Stephen G Kaler

List of Publications by Year
in descending order

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Version: 2024-02-01

48 papers	3,741 citations	236925 25 h-index	233421 45 g-index
49 all docs	49 docs citations	49 times ranked	3038 citing authors

#	Article	IF	Citations
1	Connecting copper and cancer: from transition metal signalling to metalloplasia. Nature Reviews Cancer, 2022, 22, 102-113.	28.4	519
2	Expanding the Clinical Phenotype of FGFR1 Internal Tandem Duplication. Journal of Physical Education and Sports Management, 2022, , mcs.a006174.	1.2	4
3	Inherited Disorders of Human Copper Metabolism. , 2021, , 413-443.		0
4	Targeted next generation sequencing for newborn screening of Menkes disease. Molecular Genetics and Metabolism Reports, 2020, 24, 100625.	1.1	7
5	Estimated birth prevalence of Menkes disease and ATP7A-related disorders based on the Genome Aggregation Database (gnomAD). Molecular Genetics and Metabolism Reports, 2020, 24, 100602.	1.1	20
6	Neck masses due to internal jugular vein phlebectasia: Frequency in Menkes disease and literature review of 85 pediatric subjects. American Journal of Medical Genetics, Part A, 2020, 182, 1364-1377.	1.2	4
7	Biallelic HEPHL1 variants impair ferroxidase activity and cause an abnormal hair phenotype. PLoS Genetics, 2019, 15, e1008143.	3.5	23
8	Interaction between the AAA ATPase p97/VCP and a concealed UBX domain in the copper transporter ATP7A is associated with motor neuron degeneration. Journal of Biological Chemistry, 2018, 293, 7606-7617.	3.4	9
9	Cerebrospinal Fluid-Directed rAAV9-rsATP7A Plus Subcutaneous Copper Histidinate Advance Survival and Outcomes in a Menkes Disease Mouse Model. Molecular Therapy - Methods and Clinical Development, 2018, 10, 165-178.	4.1	17
10	Copper Regulates Maturation and Expression of an MITF:Tryptase Axis in Mast Cells. Journal of Immunology, 2017, 199, 4132-4141.	0.8	18
11	Pregnancy outcomes in Liberian women who conceived after recovery from Ebola virus disease. The Lancet Global Health, 2016, 4, e678-e679.	6.3	30
12	The Activity of Menkes Disease Protein ATP7A Is Essential for Redox Balance in Mitochondria. Journal of Biological Chemistry, 2016, 291, 16644-16658.	3.4	54
13	The Involvement of Copper Transport in Von Willebrand Factor Multimer Assembly. Blood, 2016, 128, 2530-2530.	1.4	0
14	Menkes disease with discordant phenotype in female monozygotic twins. American Journal of Medical Genetics, Part A, 2015, 167, 2826-2829.	1.2	6
15	Direct interactions of adaptor protein complexes 1 and 2 with the copper transporter ATP7A mediate its anterograde and retrograde trafficking. Human Molecular Genetics, 2015, 24, 2411-2425.	2.9	33
16	Persistent Lethargy, Hypothermia, and Failure to Thrive in a Neonate. Hospital Pediatrics, 2015, 5, 234-237.	1.3	1
17	Wilson's disease and other neurological copper disorders. Lancet Neurology, The, 2015, 14, 103-113.	10.2	751
18	Translational research investigations on ATP7A: an important human copper ATPase. Annals of the New York Academy of Sciences, 2014, 1314, 64-68.	3.8	25

