

# Jean Muller

## List of Publications by Year in descending order

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94  
papers

18,313  
citations

87723

38  
h-index

38300

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. <i>Nature</i> , 2011, 473, 174-180.	13.7	5,800
2	The STRING database in 2011: functional interaction networks of proteins, globally integrated and scored. <i>Nucleic Acids Research</i> , 2011, 39, D561-D568.	6.5	3,014
3	STRING 8—a global view on proteins and their functional interactions in 630 organisms. <i>Nucleic Acids Research</i> , 2009, 37, D412-D416.	6.5	2,195
4	The Ecoresponsive Genome of <i>Daphnia pulex</i> . <i>Science</i> , 2011, 331, 555-561.	6.0	1,086
5	Functional and Evolutionary Insights from the Genomes of Three Parasitoid <i>Nasonia</i> Species. <i>Science</i> , 2010, 327, 343-348.	6.0	808
6	eggNOG v3.0: orthologous groups covering 1133 organisms at 41 different taxonomic ranges. <i>Nucleic Acids Research</i> , 2012, 40, D284-D289.	6.5	490
7	eggNOG: automated construction and annotation of orthologous groups of genes. <i>Nucleic Acids Research</i> , 2007, 36, D250-D254.	6.5	428
8	BBS10 encodes a vertebrate-specific chaperonin-like protein and is a major BBS locus. <i>Nature Genetics</i> , 2006, 38, 521-524.	9.4	259
9	AnnotSV: an integrated tool for structural variations annotation. <i>Bioinformatics</i> , 2018, 34, 3572-3574.	1.8	231
10	Efficient strategy for the molecular diagnosis of intellectual disability using targeted high-throughput sequencing. <i>Journal of Medical Genetics</i> , 2014, 51, 724-736.	1.5	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. <i>American Journal of Human Genetics</i> , 2007, 80, 1-11.	2.6	219
12	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.	6.5	202
13	DPY19L2 Deletion as a Major Cause of Globozoospermia. <i>American Journal of Human Genetics</i> , 2011, 88, 344-350.	2.6	172
14	Exome sequencing of Bardet-Biedl syndrome patient identifies a null mutation in the BBSome subunit <i>BBIP1</i> ( <i>BBS18</i> ). <i>Journal of Medical Genetics</i> , 2014, 51, 132-136.	1.5	124
15	Orthology prediction methods: A quality assessment using curated protein families. <i>BioEssays</i> , 2011, 33, 769-780.	1.2	121
16	Alström Syndrome: Mutation Spectrum of <i>ALMS1</i> . <i>Human Mutation</i> , 2015, 36, 660-668.	1.1	117
17	Exome sequencing reveals a nonsense mutation in <i>TEX15</i> causing spermatogenic failure in a Turkish family. <i>Human Molecular Genetics</i> , 2015, 24, 5581-5588.	1.4	111
18	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.	1.8	109

