Sean David Mooney

List of Publications by Year
in descending order

147 papers	9,221 citations	71102 41 h-index	49909 87 g-index
166 all docs	166 docs citations	166 times ranked	17627 citing authors

#	Article	IF	Citations
1	A large-scale evaluation of computational protein function prediction. Nature Methods, 2013, 10, 221-227.	19.0	789
2	Automated inference of molecular mechanisms of disease from amino acid substitutions. Bioinformatics, 2009, 25, 2744-2750.	4.1	691
3	Label-free quantitative proteomics of the lysine acetylome in mitochondria identifies substrates of SIRT3 in metabolic pathways. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 6601-6606.	7.1	414
4	Genetic Correction of Huntington's Disease Phenotypes in Induced Pluripotent Stem Cells. Cell Stem Cell, 2012, 11, 253-263.	11.1	336
5	An expanded evaluation of protein function prediction methods shows an improvement in accuracy. Genome Biology, 2016, 17, 184.	8.8	308
6	Inferring the molecular and phenotypic impact of amino acid variants with MutPred2. Nature Communications, 2020, 11, 5918.	12.8	305
7	The structural and functional signatures of proteins that undergo multiple events of postâ€translational modification. Protein Science, 2014, 23, 1077-1093.	7.6	287
8	Splicing factor SFRS1 recognizes a functionally diverse landscape of RNA transcripts. Genome Research, 2009, 19, 381-394.	5.5	284
9	The CAFA challenge reports improved protein function prediction and new functional annotations for hundreds of genes through experimental screens. Genome Biology, 2019, 20, 244.	8.8	261
10	Late-life rapamycin treatment reverses age-related heart dysfunction. Aging Cell, 2013, 12, 851-862.	6.7	258
11	Indicators of retention in remote digital health studies: a cross-study evaluation of 100,000 participants. Npj Digital Medicine, 2020, 3, 21.	10.9	238
12	A Novel Recessive Mutation in Fibroblast Growth Factor-23 Causes Familial Tumoral Calcinosis. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 2424-2427.	3.6	205
13	Newborn Sequencing in Genomic Medicine and Public Health. Pediatrics, 2017, 139, .	2.1	174
14	High-performance web services for querying gene and variant annotation. Genome Biology, 2016, 17, 91.	8.8	166
15	Proteomic analysis of ageâ€dependent changes in protein solubility identifies genes that modulate lifespan. Aging Cell, 2012, 11, 120-127.	6.7	155
16	An integrated approach to inferring geneâ€“disease associations in humans. Proteins: Structure, Function and Bioinformatics, 2008, 72, 1030-1037.	2.6	153
17	Bioinformatics approaches and resources for single nucleotide polymorphism functional analysis. Briefings in Bioinformatics, 2005, 6, 44-56.	6.5	146
18	Genomeâ€wide <scp>DNA</scp> methylation changes with age in diseaseâ€free human skeletal muscle. Aging Cell, 2014, 13, 360-366.	6.7	145

