Truncating mutations in the Wilson disease gene ATP7B are associated with very low ceruloplasmin oxidase activity and an early onset of Wi

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Citation Report

#	Article	IF	Citations
1	Copper Status Abnormalities and How to Measure Them in Neurodegenerative Disorders. Recent Patents on CNS Drug Discovery, 2010, 5, 182-194.	0.9	13
2	Homozygosity for Non-H1069Q Missense Mutations in ATP7B Gene and Early Severe Liver Disease: Report of Two Families and a Meta-analysis. JIMD Reports, 2011, 4, 129-137.	0.7	9
3	Late diagnosed Wilson disease with hepatic and neurological manifestations. Hepatology Research, 2011, 41, 270-276.	1.8	8
4	Polymorphism of methylenetetrahydrofolate reductase as disease modifier â€“ A dÃ©jÃ-vu in Wilson disease?. Journal of Hepatology, 2011, 55, 753-755.	1.8	5
5	Clinical presentation and mutations in Danish patients with Wilson disease. European Journal of Human Genetics, 2011, 19, 935-941.	1.4	42
7	Nutritional Influences in Selected Gastrointestinal Diseases. Digestive Diseases, 2011, 29, 154-165.	0.8	4
8	Systems biology approach to Wilsonâ€™s disease. BioMetals, 2011, 24, 455-466.	1.8	70
10	Phenotypic and Genetic Characterization of a Cohort of Pediatric Wilson Disease Patients. BMC Pediatrics, 2011, 11, 56.	0.7	45
12	Neurological Symptoms, Genotype-Phenotype Correlations and Ethnic-specific Differences in Bulgarian Patients With Wilson Disease. Neurologist, 2012, 18, 184-189.	0.4	24
13	Wilson Disease. Journal of Pediatric Gastroenterology and Nutrition, 2012, 54, 6-7.	0.9	2
14	Identification of a novel Wilson disease gene mutation frequent in Upper Austria: a genetic and clinical study. Journal of Human Genetics, 2012, 57, 564-567.	1.1	21
15	Genetics of Wilsonâ€™s disease: a clinical perspective. Indian Journal of Gastroenterology, 2012, 31, 285-293.	0.7	11
16	Defining Wilson Disease Phenotypes: From the Patient to the Bench and Back Again. Gastroenterology, 2012, 142, 692-696.	0.6	25
17	Using metalloproteomics to investigate the cellular physiology of copper in hepatocytes. Metallomics, 2012, 4, 633.	1.0	12
18	Genetic and metabolic liver disease. , 2012, , 157-259.		8
19	<scp>EGFP</scp> Tags Affect Cellular Localization of <scp>ATP7B</scp> Mutants. CNS Neuroscience and Therapeutics, 2013, 19, 346-351.	1.9	6
20	â€œNot so rareâ€•Wilson disease. Clinics and Research in Hepatology and Gastroenterology, 2013, 37, 219-221.	0.7	6
21	Metallostasis in Alzheimer's disease. Free Radical Biology and Medicine, 2013, 62, 76-89.	1.3	297

