

Viktor Kořich

List of Publications by Year in descending order

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134
papers

6,422
citations

71102

41
h-index

74163

75
g-index

147
all docs

147
docs citations

147
times ranked

6619
citing authors

#	ARTICLE	IF	CITATIONS
1	Postauthorization safety study of betaine anhydrous. Journal of Inherited Metabolic Disease, 2022, 45, 719-733.	3.6	5
2	Influence of early identification and therapy on long-term outcomes in early-onset MTHFR deficiency. Journal of Inherited Metabolic Disease, 2022, 45, 848-861.	3.6	7
3	Towards Achieving Equity and Innovation in Newborn Screening across Europe. International Journal of Neonatal Screening, 2022, 8, 31.	3.2	14
4	Laboratory newborn screening. Cesko-Slovenska Pediatrie, 2022, 77, 12-18.	0.2	3
5	Regulatory landscape of providing information on newborn screening to parents across Europe. European Journal of Human Genetics, 2021, 29, 67-78.	2.8	11
6	An international classification of inherited metabolic disorders (ICIMD). Journal of Inherited Metabolic Disease, 2021, 44, 164-177.	3.6	146
7	Cystathionine synthase deficiency in the ECHOD registry I: pyridoxine responsiveness as a determinant of biochemical and clinical phenotype at diagnosis. Journal of Inherited Metabolic Disease, 2021, 44, 677-692.	3.6	20
8	Clinical and molecular characterization of adult patients with late-onset MTHFR deficiency. Journal of Inherited Metabolic Disease, 2021, 44, 777-786.	3.6	4
9	Informing Parents about Newborn Screening: A European Comparison Study. International Journal of Neonatal Screening, 2021, 7, 13.	3.2	11
10	U-IMD: the first Unified European registry for inherited metabolic diseases. Orphanet Journal of Rare Diseases, 2021, 16, 95.	2.7	15
11	Neonatal Screening in Europe Revisited: An ISNS Perspective on the Current State and Developments Since 2010. International Journal of Neonatal Screening, 2021, 7, 15.	3.2	118
12	Homocystinuria patient and caregiver survey: experiences of diagnosis and patient satisfaction. Orphanet Journal of Rare Diseases, 2021, 16, 124.	2.7	9
13	Sulfur amino acid restriction, energy metabolism and obesity: a study protocol of an 8-week randomized controlled dietary intervention with whole foods and amino acid supplements. Journal of Translational Medicine, 2021, 19, 153.	4.4	12
14	Shifting landscapes of human MTHFR missense-variant effects. American Journal of Human Genetics, 2021, 108, 1283-1300.	6.2	33
15	Impact of Newborn Screening and Early Dietary Management on Clinical Outcome of Patients with Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency and Medium Chain Acyl-CoA Dehydrogenase Deficiency – A Retrospective Nationwide Study. Nutrients, 2021, 13, 2925.	4.1	4
16	Absence of MMACHC in peripheral retinal cells does not lead to an ocular phenotype in mice. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2021, 1867, 166201.	3.8	3
17	Inherited disorders of sulfur amino acid metabolism: recent advances in therapy. Current Opinion in Clinical Nutrition and Metabolic Care, 2021, 24, 62-70.	2.5	2
18	High Cysteine Diet Reduces Insulin Resistance in SHR-CRP Rats. Physiological Research, 2021, 70, 687-700.	0.9	2

