Viktor KoÅ¾ich

List of Publications by Year
in descending order

134 papers	6,422 citations	76196 40 h-index	79541 73 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.	1.7	5
2	Influence of early identification and therapy on longâ€term outcomes in earlyâ€onset <scp>MTHFR</scp> deficiency. Journal of Inherited Metabolic Disease, 2022, 45, 848-861.	1.7	7
3	Towards Achieving Equity and Innovation in Newborn Screening across Europe. International Journal of Neonatal Screening, 2022, 8, 31.	1.2	14
4	Laboratory newborn screening. Cesko-Slovenska Pediatrie, 2022, 77, 12-18.	0.0	3
5	Regulatory landscape of providing information on newborn screening to parents across Europe. European Journal of Human Genetics, 2021, 29, 67-78.	1.4	11
6	An international classification of inherited metabolic disorders (<scp>ICIMD</scp>). Journal of Inherited Metabolic Disease, 2021, 44, 164-177.	1.7	146
7	Cystathionine Î²â€synthase deficiency in the <scp>Eâ€HOD registryâ€part</scp> I: pyridoxine responsiveness as a determinant of biochemical and clinical phenotype at diagnosis. Journal of Inherited Metabolic Disease, 2021, 44, 677-692.	1.7	20
8	Clinical and molecular characterization of adult patients with lateâ€onset <scp>MTHFR</scp> deficiency. Journal of Inherited Metabolic Disease, 2021, 44, 777-786.	1.7	4
9	Informing Parents about Newborn Screening: A European Comparison Study. International Journal of Neonatal Screening, 2021, 7, 13.	1.2	11
10	U-IMD: the first Unified European registry for inherited metabolic diseases. Orphanet Journal of Rare Diseases, 2021, 16, 95.	1.2	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.	1.2	118
12	Homocystinuria patient and caregiver survey: experiences of diagnosis and patient satisfaction. Orphanet Journal of Rare Diseases, 2021, 16, 124.	1.2	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.	1.8	12
14	Shifting landscapes of human MTHFR missense-variant effects. American Journal of Human Genetics, 2021, 108, 1283-1300.	2.6	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 Deficiencyâ€”A Retrospective Nationwide Study. Nutrients, 2021, 13, 2925.	1.7	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.	1.8	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.	1.3	2
18	High Cysteine Diet Reduces Insulin Resistance in SHR-CRP Rats. Physiological Research, 2021, 70, 687-700.	0.4	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. Viruses, 2021, 13, 2504.	1.5	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. Orphanet Journal of Rare Diseases, 2020, 15, 3.	1.2	8
21	Lessons Learned from Inherited Metabolic Disorders of Sulfur-Containing Amino Acids Metabolism. Journal of Nutrition, 2020, 150, 2506S-2517S.	1.3	16
22	A homozygous deletion in the SLC19A1 gene as a cause of folate-dependent recurrent megaloblastic anemia. Blood, 2020, 135, 2427-2431.	0.6	13
23	Longâ€term uninterrupted enzyme replacement therapy prevents liver disease in murine model of severe homocystinuria. Human Mutation, 2020, 41, 1662-1670.	1.1	7
24	A proactive genotype-to-patient-phenotype map for cystathionine beta-synthase. Genome Medicine, 2020, 12, 13.	3.6	45
25	A key leader in homocystinuria research: Jan P. Kraus (1942â€“2019). Human Mutation, 2019, 40, 1909-1909.	1.1	1
26	Factors Influencing Parental Awareness about Newborn Screening. International Journal of Neonatal Screening, 2019, 5, 35.	1.2	8
27	Comprehensive characterization of ureagenesis in the spf^{ash} mouse, a model of human ornithine transcarbamylase deficiency, reveals ageâ€dependency of ammonia detoxification. Journal of Inherited Metabolic Disease, 2019, 42, 1064-1076.	1.7	9
28	Research activity and capability in the European reference network MetabERN. Orphanet Journal of Rare Diseases, 2019, 14, 119.	1.2	2
29	Comprehensive analysis of how experimental parameters affect H2S measurements by the monobromobimane method. Free Radical Biology and Medicine, 2019, 136, 146-158.	1.3	44
30	Newborn screening for homocystinurias: Recent recommendations versus current practice. Journal of Inherited Metabolic Disease, 2019, 42, 128-139.	1.7	37
