

# Eric W Klee

## List of Publications by Year in descending order

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Version: 2024-02-01

158  
papers

5,104  
citations

134610

34  
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134545

62  
g-index

164  
all docs

164  
docs citations

164  
times ranked

11014  
citing authors

#	ARTICLE	IF	CITATIONS
1	Standards and Guidelines for Validating Next-Generation Sequencing Bioinformatics Pipelines. <i>Journal of Molecular Diagnostics</i> , 2018, 20, 4-27.	1.2	341
2	Integrated Genomic Characterization Reveals Novel, Therapeutically Relevant Drug Targets in FGFR and EGFR Pathways in Sporadic Intrahepatic Cholangiocarcinoma. <i>PLoS Genetics</i> , 2014, 10, e1004135.	1.5	292
3	Preemptive Genotyping for Personalized Medicine: Design of the Right Drug, Right Dose, Right Time—Using Genomic Data to Individualize Treatment Protocol. <i>Mayo Clinic Proceedings</i> , 2014, 89, 25-33.	1.4	250
4	Preemptive Pharmacogenomic Testing for Precision Medicine. <i>Journal of Molecular Diagnostics</i> , 2016, 18, 438-445.	1.2	171
5	Genome-wide detection of tandem DNA repeats that are expanded in autism. <i>Nature</i> , 2020, 586, 80-86.	13.7	155
6	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. <i>Nature Communications</i> , 2019, 10, 3094.	5.8	150
7	Adrenomedullin is Up-regulated in Patients With Pancreatic Cancer and Causes Insulin Resistance in $\beta^2$ Cells and Mice. <i>Gastroenterology</i> , 2012, 143, 1510-1517.e1.	0.6	145
8	Sentieon DNaseq Variant Calling Workflow Demonstrates Strong Computational Performance and Accuracy. <i>Frontiers in Genetics</i> , 2019, 10, 736.	1.1	131
9	Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. <i>Neuron</i> , 2020, 106, 404-420.e8.	3.8	121
10	Clinical spectrum and genotype-phenotype associations of KCNA2-related encephalopathies. <i>Brain</i> , 2017, 140, 2337-2354.	3.7	117
11	Bioinformatics for Clinical Next Generation Sequencing. <i>Clinical Chemistry</i> , 2015, 61, 124-135.	1.5	114
12	Disruption of the ATXN1-CIC complex causes a spectrum of neurobehavioral phenotypes in mice and humans. <i>Nature Genetics</i> , 2017, 49, 527-536.	9.4	113
13	Confirming Variants in Next-Generation Sequencing Panel Testing by Sanger Sequencing. <i>Journal of Molecular Diagnostics</i> , 2015, 17, 456-461.	1.2	109
14	Bi-allelic Alterations in AEBP1 Lead to Defective Collagen Assembly and Connective Tissue Structure Resulting in a Variant of Ehlers-Danlos Syndrome. <i>American Journal of Human Genetics</i> , 2018, 102, 696-705.	2.6	105
15	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. <i>American Journal of Human Genetics</i> , 2018, 103, 666-678.	2.6	87
16	Outcome of Whole Exome Sequencing for Diagnostic Odyssey Cases of an Individualized Medicine Clinic. <i>Mayo Clinic Proceedings</i> , 2016, 91, 297-307.	1.4	83
17	Loss of the sphingolipid desaturase DEGS1 causes hypomyelinating leukodystrophy. <i>Journal of Clinical Investigation</i> , 2019, 129, 1240-1256.	3.9	68
18	Evaluating eukaryotic secreted protein prediction. <i>BMC Bioinformatics</i> , 2005, 6, 256.	1.2	67

