

Edwin Cuppen

List of Publications by Year in descending order

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Version: 2024-02-01

307
papers

36,675
citations

4388

86
h-index

4015

176
g-index

348
all docs

348
docs citations

348
times ranked

53787
citing authors

#	ARTICLE	IF	CITATIONS
1	Clinical utility of whole-genome sequencing in precision oncology. <i>Seminars in Cancer Biology</i> , 2022, 84, 32-39.	9.6	35
2	Analytical demands to use whole-genome sequencing in precision oncology. <i>Seminars in Cancer Biology</i> , 2022, 84, 16-22.	9.6	22
3	Clinical interpretation of whole-genome and whole-transcriptome sequencing for precision oncology. <i>Seminars in Cancer Biology</i> , 2022, 84, 23-31.	9.6	10
4	Patients with Rare Cancers in the Drug Rediscovery Protocol (DRUP) Benefit from Genomics-Guided Treatment. <i>Clinical Cancer Research</i> , 2022, 28, 1402-1411.	7.0	24
5	Comprehensive Molecular Characterization Reveals Genomic and Transcriptomic Subtypes of Metastatic Urothelial Carcinoma. <i>European Urology</i> , 2022, 81, 331-336.	1.9	23
6	MutationalPatterns: the one stop shop for the analysis of mutational processes. <i>BMC Genomics</i> , 2022, 23, 134.	2.8	66
7	Unscrambling cancer genomes via integrated analysis of structural variation and copy number. <i>Cell Genomics</i> , 2022, 2, 100112.	6.5	34
8	Distinct Genomic Profiles Are Associated with Treatment Response and Survival in Ovarian Cancer. <i>Cancers</i> , 2022, 14, 1511.	3.7	5
9	Different responses to DNA damage determine ageing differences between organs. <i>Aging Cell</i> , 2022, 21, e13562.	6.7	16
10	Genome-wide analysis of somatic noncoding mutation patterns in cancer. <i>Science</i> , 2022, 376, eabg5601.	12.6	33
11	Functional RECAP (REpair CAPacity) assay identifies homologous recombination deficiency undetected by DNA-based BRCAness tests. <i>Oncogene</i> , 2022, 41, 3498-3506.	5.9	9
12	Recurrent exon-deleting activating mutations in AHR act as drivers of urinary tract cancer. <i>Scientific Reports</i> , 2022, 12, .	3.3	2
13	A multi-platform reference for somatic structural variation detection. <i>Cell Genomics</i> , 2022, 2, 100139.	6.5	10
14	Machine learning-based tissue of origin classification for cancer of unknown primary diagnostics using genome-wide mutation features. <i>Nature Communications</i> , 2022, 13, .	12.8	28
15	VIRUSBreakend: Viral Integration Recognition Using Single Breakends. <i>Bioinformatics</i> , 2021, 37, 3115-3119.	4.1	15
16	Micro-costing diagnostics in oncology: from single-gene testing to whole- genome sequencing. <i>Expert Review of Pharmacoeconomics and Outcomes Research</i> , 2021, 21, 413-414.	1.4	16
17	Early technology assessment of using whole genome sequencing in personalized oncology. <i>Expert Review of Pharmacoeconomics and Outcomes Research</i> , 2021, 21, 343-351.	1.4	4
18	Learning mutational signatures and their multidimensional genomic properties with TensorSignatures. <i>Nature Communications</i> , 2021, 12, 3628.	12.8	30

