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	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
2	K _{ATP} Channels and Cardiovascular Disease. Circulation Research, 2013, 112, 1059-1072.	4.5	144
3	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
4	CantÃº Syndrome Is Caused by Mutations in ABCC9. American Journal of Human Genetics, 2012, 90, 1094-1101.	6.2	141
5	Newborn Screening for Lysosomal Storage Disorders in Illinois: The Initial 15-Month Experience. Journal of Pediatrics, 2017, 190, 130-135.	1.8	137
6	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
7	CantÃº Syndrome Resulting from Activating Mutation in theKCNJ8Gene. Human Mutation, 2014, 35, 809-813.	2.5	92
8	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
9	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
10	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
11	Fabry disease in infancy and early childhood: a systematic literature review. Genetics in Medicine, 2015, 17, 323-330.	2.4	82
12	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
13	Variability in phenylalanine control predicts IQ and executive abilities in children with phenylketonuria. Molecular Genetics and Metabolism, 2014, 111, 445-451.	1.1	59
14	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
15	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
16	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. Genetics in Medicine, 2019, 21, 663-675.	2.4	52
17	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
18	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	6.2	48

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19	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
20	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
21	Neurologic and neuroimaging manifestations of CantÃº syndrome. Neurology, 2016, 87, 270-276.	1.1	40
22	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
23	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
24	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
25	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
26	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
27	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
28	Myhre syndrome: Clinical features and restrictive cardiopulmonary complications. American Journal of Medical Genetics, Part A, 2015, 167, 2893-2901.	1.2	31
29	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
30	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
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	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
33	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
34	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
35	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
36	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

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37	Phenotypic expansion of KMT2Dâ€related disorder: Beyond Kabuki syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1053-1065.	1.2	23
38	Multigenerational autosomal dominant inheritance of 5p chromosomal deletions. American Journal of Medical Genetics, Part A, 2016, 170, 583-593.	1.2	21
39	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
40	De novo variants in HK1 associated with neurodevelopmental abnormalities and visual impairment. European Journal of Human Genetics, 2019, 27, 1081-1089.	2.8	19
41	Electrophysiologic consequences of KATP gain of function in the heart: Conduction abnormalities in Cantu syndrome. Heart Rhythm, 2015, 12, 2316-2324.	0.7	18
42	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
43	Fatal Human Herpesvirus 6â€“Associated Encephalitis in Two Boys With Underlying POLG Mitochondrial Disorders. Pediatric Neurology, 2014, 51, 448-452.	2.1	16
44	White and gray matter brain development in children and young adults with phenylketonuria. NeuroImage: Clinical, 2019, 23, 101916.	2.7	16
45	Kir6.1- and SUR2-dependent KATP overactivity disrupts intestinal motility in murine models of CantÃº syndrome. JCI Insight, 2020, 5, .	5.0	16
46	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
47	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
48	Elucidating the molecular mechanisms associated with TARS2-related mitochondrial disease. Human Molecular Genetics, 2022, 31, 523-534.	2.9	12
49	Relationship between age and white matter integrity in children with phenylketonuria. Molecular Genetics and Metabolism Reports, 2016, 7, 45-49.	1.1	11
50	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
51	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
52	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
53	Brainstem Disconnection: Two Additional Patients and Expansion of the Phenotype. Neuropediatrics, 2015, 46, 139-144.	0.6	9
54	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

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55	Threeâ€dimensional facial morphology in CantÃº syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1041-1052.	1.2	8
56	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
57	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
58	Continuous Renal Replacement Therapy for Two Neonates With Hyperammonemia. Frontiers in Pediatrics, 2021, 9, 732354.	1.9	6
59	Clinical utility gene card for: CantÃº syndrome. European Journal of Human Genetics, 2017, 25, 512-512.	2.8	5
60	Developmental Trajectories of Executive and Verbal Processes in Children with Phenylketonuria. Developmental Neuropsychology, 2018, 43, 207-218.	1.4	5
61	Siblings with a novel <scp>MED12</scp> variant and Odho syndrome with immune defects. Clinical Genetics, 2020, 98, 308-310.	2.0	5
62	Clinical and Pathological Features of a Newborn With Compound Heterozygous ANKS6 Variants. Pediatric and Developmental Pathology, 2020, 23, 235-239.	1.0	4
63	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
64	Behavioral and cognitive functioning in individuals with CantÃº syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2434-2444.	1.2	3
65	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