Jacek Majewski

List of Publications by Citations

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	Driver mutations in histone H3.3 and chromatin remodelling genes in paediatric glioblastoma. *Nature*, **2012**, 482, 226-31	50.4	1655
128	Hotspot mutations in H3F3A and IDH1 define distinct epigenetic and biological subgroups of glioblastoma. *Cancer Cell*, **2012**, 22, 425-37	24.3	1243
127	K27M mutation in histone H3.3 defines clinically and biologically distinct subgroups of pediatric diffuse intrinsic pontine gliomas. *Acta Neuropathologica*, **2012**, 124, 439-47	14.3	629
126	Recurrent somatic alterations of FGFR1 and NTRK2 in pilocytic astrocytoma. *Nature Genetics*, **2013**, 45, 927-32	36.3	550
125	Paediatric and adult glioblastoma: multiform (epi)genomic culprits emerge. *Nature Reviews Cancer*, **2014**, 14, 92-107	31.3	383
124	Germline and somatic SMARCA4 mutations characterize small cell carcinoma of the ovary, hypercalcemic type. *Nature Genetics*, **2014**, 46, 438-43	36.3	305
123	Recurrent somatic mutations in ACVR1 in pediatric midline high-grade astrocytoma. *Nature Genetics*, **2014**, 46, 462-6	36.3	296
122	Frequent ATRX mutations and loss of expression in adult diffuse astrocytic tumors carrying IDH1/IDH2 and TP53 mutations. *Acta Neuropathologica*, **2012**, 124, 615-25	14.3	295
121	Genome-wide analysis of transcript isoform variation in humans. *Nature Genetics*, **2008**, 40, 225-31	36.3	264
120	Distribution and characterization of regulatory elements in the human genome. *Genome Research*, **2002**, 12, 1827-36	9.7	228
119	Histone H3K36 mutations promote sarcomagenesis through altered histone methylation landscape. *Science*, **2016**, 352, 844-9	33.3	219
118	Biallelic mutations in BRCA1 cause a new Fanconi anemia subtype. *Cancer Discovery*, **2015**, 5, 135-42	24.4	215
117	Mutations in SETD2 and genes affecting histone H3K36 methylation target hemispheric high-grade gliomas. *Acta Neuropathologica*, **2013**, 125, 659-69	14.3	201
116	Global patterns of cis variation in human cells revealed by high-density allelic expression analysis. *Nature Genetics*, **2009**, 41, 1216-22	36.3	185
115	The study of eQTL variations by RNA-seq: from SNPs to phenotypes. *Trends in Genetics*, **2011**, 27, 72-9	8.5	176
114	FORGE Canada Consortium: outcomes of a 2-year national rare-disease gene-discovery project. *American Journal of Human Genetics*, **2014**, 94, 809-17	11	174
113	The histone mark H3K36me2 recruits DNMT3A and shapes the intergenic DNA methylation landscape. *Nature*, **2019**, 573, 281-286	50.4	161