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19	Prospective experimental treatment of colorectal cancer patients based on organoid drug responses. ESMO Open, 2021, 6, 100103.	4.5	62
20	The genomic landscape of 85 advanced neuroendocrine neoplasms reveals subtype-heterogeneity and potential therapeutic targets. Nature Communications, 2021, 12, 4612.	12.8	55
21	GRIDSS2: comprehensive characterisation of somatic structural variation using single breakend variants and structural variant phasing. Genome Biology, 2021, 22, 202.	8.8	73
22	Clinical Validation of Whole Genome Sequencing for Cancer Diagnostics. Journal of Molecular Diagnostics, 2021, 23, 816-833.	2.8	47
23	Reconstructing single-cell karyotype alterations in colorectal cancer identifies punctuated and gradual diversification patterns. Nature Genetics, 2021, 53, 1187-1195.	21.4	37
24	Limited evolution of the actionable metastatic cancer genome under therapeutic pressure. Nature Medicine, 2021, 27, 1553-1563.	30.7	41
25	Whole genome sequencing of metastatic colorectal cancer reveals prior treatment effects and specific metastasis features. Nature Communications, 2021, 12, 574.	12.8	39
26	Precancerous liver diseases do not cause increased mutagenesis in liver stem cells. Communications Biology, 2021, 4, 1301.	4.4	9
27	Biallelic variants in POLR3GL cause endosteal hyperostosis and oligodontia. European Journal of Human Genetics, 2020, 28, 31-39.	2.8	21
28	Single-cell derived tumor organoids display diversity in HLA class I peptide presentation. Nature Communications, 2020, 11, 5338.	12.8	41
29	Study protocol: Whole genome sequencing Implementation in standard Diagnostics for Every cancer patient (WIDE). BMC Medical Genomics, 2020, 13, 169.	1.5	30
30	Patient-Derived Ovarian Cancer Organoids Mimic Clinical Response and Exhibit Heterogeneous Inter- and Inpatient Drug Responses. Cell Reports, 2020, 31, 107762.	6.4	155
31	Pan-cancer landscape of homologous recombination deficiency. Nature Communications, 2020, 11, 5584.	12.8	262
32	The mutational impact of culturing human pluripotent and adult stem cells. Nature Communications, 2020, 11, 2493.	12.8	61
33	Challenges in Establishing Pure Lung Cancer Organoids Limit Their Utility for Personalized Medicine. Cell Reports, 2020, 31, 107588.	6.4	125
34	Mutational signature in colorectal cancer caused by genotoxic pks+ E. coli. Nature, 2020, 580, 269-273.	27.8	587
35	Impact of panel design and cut-off on tumour mutational burden assessment in metastatic solid tumour samples. British Journal of Cancer, 2020, 122, 953-956.	6.4	21
36	A deep learning system accurately classifies primary and metastatic cancers using passenger mutation patterns. Nature Communications, 2020, 11, 728.	12.8	140

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37	SARS-CoV-2 productively infects human gut enterocytes. <i>Science</i> , 2020, 369, 50-54.	12.6	1,347
38	Sperm DNA damage causes genomic instability in early embryonic development. <i>Science Advances</i> , 2020, 6, eaaz7602.	10.3	37
39	Human extrahepatic and intrahepatic cholangiocyte organoids show region-specific differentiation potential and model cystic fibrosis-related bile duct disease. <i>Scientific Reports</i> , 2020, 10, 21900.	3.3	43
40	Abstract 5673: In-depth analysis of the genomic landscape of 91 metastatic neuroendocrine tumors reveals subtype-heterogeneity and potential therapeutic targets. , 2020, , .		0
41	Patient-derived organoids can predict response to chemotherapy in metastatic colorectal cancer patients. <i>Science Translational Medicine</i> , 2019, 11, .	12.4	451
42	5-Fluorouracil treatment induces characteristic T>G mutations in human cancer. <i>Nature Communications</i> , 2019, 10, 4571.	12.8	143
43	The genomic landscape of metastatic breast cancer highlights changes in mutation and signature frequencies. <i>Nature Genetics</i> , 2019, 51, 1450-1458.	21.4	250
44	Molecular tumour boards and molecular diagnostics for patients with cancer in the Netherlands: experiences, challenges, and aspirations. <i>British Journal of Cancer</i> , 2019, 121, 34-36.	6.4	16
45	Deficiency of nucleotide excision repair is associated with mutational signature observed in cancer. <i>Genome Research</i> , 2019, 29, 1067-1077.	5.5	66
46	Early divergence of mutational processes in human fetal tissues. <i>Science Advances</i> , 2019, 5, eaaw1271.	10.3	24
47	Portrait of a cancer: mutational signature analyses for cancer diagnostics. <i>BMC Cancer</i> , 2019, 19, 457.	2.6	84
48	Tubuloids derived from human adult kidney and urine for personalized disease modeling. <i>Nature Biotechnology</i> , 2019, 37, 303-313.	17.5	301
49	The genomic landscape of metastatic castration-resistant prostate cancers reveals multiple distinct genotypes with potential clinical impact. <i>Nature Communications</i> , 2019, 10, 5251.	12.8	130
50	Prioritization of genes driving congenital phenotypes of patients with de novo genomic structural variants. <i>Genome Medicine</i> , 2019, 11, 79.	8.2	19
51	The Drug Rediscovery protocol facilitates the expanded use of existing anticancer drugs. <i>Nature</i> , 2019, 574, 127-131.	27.8	152
52	Pan-cancer whole-genome analyses of metastatic solid tumours. <i>Nature</i> , 2019, 575, 210-216.	27.8	722
53	Pancreatic cancer organoids recapitulate disease and allow personalized drug screening. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2019, 116, 26580-26590.	7.1	279
54	Unique Case of a Rare Mesenchymal Tumor Harboring a Somatic c.119delC VHL Mutation. <i>JCO Precision Oncology</i> , 2019, 3, 1-8.	3.0	0

