Dorothy K Grange

List of Publications by Year
in descending order

65	2,800	186265	189892
papers	citations	28 h-index	50 g-index

66	66	66	4882
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Elucidating the molecular mechanisms associated with TARS2-related mitochondrial disease. Human Molecular Genetics, 2022, 31, 523-534.	2.9	12
2	Variants in PRKAR1B cause a neurodevelopmental disorder with autism spectrum disorder, apraxia, and insensitivity to pain. Genetics in Medicine, 2021, 23, 1465-1473.	2.4	10
3	Behavioral and cognitive functioning in individuals with CantÃº syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2434-2444.	1.2	3
4	Continuous Renal Replacement Therapy for Two Neonates With Hyperammonemia. Frontiers in Pediatrics, 2021, 9, 732354.	1.9	6
5	Loss-of-Function Variants in PPP1R12A: From Isolated Sex Reversal to Holoprosencephaly Spectrum and Urogenital Malformations. American Journal of Human Genetics, 2020, 106, 121-128.	6.2	30
6	Clinical and Pathological Features of a Newborn With Compound Heterozygous ANKS6 Variants. Pediatric and Developmental Pathology, 2020, 23, 235-239.	1.0	4
7	Siblings with a novel <scp>MED12</scp> variant and Odho syndrome with immune defects. Clinical Genetics, 2020, 98, 308-310.	2.0	5
8	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	6.2	48
9	Fatal <scp>COVID</scp>â€19 infection in a patient with longâ€chain <scp>3â€hydroxyacylâ€CoA</scp> dehydrogenase deficiency: A case report. JIMD Reports, 2020, 56, 40-45.	1.5	9
10	Pegvaliase for the treatment of phenylketonuria: Results of the phase 2 dose-finding studies with long-term follow-up. Molecular Genetics and Metabolism, 2020, 130, 239-246.	1.1	21
11	Threeâ€dimensional facial morphology in CantÃº syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1041-1052.	1.2	8
12	Phenotypic expansion of KMT2Dâ€related disorder: Beyond Kabuki syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1053-1065.	1.2	23
13	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
14	Safety and immunogenicity of Fcâ€EDA, a recombinant ectodysplasin A1 replacement protein, in human subjects. British Journal of Clinical Pharmacology, 2020, 86, 2063-2069.	2.4	18
15	Kir6.1- and SUR2-dependent KATP overactivity disrupts intestinal motility in murine models of CantÃº syndrome. JCI Insight, 2020, 5, .	5.0	16
16	White and gray matter brain development in children and young adults with phenylketonuria. NeuroImage: Clinical, 2019, 23, 101916.	2.7	16
17	Population-Based Newborn Screening for Mucopolysaccharidosis Type II in Illinois: The First Year Experience. Journal of Pediatrics, 2019, 214, 165-167.e1.	1.8	14
18	Glibenclamide treatment in a CantÃº syndrome patient with a pathogenic ABCC9 gainâ€ofâ€function variant: Initial experience. American Journal of Medical Genetics, Part A, 2019, 179, 1585-1590.	1.2	30