#	Article	IF	Citations
22	Mutational analysis of ATP7B gene and the genotype-phenotype correlation in patients with Wilson's disease in Serbia. Vojnosanitetski Pregled, 2013, 70, 457-462.	0.1	18
24	Biometals in rare neurodegenerative disorders of childhood. Frontiers in Aging Neuroscience, 2013, 5, 14.	1.7	10
25	Distinct phenotype of a Wilson disease mutation reveals a novel trafficking determinant in the copper transporter ATP7B. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, E1364-73.	3.3	40
26	Pathogenesis and management of <scp>W</scp>ilson disease. Hepatology Research, 2014, 44, 395-402.	1.8	40
27	Population screening for Wilson's disease. Annals of the New York Academy of Sciences, 2014, 1315, 64-69.	1.8	36
28	Wilson disease: What is still unclear in pediatric patients?. Clinics and Research in Hepatology and Gastroenterology, 2014, 38, 268-272.	0.7	8
29	Phenotypeâ€“genotype correlations in patients with Wilson's disease. Annals of the New York Academy of Sciences, 2014, 1315, 1-5.	1.8	70
30	Identification and characterization of a novel splice-site mutation in the Wilson disease gene. Journal of the Neurological Sciences, 2014, 345, 154-158.	0.3	4
31	Polymorphisms of metal transporter genes DMT1 and ATP7A in Wilson's disease. Journal of Trace Elements in Medicine and Biology, 2014, 28, 8-12.	1.5	22
32	Novel <scp>EDA</scp> mutation in Xâ€linked hypohidrotic ectodermal dysplasia and genotypeâ€“phenotype correlation. Oral Diseases, 2015, 21, 994-1000.	1.5	17
33	Defective roles of ATP7B missense mutations in cellular copper tolerance and copper excretion. Molecular and Cellular Neurosciences, 2015, 67, 31-36.	1.0	14
34	Most frequent mutation c.3402delC (p.Ala1135GlnfsX13) among Wilson disease patients in Venezuela has a wide distribution and two old origins. European Journal of Medical Genetics, 2015, 58, 59-65.	0.7	15
35	Wilson's disease and other neurological copper disorders. Lancet Neurology, The, 2015, 14, 103-113.	4.9	751
36	ATP7B Gene Mutations in Croatian Patients with Wilson Disease. Genetic Testing and Molecular Biomarkers, 2016, 20, 112-117.	0.3	11
37	Epidemiology and introduction to the clinical presentation of Wilson disease. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2017, 142, 7-17.	1.0	39
38	The genetics of Wilson disease. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2017, 142, 19-34.	1.0	99
39	Quantification of ATP7B Protein in Dried Blood Spots by Peptide Immuno-SRM as a Potential Screen for Wilsonâ€™s Disease. Journal of Proteome Research, 2017, 16, 862-871.	1.8	49
40	Spectrum of <scp>ATP7B</scp> mutations and genotypeâ€“phenotype correlation in largeâ€scale Chinese patients with Wilson Disease. Clinical Genetics, 2017, 92, 69-79.	1.0	34

#	Article	IF	Citations
41	Wilson disease: At the crossroads between genetics and epigeneticsâ€"A review of the evidence. Liver Research, 2017, 1, 121-130.	0.5	24
42	Identification of two novel mutations in the ATP7B gene that cause Wilsonâ€™s disease. World Journal of Pediatrics, 2017, 13, 387-391.	0.8	1
43	Wilson disease in children. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2017, 142, 141-156.	1.0	31
44	Wilson disease and related copper disorders. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 147, 279-292.	1.0	51
45	Novel compound heterozygote mutations inÂthe ATP7B gene in an Iranian family with Wilson disease: a case report. Journal of Medical Case Reports, 2018, 12, 68.	0.4	8
47	Wilson disease. Nature Reviews Disease Primers, 2018, 4, 21.	18.1	466
48	Developmental and Inherited Liver Disease. , 2018, , 111-274.		4
49	Three novel mutations in the ATP7B gene of unrelated Vietnamese patients with Wilson disease. BMC Medical Genetics, 2018, 19, 104.	2.1	5
51	A review and update on the diagnosis and treatment of neuropsychiatric Wilson disease. Expert Review of Neurotherapeutics, 2019, 19, 1117-1126.	1.4	13
52	The Genetics of Wilson Disease. , 2019, , 105-114.		0
53	Biochemical markers for the diagnosis and monitoring of Wilson Disease. , 2019, 40, 59-77.		40
54	Computing the Pathogenicity of Wilsonâ€™s Disease ATP7B Mutations: Implications for Disease Prevalence. Journal of Chemical Information and Modeling, 2019, 59, 5230-5243.	2.5	10
55	Relationship between genetic mutations and clinical phenotypes in patients with Wilson disease. Medicine (United States), 2019, 98, e18284.	0.4	5
56	Wholeâ€exome sequencing identifies novel pathogenic variants across the ATP7B gene and some modifiers of Wilson's disease phenotype. Liver International, 2019, 39, 177-186.	1.9	38
57	Age and Sex but Not ATP7B Genotype Effectively Influence the Clinical Phenotype of Wilson Disease. Hepatology, 2019, 69, 1464-1476.	3.6	110
58	Management of Wilson Disease Diagnosed in Infancy. Journal of Pediatric Gastroenterology and Nutrition, 2020, 70, 547-554.	0.9	8
59	Wilson Disease. Neurologic Clinics, 2020, 38, 417-432.	0.8	76
60	Wilson disease: revision of diagnostic criteria in a clinical series with great genetic homogeneity. Journal of Gastroenterology, 2021, 56, 78-89.	2.3	15