112	Spatial and temporal homogeneity of driver mutations in diffuse intrinsic pontine glioma. *Nature Communications*, **2016**, 7, 11185	17.4	152
111	Rare variants in SOS2 and LZTR1 are associated with Noonan syndrome. *Journal of Medical Genetics*, **2015**, 52, 413-21	5.8	144
110	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, 431-7	5.8	137
109	Integrated (epi)-Genomic Analyses Identify Subgroup-Specific Therapeutic Targets in CNS Rhabdoid Tumors. *Cancer Cell*, **2016**, 30, 891-908	24.3	135
108	Impaired H3K36 methylation defines a subset of head and neck squamous cell carcinomas. *Nature Genetics*, **2017**, 49, 180-185	36.3	132
107	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	36.3	131
106	Mutations in PIK3R1 cause SHORT syndrome. *American Journal of Human Genetics*, **2013**, 93, 158-66	11	125
105	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, 886-93	11	125
104	Germline RECQL mutations are associated with breast cancer susceptibility. *Nature Genetics*, **2015**, 47, 643-6	36.3	123
103	Genetically encoded impairment of neuronal KCC2 cotransporter function in human idiopathic generalized epilepsy. *EMBO Reports*, **2014**, 15, 766-74	6.5	123
102	Germline and somatic FGFR1 abnormalities in dysembryoplastic neuroepithelial tumors. *Acta Neuropathologica*, **2016**, 131, 847-63	14.3	105
101	SLC25A46 is required for mitochondrial lipid homeostasis and cristae maintenance and is responsible for Leigh syndrome. *EMBO Molecular Medicine*, **2016**, 8, 1019-38	12	105
100	H3K27M induces defective chromatin spread of PRC2-mediated repressive H3K27me2/me3 and is essential for glioma tumorigenesis. *Nature Communications*, **2019**, 10, 1262	17.4	104
99	CARD9 deficiency and spontaneous central nervous system candidiasis: complete clinical remission with GM-CSF therapy. *Clinical Infectious Diseases*, **2014**, 59, 81-4	11.6	104
98	DNM1L-related mitochondrial fission defect presenting as refractory epilepsy. *European Journal of Human Genetics*, **2016**, 24, 1084-8	5.3	93
97	Heritability of alternative splicing in the human genome. *Genome Research*, **2007**, 17, 1210-8	9.7	93
96	Recessive osteogenesis imperfecta caused by missense mutations in SPARC. *American Journal of Human Genetics*, **2015**, 96, 979-85	11	81
95	Spatial heterogeneity in medulloblastoma. *Nature Genetics*, **2017**, 49, 780-788	36.3	80

94	Mutations in NOTCH2 in families with Hajdu-Cheney syndrome. *Human Mutation*, **2011**, 32, 1114-7	4.7	79
93	Mutations in LAMA1 cause cerebellar dysplasia and cysts with and without retinal dystrophy. *American Journal of Human Genetics*, **2014**, 95, 227-34	11	74
92	Germline HAVCR2 mutations altering TIM-3 characterize subcutaneous panniculitis-like T cell lymphomas with hemophagocytic lymphohistiocytic syndrome. *Nature Genetics*, **2018**, 50, 1650-1657	36.3	74
91	Phenotypic overlap between familial exudative vitreoretinopathy and microcephaly, lymphedema, and chorioretinal dysplasia caused by KIF11 mutations. *JAMA Ophthalmology*, **2014**, 132, 1393-9	3.9	73
90	Fine-scale variation and genetic determinants of alternative splicing across individuals. *PLoS Genetics*, **2009**, 5, e1000766	6	68
89	Cole-Carpenter syndrome is caused by a heterozygous missense mutation in P4HB. *American Journal of Human Genetics*, **2015**, 96, 425-31	11	65
88	ClinPred: Prediction Tool to Identify Disease-Relevant Nonsynonymous Single-Nucleotide Variants. *American Journal of Human Genetics*, **2018**, 103, 474-483	11	61
87	Mosaic Activating Mutations in FGFR1 Cause Encephalocraniocutaneous Lipomatosis. *American Journal of Human Genetics*, **2016**, 98, 579-587	11	60
86	Next-generation sequencing for diagnosis of rare diseases in the neonatal intensive care unit. *Cmaj*, **2016**, 188, E254-E260	3.5	59
85	Mutations in NFKB2 and potential genetic heterogeneity in patients with DAVID syndrome, having variable endocrine and immune deficiencies. *BMC Medical Genetics*, **2014**, 15, 139	2.1	56
84	Effect of polymorphisms within probe-target sequences on olignonucleotide microarray experiments. *Nucleic Acids Research*, **2008**, 36, 4417-23	20.1	55
83	Homozygous mutations in MFN2 cause multiple symmetric lipomatosis associated with neuropathy. *Human Molecular Genetics*, **2015**, 24, 5109-14	5.6	53
82	USP15 regulates type I interferon response and is required for pathogenesis of neuroinflammation. *Nature Immunology*, **2017**, 18, 54-63	19.1	51
81	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, 971-8	11	50
80	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, 735-43	11	48
79	Germline mutations in MAP3K6 are associated with familial gastric cancer. *PLoS Genetics*, **2014**, 10, e1004669		46
78	APRDX1 mutant allele causes a MMACHC secondary epimutation in cblC patients. *Nature Communications*, **2018**, 9, 67	17.4	45
77	Mutations in the enzyme glutathione peroxidase 4 cause Sedaghatian-type spondylometaphyseal dysplasia. *Journal of Medical Genetics*, **2014**, 51, 470-4	5.8	45