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55	Long-term expanding human airway organoids for disease modeling. EMBO Journal, 2019, 38, .	7.8	619
56	Abstract GS1-07: The genomic landscape of 501 metastatic breast cancer patients. , 2019, , .		2
57	The molecular genetic make-up of male breast cancer. Endocrine-Related Cancer, 2019, 26, 779-794.	3.1	27
58	Sequencing Approaches for Personalized Cancer Therapy Selection in Pathology. , 2019, , 432-438.		0
59	Abstract 2727: The premalignant state captured in the landscape of somatic mutations can reveal the cancer cell-of-origin. , 2019, , .		0
60	EP828â€œ...Whole genome sequencing and clinical drug response data of 142 ovarian cancer samples. , 2019, , .		0
61	Abstract 1738: Characterization of structural variants within MACROD2 in the pathogenesis of colorectal cancer. , 2019, , .		0
62	MutationalPatterns: comprehensive genome-wide analysis of mutational processes. Genome Medicine, 2018, 10, 33.	8.2	482
63	RAS pathway mutations as a predictive biomarker for treatment adaptation in pediatric B-cell precursor acute lymphoblastic leukemia. Leukemia, 2018, 32, 931-940.	7.2	75
64	A Living Biobank of Breast Cancer Organoids Captures Disease Heterogeneity. Cell, 2018, 172, 373-386.e10.	28.9	1,201
65	Measuring mutation accumulation in single human adult stem cells by whole-genome sequencing of organoid cultures. Nature Protocols, 2018, 13, 59-78.	12.0	52
66	In-depth assessment of metastatic prostate cancer with high tumour mutational burden. Annals of Oncology, 2018, 29, viii274.	1.2	3
67	A System-wide Approach to Monitor Responses to Synergistic BRAF and EGFR Inhibition in Colorectal Cancer Cells. Molecular and Cellular Proteomics, 2018, 17, 1892-1908.	3.8	13
68	Cancer cells copy migratory behavior and exchange signaling networks via extracellular vesicles. EMBO Journal, 2018, 37, .	7.8	58
69	The complete genomic landscape of metastatic prostate cancer pinpoints clinically targetable subgroups.. Journal of Clinical Oncology, 2018, 36, 5014-5014.	1.6	0
70	Genetic dissection of colorectal cancer progression by orthotopic transplantation of engineered cancer organoids. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E2357-E2364.	7.1	198
71	Ankyrin repeat and zinc-finger domain-containing 1 mutations are associated with infantile-onset inflammatory bowel disease. Journal of Biological Chemistry, 2017, 292, 7904-7920.	3.4	29
72	Implementation of a Multicenter Biobanking Collaboration for Next-Generation Sequencingâ€œBased Biomarker Discovery Based on Fresh Frozen Pretreatment Tumor Tissue Biopsies. Oncologist, 2017, 22, 33-40.	3.7	29

