Eric W Klee

List of Publications by Citations

157 papers	3,127 citations	30 h-index	51 g-index
164 ext. papers	4,199 ext. citations	5.7 avg, IF	4.9 L-index

#	Paper	IF	Citations
157	Integrated genomic characterization reveals novel, therapeutically relevant drug targets in FGFR and EGFR pathways in sporadic intrahepatic cholangiocarcinoma. *PLoS Genetics*, **2014**, 10, e1004135	6	239
156	Preemptive genotyping for personalized medicine: design of the right drug, right dose, right time-using genomic data to individualize treatment protocol. *Mayo Clinic Proceedings*, **2014**, 89, 25-33	6.4	213
155	Standards and Guidelines for Validating Next-Generation Sequencing Bioinformatics Pipelines: A Joint Recommendation of the Association for Molecular Pathology and the College of American Pathologists. *Journal of Molecular Diagnostics*, **2018**, 20, 4-27	5.1	205
154	Adrenomedullin is up-regulated in patients with pancreatic cancer and causes insulin resistance in cells and mice. *Gastroenterology*, **2012**, 143, 1510-1517.e1	13.3	116
153	Preemptive Pharmacogenomic Testing for Precision Medicine: A Comprehensive Analysis of Five Actionable Pharmacogenomic Genes Using Next-Generation DNA Sequencing and a Customized CYP2D6 Genotyping Cascade. *Journal of Molecular Diagnostics*, **2016**, 18, 438-445	5.1	108
152	Confirming Variants in Next-Generation Sequencing Panel Testing by Sanger Sequencing. *Journal of Molecular Diagnostics*, **2015**, 17, 456-61	5.1	82
151	Bioinformatics for clinical next generation sequencing. *Clinical Chemistry*, **2015**, 61, 124-35	5.5	79
150	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. *Nature Communications*, **2019**, 10, 3094	17.4	76
149	Disruption of the ATXN1-CIC complex causes a spectrum of neurobehavioral phenotypes in mice and humans. *Nature Genetics*, **2017**, 49, 527-536	36.3	71
148	Clinical spectrum and genotype-phenotype associations of KCNA2-related encephalopathies. *Brain*, **2017**, 140, 2337-2354	11.2	71
147	Bi-allelic Alterations in AEBP1 Lead to Defective Collagen Assembly and Connective Tissue Structure Resulting in a Variant of Ehlers-Danlos Syndrome. *American Journal of Human Genetics*, **2018**, 102, 696-705	11	65
146	Outcome of Whole Exome Sequencing for Diagnostic Odyssey Cases of an Individualized Medicine Clinic: The Mayo Clinic Experience. *Mayo Clinic Proceedings*, **2016**, 91, 297-307	6.4	63
145	Genome-wide reverse genetics framework to identify novel functions of the vertebrate secretome. *PLoS ONE*, **2006**, 1, e104	3.7	63
144	Genome-wide detection of tandem DNA repeats that are expanded in autism. *Nature*, **2020**, 586, 80-86	50.4	58
143	Evaluating eukaryotic secreted protein prediction. *BMC Bioinformatics*, **2005**, 6, 256	3.6	56
142	Zebrafish for the study of the biological effects of nicotine. *Nicotine and Tobacco Research*, **2011**, 13, 301-12	4.9	50
141	TREAT: a bioinformatics tool for variant annotations and visualizations in targeted and exome sequencing data. *Bioinformatics*, **2012**, 28, 277-8	7.2	50

140	Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. *Neuron*, **2020**, 106, 404-420.e8	13.9	49
139	Implementing individualized medicine into the medical practice. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2014**, 166C, 15-23	3.1	46
138	Candidate serum biomarkers for prostate adenocarcinoma identified by mRNA differences in prostate tissue and verified with protein measurements in tissue and blood. *Clinical Chemistry*, **2012**, 58, 599-609	5.5	46
137	RNA sequencing shows transcriptomic changes in rectosigmoid mucosa in patients with irritable bowel syndrome-diarrhea: a pilot case-control study. *American Journal of Physiology - Renal Physiology*, **2014**, 306, G1089-98	5.1	44
136	Expanding DNA diagnostic panel testing: is more better?. *Expert Review of Molecular Diagnostics*, **2011**, 11, 703-9	3.8	44
