Monkol Lek

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/7768564/publications.pdf

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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	Analysis of protein-coding genetic variation in 60,706 humans. Nature, 2016, 536, 285-291.	27.8	9,051
2	The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2020, 581, 434-443.	27.8	6,140
3	High-throughput discovery of novel developmental phenotypes. Nature, 2016, 537, 508-514.	27.8	1,001
4	The ExAC browser: displaying reference data information from over 60 000 exomes. Nucleic Acids Research, 2017, 45, D840-D845.	14.5	587
5	Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. Science Translational Medicine, 2017, 9, .	12.4	516
6	Distribution and Medical Impact of Loss-of-Function Variants in the Finnish Founder Population. PLoS Genetics, 2014, 10, e1004494.	3.5	351
7	Quantifying prion disease penetrance using large population control cohorts. Science Translational Medicine, 2016, 8, 322ra9.	12.4	289
8	Health and population effects of rare gene knockouts in adult humans with related parents. Science, 2016, 352, 474-477.	12.6	272
9	Effect of predicted protein-truncating genetic variants on the human transcriptome. Science, 2015, 348, 666-669.	12.6	252
10	Resolving the full spectrum of human genome variation using Linked-Reads. Genome Research, 2019, 29, 635-645.	5.5	182
11	Single-nucleus cross-tissue molecular reference maps toward understanding disease gene function. Science, 2022, 376, eabl4290.	12.6	180
12	Use of Whole-Exome Sequencing for Diagnosis of Limb-Girdle Muscular Dystrophy. JAMA Neurology, 2015, 72, 1424.	9.0	164
13	Clozapine-induced agranulocytosis is associated with rare HLA-DQB1 and HLA-B alleles. Nature Communications, 2014, 5, 4757.	12.8	153
14	Large-scale GWAS identifies multiple loci for hand grip strength providing biological insights into muscular fitness. Nature Communications, 2017, 8, 16015.	12.8	149
15	Whole-Exome Sequencing Identifies Causative Mutations in Families with Congenital Anomalies of the Kidney and Urinary Tract. Journal of the American Society of Nephrology: JASN, 2018, 29, 2348-2361.	6.1	147
16	STRetch: detecting and discovering pathogenic short tandem repeat expansions. Genome Biology, 2018, 19, 121.	8.8	117
17	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	5.3	93
18	BCL11B mutations in patients affected by a neurodevelopmental disorder with reduced type 2 innate lymphoid cells. Brain, 2018, 141, 2299-2311.	7.6	81

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19	Biallelic Variants in TTLL5, Encoding a Tubulin Glutamylase, Cause Retinal Dystrophy. American Journal of Human Genetics, 2014, 94, 760-769.	6.2	67
20	Mutations inPIGY: expanding the phenotype of inherited glycosylphosphatidylinositol deficiencies. Human Molecular Genetics, 2015, 24, 6146-6159.	2.9	64
21	Sequential targeted exome sequencing of 1001 patients affected by unexplained limb-girdle weakness. Genetics in Medicine, 2020, 22, 1478-1488.	2.4	62
22	Allelic Expression of Deleterious Protein-Coding Variants across Human Tissues. PLoS Genetics, 2014, 10, e1004304.	3.5	60
23	The Pediatric Cell Atlas: Defining the Growth Phase of Human Development at Single-Cell Resolution. Developmental Cell, 2019, 49, 10-29.	7.0	57
24	Estimating prevalence for limb-girdle muscular dystrophy based on public sequencing databases. Genetics in Medicine, 2019, 21, 2512-2520.	2.4	56
25	Expanding the phenotype of GMPPB mutations. Brain, 2015, 138, 836-844.	7.6	54
26	A three-year follow-up study evaluating clinical utility of exome sequencing and diagnostic potential of reanalysis. Npj Genomic Medicine, 2020, 5, 37.	3.8	54
27	Concept and design of a genome-wide association genotyping array tailored for transplantation-specific studies. Genome Medicine, 2015, 7, 90.	8.2	49
28	Variants in SLC18A3 , vesicular acetylcholine transporter, cause congenital myasthenic syndrome. Neurology, 2016, 87, 1442-1448.	1.1	46
29	Variants in the Oxidoreductase PYROXD1 Cause Early-Onset Myopathy with Internalized Nuclei and Myofibrillar Disorganization. American Journal of Human Genetics, 2016, 99, 1086-1105.	6.2	45
30	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2021, 597, E3-E4.	27.8	45
31	Exome sequences versus sequential gene testing in the UK highly specialised Service for Limb Girdle Muscular Dystrophy. Orphanet Journal of Rare Diseases, 2017, 12, 151.	2.7	44
32	Applying genome-wide CRISPR-Cas9 screens for therapeutic discovery in facioscapulohumeral muscular dystrophy. Science Translational Medicine, 2020, 12, .	12.4	44
33	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	2.4	44
34	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. Skeletal Muscle, 2018, 8, 23.	4.2	40
35	TOR1AIP1 as a cause of cardiac failure and recessive limb-girdle muscular dystrophy. Neuromuscular Disorders, 2016, 26, 500-503.	0.6	38
36	P4HA1 mutations cause a unique congenital disorder of connective tissue involving tendon, bone, muscle and the eye. Human Molecular Genetics, 2017, 26, 2207-2217.	2.9	37

