Dorota Monies

List of Publications by Year in descending order

48 papers	1,965 citations	279798 23 h-index	265206 42 g-index
51 all docs	51 docs citations	51 times ranked	4601 citing authors

#	Article	IF	Citations
1	Prenatal exome sequencing and chromosomal microarray analysis in fetal structural anomalies in a highly consanguineous population reveals a propensity of ciliopathy genes causing multisystem phenotypes. Human Genetics, 2022, 141, 101-126.	3.8	6
2	Identification of pharmacogenetic variants from large scale next generation sequencing data in the Saudi population. PLoS ONE, 2022, 17, e0263137.	2.5	10
3	Clinical, Neurophysiological, Radiological, Pathological, and Genetic Features of Dysferlinopathy in Saudi Arabia. Frontiers in Neuroscience, 2022, 16, 815556.	2.8	5
4	New or vanishing frontiers: LACC1-associated juvenile arthritis. International Journal of Pediatrics and Adolescent Medicine, 2021, 8, 44-47.	1.2	7
5	Bi-allelic variants in HOPS complex subunit VPS41 cause cerebellar ataxia and abnormal membrane trafficking. Brain, 2021, 144, 769-780.	7.6	33
6	Hematological findings associated with tubulinâ€folding cofactors Dâ€related encephalopathy: Expanding the phenotype. Clinical Genetics, 2021, 99, 724-731.	2.0	0
7	SARS-CoV-2â€“Related Acute Respiratory Distress Syndrome Uncovers a Patient with Severe Combined Immunodeficiency Disease. Journal of Clinical Immunology, 2021, 41, 1507-1510.	3.8	5
8	Higher PD-L1 Immunohistochemical Detection Signal in Frozen Compared to Matched Paraffin-Embedded Formalin-Fixed Tissues. Antibodies, 2021, 10, 24.	2.5	2
9	Implications of mosaicism in variant interpretation: A case of a de novo homozygous NF1 variant. European Journal of Medical Genetics, 2021, 64, 104236.	1.3	0
10	Familial Clustering of Juvenile Psoriatic Arthritis Associated with a Hemizygous FOXP3 Mutation. Current Rheumatology Reports, 2021, 23, 64.	4.7	2
11	Established and candidate transthyretin amyloidosis variants identified in the Saudi population by data mining. Human Genomics, 2021, 15, 52.	2.9	6
12	<scp>PLACK</scp> syndrome is potentially treatable with intralipids. Clinical Genetics, 2021, 99, 572-576.	2.0	3
13	Biallelic Mutations in Tetratricopeptide Repeat Domain 26 (Intraflagellar Transport 56) Cause Severe Biliary Ciliopathy in Humans. Hepatology, 2020, 71, 2067-2079.	7.3	28
14	Analysis of transcript-deleterious variants in Mendelian disorders: implications for RNA-based diagnostics. Genome Biology, 2020, 21, 145.	8.8	59
15	Exploiting the Autozygome to Support Previously Published Mendelian Gene-Disease Associations: An Update. Frontiers in Genetics, 2020, 11, 580484.	2.3	13
16	De novo truncating variants in WHSC1 recapitulate the Wolfâ€“Hirschhorn (4p16.3 microdeletion) syndrome phenotype. Genetics in Medicine, 2019, 21, 185-188.	2.4	24
17	Î²1 Integrin is essential for fascinâ€mediated breast cancer stem cell function and disease progression. International Journal of Cancer, 2019, 145, 830-841.	5.1	39
18	Lessons Learned from Large-Scale, First-Tier Clinical Exome Sequencing in a Highly Consanguineous Population. American Journal of Human Genetics, 2019, 104, 1182-1201.	6.2	184