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19	Dichotomous roles of TBX3 in the establishment of atrioventricular conduction pathways in the human heart. HeartRhythm Case Reports, 2019, 5, 109-111.	0.4	3
20	Variants in TCF20 in neurodevelopmental disability: description of 27 new patients and review of literature. Genetics in Medicine, 2019, 21, 2036-2042.	2.4	23
21	De novo variants in HK1 associated with neurodevelopmental abnormalities and visual impairment. European Journal of Human Genetics, 2019, 27, 1081-1089.	2.8	19
22	CantÃº syndrome: Findings from 74 patients in the International CantÃº Syndrome Registry. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 658-681.	1.6	50
23	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. Genetics in Medicine, 2019, 21, 663-675.	2.4	52
24	Complex Compound Inheritance of Lethal Lung Developmental Disorders Due to Disruption of the TBX-FGF Pathway. American Journal of Human Genetics, 2019, 104, 213-228.	6.2	90
25	Developmental Trajectories of Executive and Verbal Processes in Children with Phenylketonuria. Developmental Neuropsychology, 2018, 43, 207-218.	1.4	5
26	Cantu syndromeâ€“associated SUR2 (ABCC9) mutations in distinct structural domains result in KATP channel gain-of-function by differential mechanisms. Journal of Biological Chemistry, 2018, 293, 2041-2052.	3.4	34
27	Bi-allelic POLR3A Loss-of-Function Variants Cause Autosomal-Recessive Wiedemann-Rautenstrauch Syndrome. American Journal of Human Genetics, 2018, 103, 968-975.	6.2	43
28	FARS2 deficiency; new cases, review of clinical, biochemical, and molecular spectra, and variants interpretation based on structural, functional, and evolutionary significance. Molecular Genetics and Metabolism, 2018, 125, 281-291.	1.1	28
29	Phenotypic spectrum of Auâ€“Kline syndrome: a report of six new cases and review of the literature. European Journal of Human Genetics, 2018, 26, 1272-1281.	2.8	26
30	Classification of Genes: Standardized Clinical Validity Assessment of Gene-Disease Associations Aids Diagnostic Exome Analysis and Reclassifications. Human Mutation, 2017, 38, 600-608.	2.5	83
31	A postnatal role for embryonic myosin revealed by MYH3 mutations that alter TGFÎ² signaling and cause autosomal dominant spondylocarpotarsal synostosis. Scientific Reports, 2017, 7, 41803.	3.3	29
32	The Exome Clinic and the role of medical genetics expertise in the interpretation of exome sequencing results. Genetics in Medicine, 2017, 19, 1040-1048.	2.4	85
33	Clinical utility gene card for: CantÃº syndrome. European Journal of Human Genetics, 2017, 25, 512-512.	2.8	5
34	Pretreatment cognitive and neural differences between sapropterin dihydrochloride responders and non-responders with phenylketonuria. Molecular Genetics and Metabolism Reports, 2017, 12, 8-13.	1.1	2
35	Loss-of-Function Mutations in YY1AP1 Lead to Grange Syndrome and a Fibromuscular Dysplasia-Like Vascular Disease. American Journal of Human Genetics, 2017, 100, 21-30.	6.2	54
36	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	6.2	136

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37	Automatic recognition of the <scp>XLHED</scp> phenotype from facial images. American Journal of Medical Genetics, Part A, 2017, 173, 2408-2414.	1.2	35
38	Newborn Screening for Lysosomal Storage Disorders in Illinois: The Initial 15-Month Experience. Journal of Pediatrics, 2017, 190, 130-135.	1.8	137
39	Phenotypic and molecular characterisation of CDK13-related congenital heart defects, dysmorphic facial features and intellectual developmental disorders. Genome Medicine, 2017, 9, 73.	8.2	39
40	Clinical and radiographic delineation of Bent Bone Dysplasiaâ€FGFR2 type or Bent Bone Dysplasia with Distinctive Clavicles and Angelâ€shaped Phalanges. American Journal of Medical Genetics, Part A, 2016, 170, 2652-2661.	1.2	6
41	Multigenerational autosomal dominant inheritance of 5p chromosomal deletions. American Journal of Medical Genetics, Part A, 2016, 170, 583-593.	1.2	21
42	Relationship between age and white matter integrity in children with phenylketonuria. Molecular Genetics and Metabolism Reports, 2016, 7, 45-49.	1.1	11
43	A Syndromic Intellectual Disability Disorder Caused by Variants in TELO2 , a Gene Encoding a Component of the TTT Complex. American Journal of Human Genetics, 2016, 98, 909-918.	6.2	35
44	Brain White Matter Integrity Mediates the Relationship Between Phenylalanine Control and Executive Abilities in Children with Phenylketonuria. JIMD Reports, 2016, 33, 41-47.	1.5	10
45	Status dystonicus in two patients with SOX2â€anophthalmia syndrome and nonsense mutations. American Journal of Medical Genetics, Part A, 2016, 170, 3048-3050.	1.2	7
46	A Recurrent Mosaic Mutation in SMO , Encoding the Hedgehog Signal Transducer Smoothened, Is the Major Cause of Curry-Jones Syndrome. American Journal of Human Genetics, 2016, 98, 1256-1265.	6.2	70
47	Neurologic and neuroimaging manifestations of CantÃº syndrome. Neurology, 2016, 87, 270-276.	1.1	40
48	Myhre syndrome: Clinical features and restrictive cardiopulmonary complications. American Journal of Medical Genetics, Part A, 2015, 167, 2893-2901.	1.2	31
49	Electrophysiologic consequences of KATP gain of function in the heart: Conduction abnormalities in Cantu syndrome. Heart Rhythm, 2015, 12, 2316-2324.	0.7	18
50	Brainstem Disconnection: Two Additional Patients and Expansion of the Phenotype. Neuropediatrics, 2015, 46, 139-144.	0.6	9
51	Prolonged exposure to high and variable phenylalanine levels over the lifetime predicts brain white matter integrity in children with phenylketonuria. Molecular Genetics and Metabolism, 2015, 114, 19-24.	1.1	39
52	Fabry disease in infancy and early childhood: a systematic literature review. Genetics in Medicine, 2015, 17, 323-330.	2.4	82
53	Overnight Pulse Oximetry for Evaluation of Sleep Apnea among Children with Trisomy 21. Journal of Clinical Sleep Medicine, 2014, 10, 1309-1315.	2.6	25
54	CantÃº Syndrome Resulting from Activating Mutation in theKCNJ8Gene. Human Mutation, 2014, 35, 809-813.	2.5	92