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19	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. American Journal of Human Genetics, 2016, 98, 1051-1066.	6.2	137
20	MutPred Splice: machine learning-based prediction of exonic variants that disrupt splicing. Genome Biology, 2014, 15, R19.	9.6	135
21	Analysis of protein function and its prediction from amino acid sequence. Proteins: Structure, Function and Bioinformatics, 2011, 79, 2086-2096.	2.6	124
22	Fibroblast Growth Factor-23 Mutants Causing Familial Tumoral Calcinosis Are Differentially Processed. Endocrinology, 2005, 146, 3883-3891.	2.8	119
23	Genomic Analysis Reveals Disruption of Striatal Neuronal Development and Therapeutic Targets in Human Huntingtonâ€™s Disease Neural Stem Cells. Stem Cell Reports, 2015, 5, 1023-1038.	4.8	117
24	Considering the possibilities and pitfalls of Generative Pre-trained Transformer 3 (GPT-3) in healthcare delivery. Npj Digital Medicine, 2021, 4, 93.	10.9	113
25	The role of exome sequencing in newborn screening for inborn errors of metabolism. Nature Medicine, 2020, 26, 1392-1397.	30.7	112
26	An Excess of Deleterious Variants in VEGF-A Pathway Genes in Down-Syndrome-Associated Atrioventricular Septal Defects. American Journal of Human Genetics, 2012, 91, 646-659.	6.2	99
27	Gain and loss of phosphorylation sites in human cancer. Bioinformatics, 2008, 24, i241-i247.	4.1	94
28	SILAC Analysis Reveals Increased Secretion of Hemostasis-Related Factors by Senescent Cells. Cell Reports, 2019, 28, 3329-3337.e5.	6.4	94
29	Rationale and design of the Kidney Precision Medicine Project. Kidney International, 2021, 99, 498-510.	5.2	94
30	A Large Scale Huntingtin Protein Interaction Network Implicates Rho GTPase Signaling Pathways in Huntington Disease. Journal of Biological Chemistry, 2014, 289, 6709-6726.	3.4	83
31	Using Mobile Apps to Assess and Treat Depression in Hispanic and Latino Populations: Fully Remote Randomized Clinical Trial. Journal of Medical Internet Research, 2018, 20, e10130.	4.3	82
32	Calibration of Multiple In Silico Tools for Predicting Pathogenicity of Mismatch Repair Gene Missense Substitutions. Human Mutation, 2013, 34, 255-265.	2.5	80
33	The accuracy of passive phone sensors in predicting daily mood. Depression and Anxiety, 2019, 36, 72-81.	4.1	80
34	Evaluation of features for catalytic residue prediction in novel folds. Protein Science, 2006, 16, 216-226.	7.6	61
35	Morquio <scp>A</scp> Syndromeâ€Associated Mutations: A Review of Alterations in the GALNS Gene and a New Locusâ€Specific Database. Human Mutation, 2014, 35, 1271-1279.	2.5	59
36	In silico functional profiling of human disease-associated and polymorphic amino acid substitutions. Human Mutation, 2010, 31, 335-346.	2.5	57

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37	LOSS OF POST-TRANSLATIONAL MODIFICATION SITES IN DISEASE. , 2009, , 337-347.		56
38	Bioinformatic Tools for Identifying Disease Gene and SNP Candidates. Methods in Molecular Biology, 2010, 628, 307-319.	0.9	55
39	Toward More Transparent and Reproducible Omics Studies Through a Common Metadata Checklist and Data Publications. OMICS A Journal of Integrative Biology, 2014, 18, 10-14.	2.0	54
40	When loss-of-function is loss of function: assessing mutational signatures and impact of loss-of-function genetic variants. Bioinformatics, 2017, 33, i389-i398.	4.1	53
41	Nitric-oxide Synthase (NOS) Reductase Domain Models Suggest a New Control Element in Endothelial NOS That Attenuates Calmodulin-dependent Activity. Journal of Biological Chemistry, 2003, 278, 31814-31824.	3.4	50
42	Conformational preferences of substituted prolines in the collagen triple helix. Biopolymers, 2002, 64, 63-71.	2.4	49
43	MutDB services: interactive structural analysis of mutation data. Nucleic Acids Research, 2005, 33, W311-W314.	14.5	48
44	Influence of Sequence Changes and Environment on Intrinsically Disordered Proteins. PLoS Computational Biology, 2009, 5, e1000497.	3.2	47
45	Mapping genetic variations to three-dimensional protein structures to enhance variant interpretation: a proposed framework. Genome Medicine, 2017, 9, 113.	8.2	47
46	CADD score has limited clinical validity for the identification of pathogenic variants in noncoding regions in a hereditary cancer panel. Genetics in Medicine, 2016, 18, 1269-1275.	2.4	45
47	Modelling kidney disease using ontology: insights from the Kidney Precision Medicine Project. Nature Reviews Nephrology, 2020, 16, 686-696.	9.6	45
48	A predictive tool for identification of SARS-CoV-2 PCR-negative emergency department patients using routine test results. Journal of Clinical Virology, 2020, 129, 104502.	3.1	45
49	The functional importance of disease-associated mutation. BMC Bioinformatics, 2002, 3, 24.	2.6	43
50	A Genome-Scale RNAâ€“Interference Screen Identifies RRAS Signaling as a Pathologic Feature of Huntington's Disease. PLoS Genetics, 2012, 8, e1003042.	3.5	41
51	MutDB: annotating human variation with functionally relevant data. Bioinformatics, 2003, 19, 1858-1860.	4.1	40
52	Computed Free energy differences between point mutations in a collagen-like peptide. Biopolymers, 2001, 58, 347-353.	2.4	39
53	Missense variant pathogenicity predictors generalize well across a range of functionâ€specific prediction challenges. Human Mutation, 2017, 38, 1092-1108.	2.5	39
54	Working toward precision medicine: Predicting phenotypes from exomes in the Critical Assessment of Genome Interpretation (CAGI) challenges. Human Mutation, 2017, 38, 1182-1192.	2.5	39