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19	Knock-Out of Retrovirus Receptor Gene Tva in the Chicken Confers Resistance to Avian Leukosis Virus Subgroups A and K and Affects Cobalamin (Vitamin B12)-Dependent Level of Methylmalonic Acid. <i>Viruses</i> , 2021, 13, 2504.	3.3	13
20	Availability, accessibility and delivery to patients of the 28 orphan medicines approved by the European Medicine Agency for hereditary metabolic diseases in the MetabERN network. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 3.	2.7	8
21	Lessons Learned from Inherited Metabolic Disorders of Sulfur-Containing Amino Acids Metabolism. <i>Journal of Nutrition</i> , 2020, 150, 2506S-2517S.	2.9	16
22	A homozygous deletion in the SLC19A1 gene as a cause of folate-dependent recurrent megaloblastic anemia. <i>Blood</i> , 2020, 135, 2427-2431.	1.4	13
23	Long-term uninterrupted enzyme replacement therapy prevents liver disease in murine model of severe homocystinuria. <i>Human Mutation</i> , 2020, 41, 1662-1670.	2.5	7
24	A proactive genotype-to-patient-phenotype map for cystathionine beta-synthase. <i>Genome Medicine</i> , 2020, 12, 13.	8.2	45
25	A key leader in homocystinuria research: Jan P. Kraus (1942–2019). <i>Human Mutation</i> , 2019, 40, 1909-1909.	2.5	1
26	Factors Influencing Parental Awareness about Newborn Screening. <i>International Journal of Neonatal Screening</i> , 2019, 5, 35.	3.2	8
27	Comprehensive characterization of ureagenesis in the <i>spf</i> mouse, a model of human ornithine transcarbamylase deficiency, reveals age-dependency of ammonia detoxification. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 1064-1076.	3.6	9
28	Research activity and capability in the European reference network MetabERN. <i>Orphanet Journal of Rare Diseases</i> , 2019, 14, 119.	2.7	2
29	Comprehensive analysis of how experimental parameters affect H ₂ S measurements by the monobromobimane method. <i>Free Radical Biology and Medicine</i> , 2019, 136, 146-158.	2.9	44
30	Newborn screening for homocystinurias: Recent recommendations versus current practice. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 128-139.	3.6	37
31	Phenotype, treatment practice and outcome in the cobalamin-dependent remethylation disorders and MTHFR deficiency: Data from the E-HOD registry. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 333-352.	3.6	53
32	Metabolism of sulfur compounds in homocystinurias. <i>British Journal of Pharmacology</i> , 2019, 176, 594-606.	5.4	27
33	Epidemiology of rare diseases detected by newborn screening in the Czech Republic. <i>Central European Journal of Public Health</i> , 2019, 27, 153-159.	1.1	16
34	Enzyme Replacement Therapy Ameliorates Multiple Symptoms of Murine Homocystinuria. <i>Molecular Therapy</i> , 2018, 26, 834-844.	8.2	28
35	Biogenesis of Hydrogen Sulfide and Thioethers by Cystathionine Beta-Synthase. <i>Antioxidants and Redox Signaling</i> , 2018, 28, 311-323.	5.4	47
36	Phenotype, treatment practice and outcome in the cobalamin-dependent remethylation disorders and MTHFR deficiency: data from the E-HOD registry. <i>Journal of Inherited Metabolic Disease</i> , 2018, , .	3.6	2

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37	Neonatal screening in the Czech Republic: increased prevalence of selected diseases in low birthweight neonates. <i>European Journal of Pediatrics</i> , 2018, 177, 1697-1704.	2.7	5
38	Folate-Dependent Normocytic Anemia Caused By a Hypomorphic Mutation in SLC19A1 gene. <i>Blood</i> , 2018, 132, 502-502.	1.4	0
39	Activity of the liver enzyme ornithine carbamoyltransferase (OTC) in blood: LC-MS/MS assay for non-invasive diagnosis of ornithine carbamoyltransferase deficiency. <i>Clinical Chemistry and Laboratory Medicine</i> , 2017, 55, 1168-1177.	2.3	10
40	Guidelines for diagnosis and management of the cobalamin-related remethylation disorders cblC, cblD, cblE, cblF, cblG, cblJ and MTHFR deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 21-48.	3.6	206
41	Enzyme replacement prevents neonatal death, liver damage, and osteoporosis in murine homocystinuria. <i>FASEB Journal</i> , 2017, 31, 5495-5506.	0.5	24
42	Potential Pharmacological Chaperones for Cystathionine Beta-Synthase-Deficient Homocystinuria. <i>Handbook of Experimental Pharmacology</i> , 2017, 245, 345-383.	1.8	28
43	Guidelines for the diagnosis and management of cystathionine beta-synthase deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 49-74.	3.6	229
44	Mutation Update and Review of Severe Methylenetetrahydrofolate Reductase Deficiency. <i>Human Mutation</i> , 2016, 37, 427-438.	2.5	92
45	Thioethers as markers of hydrogen sulfide production in homocystinurias. <i>Biochimie</i> , 2016, 126, 14-20.	2.6	28
46	Reply to Sajantila and Budowle. <i>European Journal of Human Genetics</i> , 2016, 24, 330-330.	2.8	1
47	Genetic Variation in Renal Expression of <i>Folate Receptor 1</i> (<i>Folr1</i>) Gene Predisposes Spontaneously Hypertensive Rats to Metabolic Syndrome. <i>Hypertension</i> , 2016, 67, 335-341.	2.7	14
48	Disorders of Sulfur Amino Acid Metabolism. , 2016, , 309-320.		8
49	Enzyme replacement with PEGylated cystathionine β -synthase ameliorates homocystinuria in murine model. <i>Journal of Clinical Investigation</i> , 2016, 126, 2372-2384.	8.2	37
50	Splice-shifting oligonucleotide (SSO) mediated blocking of an exonic splicing enhancer (ESE) created by the prevalent c.903+469T>C MTRR mutation corrects splicing and restores enzyme activity in patient cells. <i>Nucleic Acids Research</i> , 2015, 43, 4627-4639.	14.5	28
51	CDH13 promoter SNPs with pleiotropic effect on cardiometabolic parameters represent methylation QTLs. <i>Human Genetics</i> , 2015, 134, 291-303.	3.8	32
52	Clinical onset and course, response to treatment and outcome in 24 patients with the cblE or cblG remethylation defect complemented by genetic and in vitro enzyme study data. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 957-967.	3.6	40
53	Chaperone therapy for homocystinuria: the rescue of CBS mutations by heme arginate. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 287-294.	3.6	28
54	Newborn screening for homocystinurias and methylation disorders: systematic review and proposed guidelines. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 1007-1019.	3.6	100