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73	Molecular Tumor Boards: current practice and future needs. <i>Annals of Oncology</i> , 2017, 28, 3070-3075.	1.2	112
74	Use of CRISPR-modified human stem cell organoids to study the origin of mutational signatures in cancer. <i>Science</i> , 2017, 358, 234-238.	12.6	337
75	Troy/TNFRSF19 marks epithelial progenitor cells during mouse kidney development that continue to contribute to turnover in adult kidney. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2017, 114, E11190-E11198.	7.1	19
76	Mapping and phasing of structural variation in patient genomes using nanopore sequencing. <i>Nature Communications</i> , 2017, 8, 1326.	12.8	315
77	Molecular dissection of germline chromothripsis in a developmental context using patient-derived iPS cells. <i>Genome Medicine</i> , 2017, 9, 9.	8.2	25
78	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. <i>Nature Genetics</i> , 2017, 49, 36-45.	21.4	251
79	Predicting clinical benefit from everolimus in patients with advanced solid tumors, the CPCT-03 study. <i>Oncotarget</i> , 2017, 8, 55582-55592.	1.8	9
80	Targeted Next Generation Sequencing as a Reliable Diagnostic Assay for the Detection of Somatic Mutations in Tumours Using Minimal DNA Amounts from Formalin Fixed Paraffin Embedded Material. <i>PLoS ONE</i> , 2016, 11, e0149405.	2.5	79
81	The Genomic Scrapheap Challenge; Extracting Relevant Data from Unmapped Whole Genome Sequencing Reads, Including Strain Specific Genomic Segments, in Rats. <i>PLoS ONE</i> , 2016, 11, e0160036.	2.5	5
82	Deficiency or inhibition of lysophosphatidic acid receptor 1 protects against hyperoxia-induced lung injury in neonatal rats. <i>Acta Physiologica</i> , 2016, 216, 358-375.	3.8	18
83	Mechanisms of Therapy Resistance in Patient-Derived Xenograft Models of BRCA1-Deficient Breast Cancer. <i>Journal of the National Cancer Institute</i> , 2016, 108, djw148.	6.3	157
84	Systematic analysis of chromatin interactions at disease associated loci links novel candidate genes to inflammatory bowel disease. <i>Genome Biology</i> , 2016, 17, 247.	8.8	39
85	The PLETHORA Gene Regulatory Network Guides Growth and Cell Differentiation in Arabidopsis Roots. <i>Plant Cell</i> , 2016, 28, 2937-2951.	6.6	127
86	Tissue-specific mutation accumulation in human adult stem cells during life. <i>Nature</i> , 2016, 538, 260-264.	27.8	759
87	The role of the dopamine D1 receptor in social cognition: studies using a novel genetic rat model. <i>DMM Disease Models and Mechanisms</i> , 2016, 9, 1147-1158.	2.4	35
88	Generation and characterization of rat liver stem cell lines and their engraftment in a rat model of liver failure. <i>Scientific Reports</i> , 2016, 6, 22154.	3.3	50
89	Epigenomic annotation of gene regulatory alterations during evolution of the primate brain. <i>Nature Neuroscience</i> , 2016, 19, 494-503.	14.8	113
90	Genome-wide analysis reveals <i>NRP1</i> as a direct HIF1 α -E2F7 target in the regulation of motorneuron guidance <i>in vivo</i> . <i>Nucleic Acids Research</i> , 2016, 44, 3549-3566.	14.5	29

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91	Prioritization and burden analysis of rare variants in 208 candidate genes suggest they do not play a major role in CAKUT. <i>Kidney International</i> , 2016, 89, 476-486.	5.2	78
92	RAS Pathway Mutations As Predictive Biomarker for Treatment Adaptation in Pediatric B-Cell Precursor Acute Lymphoblastic Leukemia. <i>Blood</i> , 2016, 128, 4087-4087.	1.4	2
93	Abstract CT113: The time to progression ratio: a new individualized volumetric parameter for early detection of clinical benefit of targeted therapies. , 2016, , .		0
94	A novel Dock8 gene mutation confers diabetogenic susceptibility in the LEW.1AR1/Ztm-iddm rat, an animal model of human type 1 diabetes. <i>Diabetologia</i> , 2015, 58, 2800-2809.	6.3	13
95	KOUNCIL: Kidney-Oriented Understanding of Correcting Ciliopathies. <i>Cilia</i> , 2015, 4, .	1.8	0
96	Genetic Etiology of Renal Agenesis: Fine Mapping of Renag1 and Identification of Kit as the Candidate Functional Gene. <i>PLoS ONE</i> , 2015, 10, e0118147.	2.5	10
97	Lack of Major Genome Instability in Tumors of p53 Null Rats. <i>PLoS ONE</i> , 2015, 10, e0122066.	2.5	11
98	Translational regulation shapes the molecular landscape of complex disease phenotypes. <i>Nature Communications</i> , 2015, 6, 7200.	12.8	79
99	Chromothripsis in Healthy Individuals Affects Multiple Protein-Coding Genes and Can Result in Severe Congenital Abnormalities in Offspring. <i>American Journal of Human Genetics</i> , 2015, 96, 651-656.	6.2	76
100	Genomic landscape of rat strain and substrain variation. <i>BMC Genomics</i> , 2015, 16, 357.	2.8	84
101	Next generation sequencing of triple negative breast cancer to find predictors for chemotherapy response. <i>Breast Cancer Research</i> , 2015, 17, 134.	5.0	58
102	Organoid Models of Human and Mouse Ductal Pancreatic Cancer. <i>Cell</i> , 2015, 160, 324-338.	28.9	1,584
103	Long-Term Culture of Genome-Stable Bipotent Stem Cells from Adult Human Liver. <i>Cell</i> , 2015, 160, 299-312.	28.9	1,166
104	Rb and FZR1/Cdh1 determine CDK4/6-cyclin D requirement in <i>C. elegans</i> and human cancer cells. <i>Nature Communications</i> , 2015, 6, 5906.	12.8	62
105	Next-generation sequencing-based genome diagnostics across clinical genetics centers: implementation choices and their effects. <i>European Journal of Human Genetics</i> , 2015, 23, 1142-1150.	2.8	56
106	Ascl2 Acts as an R-spondin/Wnt-Responsive Switch to Control Stemness in Intestinal Crypts. <i>Cell Stem Cell</i> , 2015, 16, 158-170.	11.1	217
107	Sambamba: fast processing of NGS alignment formats. <i>Bioinformatics</i> , 2015, 31, 2032-2034.	4.1	1,487
108	Toward effective software solutions for big biology. <i>Nature Biotechnology</i> , 2015, 33, 686-687.	17.5	46

