Eric W Klee

List of Publications by Year
in descending order

158 papers	5,104 citations	117625 34 h-index	118850 62 g-index
164 all docs	164 docs citations	164 times ranked	10279 citing authors

#	Article	IF	Citations
1	De novo <scp>PBX1</scp> variant in a patient with glaucoma, kidney anomalies, and developmental delay: An expansion of the <scp>CAKUTHED</scp> phenotype. American Journal of Medical Genetics, Part A, 2022, 188, 919-925.	1.2	6
2	De novo coding variants in the AGO1 gene cause a neurodevelopmental disorder with intellectual disability. Journal of Medical Genetics, 2022, 59, 965-975.	3.2	13
3	Autosomal Recessive Cerebellar Atrophy and Spastic Ataxia in Patients With Pathogenic Biallelic Variants in GEMIN5. Frontiers in Cell and Developmental Biology, 2022, 10, 783762.	3.7	10
4	Interpretation of Dihydrorhodamine-1,2,3 Flow Cytometry in Chronic Granulomatous Disease: an Atypical Exemplar. Journal of Clinical Immunology, 2022, , 1.	3.8	0
5	Editorial: Clinical Genome Sequencing: Bioinformatics Challenges and Key Considerations. Frontiers in Genetics, 2022, 13, 896032.	2.3	0
6	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 601-617.	6.2	16
7	Heterozygous variants in PRPF8 are associated with neurodevelopmental disorders. American Journal of Medical Genetics, Part A, 2022, 188, 2750-2759.	1.2	4
8	Functional validation of a novel AAAS variant in an atypical presentation of Allgrove syndrome. Molecular Genetics & Genomic Medicine, 2022, , e1966.	1.2	1
9	Gain and loss of TASK3 channel function and its regulation by novel variation cause KCNK9 imprinting syndrome. Genome Medicine, 2022, 14, .	8.2	6
10	Next-Generation Sequencing of CYP2C19 in Stent Thrombosis: Implications for Clopidogrel Pharmacogenomics. Cardiovascular Drugs and Therapy, 2021, 35, 549-559.	2.6	6
11	Impact of integrated translational research on clinical exome sequencing. Genetics in Medicine, 2021, 23, 498-507.	2.4	24
12	Defining the genotypic and phenotypic spectrum of X-linked MSL3-related disorder. Genetics in Medicine, 2021, 23, 384-395.	2.4	4
13	Recurrent ganglioneuroma in <scp>PTPN11</scp>â€associated Noonan syndrome: A case report and literature review. American Journal of Medical Genetics, Part A, 2021, 185, 1883-1887.	1.2	2
14	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.	6.2	48
15	Epigenetic alteration contributes to the transcriptional reprogramming in T-cell prolymphocytic leukemia. Scientific Reports, 2021, 11, 8318.	3.3	3
16	A form of muscular dystrophy associated with pathogenic variants in JAG2. American Journal of Human Genetics, 2021, 108, 840-856.	6.2	15
17	LPCAT1-TERT fusions are uniquely recurrent in epithelioid trophoblastic tumors and positively regulate cell growth. PLoS ONE, 2021, 16, e0250518.	2.5	4
18	<scp>TSPEAR</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.	1.2	10