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19	Neurodevelopment and brain growth in classic Menkes disease is influenced by age and symptomatology at initiation of copper treatment. Journal of Trace Elements in Medicine and Biology, 2014, 28, 427-430.	3.0	38
20	Molecular and biochemical characterization of Mottled-dappled, an embryonic lethal Menkes disease mouse model. Molecular Genetics and Metabolism, 2014, 113, 294-300.	1.1	4
21	A Novel Two-Nucleotide Deletion in the ATP7A Gene Associated With Delayed Infantile Onset of Menkes Disease. Pediatric Neurology, 2014, 50, 417-420.	2.1	2
22	Catecholamine Metabolites Affected by the Copper-Dependent Enzyme Dopamine-Beta-Hydroxylase Provide Sensitive Biomarkers for Early Diagnosis of Menkes Disease and Viral-Mediated ATP7A Gene Therapy. Advances in Pharmacology, 2013, 68, 223-233.	2.0	17
23	Inborn errors of copper metabolism. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1745-1754.	1.8	85
24	ATP7A Gene Addition to the Choroid Plexus Results in Long-term Rescue of the Lethal Copper Transport Defect in a Menkes Disease Mouse Model. Molecular Therapy, 2011, 19, 2114-2123.	8.2	64
25	ATP7A-related copper transport diseasesâ€"emerging concepts and future trends. Nature Reviews Neurology, 2011, 7, 15-29.	10.1	494
26	Missense Mutations in the Copper Transporter Gene ATP7A Cause X-Linked Distal Hereditary Motor Neuropathy. American Journal of Human Genetics, 2010, 86, 343-352.	6.2	170
27	Molecular correlates of epilepsy in early diagnosed and treated Menkes disease. Journal of Inherited Metabolic Disease, 2010, 33, 583-589.	3.6	52
28	Translational readâ€through of a nonsense mutation in ATP7A impacts treatment outcome in Menkes disease. Annals of Neurology, 2009, 65, 108-113.	5.3	35
29	Relative Efficiencies of Plasma Catechol Levels and Ratios for Neonatal Diagnosis of Menkes Disease. Neurochemical Research, 2009, 34, 1464-1468.	3.3	44
30	Diseases of Poverty with High Mortality in Infants and Children. Annals of the New York Academy of Sciences, 2008, 1136, 28-31.	3.8	18
31	Clinical outcomes in Menkes disease patients with a copper-responsive ATP7A mutation, G727R. Molecular Genetics and Metabolism, 2008, 95, 174-181.	1.1	40
32	Neonatal Diagnosis and Treatment of Menkes Disease. New England Journal of Medicine, 2008, 358, 605-614.	27.0	269
33	Internal jugular phlebectasia in Menkes disease. International Journal of Pediatric Otorhinolaryngology, 2007, 71, 1145-1148.	1.0	30
34	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
35	Rapid and Robust Screening of the Menkes Disease/Occipital Horn Syndrome Gene. Genetic Testing and Molecular Biomarkers, 2002, 6, 255-260.	1.7	35
36	Catecholamine Phenotyping: Clues to the Diagnosis, Treatment, and Pathophysiology of Neurogenetic Disorders. Journal of Neurochemistry, 2002, 67, 1781-1790.	3.9	11

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37	Prenatal diagnosis of Menkes disease. Prenatal Diagnosis, 1998, 18, 287-289.	2.3	18
38	Expression of the Menkes disease homolog in rodent neuroglial cells. Neuroscience Research Communications, 1998, 23, 61-66.	0.2	9
39	Metabolic and Molecular Bases of Menkes Disease and Occipital Horn Syndrome. Pediatric and Developmental Pathology, 1998, 1, 85-98.	1.0	110
40	Prenatal diagnosis of Menkes disease. Prenatal Diagnosis, 1998, 18, 287-289.	2.3	1
41	Successful Early Copper Therapy in Menkes Disease Associated with a Mutant Transcript Containing a Small In-Frame Deletion. Biochemical and Molecular Medicine, 1996, 57, 37-46.	1.4	61
42	Distinctive Menkes disease variant with occipital horns: Delineation of natural history and clinical phenotype. , 1996, 65, 44-51.		51
43	Menkes disease mutations and response to early copper histidine treatment. Nature Genetics, 1996, 13, 21-22.	21.4	59
44	Early copper therapy in classic Menkes disease patients with a novel splicing mutation. Annals of Neurology, 1995, 38, 921-928.	5.3	74
45	Occipital horn syndrome and a mild Menkes phenotype associated with splice site mutations at the MNK locus. Nature Genetics, 1994, 8, 195-202.	21.4	244
46	Plasma and cerebrospinal fluid neurochemical pattern in Menkes disease. Annals of Neurology, 1993, 33, 171-175.	5.3	101
47	Gastrointestinal hemorrhage associated with gastric polyps in Menkes disease. Journal of Pediatrics, 1993, 122, 93-95.	1.8	40
48	New autosomal recessive syndrome of sparse hair, osteopenia, and mental retardation in Mennonite sisters. American Journal of Medical Genetics Part A, 1992, 43, 983-988.	2.4	4