76	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. *Human Mutation*, **2015**, 36, 1009-1014	4.7	45
75	Biallelic PADI6 variants linking infertility, miscarriages, and hydatidiform moles. *European Journal of Human Genetics*, **2018**, 26, 1007-1013	5.3	43
74	Disrupted auto-regulation of the spliceosomal gene SNRPB causes cerebro-costo-mandibular syndrome. *Nature Communications*, **2014**, 5, 4483	17.4	40
73	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. *Journal of Child Neurology*, **2015**, 30, 1037-43	2.5	39
72	Causative Mutations and Mechanism of Androgenetic Hydatidiform Moles. *American Journal of Human Genetics*, **2018**, 103, 740-751	11	39
71	SPG7 mutations explain a significant proportion of French Canadian spastic ataxia cases. *European Journal of Human Genetics*, **2016**, 24, 1016-21	5.3	38
70	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, 593-6	5.8	36
69	Molecular analyses reveal close similarities between small cell carcinoma of the ovary, hypercalcemic type and atypical teratoid/rhabdoid tumor. *Oncotarget*, **2016**, 7, 1732-40	3.3	36
68	Homozygous nonsense mutation in SYNJ1 associated with intractable epilepsy and tau pathology. *Neurobiology of Aging*, **2015**, 36, 1222.e1-5	5.6	35
67	Compound heterozygous mutations in the gene PIGP are associated with early infantile epileptic encephalopathy. *Human Molecular Genetics*, **2017**, 26, 1706-1715	5.6	33
66	A polyadenylation site variant causes transcript-specific BMP1 deficiency and frequent fractures in children. *Human Molecular Genetics*, **2015**, 24, 516-24	5.6	32
65	CCDC88B is a novel regulator of maturation and effector functions of T cells during pathological inflammation. *Journal of Experimental Medicine*, **2014**, 211, 2519-35	16.6	32
64	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	6.6	31
63	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	11.5	30
62	TRPV4 and KRAS and FGFR1 gain-of-function mutations drive giant cell lesions of the jaw. *Nature Communications*, **2018**, 9, 4572	17.4	30
61	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	4.7	29
60	RTTN Mutations Cause Primary Microcephaly and Primordial Dwarfism in Humans. *American Journal of Human Genetics*, **2015**, 97, 862-8	11	29
59	Epigenetic dysregulation: a novel pathway of oncogenesis in pediatric brain tumors. *Acta Neuropathologica*, **2014**, 128, 615-27	14.3	29