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19	CSNK2B: A broad spectrum of neurodevelopmental disability and epilepsy severity. Epilepsia, 2021, 62, e103-e109.	5.1	13
20	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.	6.2	31
21	Genomics Integration Into Nephrology Practice. Kidney Medicine, 2021, 3, 785-798.	2.0	13
22	P2T2: Protein Panoramic annoTation Tool for the interpretation of protein coding genetic variants. JAMIA Open, 2021, 4, ooab065.	2.0	1
23	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021.	21.4	44
24	Expansion of the Genotypic and Phenotypic Spectrum of WASF1-Related Neurodevelopmental Disorder. Brain Sciences, 2021, 11, 931.	2.3	7
25	HELLO: improved neural network architectures and methodologies for small variant calling. BMC Bioinformatics, 2021, 22, 404.	2.6	5
26	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.	6.2	23
27	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.	4.3	20
28	SeekFusion - A Clinically Validated Fusion Transcript Detection Pipeline for PCR-Based Next-Generation Sequencing of RNA. Frontiers in Genetics, 2021, 12, 739054.	2.3	9
29	Design considerations for workflow management systems use in production genomics research and the clinic. Scientific Reports, 2021, 11, 21680.	3.3	7
30	Improved Characterization of Complex Î²-Globin Gene Cluster Structural Variants Using Long-Read Sequencing. Journal of Molecular Diagnostics, 2021, 23, 1732-1740.	2.8	5
31	Widening of the genetic and clinical spectrum of Lambâ€“Shaffer syndrome, a neurodevelopmental disorder due to SOX5 haploinsufficiency. Genetics in Medicine, 2020, 22, 524-537.	2.4	21
32	Genetic variants in DGAT1 cause diverse clinical presentations of malnutrition through a specific molecular mechanism. European Journal of Medical Genetics, 2020, 63, 103817.	1.3	6
33	Biallelic variants in PROZ as a cause of hypercoagulability and livedo racemosa. Thrombosis Research, 2020, 195, 187-189.	1.7	1
34	An activating germline IDH1 variant associated with a tumor entity characterized by unilateral and bilateral chondrosarcoma of the mastoid. Human Genetics and Genomics Advances, 2020, 1, 100006.	1.7	3
35	Functional validation of TERT and TERC variants of uncertain significance in patients with short telomere syndromes. Blood Cancer Journal, 2020, 10, 120.	6.2	2
36	Novel loss-of-function variants in TRIO are associated with neurodevelopmental disorder: case report. BMC Medical Genetics, 2020, 21, 219.	2.1	6

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37	Expansion of PURA-Related Phenotypes and Discovery of a Novel PURA Variant: A Case Report. Child Neurology Open, 2020, 7, 2329048X2095500.	1.1	4
38	Successful Treatment of Skewed Lyonization Associated with X-Linked CGD in a Female Presenting with Recalcitrant Crohnâ€™s Disease. Journal of Clinical Immunology, 2020, 40, 1056-1061.	3.8	1
39	Nail-patella-like renal disease masquerading as Fabry disease on kidney biopsy: a case report. BMC Nephrology, 2020, 21, 341.	1.8	6
40	Best practices for the analytical validation of clinical whole-genome sequencing intended for the diagnosis of germline disease. Npj Genomic Medicine, 2020, 5, 47.	3.8	67
41	Haploinsufficiency as a disease mechanism in GNB1â€associated neurodevelopmental disorder. Molecular Genetics & Genomic Medicine, 2020, 8, e1477.	1.2	12
42	Interpretation challenges of novel dualâ€class missense and spliceâ€impacting variant in POLR3Aâ€related lateâ€onset hereditary spastic ataxia. Molecular Genetics & Genomic Medicine, 2020, 8, e1341.	1.2	5
43	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, a005165.	1.2	1
44	Genome-wide detection of tandem DNA repeats that are expanded in autism. Nature, 2020, 586, 80-86.	27.8	155
45	De novo variants of NR4A2 are associated with neurodevelopmental disorder and epilepsy. Genetics in Medicine, 2020, 22, 1413-1417.	2.4	12
46	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.	2.8	25
47	LeafCutterMD: an algorithm for outlier splicing detection in rare diseases. Bioinformatics, 2020, 36, 4609-4615.	4.1	38
48	Computational Detection of Known Pathogenic Gene Fusions in a Normal Tissue Database and Implications for Genetic Disease Research. Frontiers in Genetics, 2020, 11, 173.	2.3	7
49	Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. Neuron, 2020, 106, 404-420.e8.	8.1	121
50	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.	2.5	9
51	Functional Analysis of the SIM1 Variant p.G715V in 2 Patients With Obesity. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 355-361.	3.6	11
52	Proteinâ€elongating mutations inMYH11are implicated in a dominantly inherited smooth muscle dysmotility syndrome with severe esophageal, gastric, and intestinal disease. Human Mutation, 2020, 41, 973-982.	2.5	18
53	SPECC1L regulates palate development downstream of IRF6. Human Molecular Genetics, 2020, 29, 845-858.	2.9	18
54	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.	2.8	27