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19	Genome-Wide Reverse Genetics Framework to Identify Novel Functions of the Vertebrate Secretome. PLoS ONE, 2006, 1, e104.	1.1	67
20	Best practices for the analytical validation of clinical whole-genome sequencing intended for the diagnosis of germline disease. Npj Genomic Medicine, 2020, 5, 47.	1.7	67
21	Arterial tortuosity syndrome: 40 new families and literature review. Genetics in Medicine, 2018, 20, 1236-1245.	1.1	66
22	Zebrafish for the Study of the Biological Effects of Nicotine. Nicotine and Tobacco Research, 2011, 13, 301-312.	1.4	61
23	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.	1.5	61
24	TREAT: a bioinformatics tool for variant annotations and visualizations in targeted and exome sequencing data. Bioinformatics, 2012, 28, 277-278.	1.8	59
25	Implementing individualized medicine into the medical practice. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 15-23.	0.7	58
26	Experience with precision genomics and tumor board, indicates frequent target identification, but barriers to delivery. Oncotarget, 2017, 8, 27145-27154.	0.8	55
27	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-G1098.	1.6	52
28	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. Genetics in Medicine, 2019, 21, 2723-2733.	1.1	48
29	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an epismutation of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	2.6	48
30	Larval Zebrafish Model for FDA-Approved Drug Repositioning for Tobacco Dependence Treatment. PLoS ONE, 2014, 9, e90467.	1.1	48
31	Expanding DNA diagnostic panel testing: is more better?. Expert Review of Molecular Diagnostics, 2011, 11, 703-709.	1.5	46
32	Identifying secretomes in people, pufferfish and pigs. Nucleic Acids Research, 2004, 32, 1414-1421.	6.5	44
33	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021.	9.4	44
34	De novo <i>DDX3X</i> missense variants in males appear viable and contribute to syndromic intellectual disability. American Journal of Medical Genetics, Part A, 2019, 179, 570-578.	0.7	42
35	Zebrafish approaches enhance the translational research tackle box. Translational Research, 2014, 163, 65-78.	2.2	40
36	RINT1 Bi-allelic Variations Cause Infantile-Onset Recurrent Acute Liver Failure and Skeletal Abnormalities. American Journal of Human Genetics, 2019, 105, 108-121.	2.6	39

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37	LeafCutterMD: an algorithm for outlier splicing detection in rare diseases. <i>Bioinformatics</i> , 2020, 36, 4609-4615.	1.8	38
38	Development and Verification of an RNA Sequencing (RNA-Seq) Assay for the Detection of Gene Fusions in Tumors. <i>Journal of Molecular Diagnostics</i> , 2018, 20, 495-511.	1.2	36
39	Novel <i>NR2F1</i> 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. <i>Journal of Physical Education and Sports Management</i> , 2017, 3, a002162.	0.5	33
40	De novo variants in <i>FBXO11</i> cause a syndromic form of intellectual disability with behavioral problems and dysmorphisms. <i>European Journal of Human Genetics</i> , 2019, 27, 738-746.	1.4	32
41	Truncating <i>SRCAP</i> variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature. <i>American Journal of Human Genetics</i> , 2021, 108, 1053-1068.	2.6	31
42	Computational classification of classically secreted proteins. <i>Drug Discovery Today</i> , 2007, 12, 234-240.	3.2	30
43	Pharmacogenomic findings from clinical whole exome sequencing of diagnostic odyssey patients. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2017, 5, 269-279.	0.6	30
44	Missense Variants in the Histone Acetyltransferase Complex Component Gene <i>TRRAP</i> Cause Autism and Syndromic Intellectual Disability. <i>American Journal of Human Genetics</i> , 2019, 104, 530-541.	2.6	30
45	Impact of sample acquisition and linear amplification on gene expression profiling of lung adenocarcinoma: laser capture micro-dissection cell-sampling versus bulk tissue-sampling. <i>BMC Medical Genomics</i> , 2009, 2, 13.	0.7	29
46	"Big Data" in Laboratory Medicine. <i>Clinical Chemistry</i> , 2015, 61, 1433-1440.	1.5	29
47	Utility of DNA, RNA, Protein, and Functional Approaches to Solve Cryptic Immunodeficiencies. <i>Journal of Clinical Immunology</i> , 2018, 38, 307-319.	2.0	29
48	Next Generation Sequencing of Sporadic Vestibular Schwannoma: Necessity of Biallelic <i>NF2</i> Inactivation and Implications of Accessory Non- <i>NF2</i> Variants. <i>Otology and Neurotology</i> , 2018, 39, e860-e871.	0.7	29
49	Kinase Genotype Analysis of Gastric Gastrointestinal Stromal Tumor Cytology Samples Using Targeted Next-Generation Sequencing. <i>Clinical Gastroenterology and Hepatology</i> , 2015, 13, 202-206.	2.4	28
50	Mitochondrial 3-Hydroxy-3-Methylglutaryl-CoA Synthase Deficiency: Unique Presenting Laboratory Values and a Review of Biochemical and Clinical Features. <i>JIMD Reports</i> , 2017, 40, 63-69.	0.7	27
51	A tailored approach to fusion transcript identification increases diagnosis of rare inherited disease. <i>PLoS ONE</i> , 2019, 14, e0223337.	1.1	27
52	De novo <i>TBR1</i> variants cause a neurocognitive phenotype with ID and autistic traits: report of 25 new individuals and review of the literature. <i>European Journal of Human Genetics</i> , 2020, 28, 770-782.	1.4	27
53	Pilot study of small bowel mucosal gene expression in patients with irritable bowel syndrome with diarrhea. <i>American Journal of Physiology - Renal Physiology</i> , 2016, 311, G365-G376.	1.6	25
54	Recommendations for performance optimizations when using <i>GATK3.8</i> and <i>GATK4</i> . <i>BMC Bioinformatics</i> , 2019, 20, 557.	1.2	25