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55	Automating Construction of Machine Learning Models With Clinical Big Data: Proposal Rationale and Methods. JMIR Research Protocols, 2017, 6, e175.	1.0	38
56	Mitochondrial DNA Sequence Variation Associated with Dementia and Cognitive Function in the Elderly. Journal of Alzheimer's Disease, 2012, 32, 357-372.	2.6	37
57	Leaf: an open-source, model-agnostic, data-driven web application for cohort discovery and translational biomedical research. Journal of the American Medical Informatics Association: JAMIA, 2020, 27, 109-118.	4.4	35
58	MutDB: update on development of tools for the biochemical analysis of genetic variation. Nucleic Acids Research, 2007, 36, D815-D819.	14.5	34
59	Progress towards the integration of pharmacogenomics in practice. Human Genetics, 2015, 134, 459-465.	3.8	34
60	Pathogenicity and functional impact of non-frameshifting insertion/deletion variation in the human genome. PLoS Computational Biology, 2019, 15, e1007112.	3.2	34
61	Identifying viral integration sites using SeqMap 2.0. Bioinformatics, 2011, 27, 720-722.	4.1	33
62	Uncovering exposures responsible for birth season â€“ disease effects: a global study. Journal of the American Medical Informatics Association: JAMIA, 2018, 25, 275-288.	4.4	33
63	Overnight transduction with foamyviral vectors restores the long-term repopulating activity of Fanccâ¯/â¯ stem cells. Blood, 2008, 112, 4458-4465.	1.4	30
64	Derivation, Characterization, and Neural Differentiation of Integration-Free Induced Pluripotent Stem Cell Lines from Parkinsonâ€™s Disease Patients Carrying SNCA, LRRK2, PARK2, and GBA Mutations. PLoS ONE, 2016, 11, e0154890.	2.5	29
65	Sudden unexpected fatal encephalopathy in adults with OTC gene mutations-Clues for early diagnosis and timely treatment. Orphanet Journal of Rare Diseases, 2014, 9, 105.	2.7	28
66	Clinical relevance of short-chain acyl-CoA dehydrogenase (SCAD) deficiency: Exploring the role of new variants including the first SCAD-disease-causing allele carrying a synonymous mutation. BBA Clinical, 2016, 5, 114-119.	4.1	27
67	Tor1 regulates protein solubility in Saccharomyces cerevisiae. Molecular Biology of the Cell, 2012, 23, 4679-4688.	2.1	26
68	Optimizing the Molecular Diagnosis of GALNS: Novel Methods to Define and Characterize Morquio-A Syndrome-Associated Mutations. Human Mutation, 2015, 36, 357-368.	2.5	26
69	Tau/MAPT disease-associated variant A152T alters tau function and toxicity via impaired retrograde axonal transport. Human Molecular Genetics, 2019, 28, 1498-1514.	2.9	26
70	Target site specificity and in vivo complexity of the mammalian arginylome. Scientific Reports, 2018, 8, 16177.	3.3	25
71	SARS-CoV-2 infection and COVID-19 severity in individuals with prior seasonal coronavirus infection. Diagnostic Microbiology and Infectious Disease, 2021, 100, 115338.	1.8	25
72	Structural characterization of proteins using residue environments. Proteins: Structure, Function and Bioinformatics, 2005, 61, 741-747.	2.6	24

