High Prevalence of SLC6A8 Deficiency in X-Linked Men

American Journal of Human Genetics
75, 97-105
DOI: 10.1086/422102

Citation Report

#	Article	IF	Citations
1	Comparative Frequency of Fragile-X (FMR1) and Creatine Transporter (SLC6A8) Mutations in X-Linked Mental Retardation. American Journal of Human Genetics, 2004, 75, 730-731.	6.2	9
2	Reply to Mandel. American Journal of Human Genetics, 2004, 75, 731-732.	6.2	1
3	Advances in X-linked mental retardation. Current Opinion in Pediatrics, 2005, 17, 720-724.	2.0	17
4	Incidence of Brain Creatine Transporter Deficiency in Males with Developmental Delay Referred for Brain Magnetic Resonance Imaging. Journal of Developmental and Behavioral Pediatrics, 2005, 26, 276-282.	1.1	63
5	X-linked mental retardation: further lumping, splitting and emerging phenotypes. Clinical Genetics, 2005, 67, 451-467.	2.0	51
6	X-linked mental retardation. Nature Reviews Genetics, 2005, 6, 46-57.	16.3	420
7	Non-syndromic X-linked mental retardation: From a molecular to a clinical point of view. Journal of Cellular Physiology, 2005, 204, 8-20.	4.1	36
8	Two novel mutations inSLC6A8cause creatine transporter defect and distinctive X-linked mental retardation in two unrelated Dutch families. , 2005, 132A, 288-295.		48
9	X-linked creatine transporter deficiency. Neurogenetics, 2005, 6, 165-168.	1.4	27
11	Creatine transporter localization in developing and adult retina: importance of creatine to retinal function. American Journal of Physiology - Cell Physiology, 2005, 289, C1015-C1023.	4.6	51
12	X linked mental retardation: a clinical guide. Journal of Medical Genetics, 2005, 43, 193-200.	3.2	90
13	Genetic, functional, and histopathological evaluation of two C-terminal BRCA1 missense variants. Journal of Medical Genetics, 2005, 43, 74-83.	3.2	39
14	Substituted Cysteine Accessibility of the Third Transmembrane Domain of the Creatine Transporter. Journal of Biological Chemistry, 2005, 280, 32649-32654.	3.4	19
15	Progressive intestinal, neurological and psychiatric problems in two adult males with cerebral creatine deficiency caused by an SLC6A8 mutation. Clinical Genetics, 2005, 68, 379-381.	2.0	33
16	Mutations in the JARID1C Gene, Which Is Involved in Transcriptional Regulation and Chromatin Remodeling, Cause X-Linked Mental Retardation. American Journal of Human Genetics, 2005, 76, 227-236.	6.2	349
17	The repertoire of solute carriers of family 6: Identification of new human and rodent genes. Biochemical and Biophysical Research Communications, 2005, 336, 175-189.	2.1	68
18	Laboratory diagnosis of defects of creatine biosynthesis and transport. Clinica Chimica Acta, 2005, 361, 1-9.	1.1	58
19	Purification and characterization of the creatine transporter expressed at high levels in HEK293 cells. Protein Expression and Purification, 2005, 41, 393-401.	1.3	15

#	Article	IF	Citations
21	MR Spectroscopy of Metabolic Disorders. Neuroimaging Clinics of North America, 2006, 16, 87-116.	1.0	50
23	X-linked mental retardation: many genes for a complex disorder. Current Opinion in Genetics and Development, 2006, 16, 260-269.	3.3	147
24	Effects of N-linked glycosylation on the creatine transporter. Biochemical Journal, 2006, 393, 459-469.	3.7	33
25	Are cerebral creatine deficiency syndromes on the radar screen?. Future Neurology, 2006, 1, 637-649.	0.5	16
26	Mutations in the gene encoding GlyT2 (SLC6A5) define a presynaptic component of human startle disease. Nature Genetics, 2006, 38, 801-806.	21.4	232
27	X-linked creatine transporter defect: A report on two unrelated boys with a severe clinical phenotype. Journal of Inherited Metabolic Disease, 2006, 29, 214-219.	3.6	70
28	X-Linked creatine transporter deficiency in two patients with severe mental retardation and autism. Journal of Inherited Metabolic Disease, 2006, 29, 220-223.	3.6	67