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55	A second cohort of CHD3 patients expands the molecular mechanisms known to cause Snijders Blok-Campeau syndrome. <i>European Journal of Human Genetics</i> , 2020, 28, 1422-1431.	1.4	25
56	Impact of integrated translational research on clinical exome sequencing. <i>Genetics in Medicine</i> , 2021, 23, 498-507.	1.1	24
57	TNPO2 variants associate with human developmental delays, neurologic deficits, and dysmorphic features and alter TNPO2 activity in <i>Drosophila</i> . <i>American Journal of Human Genetics</i> , 2021, 108, 1669-1691.	2.6	23
58	Novel de novo variant in <i>EBF3</i> 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. <i>Journal of Physical Education and Sports Management</i> , 2017, 3, a001743.	0.5	22
59	<i>PCNT</i> point mutations and familial intracranial aneurysms. <i>Neurology</i> , 2018, 91, e2170-e2181.	1.5	22
60	Whole Exome Sequencing Implicates an <i>INO80D</i> Mutation in a Syndrome of Aortic Hypoplasia, Premature Atherosclerosis, and Arterial Stiffness. <i>Circulation: Cardiovascular Genetics</i> , 2014, 7, 607-614.	5.1	21
61	Silent Tyrosinemia Type I Without Elevated Tyrosine or Succinylacetone Associated with Liver Cirrhosis and Hepatocellular Carcinoma. <i>Human Mutation</i> , 2016, 37, 1097-1105.	1.1	21
62	Clinical Applications and Utility of a Precision Medicine Approach for Patients With Unexplained Cytopenias. <i>Mayo Clinic Proceedings</i> , 2019, 94, 1753-1768.	1.4	21
63	Widening of the genetic and clinical spectrum of Lambâ€“Shaffer syndrome, a neurodevelopmental disorder due to <i>SOX5</i> haploinsufficiency. <i>Genetics in Medicine</i> , 2020, 22, 524-537.	1.1	21
64	Clinical Correlates and Treatment Outcomes for Patients With Short Telomere Syndromes. <i>Mayo Clinic Proceedings</i> , 2018, 93, 834-839.	1.4	20
65	COVID-19 Mortality Prediction From Deep Learning in a Large Multistate Electronic Health Record and Laboratory Information System Data Set: Algorithm Development and Validation. <i>Journal of Medical Internet Research</i> , 2021, 23, e30157.	2.1	20
66	Bioinformatics Methods for Prioritizing Serum Biomarker Candidates. <i>Clinical Chemistry</i> , 2006, 52, 2162-2164.	1.5	19
67	Quantitating tissue specificity of human genes to facilitate biomarker discovery. <i>Bioinformatics</i> , 2007, 23, 1348-1355.	1.8	19
68	Data Mining for Biomarker Development: A Review of Tissue Specificity Analysis. <i>Clinics in Laboratory Medicine</i> , 2008, 28, 127-143.	0.7	19
69	Characterization of three ciliopathy pedigrees expands the phenotype associated with biallelic <i>C2CD3</i> variants. <i>European Journal of Human Genetics</i> , 2018, 26, 1797-1809.	1.4	19
70	Molecular modeling and molecular dynamic simulation of the effects of variants in the <i>TGFBR2</i> kinase domain as a paradigm for interpretation of variants obtained by next generation sequencing. <i>PLoS ONE</i> , 2017, 12, e0170822.	1.1	19
71	A Novel Kleefstra Syndrome-associated Variant That Affects the Conserved TPLX Motif within the Ankyrin Repeat of <i>EHMT1</i> Leads to Abnormal Protein Folding. <i>Journal of Biological Chemistry</i> , 2017, 292, 3866-3876.	1.6	18
72	Proteinâ€“elongating mutations in <i>MYH11</i> are implicated in a dominantly inherited smooth muscle dysmotility syndrome with severe esophageal, gastric, and intestinal disease. <i>Human Mutation</i> , 2020, 41, 973-982.	1.1	18