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55	Insights into Severe 5,10-Methylenetetrahydrofolate Reductase Deficiency: Molecular Genetic and Enzymatic Characterization of 76 Patients. <i>Human Mutation</i> , 2015, 36, 611-621.	2.5	63
56	Enzymatic diagnosis of homocystinuria by determination of cystathionine- γ -synthase activity in plasma using LC-MS/MS. <i>Clinica Chimica Acta</i> , 2015, 438, 261-265.	1.1	26
57	Metabolic Syndrome, Alcohol Consumption and Genetic Factors Are Associated with Serum Uric Acid Concentration. <i>PLoS ONE</i> , 2014, 9, e97646.	2.5	41
58	Recommendations for reporting results of diagnostic genetic testing (biochemical, cytogenetic and) <i>Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5</i>	2.8	109
59	Simultaneous determination of cystathionine, total homocysteine, and methionine in dried blood spots by liquid chromatography/tandem mass spectrometry and its utility for the management of patients with homocystinuria. <i>Clinica Chimica Acta</i> , 2014, 437, 211-217.	1.1	32
60	Biochemical properties of nematode O-acetylserine(thiol)lyase paralogs imply their distinct roles in hydrogen sulfide homeostasis. <i>Biochimica Et Biophysica Acta - Proteins and Proteomics</i> , 2013, 1834, 2691-2701.	2.3	20
61	The Cysteine Dioxygenase Knockout Mouse: Altered Cysteine Metabolism in Nonhepatic Tissues Leads to Excess H ₂ S/HS [•] Production and Evidence of Pancreatic and Lung Toxicity. <i>Antioxidants and Redox Signaling</i> , 2013, 19, 1321-1336.	5.4	59
62	Developmental programming of growth: Genetic variant in GH2 gene encoding placental growth hormone contributes to adult height determination. <i>Placenta</i> , 2013, 34, 995-1001.	1.5	16
63	Folate Deficiency Is Associated With Oxidative Stress, Increased Blood Pressure, and Insulin Resistance in Spontaneously Hypertensive Rats. <i>American Journal of Hypertension</i> , 2013, 26, 135-140.	2.0	76
64	Cysteine and obesity. <i>Current Opinion in Clinical Nutrition and Metabolic Care</i> , 2012, 15, 49-57.	2.5	86
65	Rare Allelic Variants Determine Folate Status in an Unsupplemented European Population. <i>Journal of Nutrition</i> , 2012, 142, 1403-1409.	2.9	9
66	Novel structural arrangement of nematode cystathionine γ -synthases: characterization of <i>Caenorhabditis elegans</i> CBS-1. <i>Biochemical Journal</i> , 2012, 443, 535-547.	3.7	33
67	Clinical picture of S-adenosylhomocysteine hydrolase deficiency resembles phosphomannomutase 2 deficiency. <i>Molecular Genetics and Metabolism</i> , 2012, 107, 611-613.	1.1	30
68	Conformational Properties of Nine Purified Cystathionine γ -Synthase Mutants. <i>Biochemistry</i> , 2012, 51, 4755-4763.	2.5	24
69	Cystathionine beta- γ -synthase mutants exhibit changes in protein unfolding: conformational analysis of misfolded variants in crude cell extracts. <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 469-477.	3.6	12
70	Cysteine and Obesity. <i>Obesity</i> , 2012, 20, 473-481.	3.0	59
71	Homocysteine and Coronary Heart Disease: Meta-analysis of MTHFR Case-Control Studies, Avoiding Publication Bias. <i>PLoS Medicine</i> , 2012, 9, e1001177.	8.4	167
72	Clinical validation of cutoff target ranges in newborn screening of metabolic disorders by tandem mass spectrometry: A worldwide collaborative project. <i>Genetics in Medicine</i> , 2011, 13, 230-254.	2.4	308

