

# Jacek Majewski

## List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

129 papers	11,592 citations	49 h-index	107 g-index
137 ext. papers	13,936 ext. citations	11.3 avg, IF	5.45 L-index

#	Paper	IF	Citations
129	Epimutations in both the TESK2 and MMACHC promoters in the Epi-cblC inherited disorder of intracellular metabolism of vitamin B.. <i>Clinical Epigenetics</i> , <b>2022</b> , 14, 52	7.7	1
128	Inherent genomic properties underlie the epigenomic heterogeneity of human induced pluripotent stem cells. <i>Cell Reports</i> , <b>2021</b> , 37, 109909	10.6	0
127	Two competing mechanisms of DNMT3A recruitment regulate the dynamics of de novo DNA methylation at PRC1-targeted CpG islands. <i>Nature Genetics</i> , <b>2021</b> , 53, 794-800	36.3	17
126	Whole-exome sequencing reveals novel vacuolar ATPase genes' variants and variants in genes involved in lysosomal biology and autophagosomal formation in oral granular cell tumors. <i>Journal of Oral Pathology and Medicine</i> , <b>2021</b> , 50, 410-417	3.3	3
125	Depletion of H3K36me2 recapitulates epigenomic and phenotypic changes induced by the H3.3K36M oncohistone mutation. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2021</b> , 118,	11.5	10
124	Mutation in Eftud2 causes craniofacial defects in mice via mis-splicing of Mdm2 and increased P53. <i>Human Molecular Genetics</i> , <b>2021</b> , 30, 739-757	5.6	3
123	Chromatin dysregulation associated with NSD1 mutation in head and neck squamous cell carcinoma. <i>Cell Reports</i> , <b>2021</b> , 34, 108769	10.6	9
122	Novel pathogenic variants in NLRP7, NLRP5, and PADI6 in patients with recurrent hydatidiform moles and reproductive failure. <i>Clinical Genetics</i> , <b>2021</b> , 99, 823-828	4	5
121	A functionally impaired missense variant identified in French Canadian families implicates FANCI as a candidate ovarian cancer-predisposing gene. <i>Genome Medicine</i> , <b>2021</b> , 13, 186	14.4	2
120	Early infantile epileptic encephalopathy due to biallelic pathogenic variants in PIGQ: Report of seven new subjects and review of the literature. <i>Journal of Inherited Metabolic Disease</i> , <b>2020</b> , 43, 1321-1332	5.4	6
119	A novel pathogenic missense ADAMTS17 variant that impairs secretion causes Weill-Marchesani Syndrome with variably dysmorphic hand features. <i>Scientific Reports</i> , <b>2020</b> , 10, 10827	4.9	6
118	Latency and interval therapy affect the evolution in metastatic colorectal cancer. <i>Scientific Reports</i> , <b>2020</b> , 10, 581	4.9	4
117	ZBTB7B (ThPOK) Is Required for Pathogenesis of Cerebral Malaria and Protection against Pulmonary Tuberculosis. <i>Infection and Immunity</i> , <b>2020</b> , 88,	3.7	2
116	A Mouse Model for Cerebro-Costo-Mandibular Syndrome (CCMS) with an Intronic Deletion in SnrpB. <i>FASEB Journal</i> , <b>2020</b> , 34, 1-1	0.9	
115	Deletion of Mouse Sf3b4 in Neural Crest Cells Causes Craniofacial Abnormalities. <i>FASEB Journal</i> , <b>2020</b> , 34, 1-1	0.9	
114	H3K27M in Gliomas Causes a One-Step Decrease in H3K27 Methylation and Reduced Spreading within the Constraints of H3K36 Methylation. <i>Cell Reports</i> , <b>2020</b> , 33, 108390	10.6	14
113	Prognostic and predictive value of circulating tumor DNA during neoadjuvant chemotherapy for triple negative breast cancer. <i>Scientific Reports</i> , <b>2020</b> , 10, 14704	4.9	12

