

Jean Muller

List of Publications by Year in descending order

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Version: 2024-02-01

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	<i>In Vitro</i> Fertilization assisted by IntraCytoplasmic Sperm Injection in a male patient with Bardet-Biedl syndrome. <i>Clinical Genetics</i> , 2022, 101, 573-574.	2.0	0
2	The Economic, Medical and Psychosocial Consequences of Whole Genome Sequencing for the Genetic Diagnosis of Patients With Intellectual Disability: The DEFIDIAG Study Protocol. <i>Frontiers in Genetics</i> , 2022, 13, 852472.	2.3	1
3	SLITRK2 variants associated with neurodevelopmental disorders impair excitatory synaptic function and cognition in mice. <i>Nature Communications</i> , 2022, 13, .	12.8	6
4	A <i>BBS1</i> SVA retrotransposon insertion is a frequent cause of Bardet-Biedl syndrome. <i>Clinical Genetics</i> , 2021, 99, 318-324.	2.0	21
5	Evaluation of a Custom Design Gene Panel as a Diagnostic Tool for Human Non-Syndromic Infertility. <i>Genes</i> , 2021, 12, 410.	2.4	5
6	Periodontal (formerly type VIII) Ehlers-Danlos syndrome: Description of 13 novel cases and expansion of the clinical phenotype. <i>Clinical Genetics</i> , 2021, 100, 206-212.	2.0	5
7	AnnotSV and knotAnnotSV: a web server for human structural variations annotations, ranking and analysis. <i>Nucleic Acids Research</i> , 2021, 49, W21-W28.	14.5	38
8	Novel TLL5 Variants Associated with Cone-Rod Dystrophy and Early-Onset Severe Retinal Dystrophy. <i>International Journal of Molecular Sciences</i> , 2021, 22, 6410.	4.1	9
9	The Data Use Ontology to streamline responsible access to human biomedical datasets. <i>Cell Genomics</i> , 2021, 1, 100028.	6.5	31
10	Comparative Natural History of Visual Function From Patients With Biallelic Variants in <i>BBS1</i> and <i>BBS10</i> . , 2021, 62, 26.		11
11	Novel <i>IQCE</i> variations confirm its role in postaxial polydactyly and cause ciliary defect phenotype in zebrafish. <i>Human Mutation</i> , 2020, 41, 240-254.	2.5	5
12	Atypical Retinal Phenotype in a Patient With Alstr�m Syndrome and Biallelic Novel Pathogenic Variants in <i>ALMS1</i> , Including a de novo Variation. <i>Frontiers in Genetics</i> , 2020, 11, 938.	2.3	6
13	Evidence for secondary-variant genetic burden and non-random distribution across biological modules in a recessive ciliopathy. <i>Nature Genetics</i> , 2020, 52, 1145-1150.	21.4	22
14	Proteasome subunit <i>PSMC3</i> variants cause neurosensory syndrome combining deafness and cataract due to proteotoxic stress. <i>EMBO Molecular Medicine</i> , 2020, 12, e11861.	6.9	43
15	Reproduction Function in Male Patients With Bardet Biedl Syndrome. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, e4417-e4429.	3.6	13
16	High prevalence of Bardet-Biedl syndrome in La R�union Island is due to a founder variant in <i>ARL6/BBS3</i> . <i>Clinical Genetics</i> , 2020, 98, 166-171.	2.0	14
17	Homozygous Splice Site Mutation in <i>ZP1</i> Causes Familial Oocyte Maturation Defect. <i>Genes</i> , 2020, 11, 382.	2.4	19
18	Rare De Novo Missense Variants in RNA Helicase <i>DDX6</i> Cause Intellectual Disability and Dysmorphic Features and Lead to P-Body Defects and RNA Dysregulation. <i>American Journal of Human Genetics</i> , 2019, 105, 509-525.	6.2	50

