

Kerstin Kutsche

List of Publications by Year in descending order

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Version: 2024-02-01

39
papers

1,614
citations

516710

16
h-index

434195

31
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39
all docs

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docs citations

39
times ranked

2920
citing authors

#	ARTICLE	IF	CITATIONS
1	Biallelic <i>FRA10A1</i> variants cause a neurodevelopmental disorder with growth retardation. <i>Brain</i> , 2022, 145, 1551-1563.	7.6	9
2	Novel biallelic variants expand the SLC5A6-related phenotypic spectrum. <i>European Journal of Human Genetics</i> , 2022, 30, 439-449.	2.8	10
3	Genotype-Phenotype Correlations in Pediatric Patients with a Heterozygous Pathogenic FBN1 Variant. <i>Thoracic and Cardiovascular Surgeon</i> , 2022, 70, .	1.0	0
4	Thoracic Aortic Disease in Patients with Heterozygous Variants in FBN2. <i>Thoracic and Cardiovascular Surgeon</i> , 2022, 70, .	1.0	0
5	A homozygous hypomorphic <i>BNIP1</i> variant causes an increase in autophagosomes and reduced autophagic flux and results in a spondyloepiphyseal dysplasia. <i>Human Mutation</i> , 2022, 43, 625-642.	2.5	3
6	Cardiofaciocardio-neurodevelopmental syndrome: Report of a novel patient and expansion of the phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2448-2453.	1.2	3
7	Autosomal dominantly inherited myopathy likely caused by the <i>TNNT1</i> variant p.(Asp65Ala). <i>Human Mutation</i> , 2022, 43, 1224-1233.	2.5	0
8	Syndromic disorders caused by gain-of-function variants in <i>KCNH1</i> , <i>KCNK4</i> , and <i>KCNN3</i> a subgroup of K ⁺ channelopathies. <i>European Journal of Human Genetics</i> , 2021, 29, 1384-1395.	2.8	21
9	Pathogenic variants in <i>GNPTAB</i> and <i>GNPTG</i> encoding distinct subunits of GlcNAc-1-phosphotransferase differentially impact bone resorption in patients with mucopolysaccharidosis type II and III. <i>Genetics in Medicine</i> , 2021, 23, 2369-2377.	2.4	2
10	Biallelic <i>MADD</i> variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. <i>Brain</i> , 2020, 143, 2437-2453.	7.6	21
11	Roberts syndrome in an Indian patient with humeroradial synostosis, congenital elbow contractures and a novel homozygous splice variant in <i>ESCO2</i> . <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2793-2796.	1.2	4
12	A homozygous missense variant in <i>CACNB4</i> encoding the auxiliary calcium channel beta4 subunit causes a severe neurodevelopmental disorder and impairs channel and non-channel functions. <i>PLoS Genetics</i> , 2020, 16, e1008625.	3.5	18
13	CantÃ syndrome versus Zimmermann-Laband syndrome: Report of nine individuals with <i>ABCC9</i> variants. <i>European Journal of Medical Genetics</i> , 2020, 63, 103996.	1.3	7
14	Biallelic loss-of-function variants in <i>TBC1D2B</i> cause a neurodevelopmental disorder with seizures and gingival overgrowth. <i>Human Mutation</i> , 2020, 41, 1645-1661.	2.5	10
15	Bi-allelic Pathogenic Variants in <i>HS2ST1</i> Cause a Syndrome Characterized by Developmental Delay and Corpus Callosum, Skeletal, and Renal Abnormalities. <i>American Journal of Human Genetics</i> , 2020, 107, 1044-1061.	6.2	11
16	Title is missing!. , 2020, 16, e1008625.		0
17	Title is missing!. , 2020, 16, e1008625.		0
18	Title is missing!. , 2020, 16, e1008625.		0