112	Exome sequencing study of partial agenesis of the corpus callosum in men with developmental delay, epilepsy, and microcephaly. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2020</b> , 8, e992	2.3	6
111	The histone mark H3K36me2 recruits DNMT3A and shapes the intergenic DNA methylation landscape. <i>Nature</i> , <b>2019</b> , 573, 281-286	50.4	161
110	Epigenomic Reordering Induced by Polycomb Loss Drives Oncogenesis but Leads to Therapeutic Vulnerabilities in Malignant Peripheral Nerve Sheath Tumors. <i>Cancer Research</i> , <b>2019</b> , 79, 3205-3219	10.1	23
109	H3K27M induces defective chromatin spread of PRC2-mediated repressive H3K27me2/me3 and is essential for glioma tumorigenesis. <i>Nature Communications</i> , <b>2019</b> , 10, 1262	17.4	104
108	CYRI/FAM49B negatively regulates RAC1-driven cytoskeletal remodelling and protects against bacterial infection. <i>Nature Microbiology</i> , <b>2019</b> , 4, 1516-1531	26.6	18
107	Circulating tumor DNA (ctDNA) during and after neoadjuvant chemotherapy and prior to surgery is a powerful prognostic factor in triple-negative breast cancer (TNBC).. <i>Journal of Clinical Oncology</i> , <b>2019</b> , 37, 594-594	2.2	5
106	EFTUD2, The Gene Responsible For Mandibulofacial Dysostosis With Microcephaly (MFDN), Is Required For Implantation And Craniofacial Development In Mouse. <i>FASEB Journal</i> , <b>2019</b> , 33, 774.20	0.9	
105	A Unique Morphological Phenotype in Chemoresistant Triple-Negative Breast Cancer Reveals Metabolic Reprogramming and PLIN4 Expression as a Molecular Vulnerability. <i>Molecular Cancer Research</i> , <b>2019</b> , 17, 2492-2507	6.6	31
104	Recessive mutation in CD2AP causes focal segmental glomerulosclerosis in humans and mice. <i>Kidney International</i> , <b>2019</b> , 95, 57-61	9.9	8
103	Biallelic Loss-of-Function Variants in AIMP1 Cause a Rare Neurodegenerative Disease. <i>Journal of Child Neurology</i> , <b>2019</b> , 34, 74-80	2.5	5
102	POLR3A variants in hereditary spastic paraplegia and ataxia. <i>Brain</i> , <b>2018</b> , 141, e1	11.2	14
101	APRX1 mutant allele causes a MMACHC secondary epimutation in cblC patients. <i>Nature Communications</i> , <b>2018</b> , 9, 67	17.4	45
100	Biallelic PADI6 variants linking infertility, miscarriages, and hydatidiform moles. <i>European Journal of Human Genetics</i> , <b>2018</b> , 26, 1007-1013	5.3	43
99	Methylome analysis and whole-exome sequencing reveal that brain tumors associated with encephalocraniocutaneous lipomatosis are midline pilocytic astrocytomas. <i>Acta Neuropathologica</i> , <b>2018</b> , 136, 657-660	14.3	12
98	Une bmutation transgationnelle du gne MMACHC produit un nouveau type d'erreur inn du mabolisme d'homme bi-cblC. <i>Bulletin De L'Academie Nationale De Medecine</i> , <b>2018</b> , 202, 1585-1596	0.1	
97	Causative Mutations and Mechanism of Androgenetic Hydatidiform Moles. <i>American Journal of Human Genetics</i> , <b>2018</b> , 103, 740-751	11	39
96	TRPV4 and KRAS and FGFR1 gain-of-function mutations drive giant cell lesions of the jaw. <i>Nature Communications</i> , <b>2018</b> , 9, 4572	17.4	30
95	Germline HAVCR2 mutations altering TIM-3 characterize subcutaneous panniculitis-like T cell lymphomas with hemophagocytic lymphohistiocytic syndrome. <i>Nature Genetics</i> , <b>2018</b> , 50, 1650-1657	36.3	74