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19	PipeAlign: a new toolkit for protein family analysis. <i>Nucleic Acids Research</i> , 2003, 31, 3829-3832.	6.5	108
20	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.	1.5	104
21	Globozoospermia is mainly due to DPY19L2 deletion via non-allelic homologous recombination involving two recombination hotspots. <i>Human Molecular Genetics</i> , 2012, 21, 3695-3702.	1.4	100
22	Sequence and Comparative Genomic Analysis of Actin-related Proteins. <i>Molecular Biology of the Cell</i> , 2005, 16, 5736-5748.	0.9	99
23	Homozygosity Mapping and Candidate Prioritization Identify Mutations, Missed by Whole-Exome Sequencing, in SMOC2, Causing Major Dental Developmental Defects. <i>American Journal of Human Genetics</i> , 2011, 89, 773-781.	2.6	88
24	Next generation sequencing for molecular diagnosis of neuromuscular diseases. <i>Acta Neuropathologica</i> , 2012, 124, 273-283.	3.9	80
25	VaRank: a simple and powerful tool for ranking genetic variants. <i>PeerJ</i> , 2015, 3, e796.	0.9	80
26	20 ans après: a second mutation in MAOA identified by targeted high-throughput sequencing in a family with altered behavior and cognition. <i>European Journal of Human Genetics</i> , 2014, 22, 776-783.	1.4	75
27	Mu-opioid receptor activation induces transcriptional plasticity in the central extended amygdala. <i>European Journal of Neuroscience</i> , 2008, 27, 2973-2984.	1.2	74
28	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.	0.3	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. <i>Diabetes Care</i> , 2014, 37, 460-467.	4.3	69
30	Identification of a novel mutation confirms the implication of IFT172 (BBS20) in Bardet-Biedl syndrome. <i>Journal of Human Genetics</i> , 2016, 61, 447-450.	1.1	64
31	Defective Membrane Remodeling in Neuromuscular Diseases: Insights from Animal Models. <i>PLoS Genetics</i> , 2012, 8, e1002595.	1.5	62
32	Osteosclerotic bone dysplasia in siblings with a Fam20C mutation. <i>Clinical Genetics</i> , 2011, 80, 177-183.	1.0	61
33	Autosomal Recessive Cerebellar Ataxia Type 3 Due to ANO10 Mutations. <i>JAMA Neurology</i> , 2014, 71, 1305.	4.5	57
34	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.	1.4	56
35	AQUA: automated quality improvement for multiple sequence alignments. <i>Bioinformatics</i> , 2010, 26, 263-265.	1.8	53
36	The Molecular Architecture of Native BBSome Obtained by an Integrated Structural Approach. <i>Structure</i> , 2019, 27, 1384-1394.e4.	1.6	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. <i>American Journal of Human Genetics</i> , 2019, 105, 509-525.	2.6	50
38	Intragenic FMR1 disease-causing variants: a significant mutational mechanism leading to Fragile-X syndrome. <i>European Journal of Human Genetics</i> , 2017, 25, 423-431.	1.4	48
39	Transcriptome analysis identifies genes with enriched expression in the mouse central extended amygdala. <i>Neuroscience</i> , 2008, 156, 950-965.	1.1	47
40	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.	3.3	43
41	Mutations in the latent TGF-beta binding protein 3 (LTBP3) gene cause brachyolmia with amelogenesis imperfecta. <i>Human Molecular Genetics</i> , 2015, 24, 3038-3049.	1.4	40
42	Genetic evaluation of patients with non-syndromic male infertility. <i>Journal of Assisted Reproduction and Genetics</i> , 2018, 35, 1939-1951.	1.2	39
43	The phenotypic and molecular genetic spectrum of Alstr�m syndrome in 44 Turkish kindreds and a literature review of Alstr�m syndrome in Turkey. <i>Journal of Human Genetics</i> , 2015, 60, 1-9.	1.1	38
44	A no-stop mutation in MAGEB4 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.	1.2	38
45	AnnotSV and knotAnnotSV: a web server for human structural variations annotations, ranking and analysis. <i>Nucleic Acids Research</i> , 2021, 49, W21-W28.	6.5	38
46	Targeted next generation sequencing application in cardiac channelopathies: Analysis of a cohort of autopsy-negative sudden unexplained deaths. <i>Forensic Science International</i> , 2015, 254, 5-11.	1.3	35
47	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.	1.0	34
48	Olfaction evaluation and correlation with brain atrophy in Bardet-Biedl syndrome. <i>Clinical Genetics</i> , 2014, 86, 521-529.	1.0	32
49	Uncommon nucleotide excision repair phenotypes revealed by targeted high-throughput sequencing. <i>Orphanet Journal of Rare Diseases</i> , 2016, 11, 26.	1.2	32
50	The Data Use Ontology to streamline responsible access to human biomedical datasets. <i>Cell Genomics</i> , 2021, 1, 100028.	3.0	31
51	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.	1.1	30
52	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.	1.0	30
53	Clinical and genetic characterization of Bardet-Biedl syndrome in Tunisia: defining a strategy for molecular diagnosis. <i>Clinical Genetics</i> , 2014, 85, 172-177.	1.0	28
54	A maximum likelihood approximation method for Dirichlet's parameter estimation. <i>Computational Statistics and Data Analysis</i> , 2008, 52, 1315-1322.	0.7	26