31	Phenotype, treatment practice and outcome in the cobalaminâ€dependent remethylation disorders and MTHFR deficiency: Data from the Eâ€HOD registry. Journal of Inherited Metabolic Disease, 2019, 42, 333-352.	1.7	53
32	Metabolism of sulfur compounds in homocystinurias. British Journal of Pharmacology, 2019, 176, 594-606.	2.7	27
33	Epidemiology of rare diseases detected by newborn screening in the Czech Republic. Central European Journal of Public Health, 2019, 27, 153-159.	0.4	16
34	Enzyme Replacement Therapy Ameliorates Multiple Symptoms of Murine Homocystinuria. Molecular Therapy, 2018, 26, 834-844.	3.7	28
35	Biogenesis of Hydrogen Sulfide and Thioethers by Cystathionine Beta-Synthase. Antioxidants and Redox Signaling, 2018, 28, 311-323.	2.5	47
36	Phenotype, treatment practice and outcome in the cobalamin-dependent remethylation disorders and MTHFR deficiency: data from the E-HOD registry. Journal of Inherited Metabolic Disease, 2018, , .	1.7	2

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37	Neonatal screening in the Czech Republic: increased prevalence of selected diseases in low birthweight neonates. European Journal of Pediatrics, 2018, 177, 1697-1704.	1.3	5
38	Folate-Dependent Normocytic Anemia Caused By a Hypomorphic Mutation in SLC19A1 gene. Blood, 2018, 132, 502-502.	0.6	0
39	Activity of the liver enzyme ornithine carbamoyltransferase (OTC) in blood: LC-MS/MS assay for non-invasive diagnosis of ornithine carbamoyltransferase deficiency. Clinical Chemistry and Laboratory Medicine, 2017, 55, 1168-1177.	1.4	10
40	Guidelines for diagnosis and management of the cobalaminâ€related remethylation disorders cblC, cblD, cblE, cblF, cblG, cblJ and MTHFR deficiency. Journal of Inherited Metabolic Disease, 2017, 40, 21-48.	1.7	206
41	Enzyme replacement prevents neonatal death, liver damage, and osteoporosis in murine homocystinuria. FASEB Journal, 2017, 31, 5495-5506.	0.2	24
42	Potential Pharmacological Chaperones for Cystathionine Beta-Synthase-Deficient Homocystinuria. Handbook of Experimental Pharmacology, 2017, 245, 345-383.	0.9	28
43	Guidelines for the diagnosis and management of cystathionine betaâ€synthase deficiency. Journal of Inherited Metabolic Disease, 2017, 40, 49-74.	1.7	229
44	Mutation Update and Review of Severe Methylenetetrahydrofolate Reductase Deficiency. Human Mutation, 2016, 37, 427-438.	1.1	92
45	Thioethers as markers of hydrogen sulfide production in homocystinurias. Biochimie, 2016, 126, 14-20.	1.3	28
46	Reply to Sajantila and Budowle. European Journal of Human Genetics, 2016, 24, 330-330.	1.4	1
47	Genetic Variation in Renal Expression of Folate Receptor 1 (Folr1) Gene Predisposes Spontaneously Hypertensive Rats to Metabolic Syndrome. Hypertension, 2016, 67, 335-341.	1.3	14
48	Disorders of Sulfur Amino Acid Metabolism. , 2016, , 309-320.		8
49	Enzyme replacement with PEGylated cystathionine Î²-synthase ameliorates homocystinuria in murine model. Journal of Clinical Investigation, 2016, 126, 2372-2384.	3.9	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. Nucleic Acids Research, 2015, 43, 4627-4639.	6.5	28
51	CDH13 promoter SNPs with pleiotropic effect on cardiometabolic parameters represent methylation QTLs. Human Genetics, 2015, 134, 291-303.	1.8	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. Journal of Inherited Metabolic Disease, 2015, 38, 957-967.	1.7	40
53	Chaperone therapy for homocystinuria: the rescue of CBS mutations by heme arginate. Journal of Inherited Metabolic Disease, 2015, 38, 287-294.	1.7	28
54	Newborn screening for homocystinurias and methylation disorders: systematic review and proposed guidelines. Journal of Inherited Metabolic Disease, 2015, 38, 1007-1019.	1.7	100

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55	Insights into Severe 5,10-Methylenetetrahydrofolate Reductase Deficiency: Molecular Genetic and Enzymatic Characterization of 76 Patients. Human Mutation, 2015, 36, 611-621.	1.1	63
56	Enzymatic diagnosis of homocystinuria by determination of cystathionine-ÃŸ-synthase activity in plasma using LC-MS/MS. Clinica Chimica Acta, 2015, 438, 261-265.	0.5	26
57	Metabolic Syndrome, Alcohol Consumption and Genetic Factors Are Associated with Serum Uric Acid Concentration. PLoS ONE, 2014, 9, e97646.	1.1	41