94	ClinPred: Prediction Tool to Identify Disease-Relevant Nonsynonymous Single-Nucleotide Variants. <i>American Journal of Human Genetics</i> , <b>2018</b> , 103, 474-483	11	61
93	Identification and functional characterization of a novel MTFMT mutation associated with selective vulnerability of the visual pathway and a mild neurological phenotype. <i>Neurogenetics</i> , <b>2017</b> , 18, 97-103	3	9
92	Impaired H3K36 methylation defines a subset of head and neck squamous cell carcinomas. <i>Nature Genetics</i> , <b>2017</b> , 49, 180-185	36.3	132
91	Matchmaking facilitates the diagnosis of an autosomal-recessive mitochondrial disease caused by biallelic mutation of the tRNA isopentenyltransferase (TRIT1) gene. <i>Human Mutation</i> , <b>2017</b> , 38, 511-516	4.7	29
90	Spatial heterogeneity in medulloblastoma. <i>Nature Genetics</i> , <b>2017</b> , 49, 780-788	36.3	80
89	Expansion of the clinical phenotype of the distal 10q26.3 deletion syndrome to include ataxia and hyperemia of the hands and feet. <i>American Journal of Medical Genetics, Part A</i> , <b>2017</b> , 173, 1611-1619	2.5	3
88	Compound heterozygous mutations in the gene PIGP are associated with early infantile epileptic encephalopathy. <i>Human Molecular Genetics</i> , <b>2017</b> , 26, 1706-1715	5.6	33
87	Novel 25 kb Deletion of MERTK Causes Retinitis Pigmentosa With Severe Progression <b>2017</b> , 58, 1736-1742		11
86	Characterizing temporal genomic heterogeneity in pediatric high-grade gliomas. <i>Acta Neuropathologica Communications</i> , <b>2017</b> , 5, 78	7.3	23
85	The Genetic Causes of Nonsyndromic Congenital Retinal Detachment: A Genetic and Phenotypic Study of Pakistani Families <b>2017</b> , 58, 1028-1036		7
84	H3.1 K36M mutation in a congenital-onset soft tissue neoplasm. <i>Pediatric Blood and Cancer</i> , <b>2017</b> , 64, e26633	3	3
83	Functionally Null Missense Mutation Associates Strongly with Ovarian Carcinoma. <i>Cancer Research</i> , <b>2017</b> , 77, 4517-4529	10.1	18
82	Bmp signaling maintains a mesoderm progenitor cell state in the mouse tailbud. <i>Development (Cambridge)</i> , <b>2017</b> , 144, 2982-2993	6.6	8
81	Recessive mutations in the kinase ZAK cause a congenital myopathy with fibre type disproportion. <i>Brain</i> , <b>2017</b> , 140, 37-48	11.2	13
80	RMND1-Related Leukoencephalopathy With Temporal Lobe Cysts and Hearing Loss-Another Mendelian Mimicker of Congenital Cytomegalovirus Infection. <i>Pediatric Neurology</i> , <b>2017</b> , 66, 59-62	2.9	10
79	USP15 regulates type I interferon response and is required for pathogenesis of neuroinflammation. <i>Nature Immunology</i> , <b>2017</b> , 18, 54-63	19.1	51
78	Longitudinal mutational analysis of a cerebellar pilocytic astrocytoma recurring as a ganglioglioma. <i>Pediatric Blood and Cancer</i> , <b>2017</b> , 64, 275-278	3	4
77	A novel multisystem disease associated with recessive mutations in the tyrosyl-tRNA synthetase (YARS) gene. <i>American Journal of Medical Genetics, Part A</i> , <b>2017</b> , 173, 126-134	2.5	27