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55	Fatal Human Herpesvirus 6â€“Associated Encephalitis in Two Boys With Underlying POLG Mitochondrial Disorders. Pediatric Neurology, 2014, 51, 448-452.	2.1	16
56	Sapropterin dihydrochloride use in pregnant women with phenylketonuria: An interim report of the PKU MOMS sub-registry. Molecular Genetics and Metabolism, 2014, 112, 9-16.	1.1	36
57	Single-dose, subcutaneous recombinant phenylalanine ammonia lyase conjugated with polyethylene glycol in adult patients with phenylketonuria: an open-label, multicentre, phase 1 dose-escalation trial. Lancet, The, 2014, 384, 37-44.	13.7	142
58	Variability in phenylalanine control predicts IQ and executive abilities in children with phenylketonuria. Molecular Genetics and Metabolism, 2014, 111, 445-451.	1.1	59
59	K _{ATP} Channels and Cardiovascular Disease. Circulation Research, 2013, 112, 1059-1072.	4.5	144
60	CantÃº Syndrome Is Caused by Mutations in ABCC9. American Journal of Human Genetics, 2012, 90, 1094-1101.	6.2	141
61	Efficacy of Sapropterin Dihydrochloride in Increasing Phenylalanine Tolerance in Children with Phenylketonuria: A Phase III, Randomized, Double-Blind, Placebo-Controlled Study. Journal of Pediatrics, 2009, 154, 700-707.e1.	1.8	170
62	Two new patients with Curryâ€“Jones syndrome with trichoblastoma and medulloblastoma suggest an etiologic role of the sonic hedgehogâ€patchedâ€GLI pathway. American Journal of Medical Genetics, Part A, 2008, 146A, 2589-2597.	1.2	12
63	Cantu syndrome in a woman and her two daughters: Further confirmation of autosomal dominant inheritance and review of the cardiac manifestations. American Journal of Medical Genetics, Part A, 2006, 140A, 1673-1680.	1.2	54
64	Trisomy 20q13 â† 20qter in a girl with multiple congenital malformations and a recombinant chromosome 20 inherited from a paternal inversion (20)(p13q13.1): Clinical report and review of the trisomy 20q phenotype. American Journal of Medical Genetics, Part A, 2005, 137A, 308-312.	1.2	10
65	Severe bilateral panlobular emphysema and pulmonary arterial hypoplasia: Unusual manifestations of Menkes disease. American Journal of Medical Genetics, Part A, 2005, 139A, 151-155.	1.2	40