29	Overexpression of wild-type creatine transporter (SLC6A8) restores creatine uptake in primary SLC6A8-deficient fibroblasts. Journal of Inherited Metabolic Disease, 2006, 29, 345-346.	3.6	7
30	X-linked creatine transporter (SLC6A8) mutations in about 1% of males with mental retardation of unknown etiology. Human Genetics, 2006, 119, 604-610.	3.8	131
32	Brain pseudoatrophy and mental regression on valproate and a mitochondrial DNA mutation. Neurology, 2006, 67, 1715-1717.	1.1	36
33	High frequency of creatine deficiency syndromes in patients with unexplained mental retardation. Neurology, 2006, 67, 1713-1714.	1.1	86
34	Absence of cognitive, behavioral, or emotional dysfunction in progressive muscular atrophy. Neurology, 2006, 67, 1718-1719.	1.1	22
35	Thyroid replacement therapy and atrial fibrillation in acute ischemic stroke. Neurology, 2006, 67, 1714-1715.	1.1	4
36	Autism-lessons from the X chromosome. Social Cognitive and Affective Neuroscience, 2006, 1, 183-193.	3.0	74
37	Infusion-related hypersensitivity reactions during natalizumab treatment. Neurology, 2006, 67, 1717-1718.	1.1	47
38	The Functional Impact of SLC6 Transporter Genetic Variation. Annual Review of Pharmacology and Toxicology, 2007, 47, 401-441.	9.4	114
39	Selective Amino Acid Substitutions Convert the Creatine Transporter to a Î³-Aminobutyric Acid Transporter. Journal of Biological Chemistry, 2007, 282, 15528-15533.	3.4	42
40	Screening of Male Patients with Autism Spectrum Disorder for Creatine Transporter Deficiency. Neuropediatrics, 2007, 38, 310-312.	0.6	16

#	Article	IF	Citations
41	Quantification of Creatine and Guanidinoacetate Using GCâ€MS and LCâ€MS/MS for the Detection of Cerebral Creatine Deficiency Syndromes. Current Protocols in Human Genetics, 2007, 54, Unit 17.3.	3.5	35
42	Ontogeny up-regulates renal Na+/Clâ˜/creatine transporter in rat. Biochimica Et Biophysica Acta - Biomembranes, 2007, 1768, 2841-2848.	2.6	15
43	The Neuroprotective Role of Creatine. , 2007, 46, 205-243.		43
44	Mental retardation and verbal dyspraxia in a new patient with de novo creatine transporter (SLC6A8) mutation. American Journal of Medical Genetics, Part A, 2007, 143A, 1771-1774.	1.2	27
45	Functional characterization of missense variants in the creatine transporter gene (SLC6A8): improved diagnostic application. Human Mutation, 2007, 28, 890-896.	2.5	53
46	Creatine transporter deficiency: Prevalence among patients with mental retardation and pitfalls in metabolite screening. Clinical Biochemistry, 2007, 40, 1328-1331.	1.9	67
47	Severe Epilepsy in X-Linked Creatine Transporter Defect (CRTR-D). Epilepsia, 2007, 48, 1211-1213.	5.1	38
48	Detection of low-level somatic and germline mosaicism by denaturing high-performance liquid chromatography in a EURO-MRX family with SLC6A8 deficiency. Neurogenetics, 2008, 9, 183-190.	1.4	32
49	Yield of additional metabolic studies in neurodevelopmental disorders. Annals of Neurology, 2008, 64, 212-217.	5.3	40
50	Finding new etiologies of mental retardation and hypotonia: X marks the spot. Developmental Medicine and Child Neurology, 2008, 50, 104-111.	2.1	8
51	Session 2: Thursday 17 January 2008. Developmental Medicine and Child Neurology, 2008, 50, 6-32.	2.1	6
52	Expression and possible role of creatine transporter in the brain and at the bloodâ€cerebrospinal fluid barrier as a transporting protein of guanidinoacetate, an endogenous convulsant. Journal of Neurochemistry, 2008, 107, 768-778.	3.9	49
53	Autism and Metabolic Diseases. Journal of Child Neurology, 2008, 23, 307-314.	1.4	103
54	Laboratory Guide to the Methods in Biochemical Genetics. , 2008, , .		76
55	Treatment with<scp>l</scp>-Arginine improves neuropsychological disorders in a child with Creatine transporter defect. Neurocase, 2008, 14, 151-161.	0.6	44