135	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. *American Journal of Human Genetics*, **2018**, 103, 666-678	11	44
134	Sentieon DNASeq Variant Calling Workflow Demonstrates Strong Computational Performance and Accuracy. *Frontiers in Genetics*, **2019**, 10, 736	4.5	43
133	Arterial tortuosity syndrome: 40 new families and literature review. *Genetics in Medicine*, **2018**, 20, 1236-1245	8.2	40
132	Experience with precision genomics and tumor board, indicates frequent target identification, but barriers to delivery. *Oncotarget*, **2017**, 8, 27145-27154	3.3	40
131	Loss of the sphingolipid desaturase DEGS1 causes hypomyelinating leukodystrophy. *Journal of Clinical Investigation*, **2019**, 129, 1240-1256	15.9	37
130	Identifying secretomes in people, pufferfish and pigs. *Nucleic Acids Research*, **2004**, 32, 1414-21	20.1	36
129	Zebrafish approaches enhance the translational research tackle box. *Translational Research*, **2014**, 163, 65-78	11	34
128	Larval zebrafish model for FDA-approved drug repositioning for tobacco dependence treatment. *PLoS ONE*, **2014**, 9, e90467	3.7	34
127	Kinase genotype analysis of gastric gastrointestinal stromal tumor cytology samples using targeted next-generation sequencing. *Clinical Gastroenterology and Hepatology*, **2015**, 13, 202-6	6.9	28
126	Development and Verification of an RNA Sequencing (RNA-Seq) Assay for the Detection of Gene Fusions in Tumors. *Journal of Molecular Diagnostics*, **2018**, 20, 495-511	5.1	28
125	Computational classification of classically secreted proteins. *Drug Discovery Today*, **2007**, 12, 234-40	8.8	28
124	Best practices for the analytical validation of clinical whole-genome sequencing intended for the diagnosis of germline disease. *Npj Genomic Medicine*, **2020**, 5, 47	6.2	22
123	Utility of DNA, RNA, Protein, and Functional Approaches to Solve Cryptic Immunodeficiencies. *Journal of Clinical Immunology*, **2018**, 38, 307-319	5.7	21

122	"Big Data" in Laboratory Medicine. *Clinical Chemistry*, **2015**, 61, 1433-40	5.5	21
121	Impact of sample acquisition and linear amplification on gene expression profiling of lung adenocarcinoma: laser capture micro-dissection cell-sampling versus bulk tissue-sampling. *BMC Medical Genomics*, **2009**, 2, 13	3.7	21
120	Mitochondrial 3-Hydroxy-3-Methylglutaryl-CoA Synthase Deficiency: Unique Presenting Laboratory Values and a Review of Biochemical and Clinical Features. *JIMD Reports*, **2018**, 40, 63-69	1.9	20
119	Novel variants likely disrupt DNA binding: molecular modeling in two cases, review of published cases, genotype-phenotype correlation, and phenotypic expansion of the Bosch-Boonstra-Schaaf optic atrophy syndrome. *Journal of Physical Education and Sports Management*, **2017**, 3,	2.8	19
118	De novo DDX3X missense variants in males appear viable and contribute to syndromic intellectual disability. *American Journal of Medical Genetics, Part A*, **2019**, 179, 570-578	2.5	19
117	Pilot study of small bowel mucosal gene expression in patients with irritable bowel syndrome with diarrhea. *American Journal of Physiology - Renal Physiology*, **2016**, 311, G365-76	5.1	19
116	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. *Genetics in Medicine*, **2019**, 21, 2723-2733	8.1	18
115	RINT1 Bi-allelic Variations Cause Infantile-Onset Recurrent Acute Liver Failure and Skeletal Abnormalities. *American Journal of Human Genetics*, **2019**, 105, 108-121	11	18
114	Bioinformatics methods for prioritizing serum biomarker candidates. *Clinical Chemistry*, **2006**, 52, 2162-4	5.5	18
113	Silent Tyrosinemia Type I Without Elevated Tyrosine or Succinylacetone Associated with Liver Cirrhosis and Hepatocellular Carcinoma. *Human Mutation*, **2016**, 37, 1097-105	4.7	17