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73	Restoring assembly and activity of cystathionine Î²â€synthase mutants by ligands and chemical chaperones. <i>Journal of Inherited Metabolic Disease</i> , 2011, 34, 39-48.	3.6	46
74	Vascular presentation of cystathionine betaâ€synthase deficiency in adulthood. <i>Journal of Inherited Metabolic Disease</i> , 2011, 34, 33-37.	3.6	47
75	Determination of cystathionine beta-synthase activity in human plasma by LC-MS/MS: potential use in diagnosis of CBS deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2011, 34, 49-55.	3.6	29
76	Foreword to special issue on homocysteine disorders. <i>Journal of Inherited Metabolic Disease</i> , 2011, 34, 1-2.	3.6	0
77	Novel polymorphic AluYb8 insertion in the WNK1 gene is associated with blood pressure variation in Europeans. <i>Human Mutation</i> , 2011, 32, 806-814.	2.5	23
78	The birth prevalence of lysosomal storage disorders in the Czech Republic: comparison with data in different populations. <i>Journal of Inherited Metabolic Disease</i> , 2010, 33, 387-396.	3.6	157
79	The deep intronic c.903+469T>C mutation in the <i>MTRR</i> gene creates an SF2/ASF binding exonic splicing enhancer, which leads to pseudoexon activation and causes the cbIE type of homocystinuria. <i>Human Mutation</i> , 2010, 31, 437-444.	2.5	53
80	Cystathionine Î²-synthase mutations: effect of mutation topology on folding and activity. <i>Human Mutation</i> , 2010, 31, 809-819.	2.5	50
81	Hypervariable intronic region in NCX1 is enriched in short insertion-deletion polymorphisms and showed association with cardiovascular traits. <i>BMC Medical Genetics</i> , 2010, 11, 15.	2.1	8
82	Ancient origin of the <i>CTH</i> allele carrying the c.200C>T (p.T67I) variant in patients with cystathioninuria. <i>Clinical Genetics</i> , 2010, 78, 554-559.	2.0	10
83	Cross-Talk between the Catalytic Core and the Regulatory Domain in Cystathionine Î²-Synthase: Study by Differential Covalent Labeling and Computational Modeling. <i>Biochemistry</i> , 2010, 49, 10526-10534.	2.5	15
84	Cystathionine beta-synthase null homocystinuric mice fail to exhibit altered hemostasis or lowering of plasma homocysteine in response to betaine treatment. <i>Molecular Genetics and Metabolism</i> , 2010, 101, 163-171.	1.1	57
85	A novel transgenic mouse model of CBS-deficient homocystinuria does not incur hepatic steatosis or fibrosis and exhibits a hypercoagulative phenotype that is ameliorated by betaine treatment. <i>Molecular Genetics and Metabolism</i> , 2010, 101, 153-162.	1.1	60
86	Birth Prevalence of Homocystinuria in Central Europe: Frequency and Pathogenicity of Mutation c.1105C>T (p.R369C) in the Cystathionine Beta-Synthase Gene. <i>Journal of Pediatrics</i> , 2009, 154, 431-437.	1.8	40
87	Determination of S-Adenosylmethionine and S-Adenosylhomocysteine by LCâ€MS/MS and evaluation of their stability in mice tissues. <i>Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences</i> , 2009, 877, 2061-2066.	2.3	48
88	Cystathionine Î³-lyase: Clinical, metabolic, genetic, and structural studies. <i>Molecular Genetics and Metabolism</i> , 2009, 97, 250-259.	1.1	57
89	Quality of analytical performance in inherited metabolic disorders: the role of ERNDIM. <i>Journal of Inherited Metabolic Disease</i> , 2008, 31, 680-689.	3.6	23
90	Cystathionine Î²-synthase p.S466L mutation causes hyperhomocysteinemia in mice. <i>Human Mutation</i> , 2008, 29, 1048-1054.	2.5	28