58	Loss-of-Function Mutation in APC2 Causes Sotos Syndrome Features. *Cell Reports*, **2015**, 10, 1585-1598	10.6	28
57	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, 6293-300	5.6	28
56	A novel multisystem disease associated with recessive mutations in the tyrosyl-tRNA synthetase (YARS) gene. *American Journal of Medical Genetics, Part A*, **2017**, 173, 126-134	2.5	27
55	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	10.1	23
54	Characterizing temporal genomic heterogeneity in pediatric high-grade gliomas. *Acta Neuropathologica Communications*, **2017**, 5, 78	7.3	23
53	Nonsense mutation in the WDR73 gene is associated with Galloway-Mowat syndrome. *Journal of Medical Genetics*, **2015**, 52, 381-90	5.8	23
52	Inborn Error of Cobalamin Metabolism Associated with the Intracellular Accumulation of Transcobalamin-Bound Cobalamin and Mutations in ZNF143, Which Codes for a Transcriptional Activator. *Human Mutation*, **2016**, 37, 976-82	4.7	21
51	Gain-of-function mutation in TRPV4 identified in patients with osteonecrosis of the femoral head. *Journal of Medical Genetics*, **2016**, 53, 705-9	5.8	19
50	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, 114-6	3.2	18
49	CYRI/FAM49B negatively regulates RAC1-driven cytoskeletal remodelling and protects against bacterial infection. *Nature Microbiology*, **2019**, 4, 1516-1531	26.6	18
48	Functionally Null Missense Mutation Associates Strongly with Ovarian Carcinoma. *Cancer Research*, **2017**, 77, 4517-4529	10.1	18
47	Autosomal recessive cerebellar ataxia caused by a homozygous mutation in PMPCA. *Brain*, **2016**, 139, e19	11.2	17
46	Meconium ileus in a Lebanese family secondary to mutations in the GUCY2C gene. *European Journal of Human Genetics*, **2015**, 23, 990-2	5.3	17
45	LoLoPicker: detecting low allelic-fraction variants from low-quality cancer samples. *Oncotarget*, **2017**, 8, 37032-37040	3.3	17
44	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	36.3	17
43	THEMIS is required for pathogenesis of cerebral malaria and protection against pulmonary tuberculosis. *Infection and Immunity*, **2015**, 83, 759-68	3.7	14
42	ExomeAI: detection of recurrent allelic imbalance in tumors using whole-exome sequencing data. *Bioinformatics*, **2015**, 31, 429-31	7.2	14
41	POLR3A variants in hereditary spastic paraplegia and ataxia. *Brain*, **2018**, 141, e1	11.2	14

40	Syndrome disintegration: Exome sequencing reveals that Fitzsimmons syndrome is a co-occurrence of multiple events. *American Journal of Medical Genetics, Part A*, **2016**, 170, 1820-5	2.5	14
39	Choroideremia Is a Systemic Disease With Lymphocyte Crystals and Plasma Lipid and RBC Membrane Abnormalities **2015**, 56, 8158-65		14
38	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	10.6	14
37	Recessive mutations in the kinase ZAK cause a congenital myopathy with fibre type disproportion. *Brain*, **2017**, 140, 37-48	11.2	13
36	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	12
35	Methylome analysis and whole-exome sequencing reveal that brain tumors associated with encephalocraniocutaneous lipomatosis are midline pilocytic astrocytomas. *Acta Neuropathologica*, **2018**, 136, 657-660	14.3	12
34	Prognostic and predictive value of circulating tumor DNA during neoadjuvant chemotherapy for triple negative breast cancer. *Scientific Reports*, **2020**, 10, 14704	4.9	12
33	Novel 25 kb Deletion of MERTK Causes Retinitis Pigmentosa With Severe Progression **2017**, 58, 1736-1742		11
32	Exome and whole-genome sequencing for gene discovery: the future is now!. *Human Mutation*, **2012**, 33, 591-2	4.7	11
31	Mutations in GALC cause late-onset Krabbe disease with predominant cerebellar ataxia. *Neurogenetics*, **2016**, 17, 137-41	3	11
30	Atypical fibrodysplasia ossificans progressiva diagnosed by whole-exome sequencing. *American Journal of Medical Genetics, Part A*, **2015**, 167, 1337-41	2.5	10
29	Autosomal recessive axonal polyneuropathy in a sibling pair due to a novel homozygous mutation in IGHMBP2. *Neuromuscular Disorders*, **2015**, 25, 794-9	2.9	10
28	RMND1-Related Leukoencephalopathy With Temporal Lobe Cysts and Hearing Loss-Another Mendelian Mimicker of Congenital Cytomegalovirus Infection. *Pediatric Neurology*, **2017**, 66, 59-62	2.9	10
27	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,	11.5	10
26	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	3	9
25	Chromatin dysregulation associated with NSD1 mutation in head and neck squamous cell carcinoma. *Cell Reports*, **2021**, 34, 108769	10.6	9
24	Bmp signaling maintains a mesoderm progenitor cell state in the mouse tailbud. *Development (Cambridge)*, **2017**, 144, 2982-2993	6.6	8
23	Recessive mutation in CD2AP causes focal segmental glomerulosclerosis in humans and mice. *Kidney International*, **2019**, 95, 57-61	9.9	8