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109	Sequential cancer mutations in cultured human intestinal stem cells. <i>Nature</i> , 2015, 521, 43-47.	27.8	853
110	Characteristics of de novo structural changes in the human genome. <i>Genome Research</i> , 2015, 25, 792-801.	5.5	115
111	Detailed imaging and genetic analysis reveal a secondary <i>BRAF</i> ^L resistance mutation and extensive inpatient heterogeneity in metastatic <i>BRAF</i> mutant melanoma patients treated with vemurafenib. <i>Pigment Cell and Melanoma Research</i> , 2015, 28, 318-323.	3.3	20
112	Preserved genetic diversity in organoids cultured from biopsies of human colorectal cancer metastases. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, 13308-13311.	7.1	356
113	Towards personalized therapy in pediatric acute lymphoblastic leukemia: RAS mutations and prednisolone resistance. <i>Haematologica</i> , 2015, 100, e132-e136.	3.5	29
114	Simultaneous Detection of Clinically Relevant Mutations and Amplifications for Routine Cancer Pathology. <i>Journal of Molecular Diagnostics</i> , 2015, 17, 10-18.	2.8	35
115	Pluripotency in the light of the developmental hourglass. <i>Biological Reviews</i> , 2015, 90, 428-443.	10.4	6
116	Deficiency or inhibition of lysophosphatidic acid receptor 1 protects against hyperoxia-induced lung injury in neonatal rats. , 2015, , .		0
117	Abstract 16461: Genetic Basis of Severe Pulmonary Arterial Hypertension in a Colony of SU5416 "Hyper-responsive" Sprague-dawley Rats. <i>Circulation</i> , 2015, 132, .	1.6	0
118	Effective Therapeutic Intervention and Comprehensive Genetic Analysis of mTOR Signaling in PEComa: A Case Report. <i>Anticancer Research</i> , 2015, 35, 3399-403.	1.1	2
119	Genomic and Functional Overlap between Somatic and Germline Chromosomal Rearrangements. <i>Cell Reports</i> , 2014, 9, 2001-2010.	6.4	21
120	Large-Scale Identification of Coregulated Enhancer Networks in the Adult Human Brain. <i>Cell Reports</i> , 2014, 9, 767-779.	6.4	78
121	Identification of <i>Srp9</i> as a febrile seizure susceptibility gene. <i>Annals of Clinical and Translational Neurology</i> , 2014, 1, 239-250.	3.7	18
122	Expanding the spectrum of phenotypes associated with germline <i>PIGA</i> mutations: A child with developmental delay, accelerated linear growth, facial dysmorphisms, elevated alkaline phosphatase, and progressive CNS abnormalities. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 29-35.	1.2	37
123	Extensive localization of long noncoding RNAs to the cytosol and mono- and polyribosomal complexes. <i>Genome Biology</i> , 2014, 15, R6.	9.6	305
124	Targeted next-generation sequencing: A novel diagnostic tool for primary immunodeficiencies. <i>Journal of Allergy and Clinical Immunology</i> , 2014, 133, 529-534.e1.	2.9	143
125	Genomic and transcriptomic plasticity in treatment-naïve ovarian cancer. <i>Genome Research</i> , 2014, 24, 200-211.	5.5	72
126	<i>MEFV</i> mutations affecting pyrin amino acid 577 cause autosomal dominant autoinflammatory disease. <i>Annals of the Rheumatic Diseases</i> , 2014, 73, 455-461.	0.9	101

