MIM# 271250 and SCABD2). European Journal of Human Genetics, 2016, 24, 1154-1159.	2.8	15
72	PLCB3 Loss of Function Reduces Pseudomonas aeruginosaâ€“Dependent IL-8 Release in Cystic Fibrosis. American Journal of Respiratory Cell and Molecular Biology, 2018, 59, 428-436.	2.9	15

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73	Mutations inKARScause a severe neurological and neurosensory disease with optic neuropathy. Human Mutation, 2019, 40, 1826-1840.	2.5	15
74	High prevalence of <scp>Bardetâ€Biedl</scp> syndrome in La RÃ©union<scp>Island</scp> is due to a founder variant in ARL6/BBS3. Clinical Genetics, 2020, 98, 166-171.	2.0	14
75	Value of <scp>MRI</scp> olfactory bulb evaluation in the assessment of olfactory dysfunction in Bardetâ€“Biedl syndrome. Clinical Genetics, 2016, 90, 79-83.	2.0	13
76	CoDE-seq, an augmented whole-exome sequencing, enables the accurate detection of CNVs and mutations in Mendelian obesity and intellectual disability. Molecular Metabolism, 2018, 13, 1-9.	6.5	13
77	Reproduction Function in Male Patients With Bardet Biedl Syndrome. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e4417-e4429.	3.6	13
78	Protocol GenoDENT: Implementation of a New NGS Panel for Molecular Diagnosis of Genetic Disorders with Orodental Involvement. Methods in Molecular Biology, 2019, 1922, 407-452.	0.9	12
79	Autosomal mutations and human spermatogenic failure. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 1873-1879.	3.8	11
80	Comparative Natural History of Visual Function From Patients With Biallelic Variants inBBS1andBBS10. , 2021, 62, 26.		11
81	Long-Term Follow-Up and Molecular Characterization of a Patient with a RECQL4 Mutation Spectrum Disorder. Dermatology, 2013, 226, 353-357.	2.1	10
82	Genetic Evidence Supporting the Role of the Calcium Channel, CACNA1S, in Tooth Cusp and Root Patterning. Frontiers in Physiology, 2018, 9, 1329.	2.8	10
83	SM2PH-db: an interactive system for the integrated analysis of phenotypic consequences of missense mutations in proteins involved in human genetic diseases. Human Mutation, 2010, 31, 127-135.	2.5	9
84	Novel TTLL5 Variants Associated with Cone-Rod Dystrophy and Early-Onset Severe Retinal Dystrophy. International Journal of Molecular Sciences, 2021, 22, 6410.	4.1	9
85	SCA13 causes dominantly inherited non-progressive myoclonus ataxia. Parkinsonism and Related Disorders, 2017, 38, 80-84.	2.2	8
86	Atypical Retinal Phenotype in a Patient With AlstrÃ¶m Syndrome and Biallelic Novel Pathogenic Variants in ALMS1, Including a de novo Variation. Frontiers in Genetics, 2020, 11, 938.	2.3	6
87	SLITRK2 variants associated with neurodevelopmental disorders impair excitatory synaptic function and cognition in mice. Nature Communications, 2022, 13, .	12.8	6
88	Design and evaluation of Actichip, a thematic microarray for the study of the actin cytoskeleton. BMC Genomics, 2007, 8, 294.	2.8	5
89	NovelIQCEvariations confirm its role in postaxial polydactyly and cause ciliary defect phenotype in zebrafish. Human Mutation, 2020, 41, 240-254.	2.5	5
90	Evaluation of a Custom Design Gene Panel as a Diagnostic Tool for Human Non-Syndromic Infertility. Genes, 2021, 12, 410.	2.4	5

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91	Periodontal (formerly type <scp>VIII</scp>) <scp>Ehlersâ€“Danlos</scp> syndrome: Description of 13 novel cases and expansion of the clinical phenotype. Clinical Genetics, 2021, 100, 206-212.	2.0	5
92	DPY19L2 Deletion as a Major Cause of Globozoospermia. American Journal of Human Genetics, 2011, 88, 517.	6.2	1
93	The Economic, Medical and Psychosocial Consequences of Whole Genome Sequencing for the Genetic Diagnosis of Patients With Intellectual Disability: The DEFIDIAG Study Protocol. Frontiers in Genetics, 2022, 13, 852472.	2.3	1
94	In Vitro Fertilization assisted by IntraCytoplasmic Sperm Injection in a male patient with Bardetâ€“Biedl syndrome. Clinical Genetics, 2022, 101, 573-574.	2.0	0