76	LoLoPicker: detecting low allelic-fraction variants from low-quality cancer samples. <i>Oncotarget</i> , <b>2017</b> , 8, 37032-37040	3.3	17
75	Spatial and temporal homogeneity of driver mutations in diffuse intrinsic pontine glioma. <i>Nature Communications</i> , <b>2016</b> , 7, 11185	17.4	152
74	Next-generation sequencing for diagnosis of rare diseases in the neonatal intensive care unit. <i>Cmaj</i> , <b>2016</b> , 188, E254-E260	3.5	59
73	Syndrome disintegration: Exome sequencing reveals that Fitzsimmons syndrome is a co-occurrence of multiple events. <i>American Journal of Medical Genetics, Part A</i> , <b>2016</b> , 170, 1820-5	2.5	14
72	DNM1L-related mitochondrial fission defect presenting as refractory epilepsy. <i>European Journal of Human Genetics</i> , <b>2016</b> , 24, 1084-8	5.3	93
71	SPG7 mutations explain a significant proportion of French Canadian spastic ataxia cases. <i>European Journal of Human Genetics</i> , <b>2016</b> , 24, 1016-21	5.3	38
70	Autosomal recessive cerebellar ataxia caused by a homozygous mutation in PMPCA. <i>Brain</i> , <b>2016</b> , 139, e19	11.2	17
69	Germline and somatic FGFR1 abnormalities in dysembryoplastic neuroepithelial tumors. <i>Acta Neuropathologica</i> , <b>2016</b> , 131, 847-63	14.3	105
68	Mosaic Activating Mutations in FGFR1 Cause Encephalocraniocutaneous Lipomatosis. <i>American Journal of Human Genetics</i> , <b>2016</b> , 98, 579-587	11	60
67	Molecular analyses reveal close similarities between small cell carcinoma of the ovary, hypercalcemic type and atypical teratoid/rhabdoid tumor. <i>Oncotarget</i> , <b>2016</b> , 7, 1732-40	3.3	36
66	Integrated (epi)-Genomic Analyses Identify Subgroup-Specific Therapeutic Targets in CNS Rhabdoid Tumors. <i>Cancer Cell</i> , <b>2016</b> , 30, 891-908	24.3	135
65	Gain-of-function mutation in TRPV4 identified in patients with osteonecrosis of the femoral head. <i>Journal of Medical Genetics</i> , <b>2016</b> , 53, 705-9	5.8	19
64	SLC25A46 is required for mitochondrial lipid homeostasis and cristae maintenance and is responsible for Leigh syndrome. <i>EMBO Molecular Medicine</i> , <b>2016</b> , 8, 1019-38	12	105
63	Biallelic Mutations in PDE10A Lead to Loss of Striatal PDE10A and a Hyperkinetic Movement Disorder with Onset in Infancy. <i>American Journal of Human Genetics</i> , <b>2016</b> , 98, 735-43	11	48
62	Mutations in GALC cause late-onset Krabbe disease with predominant cerebellar ataxia. <i>Neurogenetics</i> , <b>2016</b> , 17, 137-41	3	11
61	Histone H3K36 mutations promote sarcomagenesis through altered histone methylation landscape. <i>Science</i> , <b>2016</b> , 352, 844-9	33.3	219
60	Inborn Error of Cobalamin Metabolism Associated with the Intracellular Accumulation of Transcobalamin-Bound Cobalamin and Mutations in ZNF143, Which Codes for a Transcriptional Activator. <i>Human Mutation</i> , <b>2016</b> , 37, 976-82	4.7	21
59	Biallelic mutations in BRCA1 cause a new Fanconi anemia subtype. <i>Cancer Discovery</i> , <b>2015</b> , 5, 135-42	24.4	215

58	Homozygous mutations in MFN2 cause multiple symmetric lipomatosis associated with neuropathy. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 5109-14	5.6	53
57	Germline RECQL mutations are associated with breast cancer susceptibility. <i>Nature Genetics</i> , <b>2015</b> , 47, 643-6	36.3	123
56	Whole exome sequencing identifies the TNNI3K gene as a cause of familial conduction system disease and congenital junctional ectopic tachycardia. <i>International Journal of Cardiology</i> , <b>2015</b> , 185, 114-6	3.2	18
55	Atypical fibrodysplasia ossificans progressiva diagnosed by whole-exome sequencing. <i>American Journal of Medical Genetics, Part A</i> , <b>2015</b> , 167, 1337-41	2.5	10
54	The clinical application of genome-wide sequencing for monogenic diseases in Canada: Position Statement of the Canadian College of Medical Geneticists. <i>Journal of Medical Genetics</i> , <b>2015</b> , 52, 431-7	5.8	137
53	THEMIS is required for pathogenesis of cerebral malaria and protection against pulmonary tuberculosis. <i>Infection and Immunity</i> , <b>2015</b> , 83, 759-68	3.7	14
52	Rare variants in SOS2 and LZTR1 are associated with Noonan syndrome. <i>Journal of Medical Genetics</i> , <b>2015</b> , 52, 413-21	5.8	144
51	Congenital Visual Impairment and Progressive Microcephaly Due to Lysyl-Transfer Ribonucleic Acid (RNA) Synthetase (KARS) Mutations: The Expanding Phenotype of Aminoacyl-Transfer RNA Synthetase Mutations in Human Disease. <i>Journal of Child Neurology</i> , <b>2015</b> , 30, 1037-43	2.5	39
50	A novel CCBE1 mutation leading to a mild form of hennekam syndrome: case report and review of the literature. <i>BMC Medical Genetics</i> , <b>2015</b> , 16, 28	2.1	12
49	Autosomal recessive axonal polyneuropathy in a sibling pair due to a novel homozygous mutation in IGHMBP2. <i>Neuromuscular Disorders</i> , <b>2015</b> , 25, 794-9	2.9	10
48	Loss-of-Function Mutation in APC2 Causes Sotos Syndrome Features. <i>Cell Reports</i> , <b>2015</b> , 10, 1585-1598	10.6	28
47	Homozygous mutation in the eukaryotic translation initiation factor 2alpha phosphatase gene, PPP1R15B, is associated with severe microcephaly, short stature and intellectual disability. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 6293-300	5.6	28
46	RTTN Mutations Cause Primary Microcephaly and Primordial Dwarfism in Humans. <i>American Journal of Human Genetics</i> , <b>2015</b> , 97, 862-8	11	29
45	ExomeAI: detection of recurrent allelic imbalance in tumors using whole-exome sequencing data. <i>Bioinformatics</i> , <b>2015</b> , 31, 429-31	7.2	14
44	An inherited immunoglobulin class-switch recombination deficiency associated with a defect in the INO80 chromatin remodeling complex. <i>Journal of Allergy and Clinical Immunology</i> , <b>2015</b> , 135, 998-1007.e6	11.5	30
43	A polyadenylation site variant causes transcript-specific BMP1 deficiency and frequent fractures in children. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 516-24	5.6	32
42	Homozygous nonsense mutation in SYNJ1 associated with intractable epilepsy and tau pathology. <i>Neurobiology of Aging</i> , <b>2015</b> , 36, 1222.e1-5	5.6	35
41	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 in HNRNPK. <i>Human Mutation</i> , <b>2015</b> , 36, 1009-1014	4.7	45