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127	Wnt-induced transcriptional activation is exclusively mediated by TCF/LEF. EMBO Journal, 2014, 33, 146-156.	7.8	157
128	Identification of Multipotent Luminal Progenitor Cells in Human Prostate Organoid Cultures. Cell, 2014, 159, 163-175.	28.9	609
129	Mate pair sequencing for the detection of chromosomal aberrations in patients with intellectual disability and congenital malformations. European Journal of Human Genetics, 2014, 22, 652-659.	2.8	32
130	Genome-wide survey indicates involvement of loci on canine chromosomes 7 and 31 in patellar luxation in flat-coated retrievers. BMC Genetics, 2014, 15, 64.	2.7	16
131	Genome sequencing of normal cells reveals developmental lineages and mutational processes. Nature, 2014, 513, 422-425.	27.8	315
132	Natural variation of histone modification and its impact on gene expression in the rat genome. Genome Research, 2014, 24, 942-953.	5.5	53
133	Many Inflammatory Bowel Disease Risk Loci Include Regions That Regulate Gene Expression in Immune Cells and the Intestinal Epithelium. Gastroenterology, 2014, 146, 1040-1047.	1.3	92
134	Genomes and phenomes of a population of outbred rats and its progenitors. Scientific Data, 2014, 1, 140011.	5.3	25
135	Systemic miRNA-7 delivery inhibits tumor angiogenesis and growth in murine xenograft glioblastoma. Oncotarget, 2014, 5, 6687-6700.	1.8	105
136	Towards Personalized Therapy in Pediatric Acute Lymphoblastic Leukemia; Ras Mutations and Prednisolone Resistance. Blood, 2014, 124, 372-372.	1.4	0
137	Systematic biases in DNA copy number originate from isolation procedures. Genome Biology, 2013, 14, R33.	9.6	39
138	Nucleosomal DNA binding drives the recognition of H3K36-methylated nucleosomes by the PSIP1-PWWP domain. Epigenetics and Chromatin, 2013, 6, 12.	3.9	141
139	Improving mammalian genome scaffolding using large insert mate-pair next-generation sequencing. BMC Genomics, 2013, 14, 257.	2.8	35
140	Functional Repair of CFTR by CRISPR/Cas9 in Intestinal Stem Cell Organoids of Cystic Fibrosis Patients. Cell Stem Cell, 2013, 13, 653-658.	11.1	1,149
141	Genome Sequencing Reveals Loci under Artificial Selection that Underlie Disease Phenotypes in the Laboratory Rat. Cell, 2013, 154, 691-703.	28.9	154
142	Quantitative and Qualitative Proteome Characteristics Extracted from In-Depth Integrated Genomics and Proteomics Analysis. Cell Reports, 2013, 5, 1469-1478.	6.4	113
143	Genome-wide analysis of FOXO3 mediated transcription regulation through RNA polymerase II profiling. Molecular Systems Biology, 2013, 9, 638.	7.2	104
144	Ethical, Legal, and Counseling Challenges Surrounding the Return of Genetic Results in Oncology. Journal of Clinical Oncology, 2013, 31, 1842-1848.	1.6	85