56	Simultaneous determination of guanidinoacetate, creatine and creatinine in urine and plasma by un-derivatized liquid chromatography-tandem mass spectrometry. Annals of Clinical Biochemistry, 2008, 45, 575-584.	1.6	34
57	Mental retardation and inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2009, 32, 597-608.	3.6	43
58	Mutation screening in 86 known X-linked mental retardation genes by droplet-based multiplex PCR and massive parallel sequencing. The HUGO Journal, 2009, 3, 41-49.	4.1	48

#	Article	IF	Citations
60	Structural variation in Xq28: MECP2 duplications in 1% of patients with unexplained XLMR and in 2% of male patients with severe encephalopathy. European Journal of Human Genetics, 2009, 17, 444-453.	2.8	130
61	Immunohistochemical localisation of the creatine transporter in the rat brain. Neuroscience, 2009, 163, 571-585.	2.3	52
62	Screening for X-linked creatine transporter (SLC6A8) deficiency via simultaneous determination of urinary creatine to creatinine ratio by tandem mass-spectrometry. Molecular Genetics and Metabolism, 2009, 96, 273-275.	1.1	23
63	Creatine transporter deficiency in two adult patients with static encephalopathy. Journal of Inherited Metabolic Disease, 2009, 32, 91-96.	3.6	13
64	Controlling the Flow of Energy: Inhibition and Stimulation of the Creatine Transporter. Current Enzyme Inhibition, 2009, 5, 223-233.	0.4	7
65	Creatine transporter deficiency in two halfâ€brothers. American Journal of Medical Genetics, Part A, 2010, 152A, 1979-1983.	1.2	26
66	GC/MS determination of guanidinoacetate and creatine in urine: A routine method for creatine deficiency syndrome diagnosis. Clinical Biochemistry, 2010, 43, 1356-1361.	1.9	13
67	A new case of creatine transporter deficiency associated with mild clinical phenotype and a novel mutation in the SLC6A8 gene. Developmental Medicine and Child Neurology, 2010, 52, 215-217.	2.1	14
68	Synthesis and transport of creatine in the CNS: importance for cerebral functions. Journal of Neurochemistry, 2010, 115, 297-313.	3.9	145
69	Functional and immunocytochemical characterization of the creatine transporter in rat hippocampal neurons. Journal of Neurochemistry, 2010, 115, 684-693.	3.9	20
70	X-Linked Creatine Transporter Deficiency Presenting as a Mitochondrial Disorder. Journal of Child Neurology, 2010, 25, 1009-1012.	1.4	8
71	Creatine transporter expression after antidepressant therapy in rats bred for learned helplessness. World Journal of Biological Psychiatry, 2010, 11, 329-333.	2.6	9
72	The screening of SLC6A8 deficiency among Estonian families with Xâ€linked mental retardation. Journal of Inherited Metabolic Disease, 2010, 33, 5-11.	3.6	40
73	Creatine and Creatine Deficiency Syndromes: Biochemical and Clinical Aspects. Pediatric Neurology, 2010, 42, 163-171.	2.1	89
74	Response to creatine analogs in fibroblasts and patients with creatine transporter deficiency. Molecular Genetics and Metabolism, 2010, 99, 296-299.	1.1	27
76	Transport characteristics of guanidino compounds at the blood-brain barrier and blood-cerebrospinal fluid barrier: relevance to neural disorders. Fluids and Barriers of the CNS, 2011, 8, 13.	5.0	71
77	Language disorder with mild intellectual disability in a child affected by a novel mutation of SLC6A8 gene. Molecular Genetics and Metabolism, 2011, 102, 153-156.	1.1	23
78	Autism and Genetic Syndromes. , 0, , .		1

#	Article	IF	Citations
79	Should Metabolic Diseases Be Systematically Screened in Nonsyndromic Autism Spectrum Disorders?. PLoS ONE, 2011, 6, e21932.	2.5	31
80	Clinical features and X-inactivation in females heterozygous for creatine transporter defect. Clinical Genetics, 2011, 79, 264-272.	2.0	65
81	Characterization of novel SLC6A8 variants with the use of splice-site analysis tools and implementation of a newly developed LOVD database. European Journal of Human Genetics, 2011, 19, 56-63.	2.8	18