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73	SPECC1L regulates palate development downstream of IRF6. <i>Human Molecular Genetics</i> , 2020, 29, 845-858.	1.4	18
74	The prevalence of diseases caused by lysosome-related genes in a cohort of undiagnosed patients. <i>Molecular Genetics and Metabolism Reports</i> , 2017, 13, 46-51.	0.4	17
75	RNA-seq detects a <i>SAMD12</i> - <i>EXT1</i> fusion transcript and leads to the discovery of an <i>EXT1</i> deletion in a child with multiple osteochondromas. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2019, 7, e00560.	0.6	17
76	The Zebrafish Secretome. <i>Zebrafish</i> , 2008, 5, 131-138.	0.5	16
77	Germline variants in tumor suppressor <i>FBXW7</i> lead to impaired ubiquitination and a neurodevelopmental syndrome. <i>American Journal of Human Genetics</i> , 2022, 109, 601-617.	2.6	16
78	Variants in <i>DOCK3</i> cause developmental delay and hypotonia. <i>European Journal of Human Genetics</i> , 2019, 27, 1225-1234.	1.4	15
79	A form of muscular dystrophy associated with pathogenic variants in <i>JAG2</i> . <i>American Journal of Human Genetics</i> , 2021, 108, 840-856.	2.6	15
80	A novel <i>ANO3</i> variant identified in a 53-year-old woman presenting with hyperkinetic dysarthria, blepharospasm, hyperkinesias, and complex motor tics. <i>BMC Medical Genetics</i> , 2016, 17, 93.	2.1	14
81	Comparative analysis of de novo assemblers for variation discovery in personal genomes. <i>Briefings in Bioinformatics</i> , 2018, 19, 893-904.	3.2	14
82	A case of <i>YY1</i> -associated syndromic learning disability or Gabriele de Vries syndrome with myasthenia gravis. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2846-2849.	0.7	14
83	Molecular characterization of known and novel <i>ACVR1</i> variants in phenotypes of aberrant ossification. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1764-1777.	0.7	13
84	<i>CSNK2B</i> : A broad spectrum of neurodevelopmental disability and epilepsy severity. <i>Epilepsia</i> , 2021, 62, e103-e109.	2.6	13
85	Genomics Integration Into Nephrology Practice. <i>Kidney Medicine</i> , 2021, 3, 785-798.	1.0	13
86	De novo coding variants in the <i>AGO1</i> gene cause a neurodevelopmental disorder with intellectual disability. <i>Journal of Medical Genetics</i> , 2022, 59, 965-975.	1.5	13
87	Assessing Human Genetic Variations in Glucose Transporter <i>SLC2A10</i> and Their Role in Altering Structural and Functional Properties. <i>Frontiers in Genetics</i> , 2018, 9, 276.	1.1	12
88	Haploinsufficiency as a disease mechanism in <i>GNB1</i> -associated neurodevelopmental disorder. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2020, 8, e1477.	0.6	12
89	De novo variants of <i>NR4A2</i> are associated with neurodevelopmental disorder and epilepsy. <i>Genetics in Medicine</i> , 2020, 22, 1413-1417.	1.1	12
90	AMOD: a morpholino oligonucleotide selection tool. <i>Nucleic Acids Research</i> , 2005, 33, W506-W511.	6.5	11