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19	The Molecular Architecture of Native BBSome Obtained by an Integrated Structural Approach. <i>Structure</i> , 2019, 27, 1384-1394.e4.	3.3	51
20	Mutations in <i>KARS</i> cause a severe neurological and neurosensory disease with optic neuropathy. <i>Human Mutation</i> , 2019, 40, 1826-1840.	2.5	15
21	Protocol GenoDENT: Implementation of a New NGS Panel for Molecular Diagnosis of Genetic Disorders with Orofacial Involvement. <i>Methods in Molecular Biology</i> , 2019, 1922, 407-452.	0.9	12
22	Identification and Characterization of Known Biallelic Mutations in the IFT27 (BBS19) Gene in a Novel Family With Bardet-Biedl Syndrome. <i>Frontiers in Genetics</i> , 2019, 10, 21.	2.3	30
23	Bardet-Biedl syndrome: Antenatal presentation of forty-five fetuses with biallelic pathogenic variants in known Bardet-Biedl syndrome genes. <i>Clinical Genetics</i> , 2019, 95, 384-397.	2.0	30
24	PLCB3 Loss of Function Reduces <i>Pseudomonas aeruginosa</i> -Dependent IL-8 Release in Cystic Fibrosis. <i>American Journal of Respiratory Cell and Molecular Biology</i> , 2018, 59, 428-436.	2.9	15
25	AnnotSV: an integrated tool for structural variations annotation. <i>Bioinformatics</i> , 2018, 34, 3572-3574.	4.1	231
26	Whole-genome sequencing in patients with ciliopathies uncovers a novel recurrent tandem duplication in <i>IFT140</i> . <i>Human Mutation</i> , 2018, 39, 983-992.	2.5	21
27	Genetic evaluation of patients with non-syndromic male infertility. <i>Journal of Assisted Reproduction and Genetics</i> , 2018, 35, 1939-1951.	2.5	39
28	Genetic Evidence Supporting the Role of the Calcium Channel, <i>CACNA1S</i> , in Tooth Cusp and Root Patterning. <i>Frontiers in Physiology</i> , 2018, 9, 1329.	2.8	10
29	CoDE-seq, an augmented whole-exome sequencing, enables the accurate detection of CNVs and mutations in Mendelian obesity and intellectual disability. <i>Molecular Metabolism</i> , 2018, 13, 1-9.	6.5	13
30	SCA13 causes dominantly inherited non-progressive myoclonus ataxia. <i>Parkinsonism and Related Disorders</i> , 2017, 38, 80-84.	2.2	8
31	Intragenic <i>FMR1</i> disease-causing variants: a significant mutational mechanism leading to Fragile-X syndrome. <i>European Journal of Human Genetics</i> , 2017, 25, 423-431.	2.8	48
32	A no-stop mutation in <i>MAGEB4</i> is a possible cause of rare X-linked azoospermia and oligozoospermia in a consanguineous Turkish family. <i>Journal of Assisted Reproduction and Genetics</i> , 2017, 34, 683-694.	2.5	38
33	Validation of a clinical practice-based algorithm for the diagnosis of autosomal recessive cerebellar ataxias based on NGS identified cases. <i>Journal of Neurology</i> , 2016, 263, 1314-1322.	3.6	15
34	Expanding the spectrum of <i>PEX10</i> -related peroxisomal biogenesis disorders: slowly progressive recessive ataxia. <i>Journal of Neurology</i> , 2016, 263, 1552-1558.	3.6	26
35	Value of MRI olfactory bulb evaluation in the assessment of olfactory dysfunction in Bardet-Biedl syndrome. <i>Clinical Genetics</i> , 2016, 90, 79-83.	2.0	13
36	Uncommon nucleotide excision repair phenotypes revealed by targeted high-throughput sequencing. <i>Orphanet Journal of Rare Diseases</i> , 2016, 11, 26.	2.7	32