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55	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.	1.3	3
56	Genomic Profiling Reveals Molecular Heterogeneity in Patients with Richter's Syndrome (RS) and Progressive Chronic Lymphocytic Leukemia (CLL). Blood, 2020, 136, 16-17.	1.4	1
57	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
58	Molecular characterization of known and novel ACVR1 variants in phenotypes of aberrant ossification. American Journal of Medical Genetics, Part A, 2019, 179, 1764-1777.	1.2	13
59	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. Genetics in Medicine, 2019, 21, 2723-2733.	2.4	48
60	RINT1 Bi-allelic Variations Cause Infantile-Onset Recurrent Acute Liver Failure and Skeletal Abnormalities. American Journal of Human Genetics, 2019, 105, 108-121.	6.2	39
61	Clinical Applications and Utility of a Precision Medicine Approach for Patients With Unexplained Cytopenias. Mayo Clinic Proceedings, 2019, 94, 1753-1768.	3.0	21
62	Three rare disease diagnoses in one patient through exome sequencing. Journal of Physical Education and Sports Management, 2019, 5, a004390.	1.2	5
63	Sentieon DNASeq Variant Calling Workflow Demonstrates Strong Computational Performance and Accuracy. Frontiers in Genetics, 2019, 10, 736.	2.3	131
64	A tailored approach to fusion transcript identification increases diagnosis of rare inherited disease. PLoS ONE, 2019, 14, e0223337.	2.5	27
65	Familial chronic megacolon presenting in childhood or adulthood: Seeking the presumed gene association. Neurogastroenterology and Motility, 2019, 31, e13550.	3.0	8
66	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.	2.8	32
67	Developmental delay, coarse facial features, and epilepsy in a patient with EXT2 gene variants. Clinical Case Reports (discontinued), 2019, 7, 632-637.	0.5	6
68	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.	2.8	8
69	X-Linked Lymphoproliferative Syndrome Presenting as Adult-Onset Multi-Infarct Dementia. Journal of Neuropathology and Experimental Neurology, 2019, 78, 460-466.	1.7	6
70	Variants in DOCK3 cause developmental delay and hypotonia. European Journal of Human Genetics, 2019, 27, 1225-1234.	2.8	15
71	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.	1.2	42
72	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.	6.2	30

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73	Molecular modeling of LDLR aids interpretation of genomic variants. Journal of Molecular Medicine, 2019, 97, 533-540.	3.9	10
74	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, 010-017.	0.8	1
75	An intragenic duplication of TRPS1 leading to abnormal transcripts and causing trichorhinophalangeal syndrome type I. Journal of Physical Education and Sports Management, 2019, 5, a004655.	1.2	5
76	Novel biallelic variants in MSTO1 associated with mitochondrial myopathy. Journal of Physical Education and Sports Management, 2019, 5, a004309.	1.2	5
77	Recommendations for performance optimizations when using GATK3.8 and GATK4. BMC Bioinformatics, 2019, 20, 557.	2.6	25
78	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.	1.3	9
79	RNAâ€Seq detects a SAMD12â€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.	1.2	17
80	Exome sequencing confirms diagnosis of kabuki syndrome in an-adult with hodgkin lymphoma and unusually severe multisystem phenotype. Clinical Immunology, 2019, 207, 55-57.	3.2	6
81	Loss of the sphingolipid desaturase DEGS1 causes hypomyelinating leukodystrophy. Journal of Clinical Investigation, 2019, 129, 1240-1256.	8.2	68
82	Long Range Sequencing Shows Improved Resolution in the Detection of Beta Globin Cluster Variants. Blood, 2019, 134, 3548-3548.	1.4	1
83	Utility of DNA, RNA, Protein, and Functional Approaches to Solve Cryptic Immunodeficiencies. Journal of Clinical Immunology, 2018, 38, 307-319.	3.8	29
84	Arterial tortuosity syndrome: 40 new families and literature review. Genetics in Medicine, 2018, 20, 1236-1245.	2.4	66
85	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.	6.2	105
86	Comparative analysis of de novo assemblers for variation discovery in personal genomes. Briefings in Bioinformatics, 2018, 19, 893-904.	6.5	14
87	Standards and Guidelines for Validating Next-Generation Sequencing Bioinformatics Pipelines. Journal of Molecular Diagnostics, 2018, 20, 4-27.	2.8	341
88	PCNT point mutations and familial intracranial aneurysms. Neurology, 2018, 91, e2170-e2181.	1.1	22
89	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.	1.2	7
90	Clinical Correlates and Treatment Outcomes for Patients With Short Telomere Syndromes. Mayo Clinic Proceedings, 2018, 93, 834-839.	3.0	20

