Jacek Majewski

List of Publications by Year
in descending order

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Version: 2024-02-01

133	15,588	28274	18130
papers	citations	55 h-index	120 g-index

137	137	137	24775
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	HSP90 inhibitors induce GPNMB cell-surface expression by modulating lysosomal positioning and sensitize breast cancer cells to glembatumumab vedotin. Oncogene, 2022, 41, 1701-1717.	5.9	8
2	H3K36 dimethylation shapes the epigenetic interaction landscape by directing repressive chromatin modifications in embryonic stem cells. Genome Research, 2022, , gr.276383.121.	5.5	17
3	Epimutations in both the TESK2 and MMACHC promoters in the Epi-cblC inherited disorder of intracellular metabolism of vitamin B12. Clinical Epigenetics, 2022, 14, 52.	4.1	10
4	Snrpb is required in murine neural crest cells for proper splicing and craniofacial morphogenesis. DMM Disease Models and Mechanisms, 2022, 15, .	2.4	9
5	Nucleome programming is required for the foundation of totipotency in mammalian germline development. EMBO Journal, 2022, 41, .	7.8	9
6	Wholeâ€exome sequencing reveals novel vacuolar ATPase genesâ€™ variants and variants in genes involved in lysosomal biology and autophagosomal formation in oral granular cell tumors. Journal of Oral Pathology and Medicine, 2021, 50, 410-417.	2.7	5
7	Depletion of H3K36me2 recapitulates epigenomic and phenotypic changes induced by the H3.3K36M oncohistone mutation. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	7.1	31
8	Mutation in Eftud2 causes craniofacial defects in mice via mis-splicing of Mdm2 and increased P53. Human Molecular Genetics, 2021, 30, 739-757.	2.9	20
9	Chromatin dysregulation associated with NSD1 mutation in head and neck squamous cell carcinoma. Cell Reports, 2021, 34, 108769.	6.4	42
10	Novel pathogenic variants in <scp>NLRP7</scp>, <scp>NLRP5,</scp> and <scp>PADI6</scp> in patients with recurrent hydatidiform moles and reproductive failure. Clinical Genetics, 2021, 99, 823-828.	2.0	17
11	Two competing mechanisms of DNMT3A recruitment regulate the dynamics of de novo DNA methylation at PRC1-targeted CpG islands. Nature Genetics, 2021, 53, 794-800.	21.4	59
12	Inherent genomic properties underlie the epigenomic heterogeneity of human induced pluripotent stem cells. Cell Reports, 2021, 37, 109909.	6.4	14
13	A functionally impaired missense variant identified in French Canadian families implicates FANCI as a candidate ovarian cancer-predisposing gene. Genome Medicine, 2021, 13, 186.	8.2	12
14	Exome sequencing study of partial agenesis of the corpus callosum in men with developmental delay, epilepsy, and microcephaly. Molecular Genetics & Genomic Medicine, 2020, 8, e992.	1.2	11
15	H3K27M in Gliomas Causes a One-Step Decrease in H3K27 Methylation and Reduced Spreading within the Constraints of H3K36 Methylation. Cell Reports, 2020, 33, 108390.	6.4	50
16	Prognostic and predictive value of circulating tumor DNA during neoadjuvant chemotherapy for triple negative breast cancer. Scientific Reports, 2020, 10, 14704.	3.3	41
17	Early infantile epileptic encephalopathy due to biallelic pathogenic variants in <scp>PIGQ</scp>: Report of seven new subjects and review of the literature. Journal of Inherited Metabolic Disease, 2020, 43, 1321-1332.	3.6	15
18	A novel pathogenic missense ADAMTS17 variant that impairs secretion causes Weill-Marchesani Syndrome with variably dysmorphic hand features. Scientific Reports, 2020, 10, 10827.	3.3	13