112	Characterization of three ciliopathy pedigrees expands the phenotype associated with biallelic C2CD3 variants. *European Journal of Human Genetics*, **2018**, 26, 1797-1809	5.3	17
111	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. *American Journal of Human Genetics*, **2019**, 104, 530-541	11	17
110	Next Generation Sequencing of Sporadic Vestibular Schwannoma: Necessity of Biallelic NF2 Inactivation and Implications of Accessory Non-NF2 Variants. *Otology and Neurotology*, **2018**, 39, e860-e871	2.6	17
109	Pharmacogenomic findings from clinical whole exome sequencing of diagnostic odyssey patients. *Molecular Genetics & Genomic Medicine*, **2017**, 5, 269-279	2.3	15
108	Data mining for biomarker development: a review of tissue specificity analysis. *Clinics in Laboratory Medicine*, **2008**, 28, 127-43, viii	2.1	15
107	Molecular modeling and molecular dynamic simulation of the effects of variants in the TGFBR2 kinase domain as a paradigm for interpretation of variants obtained by next generation sequencing. *PLoS ONE*, **2017**, 12, e0170822	3.7	15
106	Novel de novo variant in is likely to impact DNA binding in a patient with a neurodevelopmental disorder and expanded phenotypes: patient report, in silico functional assessment, and review of published cases. *Journal of Physical Education and Sports Management*, **2017**, 3, a001743	2.8	14
105	A tailored approach to fusion transcript identification increases diagnosis of rare inherited disease. *PLoS ONE*, **2019**, 14, e0223337	3.7	14

104	Whole exome sequencing implicates an INO80D mutation in a syndrome of aortic hypoplasia, premature atherosclerosis, and arterial stiffness. *Circulation: Cardiovascular Genetics*, **2014**, 7, 607-14		14
103	De novo TBR1 variants cause a neurocognitive phenotype with ID and autistic traits: report of 25 new individuals and review of the literature. *European Journal of Human Genetics*, **2020**, 28, 770-782	5.3	13
102	Clinical Applications and Utility of a Precision Medicine Approach for Patients With Unexplained Cytopenias. *Mayo Clinic Proceedings*, **2019**, 94, 1753-1768	6.4	13
101	Quantitating tissue specificity of human genes to facilitate biomarker discovery. *Bioinformatics*, **2007**, 23, 1348-55	7.2	13
100	point mutations and familial intracranial aneurysms. *Neurology*, **2018**, 91, e2170-e2181	6.5	13
99	Clinical Correlates and Treatment Outcomes for Patients With Short Telomere Syndromes. *Mayo Clinic Proceedings*, **2018**, 93, 834-839	6.4	13
98	The prevalence of diseases caused by lysosome-related genes in a cohort of undiagnosed patients. *Molecular Genetics and Metabolism Reports*, **2017**, 13, 46-51	1.8	12
97	The zebrafish secretome. *Zebrafish*, **2008**, 5, 131-8	2	12
96	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. *American Journal of Human Genetics*, **2021**, 108, 502-516	11	12
95	A Novel Kleefstra Syndrome-associated Variant That Affects the Conserved TPL Motif within the Ankyrin Repeat of EHMT1 Leads to Abnormal Protein Folding. *Journal of Biological Chemistry*, **2017**, 292, 3866-3876	5.4	11
94	De novo variants in FBXO11 cause a syndromic form of intellectual disability with behavioral problems and dysmorphisms. *European Journal of Human Genetics*, **2019**, 27, 738-746	5.3	11
93	RNA-Seq detects a SAMD12-EXT1 fusion transcript and leads to the discovery of an EXT1 deletion in a child with multiple osteochondromas. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e00560	2.3	11
92	A second cohort of CHD3 patients expands the molecular mechanisms known to cause Snijders Blok-Campeau syndrome. *European Journal of Human Genetics*, **2020**, 28, 1422-1431	5.3	10
91	LeafCutterMD: an algorithm for outlier splicing detection in rare diseases. *Bioinformatics*, **2020**, 36, 4609-4615	7.2	10