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37	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
38	Identification of a novel mutation confirms the implication of IFT172 (BBS20) in Bardet-Biedl syndrome. Journal of Human Genetics, 2016, 61, 447-450.	2.3	64
39	Alström Syndrome: Mutation Spectrum of <i>ALMS1</i> . Human Mutation, 2015, 36, 660-668.	2.5	117
40	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
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	Exome sequencing reveals a nonsense mutation in <i>TEX15</i> causing spermatogenic failure in a Turkish family. Human Molecular Genetics, 2015, 24, 5581-5588.	2.9	111
43	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
44	Molecular and clinical study of a cohort of 110 Algerian patients with autosomal recessive ataxia. BMC Medical Genetics, 2015, 16, 36.	2.1	25
45	The phenotypic and molecular genetic spectrum of Alström syndrome in 44 Turkish kindreds and a literature review of Alström syndrome in Turkey. Journal of Human Genetics, 2015, 60, 1-9.	2.3	38
46	VaRank: a simple and powerful tool for ranking genetic variants. PeerJ, 2015, 3, e796.	2.0	80
47	Olfaction evaluation and correlation with brain atrophy in Bardet-Biedl syndrome. Clinical Genetics, 2014, 86, 521-529.	2.0	32
48	Autosomal Recessive Cerebellar Ataxia Type 3 Due to <i>ANO10</i> Mutations. JAMA Neurology, 2014, 71, 1305.	9.0	57
49	A Novel Mutation in the <i>ROGDI</i> Gene in a Patient with Kohlschütter-Törz Syndrome. Molecular Syndromology, 2014, 5, 293-298.	0.8	18
50	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
51	Clinical and genetic characterization of Bardet-Biedl syndrome in Tunisia: defining a strategy for molecular diagnosis. Clinical Genetics, 2014, 85, 172-177.	2.0	28
52	Exome sequencing of Bardet-Biedl syndrome patient identifies a null mutation in the BBSome subunit <i>BBIP1</i> (<i>BBS18</i>). Journal of Medical Genetics, 2014, 51, 132-136.	3.2	124
53	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
54	20 ans après: 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

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55	Mesoaxial polydactyly is a major feature in Bardet-Biedl syndrome patients with <i>LZTFL1</i> (<i>BBS17</i>) mutations. <i>Clinical Genetics</i> , 2014, 85, 476-481.	2.0	34
56	Long-Term Follow-Up and Molecular Characterization of a Patient with a <i>RECQL4</i> Mutation Spectrum Disorder. <i>Dermatology</i> , 2013, 226, 353-357.	2.1	10
57	Defective Membrane Remodeling in Neuromuscular Diseases: Insights from Animal Models. <i>PLoS Genetics</i> , 2012, 8, e1002595.	3.5	62
58	eggNOG v3.0: orthologous groups covering 1133 organisms at 41 different taxonomic ranges. <i>Nucleic Acids Research</i> , 2012, 40, D284-D289.	14.5	490
59	Targeted high-throughput sequencing for diagnosis of genetically heterogeneous diseases: efficient mutation detection in Bardet-Biedl and Alström Syndromes. <i>Journal of Medical Genetics</i> , 2012, 49, 502-512.	3.2	104
60	KD4v: comprehensible knowledge discovery system for missense variant. <i>Nucleic Acids Research</i> , 2012, 40, W71-W75.	14.5	26
61	MSV3d: database of human MisSense variants mapped to 3D protein structure. <i>Database: the Journal of Biological Databases and Curation</i> , 2012, 2012, bas018-bas018.	3.0	24
62	Globozoospermia is mainly due to <i>DPY19L2</i> deletion via non-allelic homologous recombination involving two recombination hotspots. <i>Human Molecular Genetics</i> , 2012, 21, 3695-3702.	2.9	100
63	Autosomal mutations and human spermatogenic failure. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2012, 1822, 1873-1879.	3.8	11
64	Next generation sequencing for molecular diagnosis of neuromuscular diseases. <i>Acta Neuropathologica</i> , 2012, 124, 273-283.	7.7	80
65	The STRING database in 2011: functional interaction networks of proteins, globally integrated and scored. <i>Nucleic Acids Research</i> , 2011, 39, D561-D568.	14.5	3,014
66	Osteosclerotic bone dysplasia in siblings with a <i>Fam20C</i> mutation. <i>Clinical Genetics</i> , 2011, 80, 177-183.	2.0	61
67	Enterotypes of the human gut microbiome. <i>Nature</i> , 2011, 473, 174-180.	27.8	5,800
68	The Ecoresponsive Genome of <i>Daphnia pulex</i> . <i>Science</i> , 2011, 331, 555-561.	12.6	1,086
69	<i>DPY19L2</i> Deletion as a Major Cause of Globozoospermia. <i>American Journal of Human Genetics</i> , 2011, 88, 344-350.	6.2	172
70	<i>DPY19L2</i> Deletion as a Major Cause of Globozoospermia. <i>American Journal of Human Genetics</i> , 2011, 88, 517.	6.2	1
71	Homozygosity Mapping and Candidate Prioritization Identify Mutations, Missed by Whole-Exome Sequencing, in <i>SMOC2</i> , Causing Major Dental Developmental Defects. <i>American Journal of Human Genetics</i> , 2011, 89, 773-781.	6.2	88
72	Orthology prediction methods: A quality assessment using curated protein families. <i>BioEssays</i> , 2011, 33, 769-780.	2.5	121