22	The Genetic Causes of Nonsyndromic Congenital Retinal Detachment: A Genetic and Phenotypic Study of Pakistani Families **2017**, 58, 1028-1036		7
21	Early infantile epileptic encephalopathy due to biallelic pathogenic variants in PIGQ: Report of seven new subjects and review of the literature. *Journal of Inherited Metabolic Disease*, **2020**, 43, 1321-1332	5.4	6
20	A novel pathogenic missense ADAMTS17 variant that impairs secretion causes Weill-Marchesani Syndrome with variably dysmorphic hand features. *Scientific Reports*, **2020**, 10, 10827	4.9	6
19	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	2.3	6
18	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	2.2	5
17	Biallelic Loss-of-Function Variants in AIMP1 Cause a Rare Neurodegenerative Disease. *Journal of Child Neurology*, **2019**, 34, 74-80	2.5	5
16	Novel pathogenic variants in NLRP7, NLRP5, and PADI6 in patients with recurrent hydatidiform moles and reproductive failure. *Clinical Genetics*, **2021**, 99, 823-828	4	5
15	Latency and interval therapy affect the evolution in metastatic colorectal cancer. *Scientific Reports*, **2020**, 10, 581	4.9	4
14	Longitudinal mutational analysis of a cerebellar pilocytic astrocytoma recurring as a ganglioglioma. *Pediatric Blood and Cancer*, **2017**, 64, 275-278	3	4
13	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	2.5	3
12	H3.1 K36M mutation in a congenital-onset soft tissue neoplasm. *Pediatric Blood and Cancer*, **2017**, 64, e26633	3	3
11	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. *Journal of Oral Pathology and Medicine*, **2021**, 50, 410-417	3.3	3
10	Mutation in Eftud2 causes craniofacial defects in mice via mis-splicing of Mdm2 and increased P53. *Human Molecular Genetics*, **2021**, 30, 739-757	5.6	3
9	ZBTB7B (ThPOK) Is Required for Pathogenesis of Cerebral Malaria and Protection against Pulmonary Tuberculosis. *Infection and Immunity*, **2020**, 88,	3.7	2
8	A functionally impaired missense variant identified in French Canadian families implicates FANCI as a candidate ovarian cancer-predisposing gene. *Genome Medicine*, **2021**, 13, 186	14.4	2
7	Epimutations in both the TESK2 and MMACHC promoters in the Epi-cblC inherited disorder of intracellular metabolism of vitamin B.. *Clinical Epigenetics*, **2022**, 14, 52	7.7	1
6	Nucleome programming is required for the foundation of totipotency in mammalian germline development. *EMBO Journal*,	13	1
5	Inherent genomic properties underlie the epigenomic heterogeneity of human induced pluripotent stem cells. *Cell Reports*, **2021**, 37, 109909	10.6	0

List of Publications

4	A Mouse Model for Cerebro-Costo-Mandibular Syndrome (CCMS) with an Intronic Deletion in SnrpB. *FASEB Journal*, **2020**, 34, 1-1	0.9
3	Deletion of Mouse Sf3b4 in Neural Crest Cells Causes Craniofacial Abnormalities. *FASEB Journal*, **2020**, 34, 1-1	0.9
2	Une mutation transgénérationnelle du gène MMACHC produit un nouveau type d'erreur innée du métabolisme d'homme bi-cblC. *Bulletin De L'Academie Nationale De Medecine*, **2018**, 202, 1585-1596	0.1
1	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	0.9