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37	Quantifying unobserved protein-coding variants in human populations provides a roadmap for large-scale sequencing projects. Nature Communications, 2016, 7, 13293.	12.8	35
38	Analysis of theACTN3heterozygous genotype suggests that Î±-actinin-3 controls sarcomeric composition and muscle function in a dose-dependent fashion. Human Molecular Genetics, 2016, 25, 866-877.	2.9	35
39	A recurrent COL6A1 pseudoexon insertion causes muscular dystrophy and is effectively targeted by splice-correction therapies. JCI Insight, 2019, 4, .	5.0	33
40	The Challenge of Next Generation Sequencing in the Context of Neuromuscular Diseases. Journal of Neuromuscular Diseases, 2014, 1, 135-149.	2.6	25
41	Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. American Journal of Human Genetics, 2020, 107, 727-742.	6.2	25
42	Therapeutic Approaches in Facioscapulohumeral Muscular Dystrophy. Trends in Molecular Medicine, 2021, 27, 123-137.	6.7	23
43	Identification of GAA variants through whole exome sequencing targeted to a cohort of 606 patients with unexplained limb-girdle muscle weakness. Orphanet Journal of Rare Diseases, 2017, 12, 173.	2.7	21
44	Limb girdle muscular dystrophy due to mutations in POMT2. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 506-512.	1.9	21
45	A novel recessive TTN founder variant is a common cause of distal myopathy in the Serbian population. European Journal of Human Genetics, 2017, 25, 572-581.	2.8	18
46	The Effect of ACTN3 Gene Doping on Skeletal Muscle Performance. American Journal of Human Genetics, 2018, 102, 845-857.	6.2	17
47	Exome sequencing in paediatric patients with movement disorders. Orphanet Journal of Rare Diseases, 2021, 16, 32.	2.7	15
48	COL4A1 mutations as a potential novel cause of autosomal dominant CAKUT in humans. Human Genetics, 2019, 138, 1105-1115.	3.8	13
49	A novel compound heterozygous mutation in the POMK gene causing limb-girdle muscular dystrophy-dystroglycanopathy in a sib pair. Neuromuscular Disorders, 2018, 28, 614-618.	0.6	11
50	Extending the clinical and mutational spectrum ofTRIM32-related myopathies in a non-Hutterite population. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 490-493.	1.9	11
51	The Challenge of Next Generation Sequencing in the Context of Neuromuscular Diseases. Journal of Neuromuscular Diseases, 2014, 1, 135-149.	2.6	10
52	Whole-Exome Sequencing Reveals FAT4 Mutations in a Clinically Unrecognizable Patient with Syndromic CAKUT: A Case Report. Molecular Syndromology, 2017, 8, 272-277.	0.8	7
53	Cellular and animal models for facioscapulohumeral muscular dystrophy. DMM Disease Models and Mechanisms, 2020, 13, .	2.4	7
54	Whole-exome sequencing identifies mutations in MYMK in a mild form of Carey-Fineman-Ziter syndrome. Neurology: Genetics, 2018, 4, e226.	1.9	6

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55	Identification of a Novel Deep Intronic Mutation in CAPN3 Presenting a Promising Target for Therapeutic Splice Modulation. Journal of Neuromuscular Diseases, 2019, 6, 475-483.	2.6	6
56	Envisioning the next human genome reference. DMM Disease Models and Mechanisms, 2021, 14, .	2.4	5
57	Case Report: Two Families With HPDL Related Neurodegeneration. Frontiers in Genetics, 2022, 13, 780764.	2.3	4
58	Genetic variance in human disease â€“ modelling the future of genomic medicine. DMM Disease Models and Mechanisms, 2022, 15, .	2.4	3
59	Nuclease-Deficient Clustered Regularly Interspaced Short Palindromic Repeat-Based Approaches for In Vitro and In Vivo Gene Activation. Human Gene Therapy, 2021, 32, 260-274.	2.7	2
60	Editorial: Application of Omics Approaches to the Diagnosis of Genetic Neurological Disorders. Frontiers in Neurology, 2021, 12, 712010.	2.4	2
61	Decoding the genetics of rare disease: an interview with Monkol Lek. DMM Disease Models and Mechanisms, 2022, 15, .	2.4	2
62	A new phenotype of syndromic retinitis pigmentosa with myopathy is caused by mutations in retinol dehydrogenase 11. Clinical Genetics, 2022, 101, 448-453.	2.0	1
63	MitoVisualize: a resource for analysis of variants in human mitochondrial RNAs and DNA. Bioinformatics, 2022, 38, 2967-2969.	4.1	1