90	AMOD: a morpholino oligonucleotide selection tool. *Nucleic Acids Research*, **2005**, 33, W506-11	20.1	10
89	Target selection for Danio rerio functional genomics. *Genesis*, **2001**, 30, 123-5	1.9	10
88	Multigenerational pedigree with STAR syndrome: A novel FAM58A variant and expansion of the phenotype. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1328-1333	2.5	9
87	A novel ANO3 variant identified in a 53-year-old woman presenting with hyperkinetic dysarthria, blepharospasm, hyperkinesias, and complex motor tics. *BMC Medical Genetics*, **2016**, 17, 93	2.1	9

86	Widening of the genetic and clinical spectrum of Lamb-Shaffer syndrome, a neurodevelopmental disorder due to SOX5 haploinsufficiency. *Genetics in Medicine*, **2020**, 22, 524-537	8.1	9
85	Assessing Human Genetic Variations in Glucose Transporter SLC2A10 and Their Role in Altering Structural and Functional Properties. *Frontiers in Genetics*, **2018**, 9, 276	4.5	9
84	Protein-elongating mutations in MYH11 are implicated in a dominantly inherited smooth muscle dysmotility syndrome with severe esophageal, gastric, and intestinal disease. *Human Mutation*, **2020**, 41, 973-982	4.7	8
83	Pathogenic Variant in , p.Arg183Trp, Causes Juvenile-Onset Dystonia, Hearing Loss, and Developmental Delay without Midline Malformation. *Case Reports in Genetics*, **2017**, 2017, 9184265	0.7	8
82	Molecular characterization of known and novel ACVR1 variants in phenotypes of aberrant ossification. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1764-1777	2.5	8
81	Variability in assigning pathogenicity to incidental findings: insights from LDLR sequence linked to the electronic health record in 1013 individuals. *European Journal of Human Genetics*, **2017**, 25, 410-415	5.3	7
80	Clinical characteristics and platelet phenotype in a family with RUNX1 mutated thrombocytopenia. *Leukemia and Lymphoma*, **2017**, 58, 1963-1967	1.9	7
79	Haploinsufficiency as a disease mechanism in GNB1-associated neurodevelopmental disorder. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1477	2.3	7
78	Recommendations for performance optimizations when using GATK3.8 and GATK4. *BMC Bioinformatics*, **2019**, 20, 557	3.6	7
77	Familial chronic megacolon presenting in childhood or adulthood: Seeking the presumed gene association. *Neurogastroenterology and Motility*, **2019**, 31, e13550	4	6
76	Variants in DOCK3 cause developmental delay and hypotonia. *European Journal of Human Genetics*, **2019**, 27, 1225-1234	5.3	6
75	Frequency of mitogen-activated protein kinase and phosphoinositide 3-kinase signaling pathway pathogenic alterations in EUS-FNA sampled malignant lymph nodes in rectal cancer with theranostic potential. *Gastrointestinal Endoscopy*, **2015**, 82, 550-6.e1	5.2	6
74	SPECC1L regulates palate development downstream of IRF6. *Human Molecular Genetics*, **2020**, 29, 845-858	3.8	6
73	A systematic method for selection of promising serum protein biomarkers to improve prostate cancer (PCa1) detection. *Clinical Chemistry*, **2006**, 52, 2159-62	5.5	6
72	Molecular modeling of LDLR aids interpretation of genomic variants. *Journal of Molecular Medicine*, **2019**, 97, 533-540	5.5	6
71	Novel germline missense DDX41 variant in a patient with an adult-onset myeloid neoplasm with excess blasts without dysplasia. *Leukemia and Lymphoma*, **2019**, 60, 1337-1339	1.9	6
70	Impact of integrated translational research on clinical exome sequencing. *Genetics in Medicine*, **2021**, 23, 498-507	8.1	6
69	A case of YY1-associated syndromic learning disability or Gabriele-de Vries syndrome with myasthenia gravis. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2846-2849	2.5	6

68	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. *Nature Genetics*, **2021**, 53, 1006-1021	36.3	6