82	Hybridisation-based resequencing of 17 X-linked intellectual disability genes in 135 patients reveals novel mutations in ATRX, SLC6A8 and PQBP1. European Journal of Human Genetics, 2011, 19, 717-720.	2.8	21
83	Creatine deficiency syndromes and the importance of creatine synthesis in the brain. Amino Acids, 2011, 40, 1315-1324.	2.7	236
84	Creatine transporter defect diagnosed by proton NMR spectroscopy in males with intellectual disability. American Journal of Medical Genetics, Part A, 2011, 155, 2446-2452.	1.2	19
85	Disorders of creatine transport and metabolism. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2011, 157, 72-78.	1.6	88
86	Phenotypic Variability in a Portuguese Family With X-Linked Creatine Transport Deficiency. Pediatric Neurology, 2012, 46, 39-41.	2.1	8
87	Contiguous deletion of SLC6A8 and BAP31 in a patient with severe dystonia and sensorineural deafness. Molecular Genetics and Metabolism, 2012, 106, 43-47.	1.1	33
88	A novel c.2TÂ>ÂC mutation of the KDM5C/JARID1C gene in one large family with X-linked intellectual disability. European Journal of Medical Genetics, 2012, 55, 178-184.	1.3	42
89	Neuropsychological profile and clinical effects of arginine treatment in children with creatine transport deficiency. Orphanet Journal of Rare Diseases, 2012, 7, 43.	2.7	16
90	Screening for primary creatine deficiencies in French patients with unexplained neurological symptoms. Orphanet Journal of Rare Diseases, 2012, 7, 96.	2.7	33
91	Creatine Deficiency Syndromes. , 2012, , 239-247.		10
92	Expression and distribution of creatine transporter and creatine kinase (brain isoform) in developing and mature rat cochlear tissues. Histochemistry and Cell Biology, 2012, 137, 599-613.	1.7	9
93	A simple screening method using ion chromatography for the diagnosis of cerebral creatine deficiency syndromes. Amino Acids, 2012, 43, 993-997.	2.7	11
94	Deletion Xq27.3q28 in female patient with global developmental delays and skewed X-inactivation. BMC Medical Genetics, 2013, 14, 49.	2.1	10
95	Fragile X Syndrome and X-linked Intellectual Disability. , 2013, , 1-27.		1
96	Detection of a novel intragenic rearrangement in the creatine transporter gene by next generation sequencing. Molecular Genetics and Metabolism, 2013, 110, 465-471.	1.1	14

#	Article	IF	Citations
97	Creatine and guanidinoacetate reference values in a French population. Molecular Genetics and Metabolism, 2013, 110, 263-267.	1.1	32
98	Inborn errors of creatine metabolism and epilepsy. Epilepsia, 2013, 54, 217-227.	5.1	54
99	Functional and electrophysiological characterization of four nonâ€truncating mutations responsible for creatine transporter (SLC6A8) deficiency syndrome. Journal of Inherited Metabolic Disease, 2013, 36, 103-112.	3.6	14
100	Phenotype and genotype in 101 males with X-linked creatine transporter deficiency. Journal of Medical Genetics, 2013, 50, 463-472.	3.2	122
101	Biochemical, molecular, and clinical diagnoses of patients with cerebral creatine deficiency syndromes. Molecular Genetics and Metabolism, 2013, 109, 260-268.	1.1	34
102	A Novel SLC6A8 Mutation in a Large Family with X-Linked Intellectual Disability: Clinical and Proton Magnetic Resonance Spectroscopy Data of Both Hemizygous Males and Heterozygous Females. JIMD Reports, 2013, 13, 91-99.	1.5	10
103	MR Spectroscopy of Pediatric Brain Disorders. , 2013, , .		15
104	Membrane transporters and the diseases corresponding to functional defects. , 2013, , 1-146.		2
105	Cerebral Creatine Deficiencies: A Group of Treatable Intellectual Developmental Disorders. Seminars in Neurology, 2014, 34, 350-356.	1.4	48
106	The Biochemistry of Creatine. , 2014, , 91-103.		9
107	Use of MRS in Inborn Errors of Metabolism. , 2014, , 196-221.		0
108	Cloning and characterization of the promoter regions from the parent and paralogous creatine transporter genes. Gene, 2014, 533, 488-493.	2.2	10