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91	Chemical chaperone rescue of mutant human cystathionine Î²-synthase. <i>Molecular Genetics and Metabolism</i> , 2007, 91, 335-342.	1.1	70
92	Diversity of cystathionine Î²-synthase haplotypes bearing the most common homocystinuria mutation c.833T>C: a possible role for gene conversion. <i>Human Mutation</i> , 2007, 28, 255-264.	2.5	20
93	Resequencing PNMT in European hypertensive and normotensive individuals: no common susceptibility variants for hypertension and purifying selection on intron 1. <i>BMC Medical Genetics</i> , 2007, 8, 47.	2.1	12
94	Single-nucleotide polymorphisms in genes relating to homocysteine metabolism: how applicable are public SNP databases to a typical European population?. <i>European Journal of Human Genetics</i> , 2005, 13, 86-95.	2.8	18
95	cblE type of homocystinuria due to methionine synthase reductase deficiency: Functional correction by minigene expression. <i>Human Mutation</i> , 2005, 25, 239-247.	2.5	44
96	The cblD Defect Causes Either Isolated or Combined Deficiency of Methylcobalamin and Adenosylcobalamin Synthesis. <i>Journal of Biological Chemistry</i> , 2004, 279, 42742-42749.	3.4	103
97	Identification and functional analysis of two novel mutations in the CBS gene in Polish patients with homocystinuria. <i>Human Mutation</i> , 2004, 23, 631-631.	2.5	8
98	The cystathionine Î²-synthase (CBS) mutation c.1224-2A>C in Central Europe: Vitamin B6 nonresponsiveness and a common ancestral haplotype. <i>Human Mutation</i> , 2004, 24, 352-353.	2.5	16
99	CblE type of homocystinuria: Mild clinical phenotype in two patients homozygous for a novel mutation in the MTRR gene. <i>Journal of Inherited Metabolic Disease</i> , 2003, 26, 361-369.	3.6	29
100	Homocystinuria due to cystathionine Î²-synthase deficiency: Novel biochemical findings and treatment efficacy. <i>Journal of Inherited Metabolic Disease</i> , 2003, 26, 761-773.	3.6	37
101	Effect of folic acid on fenofibrate-induced elevation of homocysteine and cysteine. <i>American Heart Journal</i> , 2003, 146, 110A-115A.	2.7	22
102	Genetic variants of homocysteine metabolizing enzymes and the risk of coronary artery disease. <i>Molecular Genetics and Metabolism</i> , 2003, 79, 167-175.	1.1	38
103	MTHFR 677Câ†’T Polymorphism and Risk of Coronary Heart Disease. <i>JAMA - Journal of the American Medical Association</i> , 2002, 288, 2023.	7.4	819
104	Essential hypertension in adolescents: association with insulin resistance and with metabolism of homocysteine and vitamins. <i>American Journal of Hypertension</i> , 2002, 15, 857-864.	2.0	80
105	Methionine-loading test: evaluation of adverse effects and safety in an epidemiological study. <i>Clinical Nutrition</i> , 2002, 21, 151-156.	5.0	12
106	High homocysteine and thrombosis without connective tissue disorders are associated with a novel class of cystathionine Î²-synthase (CBS) mutations. <i>Human Mutation</i> , 2002, 19, 641-655.	2.5	64
107	Cystathionine Î²-synthase is coordinately regulated with proliferation through a redox-sensitive mechanism in cultured human cells and <i>Saccharomyces cerevisiae</i> . <i>Journal of Cellular Physiology</i> , 2002, 192, 81-92.	4.1	60
108	CblE type of homocystinuria due to methionine synthase reductase deficiency: Clinical and molecular studies and prenatal diagnosis in two families. <i>Journal of Inherited Metabolic Disease</i> , 2002, 25, 461-476.	3.6	38