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73	Identification of similar regions of protein structures using integrated sequence and structure analysis tools. BMC Structural Biology, 2006, 6, 4.	2.3	21
74	Prediction of functional regulatory SNPs in monogenic and complex disease. Human Mutation, 2011, 32, 1183-1190.	2.5	21
75	A functional analysis of disease-associated mutations in the androgen receptor gene. Nucleic Acids Research, 2003, 31, 42e-42.	14.5	20
76	Extensible open source content management systems and frameworks: a solution for many needs of a bioinformatics group. Briefings in Bioinformatics, 2007, 9, 69-74.	6.5	20
77	Structural Models of Osteogenesis Imperfecta-associated Variants in the COL1A1 Gene. Molecular and Cellular Proteomics, 2002, 1, 868-875.	3.8	19
78	Smartphone-based passive assessment of mobility in depression: Challenges and opportunities. Mental Health and Physical Activity, 2018, 14, 136-139.	1.8	19
79	Assessment of blind predictions of the clinical significance of BRCA1 and BRCA2 variants. Human Mutation, 2019, 40, 1546-1556.	2.5	19
80	A genome-wide screen of bacterial mutants that enhance dauer formation in C. elegans. Scientific Reports, 2016, 6, 38764.	3.3	18
81	Forecasting Future Asthma Hospital Encounters of Patients With Asthma in an Academic Health Care System: Predictive Model Development and Secondary Analysis Study. Journal of Medical Internet Research, 2021, 23, e22796.	4.3	18
82	STOP using just GO: a multi-ontology hypothesis generation tool for high throughput experimentation. BMC Bioinformatics, 2013, 14, 53.	2.6	17
83	The Loss and Gain of Functional Amino Acid Residues Is a Common Mechanism Causing Human Inherited Disease. PLoS Computational Biology, 2016, 12, e1005091.	3.2	16
84	Structure-based kernels for the prediction of catalytic residues and their involvement in human inherited disease. Bioinformatics, 2010, 26, 1975-1982.	4.1	15
85	NewDrosophilaLong-Term Memory Genes Revealed by Assessing Computational Function Prediction Methods. G3: Genes, Genomes, Genetics, 2019, 9, 251-267.	1.8	15
86	Matching phenotypes to whole genomes: Lessons learned from four iterations of the personal genome project community challenges. Human Mutation, 2017, 38, 1266-1276.	2.5	14
87	Mitochondrial DNA sequence variation is associated with free-living activity energy expenditure in the elderly. Biochimica Et Biophysica Acta - Bioenergetics, 2012, 1817, 1691-1700.	1.0	13
88	Intrinsic size parameters for palmitoylated and carboxyamidomethylated peptides. International Journal of Mass Spectrometry, 2014, 368, 6-14.	1.5	13
89	Regulatory Single-Nucleotide Variant Predictor Increases Predictive Performance of Functional Regulatory Variants. Human Mutation, 2016, 37, 1137-1143.	2.5	13
90	An ontology-neutral framework for enrichment analysis. AMIA ... Annual Symposium proceedings, 2010, 2010, 797-801.	0.2	13