67	A novel frameshift deletion in in a patient with Kleefstra Syndrome results in decreased H3K9 dimethylation. *Molecular Genetics & Genomic Medicine*, **2017**, 5, 141-146	2.3	5
66	Variable expressivity of syndromic BMP4-related eye, brain, and digital anomalies: A review of the literature and description of three new cases. *European Journal of Human Genetics*, **2019**, 27, 1379-1388	5.3	5
65	X-Linked Lymphoproliferative Syndrome Presenting as Adult-Onset Multi-Infarct Dementia. *Journal of Neuropathology and Experimental Neurology*, **2019**, 78, 460-466	3.1	5
64	Comparative analysis of de novo assemblers for variation discovery in personal genomes. *Briefings in Bioinformatics*, **2018**, 19, 893-904	13.4	5
63	CSNK2B: A broad spectrum of neurodevelopmental disability and epilepsy severity. *Epilepsia*, **2021**, 62, e103-e109	6.4	5
62	Familial Creutzfeldt-Jakob Disease: Case report and role of genetic counseling in post mortem testing. *Prion*, **2016**, 10, 502-506	2.3	5
61	Three rare disease diagnoses in one patient through exome sequencing. *Journal of Physical Education and Sports Management*, **2019**, 5,	2.8	4
60	Computational Detection of Known Pathogenic Gene Fusions in a Normal Tissue Database and Implications for Genetic Disease Research. *Frontiers in Genetics*, **2020**, 11, 173	4.5	4
59	Aetiology and outcomes of secondary myelofibrosis occurring in the context of inherited platelet disorders: A single institutional study of four patients. *British Journal of Haematology*, **2020**, 190, e316-e320	4.5	4
58	Functional Analysis of the SIM1 Variant p.G715V in 2 Patients With Obesity. *Journal of Clinical Endocrinology and Metabolism*, **2020**, 105,	5.6	4
57	Functional validation reveals the novel missense V419L variant in associated with Loeys-Dietz syndrome (LDS) impairs canonical TGF-□signaling. *Journal of Physical Education and Sports Management*, **2017**, 3,	2.8	4
56	An interlaboratory study of complex variant detection		4
55	Interpretation challenges of novel dual-class missense and splice-impacting variant in POLR3A-related late-onset hereditary spastic ataxia. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1341	2.3	4
54	Exome sequencing confirms diagnosis of kabuki syndrome in an-adult with hodgkin lymphoma and unusually severe multisystem phenotype. *Clinical Immunology*, **2019**, 207, 55-57	9	4
53	Co-occurrence of a maternally inherited duplication and a paternally inherited pathogenic variant in in a child with growth retardation and severe short stature: atypical Weaver syndrome or evidence of a dosage effect?. *Journal of Physical Education and Sports Management*, **2018**, 4,	2.8	4
52	TNPO2 variants associate with human developmental delays, neurologic deficits, and dysmorphic features and alter TNPO2 activity in Drosophila. *American Journal of Human Genetics*, **2021**, 108, 1669-1691	11	4
51	COVID-19 Mortality Prediction From Deep Learning in a Large Multistate Electronic Health Record and Laboratory Information System Data Set: Algorithm Development and Validation. *Journal of Medical Internet Research*, **2021**, 23, e30157	7.6	4

50	Preemptive sequencing in the genomic medicine era. *Expert Review of Precision Medicine and Drug Development*, **2017**, 2, 91-98	1.6	3
49	Novel Pathogenic Variant in TGFBR2 Confirmed by Molecular Modeling Is a Rare Cause of Loeys-Dietz Syndrome. *Case Reports in Genetics*, **2017**, 2017, 7263780	0.7	3
48	Functional characterization of a GFAP variant of uncertain significance in an Alexander disease case within the setting of an individualized medicine clinic. *Clinical Case Reports (discontinued)*, **2016**, 4, 885-95	0.7	3
47	Improving Single-Nucleotide Polymorphism-Based Fetal Fraction Estimation of Maternal Plasma Circulating Cell-Free DNA Using Bayesian Hierarchical Models. *Journal of Computational Biology*, **2018**, 25, 1040-1049	1.7	3