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19	Latency and interval therapy affect the evolution in metastatic colorectal cancer. Scientific Reports, 2020, 10, 581.	3.3	6
20	ZBTB7B (ThPOK) Is Required for Pathogenesis of Cerebral Malaria and Protection against Pulmonary Tuberculosis. Infection and Immunity, 2020, 88, .	2.2	6
21	A Mouse Model for Cerebroâ€Costoâ€Mandibular Syndrome (CCMS) with an Intronic Deletion in SnrpB. FASEB Journal, 2020, 34, 1-1.	0.5	0
22	Deletion of Mouse Sf3b4 in Neural Crest Cells Causes Craniofacial Abnormalities. FASEB Journal, 2020, 34, 1-1.	0.5	0
23	CYRI/FAM49B negatively regulates RAC1-driven cytoskeletal remodelling and protects against bacterial infection. Nature Microbiology, 2019, 4, 1516-1531.	13.3	37
24	The histone mark H3K36me2 recruits DNMT3A and shapes the intergenic DNA methylation landscape. Nature, 2019, 573, 281-286.	27.8	338
25	Epigenomic Reordering Induced by Polycomb Loss Drives Oncogenesis but Leads to Therapeutic Vulnerabilities in Malignant Peripheral Nerve Sheath Tumors. Cancer Research, 2019, 79, 3205-3219.	0.9	38
26	H3K27M induces defective chromatin spread of PRC2-mediated repressive H3K27me2/me3 and is essential for glioma tumorigenesis. Nature Communications, 2019, 10, 1262.	12.8	215
27	A Unique Morphological Phenotype in Chemoresistant Triple-Negative Breast Cancer Reveals Metabolic Reprogramming and PLIN4 Expression as a Molecular Vulnerability. Molecular Cancer Research, 2019, 17, 2492-2507.	3.4	63
28	Recessive mutation in CD2AP causes focal segmental glomerulosclerosis in humans and mice. Kidney International, 2019, 95, 57-61.	5.2	11
29	Biallelic Loss-of-Function Variants in AIMP1 Cause a Rare Neurodegenerative Disease. Journal of Child Neurology, 2019, 34, 74-80.	1.4	9
30	Circulating tumor DNA (ctDNA) during and after neoadjuvant chemotherapy and prior to surgery is a powerful prognostic factor in triple-negative breast cancer (TNBC).. Journal of Clinical Oncology, 2019, 37, 594-594.	1.6	6
31	EFTUD2 , The Gene Responsible For Mandibulofacial Dysostosis With Microcephaly (MFDM), Is Required For Implantation And Craniofacial Development In Mouse. FASEB Journal, 2019, 33, 774.20.	0.5	0
32	POLR3A variants in hereditary spastic paraplegia and ataxia. Brain, 2018, 141, e1-e1.	7.6	17
33	A PRDX1 mutant allele causes a MMACHC secondary epimutation in cblC patients. Nature Communications, 2018, 9, 67.	12.8	64
34	Biallelic PADI6 variants linking infertility, miscarriages, and hydatidiform moles. European Journal of Human Genetics, 2018, 26, 1007-1013.	2.8	69
35	Causative Mutations and Mechanism of Androgenetic Hydatidiform Moles. American Journal of Human Genetics, 2018, 103, 740-751.	6.2	69
36	TRPV4 and KRAS and FGFR1 gain-of-function mutations drive giant cell lesions of the jaw. Nature Communications, 2018, 9, 4572.	12.8	58