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91	A case of YY1â€associated syndromic learning disability or Gabrieleâ€de Vries syndrome with myasthenia gravis. American Journal of Medical Genetics, Part A, 2018, 176, 2846-2849.	1.2	14
92	GFAP canonical transcript may not be suitable for the diagnosis of adult-onset Alexander disease. Acta Neuropathologica Communications, 2018, 6, 112.	5.2	4
93	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.	6.2	87
94	Assessing Human Genetic Variations in Glucose Transporter SLC2A10 and Their Role in Altering Structural and Functional Properties. Frontiers in Genetics, 2018, 9, 276.	2.3	12
95	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.	1.3	29
96	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?. Journal of Physical Education and Sports Management, 2018, 4, a002899.	1.2	7
97	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.	1.6	3
98	Characterization of three ciliopathy pedigrees expands the phenotype associated with biallelic C2CD3 variants. European Journal of Human Genetics, 2018, 26, 1797-1809.	2.8	19
99	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.	2.8	36
100	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.	2.8	10
101	A novel de novo frameshift deletion in <scp>EHMT</scp>1 in a patient with Kleefstra Syndrome results in decreased H3K9 dimethylation. Molecular Genetics & Genomic Medicine, 2017, 5, 141-146.	1.2	8
102	Multigenerational pedigree with STAR syndrome: A novel FAM58A variant and expansion of the phenotype. , 2017, 173, 1328-1333.		11
103	A Novel Kleefstra Syndrome-associated Variant That Affects the Conserved TPLX Motif within the Ankyrin Repeat of EHMT1 Leads to Abnormal Protein Folding. Journal of Biological Chemistry, 2017, 292, 3866-3876.	3.4	18
104	Preemptive sequencing in the genomic medicine era. Expert Review of Precision Medicine and Drug Development, 2017, 2, 91-98.	0.7	3
105	Pharmacogenomic findings from clinical whole exome sequencing of diagnostic odyssey patients. Molecular Genetics & Genomic Medicine, 2017, 5, 269-279.	1.2	30
106	Novel de novo variant in EBF3 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.	1.2	22
107	Disruption of the ATXN1â€“CIC complex causes a spectrum of neurobehavioral phenotypes in mice and humans. Nature Genetics, 2017, 49, 527-536.	21.4	113
108	Clinical characteristics and platelet phenotype in a family withRUNX1mutated thrombocytopenia. Leukemia and Lymphoma, 2017, 58, 1963-1967.	1.3	10

