

Monkol Lek

List of Publications by Year in descending order

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Version: 2024-02-01

63
papers

22,064
citations

117625

34
h-index

118850

62
g-index

69
all docs

69
docs citations

69
times ranked

46159
citing authors

#	ARTICLE	IF	CITATIONS
1	A new phenotype of syndromic retinitis pigmentosa with myopathy is caused by mutations in retinol dehydrogenase 11. <i>Clinical Genetics</i> , 2022, 101, 448-453.	2.0	1
2	Centers for Mendelian Genomics: A decade of facilitating gene discovery. <i>Genetics in Medicine</i> , 2022, 24, 784-797.	2.4	44
3	Case Report: Two Families With HPDL Related Neurodegeneration. <i>Frontiers in Genetics</i> , 2022, 13, 780764.	2.3	4
4	MitoVisualize: a resource for analysis of variants in human mitochondrial RNAs and DNA. <i>Bioinformatics</i> , 2022, 38, 2967-2969.	4.1	1
5	Single-nucleus cross-tissue molecular reference maps toward understanding disease gene function. <i>Science</i> , 2022, 376, eabl4290.	12.6	180
6	Decoding the genetics of rare disease: an interview with Monkol Lek. <i>DMM Disease Models and Mechanisms</i> , 2022, 15, .	2.4	2
7	Genetic variance in human disease “ modelling the future of genomic medicine. <i>DMM Disease Models and Mechanisms</i> , 2022, 15, .	2.4	3
8	Therapeutic Approaches in Facioscapulohumeral Muscular Dystrophy. <i>Trends in Molecular Medicine</i> , 2021, 27, 123-137.	6.7	23
9	Exome sequencing in paediatric patients with movement disorders. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 32.	2.7	15
10	Nuclease-Deficient Clustered Regularly Interspaced Short Palindromic Repeat-Based Approaches for In Vitro and In Vivo Gene Activation. <i>Human Gene Therapy</i> , 2021, 32, 260-274.	2.7	2
11	Editorial: Application of Omics Approaches to the Diagnosis of Genetic Neurological Disorders. <i>Frontiers in Neurology</i> , 2021, 12, 712010.	2.4	2
12	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. <i>Nature</i> , 2021, 597, E3-E4.	27.8	45
13	Envisioning the next human genome reference. <i>DMM Disease Models and Mechanisms</i> , 2021, 14, .	2.4	5
14	Cellular and animal models for facioscapulohumeral muscular dystrophy. <i>DMM Disease Models and Mechanisms</i> , 2020, 13, .	2.4	7
15	A three-year follow-up study evaluating clinical utility of exome sequencing and diagnostic potential of reanalysis. <i>Npj Genomic Medicine</i> , 2020, 5, 37.	3.8	54
16	Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. <i>American Journal of Human Genetics</i> , 2020, 107, 727-742.	6.2	25
17	The mutational constraint spectrum quantified from variation in 141,456 humans. <i>Nature</i> , 2020, 581, 434-443.	27.8	6,140
18	Sequential targeted exome sequencing of 1001 patients affected by unexplained limb-girdle weakness. <i>Genetics in Medicine</i> , 2020, 22, 1478-1488.	2.4	62

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19	Applying genome-wide CRISPR-Cas9 screens for therapeutic discovery in facioscapulohumeral muscular dystrophy. <i>Science Translational Medicine</i> , 2020, 12, .	12.4	44
20	Identification of a Novel Deep Intronic Mutation in CAPN3 Presenting a Promising Target for Therapeutic Splice Modulation. <i>Journal of Neuromuscular Diseases</i> , 2019, 6, 475-483.	2.6	6
21	COL4A1 mutations as a potential novel cause of autosomal dominant CAKUT in humans. <i>Human Genetics</i> , 2019, 138, 1105-1115.	3.8	13
22	Estimating prevalence for limb-girdle muscular dystrophy based on public sequencing databases. <i>Genetics in Medicine</i> , 2019, 21, 2512-2520.	2.4	56
23	Resolving the full spectrum of human genome variation using Linked-Reads. <i>Genome Research</i> , 2019, 29, 635-645.	5.5	182
24	The Pediatric Cell Atlas: Defining the Growth Phase of Human Development at Single-Cell Resolution. <i>Developmental Cell</i> , 2019, 49, 10-29.	7.0	57
25	Extending the clinical and mutational spectrum of <i>TRIM32</i> -related myopathies in a non-Hutterite population. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2019, 90, 490-493.	1.9	11
26	A recurrent COL6A1 pseudoexon insertion causes muscular dystrophy and is effectively targeted by splice-correction therapies. <i>JCI Insight</i> , 2019, 4, .	5.0	33
27	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. <i>Annals of Neurology</i> , 2018, 83, 1105-1124.	5.3	93
28	The Effect of ACTN3 Gene Doping on Skeletal Muscle Performance. <i>American Journal of Human Genetics</i> , 2018, 102, 845-857.	6.2	17
29	Whole-exome sequencing identifies mutations in <i>MYMK</i> in a mild form of Carey-Fineman-Ziter syndrome. <i>Neurology: Genetics</i> , 2018, 4, e226.	1.9	6
30	Limb girdle muscular dystrophy due to mutations in <i>POMT2</i> . <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 506-512.	1.9	21
31	Detection of variants in dystroglycanopathy-associated genes through the application of targeted whole-exome sequencing analysis to a large cohort of patients with unexplained limb-girdle muscle weakness. <i>Skeletal Muscle</i> , 2018, 8, 23.	4.2	40
32	BCL11B mutations in patients affected by a neurodevelopmental disorder with reduced type 2 innate lymphoid cells. <i>Brain</i> , 2018, 141, 2299-2311.	7.6	81
33	Whole-Exome Sequencing Identifies Causative Mutations in Families with Congenital Anomalies of the Kidney and Urinary Tract. <i>Journal of the American Society of Nephrology: JASN</i> , 2018, 29, 2348-2361.	6.1	147
34	STRetch: detecting and discovering pathogenic short tandem repeat expansions. <i>Genome Biology</i> , 2018, 19, 121.	8.8	117
35	A novel compound heterozygous mutation in the POMK gene causing limb-girdle muscular dystrophy-dystroglycanopathy in a sib pair. <i>Neuromuscular Disorders</i> , 2018, 28, 614-618.	0.6	11
36	The ExAC browser: displaying reference data information from over 60 000 exomes. <i>Nucleic Acids Research</i> , 2017, 45, D840-D845.	14.5	587