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37	Germline HAVCR2 mutations altering TIM-3 characterize subcutaneous panniculitis-like T cell lymphomas with hemophagocytic lymphohistiocytic syndrome. Nature Genetics, 2018, 50, 1650-1657.	21.4	151
38	ClinPred: Prediction Tool to Identify Disease-Relevant Nonsynonymous Single-Nucleotide Variants. American Journal of Human Genetics, 2018, 103, 474-483.	6.2	149
39	Methylome analysis and whole-exome sequencing reveal that brain tumors associated with encephalocraniocutaneous lipomatosis are midline pilocytic astrocytomas. Acta Neuropathologica, 2018, 136, 657-660.	7.7	18
40	Une Ã©pimutation transgÃ©nÃ©rationnelle du gÃ¨ne MMACHC produit un nouveau type dâ€™erreur innÃ©e du mÃ©tabolisme dÃ©nommÃ©e Ã©pi-cblC. Bulletin De L'Academie Nationale De Medecine, 2018, 202, 1585-1596.	0.0	0
41	Identification and functional characterization of a novel MTFMT mutation associated with selective vulnerability of the visual pathway and a mild neurological phenotype. Neurogenetics, 2017, 18, 97-103.	1.4	11
42	Impaired H3K36 methylation defines a subset of head and neck squamous cell carcinomas. Nature Genetics, 2017, 49, 180-185.	21.4	195
43	Matchmaking facilitates the diagnosis of an autosomal-recessive mitochondrial disease caused by biallelic mutation of the tRNA isopentenyltransferase (TRIT1) gene. Human Mutation, 2017, 38, 511-516.	2.5	39
44	Spatial heterogeneity in medulloblastoma. Nature Genetics, 2017, 49, 780-788.	21.4	112
45	Expansion of the clinical phenotype of the distal 10q26.3 deletion syndrome to include ataxia and hyperemia of the hands and feet. American Journal of Medical Genetics, Part A, 2017, 173, 1611-1619.	1.2	4
46	Compound heterozygous mutations in the gene PIGP are associated with early infantile epileptic encephalopathy. Human Molecular Genetics, 2017, 26, 1706-1715.	2.9	39
47	H3.1 K36M mutation in a congenitalâ€onset soft tissue neoplasm. Pediatric Blood and Cancer, 2017, 64, e26633.	1.5	7
48	Functionally Null RAD51D Missense Mutation Associates Strongly with Ovarian Carcinoma. Cancer Research, 2017, 77, 4517-4529.	0.9	34
49	Bmp signaling maintains a mesoderm progenitor cell state in the mouse tailbud. Development (Cambridge), 2017, 144, 2982-2993.	2.5	10
50	Recessive mutations in the kinase ZAK cause a congenital myopathy with fibre type disproportion. Brain, 2017, 140, 37-48.	7.6	28
51	RMND1-Related Leukoencephalopathy With Temporal Lobe Cysts and Hearing Lossâ€”Another Mendelian Mimicker of Congenital Cytomegalovirus Infection. Pediatric Neurology, 2017, 66, 59-62.	2.1	12
52	USP15 regulates type I interferon response and is required for pathogenesis of neuroinflammation. Nature Immunology, 2017, 18, 54-63.	14.5	90
53	Longitudinal mutational analysis of a cerebellar pilocytic astrocytoma recurring as a ganglioglioma. Pediatric Blood and Cancer, 2017, 64, 275-278.	1.5	4
54	A novel multisystem disease associated with recessive mutations in the tyrosylâ€tRNA synthetase (YARS) gene. American Journal of Medical Genetics, Part A, 2017, 173, 126-134.	1.2	36

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55	Novel 25 kb Deletion of MERTK Causes Retinitis Pigmentosa With Severe Progression. , 2017, 58, 1736.		17
56	Characterizing temporal genomic heterogeneity in pediatric high-grade gliomas. Acta Neuropathologica Communications, 2017, 5, 78.	5.2	48
57	The Genetic Causes of Nonsyndromic Congenital Retinal Detachment: A Genetic and Phenotypic Study of Pakistani Families. , 2017, 58, 1028.		15
58	LoLoPicker: detecting low allelic-fraction variants from low-quality cancer samples. Oncotarget, 2017, 8, 37032-37040.	1.8	23
59	Integrated (epi)-Genomic Analyses Identify Subgroup-Specific Therapeutic Targets in CNS Rhabdoid Tumors. Cancer Cell, 2016, 30, 891-908.	16.8	191
60	Gain-of-function mutation inTRPV4identified in patients with osteonecrosis of the femoral head. Journal of Medical Genetics, 2016, 53, 705-709.	3.2	20
61	<scp>SLC</scp>25A46 is required for mitochondrial lipid homeostasis and cristae maintenance and is responsible for Leigh syndrome. EMBO Molecular Medicine, 2016, 8, 1019-1038.	6.9	141
62	Biallelic Mutations in PDE10A Lead to Loss of Striatal PDE10A and a Hyperkinetic Movement Disorder with Onset in Infancy. American Journal of Human Genetics, 2016, 98, 735-743.	6.2	65
63	Mutations in GALC cause late-onset Krabbe disease with predominant cerebellar ataxia. Neurogenetics, 2016, 17, 137-141.	1.4	16
64	Histone H3K36 mutations promote sarcomagenesis through altered histone methylation landscape. Science, 2016, 352, 844-849.	12.6	327
65	Inborn Error of Cobalamin Metabolism Associated with the Intracellular Accumulation of Transcobalamin-Bound Cobalamin and Mutations inZNF143, Which Codes for a Transcriptional Activator. Human Mutation, 2016, 37, 976-982.	2.5	30
66	Spatial and temporal homogeneity of driver mutations in diffuse intrinsic pontine glioma. Nature Communications, 2016, 7, 11185.	12.8	197
67	Next-generation sequencing for diagnosis of rare diseases in the neonatal intensive care unit. Cmaj, 2016, 188, E254-E260.	2.0	86
68	Syndrome disintegration: Exome sequencing reveals that Fitzsimmons syndrome is a coâ€occurrence of multiple events. American Journal of Medical Genetics, Part A, 2016, 170, 1820-1825.	1.2	19
69	DNM1L-related mitochondrial fission defect presenting as refractory epilepsy. European Journal of Human Genetics, 2016, 24, 1084-1088.	2.8	113
70	SPG7 mutations explain a significant proportion of French Canadian spastic ataxia cases. European Journal of Human Genetics, 2016, 24, 1016-1021.	2.8	46
71	Autosomal recessive cerebellar ataxia caused by a homozygous mutation inPMPCA. Brain, 2016, 139, e19-e19.	7.6	27
72	Germline and somatic FGFR1 abnormalities in dysembryoplastic neuroepithelial tumors. Acta Neuropathologica, 2016, 131, 847-863.	7.7	143