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91	Multigenerational pedigree with STAR syndrome: A novel FAM58A variant and expansion of the phenotype. , 2017, 173, 1328-1333.		11
92	Functional Analysis of the SIM1 Variant p.G715V in 2 Patients With Obesity. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 355-361.	1.8	11
93	Pathogenic mutations in the chromokinesin KIF22 disrupt anaphase chromosome segregation. ELife, 0, 11, .	2.8	11
94	Target selection forDanio rerio functional genomics. Genesis, 2001, 30, 123-125.	0.8	10
95	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.	1.4	10
96	Clinical characteristics and platelet phenotype in a family with<i>RUNX1</i>mutated thrombocytopenia. Leukemia and Lymphoma, 2017, 58, 1963-1967.	0.6	10
97	Molecular modeling of LDLR aids interpretation of genomic variants. Journal of Molecular Medicine, 2019, 97, 533-540.	1.7	10
98	<scp><i>TSPEAR</i></scp> 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.	0.7	10
99	Autosomal Recessive Cerebellar Atrophy and Spastic Ataxia in Patients With Pathogenic Biallelic Variants in GEMIN5. Frontiers in Cell and Developmental Biology, 2022, 10, 783762.	1.8	10
100	Familial Creutzfeldt-Jakob Disease: Case report and role of genetic counseling in post mortem testing. Prion, 2016, 10, 502-506.	0.9	9
101	Pathogenic Variant in<i> ACTB</i>, p.Arg183Trp, Causes Juvenile-Onset Dystonia, Hearing Loss, and Developmental Delay without Midline Malformation. Case Reports in Genetics, 2017, 2017, 1-4.	0.1	9
102	Novel germline missense <i>DDX41</i> variant in a patient with an adult-onset myeloid neoplasm with excess blasts without dysplasia. Leukemia and Lymphoma, 2019, 60, 1337-1339.	0.6	9
103	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.	1.2	9
104	SeekFusion - A Clinically Validated Fusion Transcript Detection Pipeline for PCR-Based Next-Generation Sequencing of RNA. Frontiers in Genetics, 2021, 12, 739054.	1.1	9
105	A novel <i>de novo</i> frameshift deletion in <i><scp>EHMT</scp>1</i> in a patient with Kleefstra Syndrome results in decreased H3K9 dimethylation. Molecular Genetics & Genomic Medicine, 2017, 5, 141-146.	0.6	8
106	Familial chronic megacolon presenting in childhood or adulthood: Seeking the presumed gene association. Neurogastroenterology and Motility, 2019, 31, e13550.	1.6	8
107	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.	1.4	8
108	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, 550-556.e1.	0.5	7