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55	<i>Gene Expression Is Altered in the Lateral Hypothalamus upon Activation of the mu Opioid Receptor</i> . Annals of the New York Academy of Sciences, 2008, 1129, 175-184.	1.8	26
56	KD4v: comprehensible knowledge discovery system for missense variant. Nucleic Acids Research, 2012, 40, W71-W75.	6.5	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.	1.7	26
58	Expanding the spectrum of PEX10-related peroxisomal biogenesis disorders: slowly progressive recessive ataxia. Journal of Neurology, 2016, 263, 1552-1558.	1.8	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.	1.1	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.	1.4	24
62	Identifying Single Copy Orthologs in Metazoa. PLoS Computational Biology, 2011, 7, e1002269.	1.5	23
63	Time-resolved analysis of transcriptional events during SNAIL1-triggered epithelial to mesenchymal transition. Biochemical and Biophysical Research Communications, 2009, 385, 485-491.	1.0	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.	9.4	22
65	Whole-genome sequencing in patients with ciliopathies uncovers a novel recurrent tandem duplication in <i>IFT140</i> . Human Mutation, 2018, 39, 983-992.	1.1	21
66	A <i>BBS1</i> SVA retrotransposon insertion is a frequent cause of Bardet-Biedl syndrome. Clinical Genetics, 2021, 99, 318-324.	1.0	21
67	Homozygous Splice Site Mutation in ZP1 Causes Familial Oocyte Maturation Defect. Genes, 2020, 11, 382.	1.0	19
68	ICDS database: interrupted CoDing sequences in prokaryotic genomes. Nucleic Acids Research, 2006, 34, D338-D343.	6.5	18
69	A Novel Mutation in the <i>ROGDI</i> Gene in a Patient with Kohlschütter-Tarntzer Syndrome. Molecular Syndromology, 2014, 5, 293-298.	0.3	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.	1.8	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.	1.4	15
72	PLCB3 Loss of Function Reduces <i>Pseudomonas aeruginosa</i> -Dependent IL-8 Release in Cystic Fibrosis. American Journal of Respiratory Cell and Molecular Biology, 2018, 59, 428-436.	1.4	15

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73	Mutations in <i>KARS</i> cause a severe neurological and neurosensory disease with optic neuropathy. <i>Human Mutation</i> , 2019, 40, 1826-1840.	1.1	15
74	High prevalence of Bardet-Biedl syndrome in La Réunion Island is due to a founder variant in <i>ARL6/BBS3</i> . <i>Clinical Genetics</i> , 2020, 98, 166-171.	1.0	14
75	Value of MRI olfactory bulb evaluation in the assessment of olfactory dysfunction in Bardet-Biedl syndrome. <i>Clinical Genetics</i> , 2016, 90, 79-83.	1.0	13
76	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.	3.0	13
77	Reproduction Function in Male Patients With Bardet Biedl Syndrome. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, e4417-e4429.	1.8	13
78	Protocol GenoDENT: Implementation of a New NGS Panel for Molecular Diagnosis of Genetic Disorders with Orodonal Involvement. <i>Methods in Molecular Biology</i> , 2019, 1922, 407-452.	0.4	12
79	Autosomal mutations and human spermatogenic failure. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2012, 1822, 1873-1879.	1.8	11
80	Comparative Natural History of Visual Function From Patients With Biallelic Variants in <i>BBS1</i> and <i>BBS10</i> . , 2021, 62, 26.		11
81	Long-Term Follow-Up and Molecular Characterization of a Patient with a RECQL4 Mutation Spectrum Disorder. <i>Dermatology</i> , 2013, 226, 353-357.	0.9	10
82	Genetic Evidence Supporting the Role of the Calcium Channel, CACNA1S, in Tooth Cusp and Root Patterning. <i>Frontiers in Physiology</i> , 2018, 9, 1329.	1.3	10
83	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.	1.1	9
84	Novel TLL5 Variants Associated with Cone-Rod Dystrophy and Early-Onset Severe Retinal Dystrophy. <i>International Journal of Molecular Sciences</i> , 2021, 22, 6410.	1.8	9
85	SCA13 causes dominantly inherited non-progressive myoclonus ataxia. <i>Parkinsonism and Related Disorders</i> , 2017, 38, 80-84.	1.1	8
86	Atypical Retinal Phenotype in a Patient With Alström Syndrome and Biallelic Novel Pathogenic Variants in <i>ALMS1</i> , Including a de novo Variation. <i>Frontiers in Genetics</i> , 2020, 11, 938.	1.1	6
87	SLITRK2 variants associated with neurodevelopmental disorders impair excitatory synaptic function and cognition in mice. <i>Nature Communications</i> , 2022, 13, .	5.8	6
88	Design and evaluation of Actichip, a thematic microarray for the study of the actin cytoskeleton. <i>BMC Genomics</i> , 2007, 8, 294.	1.2	5
89	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.	1.1	5
90	Evaluation of a Custom Design Gene Panel as a Diagnostic Tool for Human Non-Syndromic Infertility. <i>Genes</i> , 2021, 12, 410.	1.0	5

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91	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.	1.0	5
92	DPY19L2 Deletion as a Major Cause of Globozoospermia. <i>American Journal of Human Genetics</i> , 2011, 88, 517.	2.6	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. <i>Frontiers in Genetics</i> , 2022, 13, 852472.	1.1	1
94	<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.	1.0	0