Lisbeth Birk MÃ¸ller

List of Publications by Year
in descending order

61	2,123	185998 28	243296 44
papers	citations	h-index	g-index

64	64	64	2716
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	The effect of casein glycomacropeptide versus free synthetic amino acids for early treatment of phenylketonuria in a mice model. PLoS ONE, 2022, 17, e0261150.	1.1	3
2	Widening the spectrum of spinocerebellar ataxia autosomal recessive type 10 (SCAR10). BMJ Case Reports, 2022, 15, e248228.	0.2	1
3	BBS Proteins Affect Ciliogenesis and Are Essential for Hedgehog Signaling, but Not for Formation of iPSC-Derived RPE-65 Expressing RPE-Like Cells. International Journal of Molecular Sciences, 2021, 22, 1345.	1.8	14
4	Occipital Horn Syndrome as a Result of Splice Site Mutations in ATP7A. No Activity of ATP7A Splice Variants Missing Exon 10 or Exon 15. Frontiers in Molecular Neuroscience, 2021, 14, 532291.	1.4	1
5	Novel Homozygous Truncating Variant Widens the Spectrum of Early-Onset Multisystemic SYNE1 Ataxia. Cerebellum, 2021, , 1.	1.4	0
6	Crosstalk between BH4, pain, and dystonia. European Journal of Human Genetics, 2021, 29, 1727-1728.	1.4	0
7	Elevated Expression of SLC6A4 Encoding the Serotonin Transporter (SERT) in Gilles de la Tourette Syndrome. Genes, 2021, 12, 86.	1.0	12
8	Crosstalk of Hedgehog and mTORC1 Pathways. Cells, 2020, 9, 2316.	1.8	38
9	Bi-Allelic Pathogenic Variations in MERTK Including Deletions Are Associated with an Early Onset Progressive Form of Retinitis Pigmentosa. Genes, 2020, 11, 1517.	1.0	8
10	Mutational analysis of TSC1 and TSC2 in Danish patients with tuberous sclerosis complex. Scientific Reports, 2020, 10, 9909.	1.6	13
11	A Missense Mutation in RAB28 in a Family with Cone-Rod Dystrophy and Postaxial Polydactyly Prevents Localization of RAB28 to the Primary Cilium. , 2020, 61, 29.		8
12	Molecular genetic analysis using targeted NGS analysis of 677 individuals with retinal dystrophy. Scientific Reports, 2019, 9, 1219.	1.6	76
13	Generation and characterization of three isogenic induced pluripotent stem cell lines from a patient with Bardet-Biedl syndrome and homozygous for the BBS5 variant. Stem Cell Research, 2019, 41, 101594.	0.3	5
14	Chelating principles in Menkes and Wilson diseases. Journal of Inorganic Biochemistry, 2019, 190, 98-112.	1.5	45
15	Heterozygous mutations in GTP-cyclohydrolase-1 reduce BH4 biosynthesis but not pain sensitivity. Pain, 2018, 159, 1012-1024.	2.0	8
16	Comparison of two different culture conditions for derivation of early hiPSC. Cell Biology International, 2018, 42, 1467-1473.	1.4	12
17	TSC1 and TSC2 regulate cilia length and canonical Hedgehog signaling via different mechanisms. Cellular and Molecular Life Sciences, 2018, 75, 2663-2680.	2.4	34
18	Generation of induced pluripotent stem cells, KCi002-A derived from a patient with Bardet-Biedl syndrome homozygous for the BBS10 variant c.271insT. Stem Cell Research, 2018, 33, 46-50.	0.3	2

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19	Generation of induced pluripotent stem cells, KCi001-A derived from a Bardet-Biedl syndrome patient compound heterozygous for the BBS1 variants c.1169T>G/c.1135G>C. Stem Cell Research, 2018, 31, 235-239.	0.3	1
20	A Novel TTBK2 De Novo Mutation in a Danish Family with Early-Onset Spinocerebellar Ataxia. Cerebellum, 2017, 16, 268-271.	1.4	15
21	Development of hypomelanotic macules is associated with constitutive activated mTORC1 in tuberous sclerosis complex. Molecular Genetics and Metabolism, 2017, 120, 384-391.	0.5	5
22	Characterization of ATP7A missense mutants suggests a correlation between intracellular trafficking and severity of Menkes disease. Scientific Reports, 2017, 7, 757.	1.6	30
23	Mutational and phenotypical spectrum of phenylalanine hydroxylase deficiency in Denmark. Clinical Genetics, 2016, 90, 247-251.	1.0	11
24	Usher syndrome in Denmark: mutation spectrum and some clinical observations. Molecular Genetics & Genomic Medicine, 2016, 4, 527-539.	0.6	24
25	Mottled Mice and Non-Mammalian Models of Menkes Disease. Frontiers in Molecular Neuroscience, 2015, 8, 72.	1.4	19
26	Small amounts of functional ATP7A protein permit mild phenotype. Journal of Trace Elements in Medicine and Biology, 2015, 31, 173-177.	1.5	14
27	Intraplantar injection of tetrahydrobiopterin induces nociception in mice. Neuroscience Letters, 2015, 584, 247-252.	1.0	10
28	Copper-transporting P-type ATPases use a unique ion-release pathway. Nature Structural and Molecular Biology, 2014, 21, 43-48.	3.6	98
29	GCH1 variants, tetrahydrobiopterin and their effects on pain sensitivity. Scandinavian Journal of Pain, 2014, 5, 121-128.	0.5	15
30	A silent nucleotide substitution in the ATP7A gene in a child with Menkes disease. Molecular Genetics and Metabolism, 2013, 110, 490-492.	0.5	5
31	Structural models of the human copper P-type ATPases ATP7A and ATP7B. Biological Chemistry, 2012, 393, 205-216.	1.2	48
32	Neonatal Erythroderma as a First Manifestation of Menkes Disease. Pediatrics, 2012, 130, e239-e242.	1.0	10
33	Prenatal Treatment of Mosaic Mice (Atp7a mo-ms) Mouse Model for Menkes Disease, with Copper Combined by Dimethyldithiocarbamate (DMDTC). PLoS ONE, 2012, 7, e40400.	1.1	14
34	Impairment of Interrelated Iron- and Copper Homeostatic Mechanisms in Brain Contributes to the Pathogenesis of Neurodegenerative Disorders. Frontiers in Pharmacology, 2012, 3, 169.	1.6	65
35	Molecular and biochemical characterization of a unique mutation in CCS, the human copper chaperone to superoxide dismutase. Human Mutation, 2012, 33, 1207-1215.	1.1	34
36	Mutations in SLC33A1 Cause a Lethal Autosomal-Recessive Disorder with Congenital Cataracts, Hearing Loss, and Low Serum Copper and Ceruloplasmin. American Journal of Human Genetics, 2012, 90, 61-68.	2.6	85