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37	Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. <i>Science Translational Medicine</i> , 2017, 9, .	12.4	516
38	A novel recessive TTN founder variant is a common cause of distal myopathy in the Serbian population. <i>European Journal of Human Genetics</i> , 2017, 25, 572-581.	2.8	18
39	Whole-Exome Sequencing Reveals FAT4 Mutations in a Clinically Unrecognizable Patient with Syndromic CAKUT: A Case Report. <i>Molecular Syndromology</i> , 2017, 8, 272-277.	0.8	7
40	P4HA1 mutations cause a unique congenital disorder of connective tissue involving tendon, bone, muscle and the eye. <i>Human Molecular Genetics</i> , 2017, 26, 2207-2217.	2.9	37
41	Large-scale GWAS identifies multiple loci for hand grip strength providing biological insights into muscular fitness. <i>Nature Communications</i> , 2017, 8, 16015.	12.8	149
42	Exome sequences versus sequential gene testing in the UK highly specialised Service for Limb Girdle Muscular Dystrophy. <i>Orphanet Journal of Rare Diseases</i> , 2017, 12, 151.	2.7	44
43	Identification of GAA variants through whole exome sequencing targeted to a cohort of 606 patients with unexplained limb-girdle muscle weakness. <i>Orphanet Journal of Rare Diseases</i> , 2017, 12, 173.	2.7	21
44	TOR1AIP1 as a cause of cardiac failure and recessive limb-girdle muscular dystrophy. <i>Neuromuscular Disorders</i> , 2016, 26, 500-503.	0.6	38
45	Variants in <i>SLC18A3</i> , vesicular acetylcholine transporter, cause congenital myasthenic syndrome. <i>Neurology</i> , 2016, 87, 1442-1448.	1.1	46
46	Variants in the Oxidoreductase PYROXD1 Cause Early-Onset Myopathy with Internalized Nuclei and Myofibrillar Disorganization. <i>American Journal of Human Genetics</i> , 2016, 99, 1086-1105.	6.2	45
47	Analysis of protein-coding genetic variation in 60,706 humans. <i>Nature</i> , 2016, 536, 285-291.	27.8	9,051
48	High-throughput discovery of novel developmental phenotypes. <i>Nature</i> , 2016, 537, 508-514.	27.8	1,001
49	Quantifying unobserved protein-coding variants in human populations provides a roadmap for large-scale sequencing projects. <i>Nature Communications</i> , 2016, 7, 13293.	12.8	35
50	Quantifying prion disease penetrance using large population control cohorts. <i>Science Translational Medicine</i> , 2016, 8, 322ra9.	12.4	289
51	Health and population effects of rare gene knockouts in adult humans with related parents. <i>Science</i> , 2016, 352, 474-477.	12.6	272
52	Analysis of the <i>ACTN3</i> heterozygous genotype suggests that $\hat{1}\pm$ -actinin-3 controls sarcomeric composition and muscle function in a dose-dependent fashion. <i>Human Molecular Genetics</i> , 2016, 25, 866-877.	2.9	35
53	Concept and design of a genome-wide association genotyping array tailored for transplantation-specific studies. <i>Genome Medicine</i> , 2015, 7, 90.	8.2	49
54	Expanding the phenotype of GMPPB mutations. <i>Brain</i> , 2015, 138, 836-844.	7.6	54

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55	Effect of predicted protein-truncating genetic variants on the human transcriptome. <i>Science</i> , 2015, 348, 666-669.	12.6	252
56	Use of Whole-Exome Sequencing for Diagnosis of Limb-Girdle Muscular Dystrophy. <i>JAMA Neurology</i> , 2015, 72, 1424.	9.0	164
57	Mutations in <i>PIGY</i> : expanding the phenotype of inherited glycosylphosphatidylinositol deficiencies. <i>Human Molecular Genetics</i> , 2015, 24, 6146-6159.	2.9	64
58	The Challenge of Next Generation Sequencing in the Context of Neuromuscular Diseases. <i>Journal of Neuromuscular Diseases</i> , 2014, 1, 135-149.	2.6	25
59	Clozapine-induced agranulocytosis is associated with rare HLA-DQB1 and HLA-B alleles. <i>Nature Communications</i> , 2014, 5, 4757.	12.8	153
60	Distribution and Medical Impact of Loss-of-Function Variants in the Finnish Founder Population. <i>PLoS Genetics</i> , 2014, 10, e1004494.	3.5	351
61	Allelic Expression of Deleterious Protein-Coding Variants across Human Tissues. <i>PLoS Genetics</i> , 2014, 10, e1004304.	3.5	60
62	Biallelic Variants in <i>TTL5</i> , Encoding a Tubulin Glutamylase, Cause Retinal Dystrophy. <i>American Journal of Human Genetics</i> , 2014, 94, 760-769.	6.2	67
63	The Challenge of Next Generation Sequencing in the Context of Neuromuscular Diseases. <i>Journal of Neuromuscular Diseases</i> , 2014, 1, 135-149.	2.6	10