40	Choroideremia Is a Systemic Disease With Lymphocyte Crystals and Plasma Lipid and RBC Membrane Abnormalities <b>2015</b> , 56, 8158-65		14
39	Recessive osteogenesis imperfecta caused by missense mutations in SPARC. <i>American Journal of Human Genetics</i> , <b>2015</b> , 96, 979-85	11	81
38	Homozygosity for frameshift mutations in XYLT2 result in a spondylo-ocular syndrome with bone fragility, cataracts, and hearing defects. <i>American Journal of Human Genetics</i> , <b>2015</b> , 96, 971-8	11	50
37	Nonsense mutation in the WDR73 gene is associated with Galloway-Mowat syndrome. <i>Journal of Medical Genetics</i> , <b>2015</b> , 52, 381-90	5.8	23
36	Meconium ileus in a Lebanese family secondary to mutations in the GUCY2C gene. <i>European Journal of Human Genetics</i> , <b>2015</b> , 23, 990-2	5.3	17
35	Autosomal-Recessive Intellectual Disability with Cerebellar Atrophy Syndrome Caused by Mutation of the Manganese and Zinc Transporter Gene SLC39A8. <i>American Journal of Human Genetics</i> , <b>2015</b> , 97, 886-93	11	125
34	Cole-Carpenter syndrome is caused by a heterozygous missense mutation in P4HB. <i>American Journal of Human Genetics</i> , <b>2015</b> , 96, 425-31	11	65
33	Paediatric and adult glioblastoma: multiform (epi)genomic culprits emerge. <i>Nature Reviews Cancer</i> , <b>2014</b> , 14, 92-107	31.3	383
32	Fusion of TTYH1 with the C19MC microRNA cluster drives expression of a brain-specific DNMT3B isoform in the embryonal brain tumor ETMR. <i>Nature Genetics</i> , <b>2014</b> , 46, 39-44	36.3	131
31	CCDC88B is a novel regulator of maturation and effector functions of T cells during pathological inflammation. <i>Journal of Experimental Medicine</i> , <b>2014</b> , 211, 2519-35	16.6	32
30	Germline and somatic SMARCA4 mutations characterize small cell carcinoma of the ovary, hypercalcemic type. <i>Nature Genetics</i> , <b>2014</b> , 46, 438-43	36.3	305
29	Genetically encoded impairment of neuronal KCC2 cotransporter function in human idiopathic generalized epilepsy. <i>EMBO Reports</i> , <b>2014</b> , 15, 766-74	6.5	123
28	CARD9 deficiency and spontaneous central nervous system candidiasis: complete clinical remission with GM-CSF therapy. <i>Clinical Infectious Diseases</i> , <b>2014</b> , 59, 81-4	11.6	104
27	Mutations in LAMA1 cause cerebellar dysplasia and cysts with and without retinal dystrophy. <i>American Journal of Human Genetics</i> , <b>2014</b> , 95, 227-34	11	74
26	Mutations in the enzyme glutathione peroxidase 4 cause Sedaghatian-type spondylometaphyseal dysplasia. <i>Journal of Medical Genetics</i> , <b>2014</b> , 51, 470-4	5.8	45
25	FORGE Canada Consortium: outcomes of a 2-year national rare-disease gene-discovery project. <i>American Journal of Human Genetics</i> , <b>2014</b> , 94, 809-17	11	174
24	Mutations in NFKB2 and potential genetic heterogeneity in patients with DAVID syndrome, having variable endocrine and immune deficiencies. <i>BMC Medical Genetics</i> , <b>2014</b> , 15, 139	2.1	56
23	Recurrent somatic mutations in ACVR1 in pediatric midline high-grade astrocytoma. <i>Nature Genetics</i> , <b>2014</b> , 46, 462-6	36.3	296