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109	Novel NR2F1 variants likely disrupt DNA binding: molecular modeling in two cases, review of published cases, genotypeâ€“phenotype correlation, and phenotypic expansion of the Boschâ€“Boonstraâ€“Schaaf optic atrophy syndrome. Journal of Physical Education and Sports Management, 2017, 3, a002162.	1.2	33
110	The prevalence of diseases caused by lysosome-related genes in a cohort of undiagnosed patients. Molecular Genetics and Metabolism Reports, 2017, 13, 46-51.	1.1	17
111	Clinical spectrum and genotypeâ€“phenotype associations of KCNA2-related encephalopathies. Brain, 2017, 140, 2337-2354.	7.6	117
112	Functional validation reveals the novel missense V419L variant in TGFBR2 associated with Loeysâ€“Dietz syndrome (LDS) impairs canonical TGF-Î² signaling. Journal of Physical Education and Sports Management, 2017, 3, a001727.	1.2	7
113	Earlyâ€onset limbâ€girdle muscular dystrophyâ€2L in a female athlete. Muscle and Nerve, 2017, 55, E19-E21.	2.2	7
114	Mitochondrial 3-Hydroxy-3-Methylglutaryl-CoA Synthase Deficiency: Unique Presenting Laboratory Values and a Review of Biochemical and Clinical Features. JIMD Reports, 2017, 40, 63-69.	1.5	27
115	Experience with precision genomics and tumor board, indicates frequent target identification, but barriers to delivery. Oncotarget, 2017, 8, 27145-27154.	1.8	55
116	Novel Pathogenic Variant in TGFBR2 Confirmed by Molecular Modeling Is a Rare Cause of Loeys-Dietz Syndrome. Case Reports in Genetics, 2017, 2017, 1-4.	0.2	4
117	Pathogenic Variant in ACTB, p.Arg183Trp, Causes Juvenile-Onset Dystonia, Hearing Loss, and Developmental Delay without Midline Malformation. Case Reports in Genetics, 2017, 2017, 1-4.	0.2	9
118	Late onset asymptomatic pancreatic neuroendocrine tumor â€“ A case report on the phenotypic expansion for MEN1. Hereditary Cancer in Clinical Practice, 2017, 15, 10.	1.5	1
119	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.	2.5	19
120	Whole Exome Sequencing Leading to the Diagnosis of Dysferlinopathy with a Novel Missense Mutation (c.959G>C). Case Reports in Genetics, 2016, 2016, 1-4.	0.2	0
121	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-G376.	3.4	25
122	Familial Creutzfeldt-Jakob Disease: Case report and role of genetic counseling in post mortem testing. Prion, 2016, 10, 502-506.	1.8	9
123	A novel ANO3 variant identified in a 53-year-old woman presenting with hyperkinetic dysarthria, blepharospasm, hyperkinesias, and complex motorÂtics. BMC Medical Genetics, 2016, 17, 93.	2.1	14
124	Functional characterization of a <scp>GFAP</scp> variant of uncertain significance in an Alexander disease case within the setting of an individualized medicine clinic. Clinical Case Reports (discontinued), 2016, 4, 885-895.	0.5	3
125	Silent Tyrosinemia Type I Without Elevated Tyrosine or Succinylacetone Associated with Liver Cirrhosis and Hepatocellular Carcinoma. Human Mutation, 2016, 37, 1097-1105.	2.5	21
126	Outcome of Whole Exome Sequencing for Diagnostic Odyssey Cases of an Individualized Medicine Clinic. Mayo Clinic Proceedings, 2016, 91, 297-307.	3.0	83