58	Recommendations for reporting results of diagnostic genetic testing (biochemical, cytogenetic and) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5	1.4	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. Clinica Chimica Acta, 2014, 437, 211-217.	0.5	32
60	Biochemical properties of nematode O-acetylserine(thiol)lyase paralogs imply their distinct roles in hydrogen sulfide homeostasis. Biochimica Et Biophysica Acta - Proteins and Proteomics, 2013, 1834, 2691-2701.	1.1	20
61	The Cysteine Dioxgenase Knockout Mouse: Altered Cysteine Metabolism in Nonhepatic Tissues Leads to Excess H₂S/HS^{â”}Production and Evidence of Pancreatic and Lung Toxicity. Antioxidants and Redox Signaling, 2013, 19, 1321-1336.	2.5	59
62	Developmental programming of growth: Genetic variant in GH2 gene encoding placental growth hormone contributes to adult height determination. Placenta, 2013, 34, 995-1001.	0.7	16
63	Folate Deficiency Is Associated With Oxidative Stress, Increased Blood Pressure, and Insulin Resistance in Spontaneously Hypertensive Rats. American Journal of Hypertension, 2013, 26, 135-140.	1.0	76
64	Cysteine and obesity. Current Opinion in Clinical Nutrition and Metabolic Care, 2012, 15, 49-57.	1.3	86
65	Rare Allelic Variants Determine Folate Status in an Unsupplemented European Population. Journal of Nutrition, 2012, 142, 1403-1409.	1.3	9
66	Novel structural arrangement of nematode cystathionine Î²-synthases: characterization of Caenorhabditis elegans CBS-1. Biochemical Journal, 2012, 443, 535-547.	1.7	33
67	Clinical picture of S-adenosylhomocysteine hydrolase deficiency resembles phosphomannomutase 2 deficiency. Molecular Genetics and Metabolism, 2012, 107, 611-613.	0.5	30
68	Conformational Properties of Nine Purified Cystathionine Î²-Synthase Mutants. Biochemistry, 2012, 51, 4755-4763.	1.2	24
69	Cystathionine betaâ€synthase mutants exhibit changes in protein unfolding: conformational analysis of misfolded variants in crude cell extracts. Journal of Inherited Metabolic Disease, 2012, 35, 469-477.	1.7	12
70	Cysteine and Obesity. Obesity, 2012, 20, 473-481.	1.5	59
71	Homocysteine and Coronary Heart Disease: Meta-analysis of MTHFR Case-Control Studies, Avoiding Publication Bias. PLoS Medicine, 2012, 9, e1001177.	3.9	167
72	Clinical validation of cutoff target ranges in newborn screening of metabolic disorders by tandem mass spectrometry: A worldwide collaborative project. Genetics in Medicine, 2011, 13, 230-254.	1.1	308

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73	Restoring assembly and activity of cystathionine Î²â€synthase mutants by ligands and chemical chaperones. Journal of Inherited Metabolic Disease, 2011, 34, 39-48.	1.7	46
74	Vascular presentation of cystathionine betaâ€synthase deficiency in adulthood. Journal of Inherited Metabolic Disease, 2011, 34, 33-37.	1.7	47
75	Determination of cystathionine beta-synthase activity in human plasma by LC-MS/MS: potential use in diagnosis of CBS deficiency. Journal of Inherited Metabolic Disease, 2011, 34, 49-55.	1.7	29
76	Foreword to special issue on homocysteine disorders. Journal of Inherited Metabolic Disease, 2011, 34, 1-2.	1.7	0
77	Novel polymorphic AluYb8 insertion in the WNK1 gene is associated with blood pressure variation in Europeans. Human Mutation, 2011, 32, 806-814.	1.1	23
78	The birth prevalence of lysosomal storage disorders in the Czech Republic: comparison with data in different populations. Journal of Inherited Metabolic Disease, 2010, 33, 387-396.	1.7	157
79	The deep intronic c.903+469T>C mutation in the MTRR gene creates an SF2/ASF binding exonic splicing enhancer, which leads to pseudoexon activation and causes the cblE type of homocystinuria. Human Mutation, 2010, 31, 437-444.	1.1	53
80	Cystathionine Î²-synthase mutations: effect of mutation topology on folding and activity. Human Mutation, 2010, 31, 809-819.	1.1	50
81	Hypervariable intronic region in NCX1 is enriched in short insertion-deletion polymorphisms and showed association with cardiovascular traits. BMC Medical Genetics, 2010, 11, 15.	2.1	8
82	Ancient origin of the CTH alelle carrying the c.200C>T (p.T67I) variant in patients with cystathioninuria. Clinical Genetics, 2010, 78, 554-559.	1.0	10