109	Female mice heterozygous for creatine transporter deficiency show moderate cognitive deficits. Journal of Inherited Metabolic Disease, 2014, 37, 63-68.	3.6	27
110	Urine screening for patients with developmental disabilities detected a patient with creatine transporter deficiency due to a novel missense mutation in SLC6A8. Brain and Development, 2014, 36, 630-633.	1.1	13
111	Effects of Amide Creatine Derivatives in Brain Hippocampal Slices, and Their Possible Usefulness for Curing Creatine Transporter Deficiency. Neurochemical Research, 2014, 39, 37-45.	3.3	12
112	Xâ€linked creatine transporter deficiency: clinical aspects and pathophysiology. Journal of Inherited Metabolic Disease, 2014, 37, 715-733.	3.6	85
113	Defining the blanks â€“ Pharmacochaperoning of SLC6 transporters and ABC transporters. Pharmacological Research, 2014, 83, 63-73.	7.1	42
114	Treatment of X-linked creatine transporter (SLC6A8) deficiency: systematic review of the literature and three new cases. Molecular Genetics and Metabolism, 2014, 112, 259-274.	1.1	54

#	Article	IF	Citations
115	A novel SLC6A8 mutation associated with motor dysfunction in a child exhibiting creatine transporter deficiency. Human Genome Variation, 2015, 2, 15037.	0.7	5
116	Diagnostic methods and recommendations for the cerebral creatine deficiency syndromes. Pediatric Research, 2015, 77, 398-405.	2.3	167
117	Estimated carrier frequency of creatine transporter deficiency in females in the general population using functional characterization of novel missense variants in the SLC6A8 gene. Gene, 2015, 565, 187-191.	2.2	25
118	Creatine biosynthesis and transport in health and disease. Biochimie, 2015, 119, 146-165.	2.6	151
119	Creatine Transporter Deficiency. Journal of Developmental and Behavioral Pediatrics, 2016, 37, 322-326.	1.1	11
120	Creatine transporter deficiency leads to increased whole body and cellular metabolism. Amino Acids, 2016, 48, 2057-2065.	2.7	35
121	Chromosome Xq28 duplication encompassing MECP2: Clinical and molecular analysis of 16 new patients from 10 families in China. European Journal of Medical Genetics, 2016, 59, 347-353.	1.3	14
122	Creatine transporter deficiency: Novel mutations and functional studies. Molecular Genetics and Metabolism Reports, 2016, 8, 20-23.	1.1	17
123	Creatine kinase in ischemic and inflammatory disorders. Clinical and Translational Medicine, 2016, 5, 31.	4.0	40
124	Creatine Defects and Central Nervous System. Seminars in Pediatric Neurology, 2016, 23, 285-289.	2.0	29
125	Inherited metabolic disorders in <scp>T</scp>urkish patients with autism spectrum disorders. Autism Research, 2016, 9, 217-223.	3.8	30
126	Creatine synthesis and exchanges between brain cells: What can be learned from human creatine deficiencies and various experimental models?. Amino Acids, 2016, 48, 1877-1895.	2.7	52
127	Liver X Receptor Agonist Modifies the DNA Methylation Profile of Synapse and Neurogenesis-Related Genes in the Triple Transgenic Mouse Model of Alzheimerâ€™s Disease. Journal of Molecular Neuroscience, 2016, 58, 243-253.	2.3	27
128	Variable White Matter Atrophy and Intellectual Development in a Family With X-linked Creatine Transporter Deficiency Despite Genotypic Homogeneity. Pediatric Neurology, 2017, 67, 45-52.	2.1	8
129	Laboratory diagnosis of creatine deficiency syndromes: a technical standard and guideline of the American College of Medical Genetics and Genomics. Genetics in Medicine, 2017, 19, 256-263.	2.4	21
130	Pharmacochaperoning in a Drosophila model system rescues human dopamine transporter variants associated with infantile/juvenile parkinsonism. Journal of Biological Chemistry, 2017, 292, 19250-19265.	3.4	41
131	Treatable Genetic Metabolic Epilepsies. Current Treatment Options in Neurology, 2017, 19, 30.	1.8	10