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145	Chromothripsis in congenital disorders and cancer: similarities and differences. <i>Current Opinion in Cell Biology</i> , 2013, 25, 341-348.	5.4	83
146	A systematic genome-wide analysis of zebrafish protein-coding gene function. <i>Nature</i> , 2013, 496, 494-497.	27.8	579
147	Combined sequence-based and genetic mapping analysis of complex traits in outbred rats. <i>Nature Genetics</i> , 2013, 45, 767-775.	21.4	176
148	Challenges for implementing next-generation sequencing-based genome diagnostics: it's also the people, not just the machines. <i>Personalized Medicine</i> , 2013, 10, 473-484.	1.5	9
149	Incomplete segregation of MYH11 variants with thoracic aortic aneurysms and dissections and patent ductus arteriosus. <i>European Journal of Human Genetics</i> , 2013, 21, 487-493.	2.8	36
150	A Deep Sequencing Approach to Uncover the miRNOME in the Human Heart. <i>PLoS ONE</i> , 2013, 8, e57800.	2.5	88
151	Pmch-Deficiency in Rats Is Associated with Normal Adipocyte Differentiation and Lower Sympathetic Adipose Drive. <i>PLoS ONE</i> , 2013, 8, e60214.	2.5	10
152	Effect of vertical sleeve gastrectomy in melanocortin receptor 4-deficient rats. <i>American Journal of Physiology - Endocrinology and Metabolism</i> , 2012, 303, E103-E110.	3.5	41
153	Discovery of variants unmasked by hemizygous deletions. <i>European Journal of Human Genetics</i> , 2012, 20, 748-753.	2.8	19
154	E2F7 represses a network of oscillating cell cycle genes to control S-phase progression. <i>Nucleic Acids Research</i> , 2012, 40, 3511-3523.	14.5	91
155	Primary Colorectal Cancers and Their Subsequent Hepatic Metastases Are Genetically Different: Implications for Selection of Patients for Targeted Treatment. <i>Clinical Cancer Research</i> , 2012, 18, 688-699.	7.0	136
156	Integrated genome-wide analysis of transcription factor occupancy, RNA polymerase II binding and steady-state RNA levels identify differentially regulated functional gene classes. <i>Nucleic Acids Research</i> , 2012, 40, 148-158.	14.5	65
157	Cell Autonomous Lipin 1 Function Is Essential for Development and Maintenance of White and Brown Adipose Tissue. <i>Molecular and Cellular Biology</i> , 2012, 32, 4794-4810.	2.3	40
158	Diabetes Risk Gene and Wnt Effector Tcf7l2/TCF4 Controls Hepatic Response to Perinatal and Adult Metabolic Demand. <i>Cell</i> , 2012, 151, 1595-1607.	28.9	202
159	Genetic basis of transcriptome differences between the founder strains of the rat HXB/BXH recombinant inbred panel. <i>Genome Biology</i> , 2012, 13, r31.	9.6	32
160	Application of exome sequencing in the search for genetic causes of rare disorders of copper metabolism. <i>Metallomics</i> , 2012, 4, 606.	2.4	6
161	<i>SMN1</i> gene duplications are associated with sporadic ALS. <i>Neurology</i> , 2012, 78, 776-780.	1.1	54
162	X-exome sequencing identifies a <i>HDAC8</i> variant in a large pedigree with X-linked intellectual disability, truncal obesity, gynaecomastia, hypogonadism and unusual face. <i>Journal of Medical Genetics</i> , 2012, 49, 539-543.	3.2	65

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163	NIPA1 polyalanine repeat expansions are associated with amyotrophic lateral sclerosis. Human Molecular Genetics, 2012, 21, 2497-2502.	2.9	49
164	Dominant missense mutations in ABCC9 cause Cantu's syndrome. Nature Genetics, 2012, 44, 793-796.	21.4	184
165	The ter Mutation in the Rat Dnd1 Gene Initiates Gonadal Teratomas and Infertility in Both Genders. PLoS ONE, 2012, 7, e38001.	2.5	25
166	miR-127 Protects Proximal Tubule Cells against Ischemia/Reperfusion: Identification of Kinesin Family Member 3B as miR-127 Target. PLoS ONE, 2012, 7, e44305.	2.5	59
167	Constitutional Chromothripsis Rearrangements Involve Clustered Double-Stranded DNA Breaks and Nonhomologous Repair Mechanisms. Cell Reports, 2012, 1, 648-655.	6.4	193
168	Resolving the Breakpoints of the 17q21.31 Microdeletion Syndrome with Next-Generation Sequencing. American Journal of Human Genetics, 2012, 90, 599-613.	6.2	22
169	Melanocortin Receptor 4 Deficiency Affects Body Weight Regulation, Grooming Behavior, and Substrate Preference in the Rat. Obesity, 2012, 20, 612-621.	3.0	77
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