46	"The molecule is the thing:" the promise of molecular modeling and dynamic simulations in aiding the prioritization and interpretation of genomic testing results. *F1000Research*, **2016**, 5, 766	3.6	3
45	One in seven pathogenic variants can be challenging to detect by NGS: An analysis of 450,000 patients with implications for clinical sensitivity and genetic test implementation		3
44	Genetic variants in DGAT1 cause diverse clinical presentations of malnutrition through a specific molecular mechanism. *European Journal of Medical Genetics*, **2020**, 63, 103817	2.6	3
43	A form of muscular dystrophy associated with pathogenic variants in JAG2. *American Journal of Human Genetics*, **2021**, 108, 840-856	11	3
42	TSPEAR variants are primarily associated with ectodermal dysplasia and tooth agenesis but not hearing loss: A novel cohort study. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2417-2433	2.5	3
41	Truncating SRCAP variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature. *American Journal of Human Genetics*, **2021**, 108, 1053-1068	11	3
40	Novel biallelic variants in associated with mitochondrial myopathy. *Journal of Physical Education and Sports Management*, **2019**, 5,	2.8	3
39	Next-Generation Sequencing of CYP2C19 in Stent Thrombosis: Implications for Clopidogrel Pharmacogenomics. *Cardiovascular Drugs and Therapy*, **2021**, 35, 549-559	3.9	3
38	Extension of the mutational and clinical spectrum of SOX2 related disorders: Description of six new cases and a novel association with suprasellar teratoma. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2710-2719	2.5	3
37	GFAP canonical transcript may not be suitable for the diagnosis of adult-onset Alexander disease. *Acta Neuropathologica Communications*, **2018**, 6, 112	7.3	3
36	Developmental delay, coarse facial features, and epilepsy in a patient with gene variants. *Clinical Case Reports (discontinued)*, **2019**, 7, 632-637	0.7	2
35	De novo variants of NR4A2 are associated with neurodevelopmental disorder and epilepsy. *Genetics in Medicine*, **2020**, 22, 1413-1417	8.1	2
34	Jumonji domain containing 1C (JMJD1C) sequence variants in seven patients with autism spectrum disorder, intellectual disability and seizures. *European Journal of Medical Genetics*, **2020**, 63, 103850	2.6	2
33	Early-onset limb-girdle muscular dystrophy-2L in a female athlete. *Muscle and Nerve*, **2017**, 55, E19-E21	3.4	2

32	Novel loss-of-function variants in TRIO are associated with neurodevelopmental disorder: case report. *BMC Medical Genetics*, **2020**, 21, 219	2.1	2
31	Expansion of -Related Phenotypes and Discovery of a Novel PURA Variant: A Case Report. *Child Neurology Open*, **2020**, 7, 2329048X20955003	1.3	2
30	Nail-patella-like renal disease masquerading as Fabry disease on kidney biopsy: a case report. *BMC Nephrology*, **2020**, 21, 341	2.7	2
29	Epigenetic alteration contributes to the transcriptional reprogramming in T-cell prolymphocytic leukemia. *Scientific Reports*, **2021**, 11, 8318	4.9	2
28	Expansion of the Genotypic and Phenotypic Spectrum of WASF1-Related Neurodevelopmental Disorder. *Brain Sciences*, **2021**, 11,	3.4	2
27	Genomic Profiling Reveals Molecular Heterogeneity in Patients with Richter's Syndrome (RS) and Progressive Chronic Lymphocytic Leukemia (CLL). *Blood*, **2020**, 136, 16-17	2.2	1
26	Long Range Sequencing Shows Improved Resolution in the Detection of Beta Globin Cluster Variants. *Blood*, **2019**, 134, 3548-3548	2.2	1
25	Biallelic variants in PROZ as a cause of hypercoagulability and livedo racemosa. *Thrombosis Research*, **2020**, 195, 187-189	8.2	1
24	An activating germline variant associated with a tumor entity characterized by unilateral and bilateral chondrosarcoma of the mastoid.. *Human Genetics and Genomics Advances*, **2020**, 1, 100006	0.8	1