83	Cross-Talk between the Catalytic Core and the Regulatory Domain in Cystathionine Î²-Synthase: Study by Differential Covalent Labeling and Computational Modeling. Biochemistry, 2010, 49, 10526-10534.	1.2	15
84	Cystathionine beta-synthase null homocystinuric mice fail to exhibit altered hemostasis or lowering of plasma homocysteine in response to betaine treatment. Molecular Genetics and Metabolism, 2010, 101, 163-171.	0.5	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. Molecular Genetics and Metabolism, 2010, 101, 153-162.	0.5	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. Journal of Pediatrics, 2009, 154, 431-437.	0.9	40
87	Determination of S-Adenosylmethionine and S-Adenosylhomocysteine by LCâ€“MS/MS and evaluation of their stability in mice tissues. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2009, 877, 2061-2066.	1.2	48
88	Cystathionine Î³-lyase: Clinical, metabolic, genetic, and structural studies. Molecular Genetics and Metabolism, 2009, 97, 250-259.	0.5	57
89	Quality of analytical performance in inherited metabolic disorders: the role of ERNDIM. Journal of Inherited Metabolic Disease, 2008, 31, 680-689.	1.7	23
90	Cystathionine Î²-synthase p.S466L mutation causes hyperhomocysteinemia in mice. Human Mutation, 2008, 29, 1048-1054.	1.1	28

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91	Chemical chaperone rescue of mutant human cystathionine Î²-synthase. Molecular Genetics and Metabolism, 2007, 91, 335-342.	0.5	70
92	Diversity of cystathionine Î²-synthase haplotypes bearing the most common homocystinuria mutation c.833T>C: a possible role for gene conversion. Human Mutation, 2007, 28, 255-264.	1.1	20
93	Resequencing PNMTin European hypertensive and normotensive individuals: no common susceptibilily variants for hypertension and purifying selection on intron 1. BMC Medical Genetics, 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?. European Journal of Human Genetics, 2005, 13, 86-95.	1.4	18
95	cblEType of homocystinuria due to methionine synthase reductase deficiency: Functional correction by minigene expression. Human Mutation, 2005, 25, 239-247.	1.1	44
96	The cblD Defect Causes Either Isolated or Combined Deficiency of Methylcobalamin and Adenosylcobalamin Synthesis. Journal of Biological Chemistry, 2004, 279, 42742-42749.	1.6	103
97	Identification and functional analysis of two novel mutations in the CBS gene in Polish patients with homocystinuria. Human Mutation, 2004, 23, 631-631.	1.1	8
98	The cystathionine ?-synthase (CBS) mutation c.1224-2A>C in Central Europe: Vitamin B6 nonresponsiveness and a common ancestral haplotype. Human Mutation, 2004, 24, 352-353.	1.1	16
99	CblE type of homocystinuria: Mild clinical phenotype in two patients homozygous for a novel mutation in the MTRR gene. Journal of Inherited Metabolic Disease, 2003, 26, 361-369.	1.7	29
100	Homocystinuria due to cystathionine Î²-synthase deficiency: Novel biochemical findings and treatment efficacy. Journal of Inherited Metabolic Disease, 2003, 26, 761-773.	1.7	37
101	Effect of folic acid on fenofibrate-induced elevation of homocysteine and cysteine. American Heart Journal, 2003, 146, 110A-115A.	1.2	22
102	Genetic variants of homocysteine metabolizing enzymes and the risk of coronary artery disease. Molecular Genetics and Metabolism, 2003, 79, 167-175.	0.5	38
103	MTHFR 677Câ†'T Polymorphism and Risk of Coronary Heart Disease. JAMA - Journal of the American Medical Association, 2002, 288, 2023.	3.8	819
104	Essential hypertension in adolescents: association with insulin resistance and with metabolism of homocysteine and vitamins. American Journal of Hypertension, 2002, 15, 857-864.	1.0	80
105	Methionine-loading test: evaluation of adverse effects and safety in an epidemiological study. Clinical Nutrition, 2002, 21, 151-156.	2.3	12
106	High homocysteine and thrombosis without connective tissue disorders are associated with a novel class of cystathionine Î²-synthase (CBS) mutations. Human Mutation, 2002, 19, 641-655.	1.1	64
107	Cystathionine ?-synthase is coordinately regulated with proliferation through a redox-sensitive mechanism in cultured human cells andSaccharomyces cerevisiae. Journal of Cellular Physiology, 2002, 192, 81-92.	2.0	60