132	Creatine in the central nervous system: From magnetic resonance spectroscopy to creatine deficiencies. Analytical Biochemistry, 2017, 529, 144-157.	2.4	88

#	Article	IF	Citations
133	Abnormal <i>N</i>-Glycosylation of a Novel Missense Creatine Transporter Mutant, G561R, Associated with Cerebral Creatine Deficiency Syndromes Alters Transporter Activity and Localization. Biological and Pharmaceutical Bulletin, 2017, 40, 49-55.	1.4	11
134	Relax, Cool Down and Scaffold: How to Restore Surface Expression of Folding-Deficient Mutant GPCRs and SLC6 Transporters. International Journal of Molecular Sciences, 2017, 18, 2416.	4.1	5
135	Variability of Creatine Metabolism Genes in Children with Autism Spectrum Disorder. International Journal of Molecular Sciences, 2017, 18, 1665.	4.1	11
136	Functional assessment of creatine transporter in control and X-linked SLC6A8-deficient fibroblasts. Molecular Genetics and Metabolism, 2018, 123, 463-471.	1.1	3
137	Cognitive deficits and increases in creatine precursors in a brainâ€specific knockout of the creatine transporter gene Slc6a8. Genes, Brain and Behavior, 2018, 17, e12461.	2.2	24
138	A novel SLC6A8 mutation associated with intellectual disabilities in a Chinese family exhibiting creatine transporter deficiency: case report. BMC Medical Genetics, 2018, 19, 193.	2.1	13
139	Rescue by 4-phenylbutyrate of several misfolded creatine transporter-1 variants linked to the creatine transporter deficiency syndrome. Neuropharmacology, 2019, 161, 107572.	4.1	29
140	Disorders of Creatine Metabolism and Epilepsy. , 2019, , 296-299.		0
141	Deletion of the Creatine Transporter (Slc6a8) in Dopaminergic Neurons Leads to Hyperactivity in Mice. Journal of Molecular Neuroscience, 2020, 70, 102-111.	2.3	9
142	Cerebral creatine deficiency: Black cat in the coal cellar. Acta Neurologica Belgica, 2021, 121, 1859-1861.	1.1	0
143	The Creatine Transporter Unfolded: A Knotty Premise in the Cerebral Creatine Deficiency Syndrome. Frontiers in Synaptic Neuroscience, 2020, 12, 588954.	2.5	28
144	Cyclocreatine Transport by SLC6A8, the Creatine Transporter, in HEK293 Cells, a Human Blood-Brain Barrier Model Cell, and CCDSs Patient-Derived Fibroblasts. Pharmaceutical Research, 2020, 37, 61.	3.5	7
145	Classification of the Molecular Defects Associated with Pathogenic Variants of the SLC6A8 Creatine Transporter. Biochemistry, 2020, 59, 1367-1377.	2.5	17
146	Studies of structural determinants of substrate binding in the Creatine Transporter (CreaT, SLC6A8) using molecular models. Scientific Reports, 2020, 10, 6241.	3.3	16
147	Determination of Intrinsic Creatine Transporter (Slc6a8) Activity and Creatine Transport Function of Leukocytes in Rats. Biological and Pharmaceutical Bulletin, 2020, 43, 474-479.	1.4	3
148	¹H-MR Spectroscopy of the Early Developmental Brain, Neonatal Encephalopathies, and Neurometabolic Disorders. Magnetic Resonance in Medical Sciences, 2022, 21, 9-28.	2.0	9
149	SLC6A8 is involved in the progression of non-small cell lung cancer through the Notch signaling pathway. Annals of Translational Medicine, 2021, 9, 264-264.	1.7	14
150	Autism: Screening of inborn errors of metabolism and unexpected results. Autism Research, 2021, 14, 887-896.	3.8	12

#	Article	IF	Citations
151	X-linked creatine transporter deficiency results in prolonged QTc and increased sudden death risk in humans and disease model. Genetics in Medicine, 2021, 23, 1864-1872.	2.4	8
152	Neuronal Cytoskeleton in Intellectual Disability: From Systems Biology and Modeling to Therapeutic Opportunities. International Journal of Molecular Sciences, 2021, 22, 6167.	4.1	13
153	Treatment experience in two adults with creatinfe transporter deficiency. Molecular Genetics and Metabolism Reports, 2021, 27, 100731.	1.1	3