23	Functional validation of TERT and TERC variants of uncertain significance in patients with short telomere syndromes. *Blood Cancer Journal*, **2020**, 10, 120	7	1
22	A novel missense variant and multiexon deletion causing a delayed presentation of xeroderma pigmentosum, group C. *Journal of Physical Education and Sports Management*, **2020**, 6,	2.8	1
21	LPCAT1-TERT fusions are uniquely recurrent in epithelioid trophoblastic tumors and positively regulate cell growth. *PLoS ONE*, **2021**, 16, e0250518	3.7	1
20	Genomics Integration Into Nephrology Practice. *Kidney Medicine*, **2021**, 3, 785-798	2.8	1
19	Proposal for Modification of Cahan's Criteria Utilizing Molecular Genetic Analyses for Cases without Baseline Histopathology: A Unique Method Applicable to Primary Radiosurgery. *Journal of Neurological Surgery, Part B: Skull Base*, **2019**, 80, 10-17	1.5	1
18	An intragenic duplication of leading to abnormal transcripts and causing trichorhinophalangeal syndrome type I. *Journal of Physical Education and Sports Management*, **2019**, 5,	2.8	1
17	HELLO: improved neural network architectures and methodologies for small variant calling. *BMC Bioinformatics*, **2021**, 22, 404	3.6	1
16	Autosomal Recessive Cerebellar Atrophy and Spastic Ataxia in Patients With Pathogenic Biallelic Variants in .. *Frontiers in Cell and Developmental Biology*, **2022**, 10, 783762	5.7	1
15	Gain and loss of TASK3 channel function and its regulation by novel variation cause KCNK9 imprinting syndrome. *Genome Medicine*, **2022**, 14,	14.4	1

14	Late onset asymptomatic pancreatic neuroendocrine tumor - A case report on the phenotypic expansion for MEN1. *Hereditary Cancer in Clinical Practice*, **2017**, 15, 10	2.3	0
13	Design considerations for workflow management systems use in production genomics research and the clinic. *Scientific Reports*, **2021**, 11, 21680	4.9	0
12	De novo PBX1 variant in a patient with glaucoma, kidney anomalies, and developmental delay: An expansion of the CAKUTHED phenotype. *American Journal of Medical Genetics, Part A*, **2021**,	2.5	0
11	Improved Characterization of Complex Globin Gene Cluster Structural Variants Using Long-Read Sequencing. *Journal of Molecular Diagnostics*, **2021**, 23, 1732-1740	5.1	0
10	SeekFusion - A Clinically Validated Fusion Transcript Detection Pipeline for PCR-Based Next-Generation Sequencing of RNA. *Frontiers in Genetics*, **2021**, 12, 739054	4.5	0
9	Recurrent ganglioneuroma in PTPN11-associated Noonan syndrome: A case report and literature review. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1883-1887	2.5	0
8	Defining the genotypic and phenotypic spectrum of X-linked MSL3-related disorder. *Genetics in Medicine*, **2021**, 23, 384-395	8.1	0
7	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome.. *American Journal of Human Genetics*, **2022**, 109, 601-617	11	0
6	Heterozygous variants in PRPF8 are associated with neurodevelopmental disorders.. *American Journal of Medical Genetics, Part A*, **2022**,	2.5	0
5	Successful Treatment of Skewed Lyonization Associated with X-Linked CGD in a Female Presenting with Recalcitrant Crohn's Disease. *Journal of Clinical Immunology*, **2020**, 40, 1056-1061	5.7	
4	PT: Protein Panoramic annoTation Tool for the interpretation of protein coding genetic variants. *JAMIA Open*, **2021**, 4, ooab065	2.9	
3	Whole Exome Sequencing Leading to the Diagnosis of Dysferlinopathy with a Novel Missense Mutation (c.959G>C). *Case Reports in Genetics*, **2016**, 2016, 9280812	0.7	
2	Interpretation of Dihydrorhodamine-1,2,3 Flow Cytometry in Chronic Granulomatous Disease: an Atypical Exemplar.. *Journal of Clinical Immunology*, **2022**, 1	5.7	
1	Functional validation of a novel AAAS variant in an atypical presentation of Allgrove syndrome.. *Molecular Genetics & Genomic Medicine*, **2022**, e1966	2.3	