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37	Clinical expression of Menkes disease in females with normal karyotype. Orphanet Journal of Rare Diseases, 2012, 7, 6.	1.2	37
38	Crystal structure of a copper-transporting PIB-type ATPase. Nature, 2011, 475, 59-64.	13.7	293
39	Investigation of Parameters that Affect the Success Rate of Microarray-Based Allele-Specific Hybridization Assays. PLoS ONE, 2011, 6, e14777.	1.1	5
40	Splice Site Mutations in the ATP7A Gene. PLoS ONE, 2011, 6, e18599.	1.1	22
41	Clinical presentation and mutations in Danish patients with Wilson disease. European Journal of Human Genetics, 2011, 19, 935-941.	1.4	42
42	Progressive cerebellar degenerative changes in the severe mental retardation syndrome caused by duplication of MECP2 and adjacent loci on Xq28. European Journal of Pediatrics, 2010, 169, 941-949.	1.3	42
43	Molecular diagnosis of Menkes disease: Genotypeâ€“phenotype correlation. Biochimie, 2009, 91, 1273-1277.	1.3	77
44	Multi-stringency wash of partially hybridized 60-mer probes reveals that the stringency along the probe decreases with distance from the microarray surface. Nucleic Acids Research, 2008, 36, e132-e132.	6.5	33
45	Mutation Detection in the Menkes Gene ATP7A Using the Protein Truncation Test. Clinical Medicine Pathology, 2008, 1, CPath.S565.	0.0	2
46	Evidence That Translation Reinitiation Leads to a Partially Functional Menkes Protein Containing Two Copper-Binding Sites. American Journal of Human Genetics, 2006, 79, 214-229.	2.6	61
47	Identification and analysis of 21 novel disease-causing amino acid substitutions in the conserved part of ATP7A. Human Mutation, 2005, 26, 84-93.	1.1	49
48	Homozygosity for a gross partial gene deletion of the C-terminal end of ATP7B in a Wilson patient with hepatic and no neurological manifestations. American Journal of Medical Genetics, Part A, 2005, 138A, 340-343.	0.7	33
49	Pre- and postnatal diagnosis of tyrosine hydroxylase deficiency. Prenatal Diagnosis, 2005, 25, 671-675.	1.1	17
50	X-Linked Menkes Disease: First Documented Report of Germ-Line Mosaicism. Genetic Testing and Molecular Biomarkers, 2004, 8, 286-291.	1.7	13
51	Variable clinical expression of an identical mutation in the ATP7A gene for Menkes disease/Occipital horn syndrome in three affected males in a single family. Journal of Pediatrics, 2004, 145, 119-121.	0.9	36
52	Screening of 383 unrelated patients affected with Menkes disease and finding of 57 gross deletions inATP7A. Human Mutation, 2003, 22, 457-464.	1.1	55
53	The Cryptic Adenine Deaminase Gene of Escherichia coli. Journal of Biological Chemistry, 2002, 277, 31373-31380.	1.6	34
54	The RihA, RihB, and RihC Ribonucleoside Hydrolases of Escherichia coli. Journal of Biological Chemistry, 2001, 276, 884-894.	1.6	70

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55	Invariance of the Nucleoside Triphosphate Pools ofEscherichia coli with Growth Rate. Journal of Biological Chemistry, 2000, 275, 3931-3935.	1.6	61
56	Characterization of the hCTR1 gene: Genomic organization, functional expression, and identification of a highly homologous processed gene. Gene, 2000, 257, 13-22.	1.0	59
57	Similar Splice-Site Mutations of the ATP7A Gene Lead to Different Phenotypes: Classical Menkes Disease or Occipital Horn Syndrome. American Journal of Human Genetics, 2000, 66, 1211-1220.	2.6	122
58	Cooperative binding of copper(I) to the metal binding domains in Menkes disease protein. BBA - Proteins and Proteomics, 1999, 1434, 103-113.	2.1	33
59	Mutation Spectrum of ATP7A, the Gene Defective in Menkes Disease. Advances in Experimental Medicine and Biology, 1999, 448, 83-95.	0.8	67
60	Structural requirements for glycosyl-phosphatidylinositol-anchor attachment in the cellular receptor for urokinase plasminogen activator. FEBS Journal, 1992, 208, 493-500.	0.2	53
61	Mosaicism in Tuberous Sclerosis Complex: A Case Report, Literature Review, and Original Data from Danish Hospitals. EMJ Dermatology, 0, , 98-105.	0.0	1