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19	Recessive Truncating Mutations in ALKBH8 Cause Intellectual Disability and Severe Impairment of Wobble Uridine Modification. American Journal of Human Genetics, 2019, 104, 1202-1209.	6.2	34
20	Autosomal recessive ADCY5-Related dystonia and myoclonus: Expanding the genetic spectrum of ADCY5-Related movement disorders. Parkinsonism and Related Disorders, 2019, 64, 145-149.	2.2	18
21	The many faces of peroxisomal disorders: Lessons from a large Arab cohort. Clinical Genetics, 2019, 95, 310-319.	2.0	12
22	Autozygome and high throughput confirmation of disease genes candidacy. Genetics in Medicine, 2019, 21, 736-742.	2.4	81
23	Expanding the phenome and variome of skeletal dysplasia. Genetics in Medicine, 2018, 20, 1609-1616.	2.4	46
24	Further delineation of Temtamy syndrome of corpus callosum and ocular abnormalities. American Journal of Medical Genetics, Part A, 2018, 176, 715-721.	1.2	7
25	Exome Sequencing: Mutilating Sensory Neuropathy with Spastic Paraplegia due to a Mutation in FAM134B Gene. Case Reports in Genetics, 2018, 2018, 1-5.	0.2	12
26	Novel CARMIL2 Mutations in Patients with Variable Clinical Dermatitis, Infections, and Combined Immunodeficiency. Frontiers in Immunology, 2018, 9, 203.	4.8	61
27	High Incidence of Severe Combined Immunodeficiency Disease in Saudi Arabia Detected Through Combined T Cell Receptor Excision Circle and Next Generation Sequencing of Newborn Dried Blood Spots. Frontiers in Immunology, 2018, 9, 782.	4.8	57
28	Phenotype heterogeneity of congenital adrenal hyperplasia due to genetic mosaicism and concomitant nephrogenic diabetes insipidus in a sibling. BMC Medical Genetics, 2018, 19, 115.	2.1	2
29	Variants in EXOSC9 Disrupt the RNA Exosome and Result in Cerebellar Atrophy with Spinal Motor Neuronopathy. American Journal of Human Genetics, 2018, 102, 858-873.	6.2	65
30	Biallelic UFM1 and UFC1 mutations expand the essential role of ufmylation in brain development. Brain, 2018, 141, 1934-1945.	7.6	70
31	LGMD1D myopathy with cytoplasmic and nuclear inclusions in a Saudi family due to DNAJB6 mutation. Acta Myologica, 2018, 37, 221-226.	1.5	5
32	The landscape of genetic diseases in Saudi Arabia based on the first 1000 diagnostic panels and exomes. Human Genetics, 2017, 136, 921-939.	3.8	209
33	Autozygosity reveals recessive mutations and novel mechanisms in dominant genes: implications in variant interpretation. Genetics in Medicine, 2017, 19, 1144-1150.	2.4	77
34	Expanding the genetic heterogeneity of intellectual disability. Human Genetics, 2017, 136, 1419-1429.	3.8	122
35	Novel phenotypes and loci identified through clinical genomics approaches to pediatric cataract. Human Genetics, 2017, 136, 205-225.	3.8	73
36	Identification of a novel genetic locus underlying tremor and dystonia. Human Genomics, 2017, 11, 25.	2.9	6

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37	Recessive VARS2 mutation underlies a novel syndrome with epilepsy, mental retardation, short stature, growth hormone deficiency, and hypogonadism. Human Genomics, 2017, 11, 28.	2.9	12
38	Clinical heterogeneity of PLA2G6-related Parkinsonism: analysis of two Saudi families. BMC Research Notes, 2016, 9, 295.	1.4	18
39	Characterizing the morbid genome of ciliopathies. Genome Biology, 2016, 17, 242.	8.8	118
40	KCNA4deficiency leads to a syndrome of abnormal striatum, congenital cataract and intellectual disability. Journal of Medical Genetics, 2016, 53, 786-792.	3.2	24
41	Genetic spectrum of Saudi Arabian patients with antenatal cystic kidney disease and ciliopathy phenotypes using a targeted renal gene panel. Journal of Medical Genetics, 2016, 53, 338-347.	3.2	28
42	A null mutation in TNIK defines a novel locus for intellectual disability. Human Genetics, 2016, 135, 773-778.	3.8	23
43	Clinical genomics can facilitate countrywide estimation of autosomal recessive disease burden. Genetics in Medicine, 2016, 18, 1244-1249.	2.4	82
44	A first-line diagnostic assay for limb-girdle muscular dystrophy and other myopathies. Human Genomics, 2016, 10, 32.	2.9	33
45	Exome-based caseâ€“control association study using extreme phenotype design reveals novel candidates with protective effect in diabetic retinopathy. Human Genetics, 2016, 135, 193-200.	3.8	45
46	Expanding the clinical, allelic, and locus heterogeneity of retinal dystrophies. Genetics in Medicine, 2016, 18, 554-562.	2.4	89
47	KIAA0556 is a novel ciliary basal body component mutated in Joubert syndrome. Genome Biology, 2015, 16, 293.	8.8	56
48	Allelic heterogeneity in inbred populations: The Saudi experience with AlstrÃ¶m syndrome as an illustrative example. American Journal of Medical Genetics, Part A, 2009, 149A, 662-665.	1.2	48