108	CblE type of homocystinuria due to methionine synthase reductase deficiency: Clinical and molecular studies and prenatal diagnosis in two families. Journal of Inherited Metabolic Disease, 2002, 25, 461-476.	1.7	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. Biochemical and Biophysical Research Communications, 2001, 284, 426-430.	1.0	97
110	Impaired Heme Binding and Aggregation of Mutant Cystathionine Î²-Synthase Subunits in Homocystinuria. American Journal of Human Genetics, 2001, 68, 1506-1513.	2.6	78
111	Cystathionine beta-synthase deficiency in Central Europe: Discrepancy between biochemical and molecular genetic screening for homocystinuric alleles. Human Mutation, 2001, 18, 548-549.	1.1	29
112	Haplotyping of wild type and I278T alleles of the human cystathionine ?-synthase gene based on a cluster of novel SNPs in IVS12. Human Mutation, 2001, 17, 350-351.	1.1	8
113	Folate supplementation prevents plasma homocysteine increase after fenofibrate therapy. Nutrition, 2001, 17, 721-723.	1.1	57
114	Is the common 844ins68 polymorphism in the cystathionine Î²-synthase gene associated with atherosclerosis?. Journal of Inherited Metabolic Disease, 1999, 22, 674-675.	1.7	16
115	Cystathionine ?-synthase mutations in homocystinuria. , 1999, 13, 362-375.		247
116	Four novel mutations in the cystathionine ?-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. American Journal of Human Genetics, 1999, 65, 327-335.	2.6	40
118	Cystathionine Î²-synthase mutations in homocystinuria. Human Mutation, 1999, 13, 362.	1.1	28
119	Unusual enzyme findings in five patients with metabolic profiles suggestive of succinic semialdehyde dehydrogenase deficiency (4-hydroxybutyric aciduria). Journal of Inherited Metabolic Disease, 1998, 21, 255-261.	1.7	7
120	The Human Cystathionine Î²-Synthase (CBS) Gene: Complete Sequence, Alternative Splicing, and Polymorphisms. Genomics, 1998, 52, 312-324.	1.3	134
121	Analysis of CBS alleles in Czech and Slovak patients with homocystinuria: Report on three novel mutations E176K, W409X and 1223 + 37 de199. Journal of Inherited Metabolic Disease, 1997, 20, 363-366.	1.7	14
122	Hyperhomocysteinemia in premature arterial disease: examination of cystathionine Î²-synthase alleles at the molecular level. Human Molecular Genetics, 1995, 4, 623-629.	1.4	60
123	Identical genotypes in siblings with different homocystinuric phenotypes: identification of three mutations in cystathionine Î²-synthase using an improved bacterial expression system. Human Molecular Genetics, 1994, 3, 1103-1108.	1.4	40
124	Molecular defect in a patient with pyridoxine-responsive homocystinuria. Human Molecular Genetics, 1993, 2, 815-816.	1.4	27
125	Molecular basis of cystathionine Î²-synthase deficiency in pyridoxine responsive and nonresponsive homocystinuria. Human Molecular Genetics, 1993, 2, 1857-1860.	1.4	99
126	Human cystathionine Î²-synthase cDNA: sequence, alternative splicing and expression in cultured cells. Human Molecular Genetics, 1993, 2, 1633-1638.	1.4	116

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127	Pyridoxine Responsive and Unresponsive Homocystinuria. Journal of Nutritional Science and Vitaminology, 1992, 38, 589-592.	0.2	2
128	Unusual clinical presentation in two boys with cytochromec oxidase deficiency. Journal of Inherited Metabolic Disease, 1992, 15, 320-322.	1.7	5
129	Screening for mutations by expressing patient cDNA segments inE. coli: Homocystinuria due to cystathionine Î²-synthase deficiency. Human Mutation, 1992, 1, 113-123.	1.1	103
130	Accumulation and impaired in vivo metabolism of di- and trihydroxycholestanoic acid in two patients. Clinica Chimica Acta, 1991, 202, 123-132.	0.5	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.	0.5	34
132	Mevalonic aciduria. Journal of Inherited Metabolic Disease, 1991, 14, 265-266.	1.7	14
133	Di- and trihydroxycholestanaemia in twin sisters. Journal of Inherited Metabolic Disease, 1991, 14, 357-360.	1.7	7
134	Newborn screening for homocystinurias: recent recommendations versus current practice. Journal of Inherited Metabolic Disease, 0, , .	1.7	1