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91	Using RNase sequence specificity to refine the identification of RNA-protein binding regions. BMC Genomics, 2008, 9, S17.	2.8	12
92	Biological and functional relevance of CASP predictions. Proteins: Structure, Function and Bioinformatics, 2018, 86, 374-386.	2.6	12
93	Comparative protein interactomics of neuroglobin and myoglobin. Journal of Neurochemistry, 2012, 123, 192-198.	3.9	11
94	In silico comparative characterization of pharmacogenomic missense variants. BMC Genomics, 2014, 15, S4.	2.8	11
95	Research informatics and the COVID-19 pandemic: Challenges, innovations, lessons learned, and recommendations. Journal of Clinical and Translational Science, 2021, 5, e110.	0.6	11
96	Connecting Protein Interaction Data, Mutations, and Disease Using Bioinformatics. Methods in Molecular Biology, 2009, 541, 449-461.	0.9	11
97	Integrated single-cell sequencing and histopathological analyses reveal diverse injury and repair responses in a participant with acute kidney injury: a clinical-molecular-pathologic correlation. Kidney International, 2022, 101, 1116-1125.	5.2	11
98	A Probabilistic Model to Predict Clinical Phenotypic Traits from Genome Sequencing. PLoS Computational Biology, 2014, 10, e1003825.	3.2	10
99	The feasibility of using smartphones to assess and remediate depression in Hispanic/Latino individuals nationally. , 2017, , .		10
100	CAGI SickKids challenges: Assessment of phenotype and variant predictions derived from clinical and genomic data of children with undiagnosed diseases. Human Mutation, 2019, 40, 1373-1391.	2.5	10
101	Assessment of predicted enzymatic activity of Î±â€•‹i›N‹/i› â€acetylglucosaminidase variants of unknown significance for CAGI 2016. Human Mutation, 2019, 40, 1519-1529.	2.5	10
102	Predicting venous thromboembolism risk from exomes in the Critical Assessment of Genome Interpretation (CAGI) challenges. Human Mutation, 2019, 40, 1314-1320.	2.5	10
103	Genetic nondiscrimination legislation: a critical prerequisite for pharmacogenomics data sharing. Pharmacogenomics, 2007, 8, 519-519.	1.3	9
104	Evaluation of resequencing on number of tag SNPs of 13 atherosclerosis-related genes in Thai population. Journal of Human Genetics, 2008, 53, 74-86.	2.3	9
105	High-resolution melting curve analysis for genotyping of common SNP in MTHFR gene using fixed-cell suspension. Molecular and Cellular Probes, 2008, 22, 329-332.	2.1	9
106	Functional organization and its implication in evolution of the human protein-protein interaction network. BMC Genomics, 2012, 13, 150.	2.8	9
107	Extending the coverage of spectral libraries: A neighbor-based approach to predicting intensities of peptide fragmentation spectra. Proteomics, 2013, 13, 756-765.	2.2	9
108	Ten Simple Rules for a Community Computational Challenge. PLoS Computational Biology, 2015, 11, e1004150.	3.2	9

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109	Unbiased identification of novel transcription factors in striatal compartmentation and striosome maturation. ELife, 2021, 10, .	6.0	9
110	Assessing the performance of in silico methods for predicting the pathogenicity of variants in the gene CHEK2, among Hispanic females with breast cancer. Human Mutation, 2019, 40, 1612-1622.	2.5	8
111	Automated retrieval, preprocessing, and visualization of gridded hydrometeorology data products for spatial-temporal exploratory analysis and intercomparison. Environmental Modelling and Software, 2019, 116, 119-130.	4.5	8
112	A Continuously Benchmarked and Crowdsourced Challenge for Rapid Development and Evaluation of Models to Predict COVID-19 Diagnosis and Hospitalization. JAMA Network Open, 2021, 4, e2124946.	5.9	8
113	Mitochondrial DNA Sequence Variation Associated With Peripheral Nerve Function in the Elderly. Journals of Gerontology - Series A Biological Sciences and Medical Sciences, 2015, 70, 1400-1408.	3.6	7
114	Structure-based kernels for the prediction of catalytic residues and their involvement in human inherited disease. BMC Bioinformatics, 2010, 11, .	2.6	6
115	Piloting a model-to-data approach to enable predictive analytics in health care through patient mortality prediction. Journal of the American Medical Informatics Association: JAMIA, 2020, 27, 1393-1400.	4.4	6
116	Information needs and priority use cases of population health researchers to improve preparedness for future hurricanes and floods. Journal of the American Medical Informatics Association: JAMIA, 2021, 28, 249-260.	4.4	6
117	Toward More Transparent and Reproducible Omics Studies Through a Common Metadata Checklist and Data Publications. Big Data, 2013, 1, 196-201.	3.4	5
118	Medium-Chain Acyl-CoA Deficiency: Outlines from Newborn Screening,In SilicoPredictions, and Molecular Studies. Scientific World Journal, The, 2013, 2013, 1-8.	2.1	5
119	Assessing computational predictions of the phenotypic effect of cystathionineâ€betaâ€synthase variants. Human Mutation, 2019, 40, 1530-1545.	2.5	5
120	Patient perspectives and involvement in precision medicine research. Kidney International, 2021, 99, 511-514.	5.2	5
121	A Comparison of Natural Language Processing Methods for the Classification of Lumbar Spine Imaging Findings Related to Lower Back Pain. Academic Radiology, 2022, 29, S188-S200.	2.5	5
122	Collaborative software for traditional and translational research. Human Genomics, 2012, 6, 21.	2.9	4
123	Biotinidase deficiency due to a de novo mutation or gonadal mosaicism in a first child. Clinica Chimica Acta, 2015, 445, 70-72.	1.1	4
124	ShareDNA: a smartphone app to facilitate family communication of genetic results. BMC Medical Genomics, 2021, 14, 10.	1.5	4
125	PRECISION MEDICINE: DATA AND DISCOVERY FOR IMPROVED HEALTH AND THERAPY. Pacific Symposium on Biocomputing Pacific Symposium on Biocomputing, 2016, 21, 243-8.	0.7	4
126	Analysis of Mutations in the COLIA1 Gene with Second-Order Rule Induction. International Journal of Pattern Recognition and Artificial Intelligence, 2003, 17, 721-740.	1.2	3