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109	Association of the Glu298Asp Polymorphism in the Endothelial Nitric Oxide Synthase Gene with Essential Hypertension Resistant to Conventional Therapy. <i>Biochemical and Biophysical Research Communications</i> , 2001, 284, 426-430.	2.1	97
110	Impaired Heme Binding and Aggregation of Mutant Cystathionine Î ² -Synthase Subunits in Homocystinuria. <i>American Journal of Human Genetics</i> , 2001, 68, 1506-1513.	6.2	78
111	Cystathionine beta-synthase deficiency in Central Europe: Discrepancy between biochemical and molecular genetic screening for homocystinuric alleles. <i>Human Mutation</i> , 2001, 18, 548-549.	2.5	29
112	Haplotyping of wild type and I278T alleles of the human cystathionine Î ² -synthase gene based on a cluster of novel SNPs in IVS12. <i>Human Mutation</i> , 2001, 17, 350-351.	2.5	8
113	Folate supplementation prevents plasma homocysteine increase after fenofibrate therapy. <i>Nutrition</i> , 2001, 17, 721-723.	2.4	57
114	Is the common 844ins68 polymorphism in the cystathionine Î ² -synthase gene associated with atherosclerosis?. <i>Journal of Inherited Metabolic Disease</i> , 1999, 22, 674-675.	3.6	16
115	Cystathionine Î ² -synthase mutations in homocystinuria. <i>Human Mutation</i> , 1999, 13, 362-375.	2.5	247
116	Four novel mutations in the cystathionine Î ² -synthase gene: Effect of a second linked mutation on the severity of the homocystinuric phenotype. , 1999, 13, 453-457.		27
117	Identification of a Mutation Cluster in Mevalonate Kinase Deficiency, Including a New Mutation in a Patient of Mennonite Ancestry. <i>American Journal of Human Genetics</i> , 1999, 65, 327-335.	6.2	40
118	Cystathionine Î ² -synthase mutations in homocystinuria. <i>Human Mutation</i> , 1999, 13, 362.	2.5	28
119	Unusual enzyme findings in five patients with metabolic profiles suggestive of succinic semialdehyde dehydrogenase deficiency (4-hydroxybutyric aciduria). <i>Journal of Inherited Metabolic Disease</i> , 1998, 21, 255-261.	3.6	7
120	The Human Cystathionine Î ² -Synthase (CBS) Gene: Complete Sequence, Alternative Splicing, and Polymorphisms. <i>Genomics</i> , 1998, 52, 312-324.	2.9	134
121	Analysis of CBS alleles in Czech and Slovak patients with homocystinuria: Report on three novel mutations E176K, W409X and 1223 + 37 de199. <i>Journal of Inherited Metabolic Disease</i> , 1997, 20, 363-366.	3.6	14
122	Hyperhomocysteinemia in premature arterial disease: examination of cystathionine Î ² -synthase alleles at the molecular level. <i>Human Molecular Genetics</i> , 1995, 4, 623-629.	2.9	60
123	Identical genotypes in siblings with different homocystinuric phenotypes: identification of three mutations in cystathionine Î ² -synthase using an improved bacterial expression system. <i>Human Molecular Genetics</i> , 1994, 3, 1103-1108.	2.9	40
124	Molecular defect in a patient with pyridoxine-responsive homocystinuria. <i>Human Molecular Genetics</i> , 1993, 2, 815-816.	2.9	27
125	Molecular basis of cystathionine Î ² -synthase deficiency in pyridoxine responsive and nonresponsive homocystinuria. <i>Human Molecular Genetics</i> , 1993, 2, 1857-1860.	2.9	99
126	Human cystathionine Î ² -synthase cDNA: sequence, alternative splicing and expression in cultured cells. <i>Human Molecular Genetics</i> , 1993, 2, 1633-1638.	2.9	116

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127	Pyridoxine Responsive and Unresponsive Homocystinuria. Journal of Nutritional Science and Vitaminology, 1992, 38, 589-592.	0.6	2
128	Unusual clinical presentation in two boys with cytochrome oxidase deficiency. Journal of Inherited Metabolic Disease, 1992, 15, 320-322.	3.6	5
129	Screening for mutations by expressing patient cDNA segments in E. coli: Homocystinuria due to cystathionine β -synthase deficiency. Human Mutation, 1992, 1, 113-123.	2.5	103
130	Accumulation and impaired in vivo metabolism of di- and trihydroxycholestanic acid in two patients. Clinica Chimica Acta, 1991, 202, 123-132.	1.1	6
131	Facts and artefacts in mevalonic aciduria: development of a stable isotope dilution GCMS assay for mevalonic acid and its application to physiological fluids, tissue samples, prenatal diagnosis and carrier detection. Clinica Chimica Acta, 1991, 198, 209-227.	1.1	34
132	Mevalonic aciduria. Journal of Inherited Metabolic Disease, 1991, 14, 265-266.	3.6	14
133	Di- and trihydroxycholestanemia in twin sisters. Journal of Inherited Metabolic Disease, 1991, 14, 357-360.	3.6	7
134	Newborn screening for homocystinurias: recent recommendations versus current practice. Journal of Inherited Metabolic Disease, 0, , .	3.6	1