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	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
2	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
3	Expanding the genetic heterogeneity of intellectual disability. Human Genetics, 2017, 136, 1419-1429.	3.8	122
4	Characterizing the morbid genome of ciliopathies. Genome Biology, 2016, 17, 242.	8.8	118
5	Expanding the clinical, allelic, and locus heterogeneity of retinal dystrophies. Genetics in Medicine, 2016, 18, 554-562.	2.4	89
6	Clinical genomics can facilitate countrywide estimation of autosomal recessive disease burden. Genetics in Medicine, 2016, 18, 1244-1249.	2.4	82
7	Autozygome and high throughput confirmation of disease genes candidacy. Genetics in Medicine, 2019, 21, 736-742.	2.4	81
8	Autozygosity reveals recessive mutations and novel mechanisms in dominant genes: implications in variant interpretation. Genetics in Medicine, 2017, 19, 1144-1150.	2.4	77
9	Novel phenotypes and loci identified through clinical genomics approaches to pediatric cataract. Human Genetics, 2017, 136, 205-225.	3.8	73
10	Biallelic UFM1 and UFC1 mutations expand the essential role of ufmylation in brain development. Brain, 2018, 141, 1934-1945.	7.6	70
11	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
12	Novel CARMIL2 Mutations in Patients with Variable Clinical Dermatitis, Infections, and Combined Immunodeficiency. Frontiers in Immunology, 2018, 9, 203.	4.8	61
13	Analysis of transcript-deleterious variants in Mendelian disorders: implications for RNA-based diagnostics. Genome Biology, 2020, 21, 145.	8.8	59
14	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
15	KIAA0556 is a novel ciliary basal body component mutated in Joubert syndrome. Genome Biology, 2015, 16, 293.	8.8	56
16	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
17	Expanding the phenome and variome of skeletal dysplasia. Genetics in Medicine, 2018, 20, 1609-1616.	2.4	46
18	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

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19	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
20	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
21	A first-line diagnostic assay for limb-girdle muscular dystrophy and other myopathies. Human Genomics, 2016, 10, 32.	2.9	33
22	Bi-allelic variants in HOPS complex subunit VPS41 cause cerebellar ataxia and abnormal membrane trafficking. Brain, 2021, 144, 769-780.	7.6	33
23	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
24	Biallelic Mutations in Tetratricopeptide Repeat Domain 26 (Intraflagellar Transport 56) Cause Severe Biliary Ciliopathy in Humans. Hepatology, 2020, 71, 2067-2079.	7.3	28
25	KCNA4deficiency leads to a syndrome of abnormal striatum, congenital cataract and intellectual disability. Journal of Medical Genetics, 2016, 53, 786-792.	3.2	24
26	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
27	A null mutation in TNIK defines a novel locus for intellectual disability. Human Genetics, 2016, 135, 773-778.	3.8	23
28	Clinical heterogeneity of PLA2G6-related Parkinsonism: analysis of two Saudi families. BMC Research Notes, 2016, 9, 295.	1.4	18
29	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
30	Exploiting the Autozygome to Support Previously Published Mendelian Gene-Disease Associations: An Update. Frontiers in Genetics, 2020, 11, 580484.	2.3	13
31	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
32	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
33	The many faces of peroxisomal disorders: Lessons from a large Arab cohort. Clinical Genetics, 2019, 95, 310-319.	2.0	12
34	Identification of pharmacogenetic variants from large scale next generation sequencing data in the Saudi population. PLoS ONE, 2022, 17, e0263137.	2.5	10
35	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
36	New or vanishing frontiers: LACC1-associated juvenile arthritis. International Journal of Pediatrics and Adolescent Medicine, 2021, 8, 44-47.	1.2	7

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37	Identification of a novel genetic locus underlying tremor and dystonia. Human Genomics, 2017, 11, 25.	2.9	6
38	Established and candidate transthyretin amyloidosis variants identified in the Saudi population by data mining. Human Genomics, 2021, 15, 52.	2.9	6
39	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
40	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
41	LGMD1D myopathy with cytoplasmic and nuclear inclusions in a Saudi family due to DNAJB6 mutation. Acta Myologica, 2018, 37, 221-226.	1.5	5
42	Clinical, Neurophysiological, Radiological, Pathological, and Genetic Features of Dysferlinopathy in Saudi Arabia. Frontiers in Neuroscience, 2022, 16, 815556.	2.8	5
43	<scp>PLACK</scp> syndrome is potentially treatable with intralipids. Clinical Genetics, 2021, 99, 572-576.	2.0	3
44	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
45	Higher PD-L1 Immunohistochemical Detection Signal in Frozen Compared to Matched Paraffin-Embedded Formalin-Fixed Tissues. Antibodies, 2021, 10, 24.	2.5	2
46	Familial Clustering of Juvenile Psoriatic Arthritis Associated with a Hemizygous FOXP3 Mutation. Current Rheumatology Reports, 2021, 23, 64.	4.7	2
47	Hematological findings associated with tubulinâ€folding cofactors Dâ€related encephalopathy: Expanding the phenotype. Clinical Genetics, 2021, 99, 724-731.	2.0	0
48	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