155	The Role of Preclinical Models in Creatine Transporter Deficiency: Neurobiological Mechanisms, Biomarkers and Therapeutic Development. Genes, 2021, 12, 1123.	2.4	8
156	6.3 Mitochondria-Nucleus Energetic Communication: Role for Phosphotransfer Networks in Processing Cellular Information. , 2007, , 641-666.		5
157	Introduction â€“ Creatine: Cheap Ergogenic Supplement with Great Potential for Health and Disease. , 2007, 46, 1-16.		23
158	Functional Insights into the Creatine Transporter. , 2007, 46, 99-118.		64
159	Cerebral Creatine Deficiency Syndromes: Clinical Aspects, Treatment and Pathophysiology. , 2007, 46, 149-166.		145
160	Diagnosis of Creatine Metabolism Disorders by Determining Creatine and Guanidinoacetate in Plasma and Urine. Methods in Molecular Biology, 2010, 603, 175-185.	0.9	5
161	Creatine Deficiency Syndromes. , 2006, , 211-217.		9
162	Creatine Disorders. , 2014, , 529-540.		6
164	Creatine transporter deficiency, an underdiagnosed cause of male intellectual disability. BMJ Case Reports, 2020, 13, e237542.	0.5	6
165	Cyclocreatine treatment improves cognition in mice with creatine transporter deficiency. Journal of Clinical Investigation, 2012, 122, 2837-2846.	8.2	86
166	SLC6A8 Knockdown Suppresses the Invasion and Migration of Human Hepatocellular Carcinoma Huh-7 and Hep3B Cells. Technology in Cancer Research and Treatment, 2020, 19, 153303382098302.	1.9	7
167	Network Analysis of Differential Expression for the Identification of Disease-Causing Genes. PLoS ONE, 2009, 4, e5526.	2.5	61
168	Kreatinmangelsyndrome. , 2007, , 412-413.		0
169	Autism Spectrum Disorders: The Role of Genetics in Diagnosis and Treatment. , 2011, , .		0
171	StÃ¶rungen des Energiestoffwechsels. , 2014, , 516-539.		0

#	Article	IF	Citations
172	Kreatinmangelsyndrome. , 2015, , 1-5.		0
173	Genetische Defekte der FettsÃurenoxidation und des Ketonstoffwechsels. , 2015, , 1-20.		0
174	Mitochondriopathien. , 2015, , 1-20.		0
176	Kreatinmangelsyndrome. Springer Reference Medizin, 2019, , 1-2.	0.0	0
177	Genetische Defekte der FettsÃurenoxidation und des Ketonstoffwechsels. Springer Reference Medizin, 2019, , 1-15.	0.0	0
178	Genetische Defekte der FettsÃurenoxidation und des Ketonstoffwechsels. Springer Reference Medizin, 2020, , 735-749.	0.0	0
179	Kreatinmangelsyndrome. Springer Reference Medizin, 2020, , 767-768.	0.0	0
180	Creatine and its Metabolites. , 2008, , 739-749.		2
181	Current and potential new treatment strategies for creatine deficiency syndromes. Molecular Genetics and Metabolism, 2022, 135, 15-26.	1.1	17
183	Identification of novel variations in SLC6A8 and GAMT genes causing cerebral creatine deficiency syndrome. Clinica Chimica Acta, 2022, 532, 29-36.	1.1	1
185	Laboratory Diagnosis of Cerebral Creatine Deficiency Syndromes by Determining Creatine and Guanidinoacetate in Plasma and Urine. Methods in Molecular Biology, 2022, , 129-140.	0.9	1
186	Need for revision of the ACMG/AMP guidelines for interpretation of X-linked variants. Intractable and Rare Diseases Research, 2022, 11, 120-124.	0.9	1
188	X-linked creatine transporter (SLC6A8) deficiency in females: Difficult to recognize, but a potentially treatable disease. Molecular Genetics and Metabolism, 2023, 140, 107694.	1.1	0
190	Forensically relevant challenging behaviors and the genetics domain. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2023, , 65-73.	1.8	0
191	Diagnosis and Treatment of X-Linked Creatine Transporter Deficiency: Case Report and Literature Review. Brain Sciences, 2023, 13, 1382.	2.3	1
192	Suggestion of creatine as a new neurotransmitter by approaches ranging from chemical analysis and biochemistry to electrophysiology. ELife, 0, 12, .	6.0	0
193	ClinGen variant curation expert panel recommendations for classification of variants in GAMT, GATM and SLC6A8 for cerebral creatine deficiency syndromes. Molecular Genetics and Metabolism, 2024, 142, 108362.	1.1	0