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73	Identifying Single Copy Orthologs in Metazoa. <i>PLoS Computational Biology</i> , 2011, 7, e1002269.	3.2	23
74	Identification of 28 novel mutations in the Bardet-Biedl syndrome genes: the burden of private mutations in an extensively heterogeneous disease. <i>Human Genetics</i> , 2010, 127, 583-593.	3.8	109
75	SM2PH-db: an interactive system for the integrated analysis of phenotypic consequences of missense mutations in proteins involved in human genetic diseases. <i>Human Mutation</i> , 2010, 31, 127-135.	2.5	9
76	AQUA: automated quality improvement for multiple sequence alignments. <i>Bioinformatics</i> , 2010, 26, 263-265.	4.1	53
77	eggNOG v2.0: extending the evolutionary genealogy of genes with enhanced non-supervised orthologous groups, species and functional annotations. <i>Nucleic Acids Research</i> , 2010, 38, D190-D195.	14.5	202
78	Mutations in SDCCAG8/NPHP10 Cause Bardet-Biedl Syndrome and Are Associated with Penetrant Renal Disease and Absent Polydactyly. <i>Molecular Syndromology</i> , 2010, 1, 273-281.	0.8	73
79	Functional and Evolutionary Insights from the Genomes of Three Parasitoid <i>Nasonia</i> Species. <i>Science</i> , 2010, 327, 343-348.	12.6	808
80	STRING 8—a global view on proteins and their functional interactions in 630 organisms. <i>Nucleic Acids Research</i> , 2009, 37, D412-D416.	14.5	2,195
81	Time-resolved analysis of transcriptional events during SNAIL-triggered epithelial to mesenchymal transition. <i>Biochemical and Biophysical Research Communications</i> , 2009, 385, 485-491.	2.1	22
82	A maximum likelihood approximation method for Dirichlet's parameter estimation. <i>Computational Statistics and Data Analysis</i> , 2008, 52, 1315-1322.	1.2	26
83	Gene Expression Is Altered in the Lateral Hypothalamus upon Activation of the mu Opioid Receptor. <i>Annals of the New York Academy of Sciences</i> , 2008, 1129, 175-184.	3.8	26
84	Mu-opioid receptor activation induces transcriptional plasticity in the central extended amygdala. <i>European Journal of Neuroscience</i> , 2008, 27, 2973-2984.	2.6	74
85	Transcriptome analysis identifies genes with enriched expression in the mouse central extended amygdala. <i>Neuroscience</i> , 2008, 156, 950-965.	2.3	47
86	A Nitrile Hydratase in the Eukaryote <i>Monosiga brevicollis</i> . <i>PLoS ONE</i> , 2008, 3, e3976.	2.5	24
87	eggNOG: automated construction and annotation of orthologous groups of genes. <i>Nucleic Acids Research</i> , 2007, 36, D250-D254.	14.5	428
88	Identification of a Novel BBS Gene (BBS12) Highlights the Major Role of a Vertebrate-Specific Branch of Chaperonin-Related Proteins in Bardet-Biedl Syndrome. <i>American Journal of Human Genetics</i> , 2007, 80, 1-11.	6.2	219
89	Design and evaluation of Actichip, a thematic microarray for the study of the actin cytoskeleton. <i>BMC Genomics</i> , 2007, 8, 294.	2.8	5
90	BBS10 encodes a vertebrate-specific chaperonin-like protein and is a major BBS locus. <i>Nature Genetics</i> , 2006, 38, 521-524.	21.4	259

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91	Pitfalls of homozygosity mapping: an extended consanguineous Bardet-Biedl syndrome family with two mutant genes (BBS2, BBS10), three mutations, but no triallelism. <i>European Journal of Human Genetics</i> , 2006, 14, 1195-1203.	2.8	56
92	ICDS database: interrupted CoDing sequences in prokaryotic genomes. <i>Nucleic Acids Research</i> , 2006, 34, D338-D343.	14.5	18
93	Sequence and Comparative Genomic Analysis of Actin-related Proteins. <i>Molecular Biology of the Cell</i> , 2005, 16, 5736-5748.	2.1	99
94	PipeAlign: a new toolkit for protein family analysis. <i>Nucleic Acids Research</i> , 2003, 31, 3829-3832.	14.5	108