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73	Mosaic Activating Mutations in FGFR1 Cause Encephalocraniocutaneous Lipomatosis. American Journal of Human Genetics, 2016, 98, 579-587.	6.2	88
74	Molecular analyses reveal close similarities between small cell carcinoma of the ovary, hypercalcemic type and atypical teratoid/rhabdoid tumor. Oncotarget, 2016, 7, 1732-1740.	1.8	42
75	GeneMatcher Aids in the Identification of a New Malformation Syndrome with Intellectual Disability, Unique Facial Dysmorphisms, and Skeletal and Connective Tissue Abnormalities Caused by De Novo Variants inHNRNPK. Human Mutation, 2015, 36, 1009-1014.	2.5	56
76	Choroideremia Is a Systemic Disease With Lymphocyte Crystals and Plasma Lipid and RBC Membrane Abnormalities. , 2015, 56, 8158.		20
77	Recessive Osteogenesis Imperfecta Caused by Missense Mutations in SPARC. American Journal of Human Genetics, 2015, 96, 979-985.	6.2	107
78	Homozygosity for Frameshift Mutations in XYLT2 Result in a Spondylo-Ocular Syndrome with Bone Fragility, Cataracts, and Hearing Defects. American Journal of Human Genetics, 2015, 96, 971-978.	6.2	65
79	Nonsense mutation in theWDR73gene is associated with Galloway-Mowat syndrome. Journal of Medical Genetics, 2015, 52, 381-390.	3.2	36
80	Meconium ileus in a Lebanese family secondary to mutations in the GUCY2C gene. European Journal of Human Genetics, 2015, 23, 990-992.	2.8	24
81	Autosomal-Recessive Intellectual Disability with Cerebellar Atrophy Syndrome Caused by Mutation of the Manganese and Zinc Transporter Gene SLC39A8. American Journal of Human Genetics, 2015, 97, 886-893.	6.2	171
82	Cole-Carpenter Syndrome Is Caused by a Heterozygous Missense Mutation in P4HB. American Journal of Human Genetics, 2015, 96, 425-431.	6.2	92
83	Biallelic Mutations in BRCA1 Cause a New Fanconi Anemia Subtype. Cancer Discovery, 2015, 5, 135-142.	9.4	251
84	Homozygous mutations inMFN2cause multiple symmetric lipomatosis associated with neuropathy. Human Molecular Genetics, 2015, 24, 5109-5114.	2.9	61
85	Germline RECQL mutations are associated with breast cancer susceptibility. Nature Genetics, 2015, 47, 643-646.	21.4	168
86	Whole exome sequencing identifies the TNNI3K gene as a cause of familial conduction system disease and congenital junctional ectopic tachycardia. International Journal of Cardiology, 2015, 185, 114-116.	1.7	29
87	Atypical fibrodysplasia ossificans progressiva diagnosed by wholeâ€exome sequencing. American Journal of Medical Genetics, Part A, 2015, 167, 1337-1341.	1.2	11
88	The clinical application of genome-wide sequencing for monogenic diseases in Canada: Position Statement of the Canadian College of Medical Geneticists. Journal of Medical Genetics, 2015, 52, 431-437.	3.2	187
89	THEMIS Is Required for Pathogenesis of Cerebral Malaria and Protection against Pulmonary Tuberculosis. Infection and Immunity, 2015, 83, 759-768.	2.2	26
90	Rare variants inSOS2andLZTR1are associated with Noonan syndrome. Journal of Medical Genetics, 2015, 52, 413-421.	3.2	187