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127	Preemptive Pharmacogenomic Testing for Precision Medicine. Journal of Molecular Diagnostics, 2016, 18, 438-445.	2.8	171
128	â€œThe moleculeâ€™s the thing:â€•the promise of molecular modeling and dynamic simulations in aiding the prioritization and interpretation of genomic testing results. F1000Research, 2016, 5, 766.	1.6	4
129	â€œBig Dataâ€•in Laboratory Medicine. Clinical Chemistry, 2015, 61, 1433-1440.	3.2	29
130	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.	1.0	7
131	Confirming Variants in Next-Generation Sequencing Panel Testing by Sanger Sequencing. Journal of Molecular Diagnostics, 2015, 17, 456-461.	2.8	109
132	Kinase Genotype Analysis of Gastric Gastrointestinal Stromal Tumor Cytology Samples Using Targeted Next-Generation Sequencing. Clinical Gastroenterology and Hepatology, 2015, 13, 202-206.	4.4	28
133	Bioinformatics for Clinical Next Generation Sequencing. Clinical Chemistry, 2015, 61, 124-135.	3.2	114
134	Whole Exome Sequencing Implicates an INO80D Mutation in a Syndrome of Aortic Hypoplasia, Premature Atherosclerosis, and Arterial Stiffness. Circulation: Cardiovascular Genetics, 2014, 7, 607-614.	5.1	21
135	Integrated Genomic Characterization Reveals Novel, Therapeutically Relevant Drug Targets in FGFR and EGFR Pathways in Sporadic Intrahepatic Cholangiocarcinoma. PLoS Genetics, 2014, 10, e1004135.	3.5	292
136	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.	3.4	52
137	Zebrafish approaches enhance the translational research tackle box. Translational Research, 2014, 163, 65-78.	5.0	40
138	Preemptive Genotyping for Personalized Medicine: Design of the Right Drug, Right Dose, Right Timeâ€”Using Genomic Data to Individualize Treatment Protocol. Mayo Clinic Proceedings, 2014, 89, 25-33.	3.0	250
139	Implementing individualized medicine into the medical practice. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 15-23.	1.6	58
140	Larval Zebrafish Model for FDA-Approved Drug Repositioning for Tobacco Dependence Treatment. PLoS ONE, 2014, 9, e90467.	2.5	48
141	TREAT: a bioinformatics tool for variant annotations and visualizations in targeted and exome sequencing data. Bioinformatics, 2012, 28, 277-278.	4.1	59
142	Adrenomedullin is Up-regulated in Patients With Pancreatic Cancer and Causes Insulin Resistance in Î² Cells and Mice. Gastroenterology, 2012, 143, 1510-1517.e1.	1.3	145
143	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.	3.2	61
144	Zebrafish for the Study of the Biological Effects of Nicotine. Nicotine and Tobacco Research, 2011, 13, 301-312.	2.6	61

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145	Expanding DNA diagnostic panel testing: is more better?. Expert Review of Molecular Diagnostics, 2011, 11, 703-709.	3.1	46
146	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.	1.5	29
147	Data Mining for Biomarker Development: A Review of Tissue Specificity Analysis. Clinics in Laboratory Medicine, 2008, 28, 127-143.	1.4	19
148	The Zebrafish Secretome. Zebrafish, 2008, 5, 131-138.	1.1	16
149	Quantitating tissue specificity of human genes to facilitate biomarker discovery. Bioinformatics, 2007, 23, 1348-1355.	4.1	19
150	Computational classification of classically secreted proteins. Drug Discovery Today, 2007, 12, 234-240.	6.4	30
151	Genome-Wide Reverse Genetics Framework to Identify Novel Functions of the Vertebrate Secretome. PLoS ONE, 2006, 1, e104.	2.5	67
152	A Systematic Method for Selection of Promising Serum Protein Biomarkers to Improve Prostate Cancer (PCa1) Detection. Clinical Chemistry, 2006, 52, 2159-2162.	3.2	6
153	Bioinformatics Methods for Prioritizing Serum Biomarker Candidates. Clinical Chemistry, 2006, 52, 2162-2164.	3.2	19
154	AMOD: a morpholino oligonucleotide selection tool. Nucleic Acids Research, 2005, 33, W506-W511.	14.5	11
155	Evaluating eukaryotic secreted protein prediction. BMC Bioinformatics, 2005, 6, 256.	2.6	67
156	Identifying secretomes in people, pufferfish and pigs. Nucleic Acids Research, 2004, 32, 1414-1421.	14.5	44
157	Target selection forDanio rerio functional genomics. Genesis, 2001, 30, 123-125.	1.6	10
158	Pathogenic mutations in the chromokinesin KIF22 disrupt anaphase chromosome segregation. ELife, 0, 11, .	6.0	11