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109	Functional validation reveals the novel missense V419L variant in <i>TGFBR2</i> associated with Loey's-Dietz syndrome (LDS) impairs canonical TGF- $\beta$ 2 signaling. <i>Journal of Physical Education and Sports Management</i> , 2017, 3, a001727.	0.5	7
110	Early-onset limb-girdle muscular dystrophy-2L in a female athlete. <i>Muscle and Nerve</i> , 2017, 55, E19-E21.	1.0	7
111	Extension of the mutational and clinical spectrum of <i>SOX2</i> related disorders: Description of six new cases and a novel association with suprasellar teratoma. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2710-2719.	0.7	7
112	Co-occurrence of a maternally inherited DNMT3A duplication and a paternally inherited pathogenic variant in EZH2 in a child with growth retardation and severe short stature: atypical Weaver syndrome or evidence of a DNMT3A dosage effect?. <i>Journal of Physical Education and Sports Management</i> , 2018, 4, a002899.	0.5	7
113	Computational Detection of Known Pathogenic Gene Fusions in a Normal Tissue Database and Implications for Genetic Disease Research. <i>Frontiers in Genetics</i> , 2020, 11, 173.	1.1	7
114	Expansion of the Genotypic and Phenotypic Spectrum of WASF1-Related Neurodevelopmental Disorder. <i>Brain Sciences</i> , 2021, 11, 931.	1.1	7
115	Design considerations for workflow management systems use in production genomics research and the clinic. <i>Scientific Reports</i> , 2021, 11, 21680.	1.6	7
116	A Systematic Method for Selection of Promising Serum Protein Biomarkers to Improve Prostate Cancer (PCa) Detection. <i>Clinical Chemistry</i> , 2006, 52, 2159-2162.	1.5	6
117	Developmental delay, coarse facial features, and epilepsy in a patient with <i>EXT2</i> gene variants. <i>Clinical Case Reports (discontinued)</i> , 2019, 7, 632-637.	0.2	6
118	X-Linked Lymphoproliferative Syndrome Presenting as Adult-Onset Multi-Infarct Dementia. <i>Journal of Neuropathology and Experimental Neurology</i> , 2019, 78, 460-466.	0.9	6
119	Exome sequencing confirms diagnosis of kabuki syndrome in an-adult with hodgkin lymphoma and unusually severe multisystem phenotype. <i>Clinical Immunology</i> , 2019, 207, 55-57.	1.4	6
120	Genetic variants in DGAT1 cause diverse clinical presentations of malnutrition through a specific molecular mechanism. <i>European Journal of Medical Genetics</i> , 2020, 63, 103817.	0.7	6
121	Novel loss-of-function variants in TRIO are associated with neurodevelopmental disorder: case report. <i>BMC Medical Genetics</i> , 2020, 21, 219.	2.1	6
122	Nail-patella-like renal disease masquerading as Fabry disease on kidney biopsy: a case report. <i>BMC Nephrology</i> , 2020, 21, 341.	0.8	6
123	Next-Generation Sequencing of CYP2C19 in Stent Thrombosis: Implications for Clopidogrel Pharmacogenomics. <i>Cardiovascular Drugs and Therapy</i> , 2021, 35, 549-559.	1.3	6
124	De novo <i>PBX1</i> variant in a patient with glaucoma, kidney anomalies, and developmental delay: An expansion of the <i>CAKUTHED</i> phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 919-925.	0.7	6
125	Gain and loss of TASK3 channel function and its regulation by novel variation cause KCNK9 imprinting syndrome. <i>Genome Medicine</i> , 2022, 14, .	3.6	6
126	Three rare disease diagnoses in one patient through exome sequencing. <i>Journal of Physical Education and Sports Management</i> , 2019, 5, a004390.	0.5	5