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91	Congenital Visual Impairment and Progressive Microcephaly Due to Lysylâ€“Transfer Ribonucleic Acid (RNA) Synthetase (KARS) Mutations. Journal of Child Neurology, 2015, 30, 1037-1043.	1.4	47
92	A novel CCBE1 mutation leading to a mild form of hennekam syndrome: case report and review of the literature. BMC Medical Genetics, 2015, 16, 28.	2.1	14
93	Autosomal recessive axonal polyneuropathy in a sibling pair due to a novel homozygous mutation in IGHMBP2. Neuromuscular Disorders, 2015, 25, 794-799.	0.6	16
94	Loss-of-Function Mutation in APC2 Causes Sotos Syndrome Features. Cell Reports, 2015, 10, 1585-1598.	6.4	40
95	Homozygous mutation in the eukaryotic translation initiation factor 2alpha phosphatase gene,PPP1R15B, is associated with severe microcephaly, short stature and intellectual disability. Human Molecular Genetics, 2015, 24, 6293-6300.	2.9	36
96	RTTN Mutations Cause Primary Microcephaly and Primordial Dwarfism in Humans. American Journal of Human Genetics, 2015, 97, 862-868.	6.2	36
97	ExomeAI: detection of recurrent allelic imbalance in tumors using whole-exome sequencing data. Bioinformatics, 2015, 31, 429-431.	4.1	18
98	An inherited immunoglobulin class-switch recombination deficiency associated with a defect in the INO80 chromatin remodeling complex. Journal of Allergy and Clinical Immunology, 2015, 135, 998-1007.e6.	2.9	37
99	A polyadenylation site variant causes transcript-specific BMP1 deficiency and frequent fractures in children. Human Molecular Genetics, 2015, 24, 516-524.	2.9	37
100	Homozygous nonsense mutation in SYNJ1 associated with intractable epilepsy and tau pathology. Neurobiology of Aging, 2015, 36, 1222.e1-1222.e5.	3.1	50
101	Mutations in NFKB2and potential genetic heterogeneity in patients with DAVID syndrome, having variable endocrine and immune deficiencies. BMC Medical Genetics, 2014, 15, 139.	2.1	84
102	Recurrent somatic mutations in ACVR1 in pediatric midline high-grade astrocytoma. Nature Genetics, 2014, 46, 462-466.	21.4	381
103	Germline Mutations in MAP3K6 Are Associated with Familial Gastric Cancer. PLoS Genetics, 2014, 10, e1004669.	3.5	57
104	Disrupted auto-regulation of the spliceosomal gene SNRPB causes cerebroâ€“costoâ€“mandibular syndrome. Nature Communications, 2014, 5, 4483.	12.8	57
105	Epigenetic dysregulation: a novel pathway of oncogenesis in pediatric brain tumors. Acta Neuropathologica, 2014, 128, 615-627.	7.7	49
106	Phenotypic Overlap Between Familial Exudative Vitreoretinopathy and Microcephaly, Lymphedema, and Chorioretinal Dysplasia Caused byKIF11Mutations. JAMA Ophthalmology, 2014, 132, 1393.	2.5	95
107	Paediatric and adult glioblastoma: multiform (epi)genomic culprits emerge. Nature Reviews Cancer, 2014, 14, 92-107.	28.4	469
108	Fusion of TTYH1 with the C19MC microRNA cluster drives expression of a brain-specific DNMT3B isoform in the embryonal brain tumor ETMR. Nature Genetics, 2014, 46, 39-44.	21.4	167

