Kerstin Kutsche

List of Publications by Year
in descending order

39 papers	1,614 citations	516710 16 h-index	434195 31 g-index
39 all docs	39 docs citations	39 times ranked	2920 citing authors

#	Article	IF	Citations
1	Mutation of SHOC2 promotes aberrant protein N-myristoylation and causes Noonan-like syndrome with loose anagen hair. Nature Genetics, 2009, 41, 1022-1026.	21.4	358
2	A restricted spectrum of NRAS mutations causes Noonan syndrome. Nature Genetics, 2010, 42, 27-29.	21.4	271
3	Mutations in KCNH1 and ATP6V1B2 cause Zimmermann-Laband syndrome. Nature Genetics, 2015, 47, 661-667.	21.4	177
4	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
5	Mutation of KCNJ8 in a patient with CantÃº 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
6	Genotype and phenotype in patients with Noonan syndrome and a RIT1 mutation. Genetics in Medicine, 2016, 18, 1226-1234.	2.4	77
7	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
8	The homozygous variant c.797G>A/p.(Cys266Tyr) inPISDis associated with a Spondyloepimetaphyseal dysplasia with large epiphyses and disturbed mitochondrial function. Human Mutation, 2019, 40, 299-309.	2.5	54
9	Gain-of-Function Mutations in KCNN3 Encoding the Small-Conductance Ca2+-Activated K+ Channel SK3 Cause Zimmermann-Laband Syndrome. American Journal of Human Genetics, 2019, 104, 1139-1157.	6.2	45
10	Structureâ€“functionâ€“behavior relationship in estrogen-induced synaptic plasticity. Hormones and Behavior, 2015, 74, 139-148.	2.1	39
11	The lysosomal storage disorders mucolipidosis type II, type III alpha/beta, and type III gamma: Update onGNPTABandGNPTGmutations. Human Mutation, 2019, 40, 842-864.	2.5	36
12	OncogenicHRASmutations cause prolonged PI3K signaling in response to epidermal growth factor in fibroblasts of patients with Costello syndrome. Human Mutation, 2009, 30, 352-362.	2.5	29
13	RIT1 controls actin dynamics via complex formation with RAC1/CDC42 and PAK1. PLoS Genetics, 2018, 14, e1007370.	3.5	25
14	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. Brain, 2020, 143, 2437-2453.	7.6	21
15	Syndromic disorders caused by gain-of-function variants in KCNH1, KCNK4, and KCNN3â€”a subgroup of K+ channelopathies. European Journal of Human Genetics, 2021, 29, 1384-1395.	2.8	21
16	Elsahyâ€“Waters syndrome is caused by biallelic mutations in CDH11. American Journal of Medical Genetics, Part A, 2018, 176, 477-482.	1.2	18
17	A homozygous missense variant in CACNB4 encoding the auxiliary calcium channel beta4 subunit causes a severe neurodevelopmental disorder and impairs channel and non-channel functions. PLoS Genetics, 2020, 16, e1008625.	3.5	18
18	The novel RAF1 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

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19	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
20	Autosomal dominant Robinow syndrome associated with a novel DVL3 splice mutation. American Journal of Medical Genetics, Part A, 2018, 176, 992-996.	1.2	14
21	Biallelic variants in SMAD6 are associated with a complex cardiovascular phenotype. Human Genetics, 2019, 138, 625-634.	3.8	12
22	Bi-allelic Pathogenic Variants in HS2ST1 Cause a Syndrome Characterized by Developmental Delay and Corpus Callosum, Skeletal, and Renal Abnormalities. American Journal of Human Genetics, 2020, 107, 1044-1061.	6.2	11
23	Biallelic lossâ€ofâ€function variants in TBC1D2B cause a neurodevelopmental disorder with seizures and gingival overgrowth. Human Mutation, 2020, 41, 1645-1661.	2.5	10
24	Novel biallelic variants expand the SLC5A6-related phenotypic spectrum. European Journal of Human Genetics, 2022, 30, 439-449.	2.8	10
25	Biallelic FRA10AC1 variants cause a neurodevelopmental disorder with growth retardation. Brain, 2022, 145, 1551-1563.	7.6	9
26	Genetic diagnostics of inherited aortic diseases. Herz, 2017, 42, 459-467.	1.1	7
27	CantÃº syndrome versus Zimmermann-Laband syndrome: Report of nine individuals with ABCC9 variants. European Journal of Medical Genetics, 2020, 63, 103996.	1.3	7
28	Roberts syndrome in an Indian patient with humeroradial synostosis, congenital elbow contractures and a novel homozygous splice variant in ESCO2. American Journal of Medical Genetics, Part A, 2020, 182, 2793-2796.	1.2	4
29	A homozygous hypomorphicBNIP1variant causes an increase in autophagosomes and reduced autophagic flux and results in a spondyloâ€epiphyseal dysplasia. Human Mutation, 2022, 43, 625-642.	2.5	3
30	Cardiofacioneurodevelopmental syndrome: Report of a novel patient and expansion of the phenotype. American Journal of Medical Genetics, Part A, 2022, 188, 2448-2453.	1.2	3
31	Pathogenic variants in GNPTAB and GNPTG encoding distinct subunits of GlcNAc-1-phosphotransferase differentially impact bone resorption in patients with mucolipidosis type II and III. Genetics in Medicine, 2021, 23, 2369-2377.	2.4	2
32	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
33	Genotypeâ€“Phenotype Correlations in Pediatric Patients with a Heterozygous Pathogenic FBN1 Variant. Thoracic and Cardiovascular Surgeon, 2022, 70, .	1.0	0
34	Thoracic Aortic Disease in Patients with Heterozygous Variants in FBN2. Thoracic and Cardiovascular Surgeon, 2022, 70, .	1.0	0
35	Title is missing!. , 2020, 16, e1008625.		0
36	Title is missing!. , 2020, 16, e1008625.		0

#	Article	IF	Citations
37	Title is missing!. , 2020, 16, e1008625.		0
38	Title is missing!. , 2020, 16, e1008625.		0
39	Autosomal dominantly inherited myopathy likely caused by the TNNT1 variant p.(Asp65Ala). Human Mutation, 2022, 43, 1224-1233.	2.5	0