#	ARTICLE	IF	CITATIONS
19	Title is missing!. , 2020, 16, e1008625.		0
20	Gain-of-Function Mutations in KCNN3 Encoding the Small-Conductance Ca ²⁺ -Activated K ⁺ Channel SK3 Cause Zimmermann-Laband Syndrome. American Journal of Human Genetics, 2019, 104, 1139-1157.	6.2	45
21	The lysosomal storage disorders mucopolipidosis type II, type III alpha/beta, and type III gamma: Update on <i>GNPTAB</i> and <i>GNPTC</i> mutations. Human Mutation, 2019, 40, 842-864.	2.5	36
22	Biallelic variants in SMAD6 are associated with a complex cardiovascular phenotype. Human Genetics, 2019, 138, 625-634.	3.8	12
23	The homozygous variant c.797G>A/p.(Cys266Tyr) in <i>PISD</i> is associated with a Spondyloepimetaphyseal dysplasia with large epiphyses and disturbed mitochondrial function. Human Mutation, 2019, 40, 299-309.	2.5	54
24	Autosomal dominant Robinow syndrome associated with a novel <i>DVL3</i> splice mutation. American Journal of Medical Genetics, Part A, 2018, 176, 992-996.	1.2	14
25	The novel <i>RAF1</i> mutation p.(Gly361Ala) located outside the kinase domain of the CR3 region in two patients with Noonan syndrome, including one with a rare brain tumor. American Journal of Medical Genetics, Part A, 2018, 176, 470-476.	1.2	17
26	Elsahyâ€Waters syndrome is caused by biallelic mutations in <i>CDH11</i> . American Journal of Medical Genetics, Part A, 2018, 176, 477-482.	1.2	18
27	Activating Mutations in PAK1, Encoding p21-Activated Kinase 1, Cause a Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 103, 579-591.	6.2	54
28	Coinheritance of biallelic SLURP1 and SLC39A4 mutations cause a severe genodermatosis with skin peeling and hair loss all over the body. British Journal of Dermatology, 2018, 179, 1192-1194.	1.5	0
29	RIT1 controls actin dynamics via complex formation with RAC1/CDC42 and PAK1. PLoS Genetics, 2018, 14, e1007370.	3.5	25
30	Genetic diagnostics of inherited aortic diseases. Herz, 2017, 42, 459-467.	1.1	7
31	Genotype and phenotype in patients with Noonan syndrome and a RIT1 mutation. Genetics in Medicine, 2016, 18, 1226-1234.	2.4	77
32	A novel multiple joint dislocation syndrome associated with a homozygous nonsense variant in the EXOC6B gene. European Journal of Human Genetics, 2016, 24, 1206-1210.	2.8	16
33	Structureâ€functionâ€behavior relationship in estrogen-induced synaptic plasticity. Hormones and Behavior, 2015, 74, 139-148.	2.1	39
34	Cancer spectrum and frequency among children with Noonan, Costello, and cardio-facio-cutaneous syndromes. British Journal of Cancer, 2015, 112, 1392-1397.	6.4	167
35	Mutations in KCNH1 and ATP6V1B2 cause Zimmermann-Laband syndrome. Nature Genetics, 2015, 47, 661-667.	21.4	177
36	Mutation of KCNJ8 in a patient with CantÃ© syndrome with unique vascular abnormalities â€ Support for the role of K(ATP) channels in this condition. European Journal of Medical Genetics, 2013, 56, 678-682.	1.3	79

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37	A restricted spectrum of NRAS mutations causes Noonan syndrome. <i>Nature Genetics</i> , 2010, 42, 27-29.	21.4	271
38	Oncogenic HRAS mutations cause prolonged PI3K signaling in response to epidermal growth factor in fibroblasts of patients with Costello syndrome. <i>Human Mutation</i> , 2009, 30, 352-362.	2.5	29
39	Mutation of SHOC2 promotes aberrant protein N-myristoylation and causes Noonan-like syndrome with loose anagen hair. <i>Nature Genetics</i> , 2009, 41, 1022-1026.	21.4	358