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127	DEFOG: discrete enrichment of functionally organized genes. Integrative Biology (United Kingdom), 2012, 4, 795.	1.3	3
128	Analysis of Features from Protein-protein Hetero-complex Structures to Predict Protein Interaction Interfaces Using Machine Learning. Procedia Technology, 2013, 10, 62-66.	1.1	3
129	Evaluation of the secondary use of electronic health records to detect seasonal, holiday-related, and rare events related to traumatic injury and poisoning. BMC Public Health, 2020, 20, 46.	2.9	3
130	Development and Validation of ARC, a Model for Anticipating Acute Respiratory Failure in Coronavirus Disease 2019 Patients. , 2021, 3, e0441.		3
131	Data science in clinical and translational research: Improving the health of the data to knowledge pipeline. Journal of Clinical and Translational Science, 2021, 5, e77.	0.6	3
132	A Participant-Centered Approach to Understanding Risks and Benefits of Participation in Research Informed by the Kidney Precision Medicine Project. American Journal of Kidney Diseases, 2022, 80, 132-138.	1.9	3
133	PRECISION MEDICINE: DATA AND DISCOVERY FOR IMPROVED HEALTH AND THERAPY. , 2016, , .		2
134	Harnessing Innovative Technologies to Train Nurses in Suicide Safety Planning With Hospitalized Patients: Protocol for Formative and Pilot Feasibility Research. JMIR Research Protocols, 2021, 10, e33695.	1.0	2
135	From the clinic to the community: Can health system data accurately estimate population obesity prevalence?. Obesity, 2021, 29, 1961-1968.	3.0	2
136	USE OF GENOME DATA IN NEWBORNS AS A STARTING POINT FOR LIFE-LONG PRECISION MEDICINE. , 2016, , .		1
137	Non-Coding Variation: The 2016 Annual Scientific Meeting of the Human Genome Variation Society. Human Mutation, 2017, 38, 460-463.	2.5	1
138	Conformational preferences of substituted prolines in the collagen triple helix. Biopolymers, 2002, 64, 63-71.	2.4	1
139	Comparative Study of Particle Swarm Approaches for the Prediction of Functionally Important Residues in Protein Structures. , 2008, , .		0
140	Predicting Protein-Disease Relationships Using Sequence, Physicochemical Properties, and Molecular Function Information. Nature Precedings, 2008, , .	0.1	0
141	Bioinformatics Approaches to the Functional Proï¬ling of Genetic Variants. , 0, , .		0
142	Continuing challenges swirl around bioinformatics service delivery. Journal of Biomedical Informatics, 2019, 94, 103209.	4.3	0
143	Session Introduction. , 2002, , .		0
144	Session Introduction. , 2004, , .		0

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145	Reconstitution of Neutrophil NADPH Oxidase Activity in Murine X-CGD Following Transplantation of Retrovirus-Transduced Marrow: Potential Impact of Submyeloablative Conditioning.. Blood, 2005, 106, 3050-3050.	1.4	0
146	Biased Engraftment of Retrovirus-Transduced Marrow in Murine X-CGD Following Submyeloablative Conditioning.. Blood, 2006, 108, 3285-3285.	1.4	0
147	Establishing the reliability of algorithms. , 2020, , .		0