22	Germline mutations in MAP3K6 are associated with familial gastric cancer. <i>PLoS Genetics</i> , <b>2014</b> , 10, e1004669	46
21	Disrupted auto-regulation of the spliceosomal gene SNRPB causes cerebro-costo-mandibular syndrome. <i>Nature Communications</i> , <b>2014</b> , 5, 4483	17.4 40
20	Epigenetic dysregulation: a novel pathway of oncogenesis in pediatric brain tumors. <i>Acta Neuropathologica</i> , <b>2014</b> , 128, 615-27	14.3 29
19	Phenotypic overlap between familial exudative vitreoretinopathy and microcephaly, lymphedema, and chorioretinal dysplasia caused by KIF11 mutations. <i>JAMA Ophthalmology</i> , <b>2014</b> , 132, 1393-9	3.9 73
18	Recurrent somatic alterations of FGFR1 and NTRK2 in pilocytic astrocytoma. <i>Nature Genetics</i> , <b>2013</b> , 45, 927-32	36.3 550
17	Mutations in PIK3R1 cause SHORT syndrome. <i>American Journal of Human Genetics</i> , <b>2013</b> , 93, 158-66	11 125
16	Mutations in SETD2 and genes affecting histone H3K36 methylation target hemispheric high-grade gliomas. <i>Acta Neuropathologica</i> , <b>2013</b> , 125, 659-69	14.3 201
15	Exome and whole-genome sequencing for gene discovery: the future is now!. <i>Human Mutation</i> , <b>2012</b> , 33, 591-2	4.7 11
14	Frequent ATRX mutations and loss of expression in adult diffuse astrocytic tumors carrying IDH1/IDH2 and TP53 mutations. <i>Acta Neuropathologica</i> , <b>2012</b> , 124, 615-25	14.3 295
13	K27M mutation in histone H3.3 defines clinically and biologically distinct subgroups of pediatric diffuse intrinsic pontine gliomas. <i>Acta Neuropathologica</i> , <b>2012</b> , 124, 439-47	14.3 629
12	Hotspot mutations in H3F3A and IDH1 define distinct epigenetic and biological subgroups of glioblastoma. <i>Cancer Cell</i> , <b>2012</b> , 22, 425-37	24.3 1243
11	Driver mutations in histone H3.3 and chromatin remodelling genes in paediatric glioblastoma. <i>Nature</i> , <b>2012</b> , 482, 226-31	50.4 1655
10	The study of eQTL variations by RNA-seq: from SNPs to phenotypes. <i>Trends in Genetics</i> , <b>2011</b> , 27, 72-9	8.5 176
9	Mutations in NOTCH2 in families with Hajdu-Cheney syndrome. <i>Human Mutation</i> , <b>2011</b> , 32, 1114-7	4.7 79
8	A new ocular phenotype associated with an unexpected but known systemic disorder and mutation: novel use of genomic diagnostics and exome sequencing. <i>Journal of Medical Genetics</i> , <b>2011</b> , 48, 593-6	5.8 36
7	Fine-scale variation and genetic determinants of alternative splicing across individuals. <i>PLoS Genetics</i> , <b>2009</b> , 5, e1000766	6 68
6	Global patterns of cis variation in human cells revealed by high-density allelic expression analysis. <i>Nature Genetics</i> , <b>2009</b> , 41, 1216-22	36.3 185
5	Genome-wide analysis of transcript isoform variation in humans. <i>Nature Genetics</i> , <b>2008</b> , 40, 225-31	36.3 264



4	Effect of polymorphisms within probe-target sequences on oligonucleotide microarray experiments. <i>Nucleic Acids Research</i> , <b>2008</b> , 36, 4417-23	20.1	55
3	Heritability of alternative splicing in the human genome. <i>Genome Research</i> , <b>2007</b> , 17, 1210-8	9.7	93
2	Distribution and characterization of regulatory elements in the human genome. <i>Genome Research</i> , <b>2002</b> , 12, 1827-36	9.7	228
1	Nucleome programming is required for the foundation of totipotency in mammalian germline development. <i>EMBO Journal</i> ,	13	1