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109	CCDC88B is a novel regulator of maturation and effector functions of T cells during pathological inflammation. Journal of Experimental Medicine, 2014, 211, 2519-2535.	8.5	44
110	Germline and somatic SMARCA4 mutations characterize small cell carcinoma of the ovary, hypercalcemic type. Nature Genetics, 2014, 46, 438-443.	21.4	383
111	Genetically encoded impairment of neuronal <scp>KCC</scp> 2 cotransporter function in human idiopathic generalized epilepsy. EMBO Reports, 2014, 15, 766-774.	4.5	163
112	CARD9 Deficiency and Spontaneous Central Nervous System Candidiasis: Complete Clinical Remission With GM-CSF Therapy. Clinical Infectious Diseases, 2014, 59, 81-84.	5.8	153
113	Mutations in LAMA1 Cause Cerebellar Dysplasia and Cysts with and without Retinal Dystrophy. American Journal of Human Genetics, 2014, 95, 227-234.	6.2	92
114	Mutations in the enzyme glutathione peroxidase 4 cause Sedaghatian-type spondylometaphyseal dysplasia. Journal of Medical Genetics, 2014, 51, 470-474.	3.2	64
115	FORGE Canada Consortium: Outcomes of a 2-Year National Rare-Disease Gene-Discovery Project. American Journal of Human Genetics, 2014, 94, 809-817.	6.2	219
116	Recurrent somatic alterations of FGFR1 and NTRK2 in pilocytic astrocytoma. Nature Genetics, 2013, 45, 927-932.	21.4	674
117	Mutations in PIK3R1 Cause SHORT Syndrome. American Journal of Human Genetics, 2013, 93, 158-166.	6.2	156
118	Mutations in SETD2 and genes affecting histone H3K36 methylation target hemispheric high-grade gliomas. Acta Neuropathologica, 2013, 125, 659-669.	7.7	250
119	Frequent ATRX mutations and loss of expression in adult diffuse astrocytic tumors carrying IDH1/IDH2 and TP53 mutations. Acta Neuropathologica, 2012, 124, 615-625.	7.7	376
120	K27M mutation in histone H3.3 defines clinically and biologically distinct subgroups of pediatric diffuse intrinsic pontine gliomas. Acta Neuropathologica, 2012, 124, 439-447.	7.7	799
121	Hotspot Mutations in H3F3A and IDH1 Define Distinct Epigenetic and Biological Subgroups of Glioblastoma. Cancer Cell, 2012, 22, 425-437.	16.8	1,551
122	Driver mutations in histone H3.3 and chromatin remodelling genes in paediatric glioblastoma. Nature, 2012, 482, 226-231.	27.8	2,129
123	Exome and whole-genome sequencing for gene discovery: The future is now!. Human Mutation, 2012, 33, 591-592.	2.5	12
124	Identification of novel genes causing autosomal recessive disorders in Qatari population using whole exome sequencing. , 2012, , .		0
125	The study of eQTL variations by RNA-seq: from SNPs to phenotypes. Trends in Genetics, 2011, 27, 72-79.	6.7	216
126	Mutations in NOTCH2 in families with Hajdu-Cheney syndrome. Human Mutation, 2011, 32, 1114-1117.	2.5	93

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127	A new ocular phenotype associated with an unexpected but known systemic disorder and mutation: novel use of genomic diagnostics and exome sequencing. Journal of Medical Genetics, 2011, 48, 593-596.	3.2	44
128	Fine-Scale Variation and Genetic Determinants of Alternative Splicing across Individuals. PLoS Genetics, 2009, 5, e1000766.	3.5	81
129	Global patterns of cis variation in human cells revealed by high-density allelic expression analysis. Nature Genetics, 2009, 41, 1216-1222.	21.4	206
130	Genome-wide analysis of transcript isoform variation in humans. Nature Genetics, 2008, 40, 225-231.	21.4	283
131	Effect of polymorphisms within probeâ€"target sequences on olignonucleotide microarray experiments. Nucleic Acids Research, 2008, 36, 4417-4423.	14.5	59
132	Heritability of alternative splicing in the human genome. Genome Research, 2007, 17, 1210-1218.	5.5	101
133	Distribution and Characterization of Regulatory Elements in the Human Genome. Genome Research, 2002, 12, 1827-1836.	5.5	261