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127	An intragenic duplication of TRPS1 leading to abnormal transcripts and causing trichorhinophalangeal syndrome type I. <i>Journal of Physical Education and Sports Management</i> , 2019, 5, a004655.	0.5	5
128	Novel biallelic variants in MSTO1 associated with mitochondrial myopathy. <i>Journal of Physical Education and Sports Management</i> , 2019, 5, a004309.	0.5	5
129	Interpretation challenges of novel dual-class missense and splice-impacting variant in POLR3A-related late-onset hereditary spastic ataxia. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2020, 8, e1341.	0.6	5
130	HELLO: improved neural network architectures and methodologies for small variant calling. <i>BMC Bioinformatics</i> , 2021, 22, 404.	1.2	5
131	Improved Characterization of Complex $\beta$ -Globin Gene Cluster Structural Variants Using Long-Read Sequencing. <i>Journal of Molecular Diagnostics</i> , 2021, 23, 1732-1740.	1.2	5
132	Novel Pathogenic Variant in TGFBR2 Confirmed by Molecular Modeling Is a Rare Cause of Loey-Dietz Syndrome. <i>Case Reports in Genetics</i> , 2017, 2017, 1-4.	0.1	4
133	GFAP canonical transcript may not be suitable for the diagnosis of adult-onset Alexander disease. <i>Acta Neuropathologica Communications</i> , 2018, 6, 112.	2.4	4
134	Expansion of <i>PURA</i> -Related Phenotypes and Discovery of a Novel <i>PURA</i> Variant: A Case Report. <i>Child Neurology Open</i> , 2020, 7, 2329048X2095500.	0.5	4
135	Defining the genotypic and phenotypic spectrum of X-linked <i>MSL3</i> -related disorder. <i>Genetics in Medicine</i> , 2021, 23, 384-395.	1.1	4
136	<i>LPCAT1-TERT</i> fusions are uniquely recurrent in epithelioid trophoblastic tumors and positively regulate cell growth. <i>PLoS ONE</i> , 2021, 16, e0250518.	1.1	4
137	"The molecule's the thing": the promise of molecular modeling and dynamic simulations in aiding the prioritization and interpretation of genomic testing results. <i>F1000Research</i> , 2016, 5, 766.	0.8	4
138	Heterozygous variants in <i>PRPF8</i> are associated with neurodevelopmental disorders. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2750-2759.	0.7	4
139	Functional characterization of a <i>GFAP</i> variant of uncertain significance in an Alexander disease case within the setting of an individualized medicine clinic. <i>Clinical Case Reports (discontinued)</i> , 2016, 4, 885-895.	0.2	3
140	Preemptive sequencing in the genomic medicine era. <i>Expert Review of Precision Medicine and Drug Development</i> , 2017, 2, 91-98.	0.4	3
141	Improving Single-Nucleotide Polymorphism-Based Fetal Fraction Estimation of Maternal Plasma Circulating Cell-Free DNA Using Bayesian Hierarchical Models. <i>Journal of Computational Biology</i> , 2018, 25, 1040-1049.	0.8	3
142	An activating germline <i>IDH1</i> variant associated with a tumor entity characterized by unilateral and bilateral chondrosarcoma of the mastoid. <i>Human Genetics and Genomics Advances</i> , 2020, 1, 100006.	1.0	3
143	Jumonji domain containing 1C ( <i>JMJD1C</i> ) sequence variants in seven patients with autism spectrum disorder, intellectual disability and seizures. <i>European Journal of Medical Genetics</i> , 2020, 63, 103850.	0.7	3
144	Epigenetic alteration contributes to the transcriptional reprogramming in T-cell prolymphocytic leukemia. <i>Scientific Reports</i> , 2021, 11, 8318.	1.6	3

#	ARTICLE	IF	CITATIONS
145	Functional validation of TERT and TERC variants of uncertain significance in patients with short telomere syndromes. <i>Blood Cancer Journal</i> , 2020, 10, 120.	2.8	2
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147	Late onset asymptomatic pancreatic neuroendocrine tumor – A case report on the phenotypic expansion for MEN1. <i>Hereditary Cancer in Clinical Practice</i> , 2017, 15, 10.	0.6	1
148	Proposal for Modification of Cahan's Criteria Utilizing Molecular Genetic Analyses for Cases without Baseline Histopathology: A Unique Method Applicable to Primary Radiosurgery. <i>Journal of Neurological Surgery, Part B: Skull Base</i> , 2019, 80, 010-017.	0.4	1
149	Biallelic variants in <i>PROZ</i> as a cause of hypercoagulability and livedo racemosa. <i>Thrombosis Research</i> , 2020, 195, 187-189.	0.8	1
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151	A novel missense variant and multiexon deletion causing a delayed presentation of xeroderma pigmentosum, group C. <i>Journal of Physical Education and Sports Management</i> , 2020, 6, a005165.	0.5	1
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157	Interpretation of Dihydrorhodamine-1,2,3 Flow Cytometry in Chronic Granulomatous Disease: an Atypical Exemplar. <i>Journal of Clinical Immunology</i> , 2022, , 1.	2.0	0
158	Editorial: Clinical Genome Sequencing: Bioinformatics Challenges and Key Considerations. <i>Frontiers in Genetics</i> , 2022, 13, 896032.	1.1	0