#	Article	IF	Citations
61	Investigation of the Wilson gene ATP7B transcriptional start site and the effect of core promoter alterations. Scientific Reports, 2021, 11, 7674.	1.6	2
62	Management Perspective of Wilsonâ€™s Disease: Early Diagnosis and Individualized Therapy. Current Neuropharmacology, 2021, 19, 465-485.	1.4	29
64	ATP7B variant spectrum in a French pediatric Wilson disease cohort. European Journal of Medical Genetics, 2021, 64, 104305.	0.7	6
65	Genotype-Phenotype Correlations in a Mountain Population Community with High Prevalence of Wilsonâ€™s Disease: Genetic and Clinical Homogeneity. PLoS ONE, 2014, 9, e98520.	1.1	38
66	Genetics and epigenetic factors of Wilson disease. Annals of Translational Medicine, 2019, 7, S58-S58.	0.7	33
67	Wilsonâ€™s Disease. , 2022, , 371-390.		0
68	Acute liver failure with hemolytic anemia in children with Wilsonâ€™s disease: Genotype-phenotype correlations?. World Journal of Hepatology, 2021, 13, 1428-1438.	0.8	7
69	Wilson's Disease among Children at King Hussein Medical Center. Journal of the Royal Medical Services, 2013, 20, 6-9.	0.0	0
70	A Biochemical Study to Estimate Malonaldehyde and Ceruloplasmin Oxidase Activity As Parameters Express Oxidation Â¢â‚¬Å“ Antioxidation Balance for Patients With Acute Hepatitis Virus Type C. International Journal of Computers & Technology, 2014, 12, 3508-3514.	0.2	0
71	Wilson HastalÄ±ÄŸÄ±: TanÄ±sal YaklaÅŸÄ±m. Ã‡ukurova Ãœniversitesi TÄ±p FakÃ¼ltesi Dergisi, 2015, 40, 345.	0.0	0
74	Wilson disease in children and young adults - State of the art. Saudi Journal of Gastroenterology, 2022, 28, 21.	0.5	10
75	Genomic medicine for liver disease. Hepatology, 2022, 76, 860-868.	3.6	7
76	Clinical and genetic characterization of a large cohort of patients with Wilsonâ€™s disease in China. Translational Neurodegeneration, 2022, 11, 13.	3.6	15
77	Assessment of the diagnostic value of serum ceruloplasmin for Wilsonâ€™s disease in children. BMC Gastroenterology, 2022, 22, 124.	0.8	3
78	Wilson Disease: Update on Pathophysiology and Treatment. Frontiers in Cell and Developmental Biology, 2022, 10, 871877.	1.8	18
80	ATP7B Genotype and Chronic Liver Disease Treatment Outcomes in Wilson Disease: Worse Survival With Loss-of-Function Variants. Clinical Gastroenterology and Hepatology, 2023, 21, 1323-1329.e4.	2.4	2
81	A multidisciplinary approach to the diagnosis and management of Wilson disease: 2022 Practice Guidance on Wilson disease from the American Association for the Study of Liver Diseases. Hepatology, 0, Publish Ahead of Print, .	3.6	24
82	Role of serum ceruloplasmin in the diagnosis of Wilson's disease: A large Chinese study. Frontiers in Neurology, 0, 13, .	1.1	2

#	Article	IF	Citations
83	Clinical spectrum and genotype-phenotype associations in Finnish patients with Wilson's disease. Journal of the Neurological Sciences, 2023, 448, 120620.	0.3	1
84	Clinical and genetic characterization of pediatric patients with Wilsonâ€™s disease from Yunnan province where ethnic minorities gather. Frontiers in Genetics, 0, 14, .	1.1	0
85	Clinical and Forensic Signs Resulting from Exposure to Heavy Metals and Other Chemical Elements of the Periodic Table. Journal of Clinical Medicine, 2023, 12, 2591.	1.0	4
86	Developmental and Inherited Liver Disease. , 2024, , 122-294.		0