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	MTHFR 677Câ†'T Polymorphism and Risk of Coronary Heart Disease. JAMA - Journal of the American Medical Association, 2002, 288, 2023.	3.8	819
2	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
3	Cystathionine ?-synthase mutations in homocystinuria. , 1999, 13, 362-375.		247
4	Guidelines for the diagnosis and management of cystathionine betaâ€synthase deficiency. Journal of Inherited Metabolic Disease, 2017, 40, 49-74.	1.7	229
5	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
6	Homocysteine and Coronary Heart Disease: Meta-analysis of MTHFR Case-Control Studies, Avoiding Publication Bias. PLoS Medicine, 2012, 9, e1001177.	3.9	167
7	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
8	An international classification of inherited metabolic disorders (<scp>ICIMD</scp>). Journal of Inherited Metabolic Disease, 2021, 44, 164-177.	1.7	146
9	The Human Cystathionine Î²-Synthase (CBS) Gene: Complete Sequence, Alternative Splicing, and Polymorphisms. Genomics, 1998, 52, 312-324.	1.3	134
10	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
11	Human cystathionine Î²-synthase cDNA: sequence, alternative splicing and expression in cultured cells. Human Molecular Genetics, 1993, 2, 1633-1638.	1.4	116
12	Recommendations for reporting results of diagnostic genetic testing (biochemical, cytogenetic and) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5	1.4	109
13	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
14	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
15	Newborn screening for homocystinurias and methylation disorders: systematic review and proposed guidelines. Journal of Inherited Metabolic Disease, 2015, 38, 1007-1019.	1.7	100
16	Molecular basis of cystathionine Î²-synthase deficiency in pyridoxine responsive and nonresponsive homocystinuria. Human Molecular Genetics, 1993, 2, 1857-1860.	1.4	99
17	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
18	Mutation Update and Review of Severe Methylenetetrahydrofolate Reductase Deficiency. Human Mutation, 2016, 37, 427-438.	1.1	92

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19	Cysteine and obesity. Current Opinion in Clinical Nutrition and Metabolic Care, 2012, 15, 49-57.	1.3	86
20	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
21	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
22	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
23	Chemical chaperone rescue of mutant human cystathionine Î²-synthase. Molecular Genetics and Metabolism, 2007, 91, 335-342.	0.5	70
24	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
25	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
26	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
27	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
28	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
29	Cysteine and Obesity. Obesity, 2012, 20, 473-481.	1.5	59
30	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
31	Folate supplementation prevents plasma homocysteine increase after fenofibrate therapy. Nutrition, 2001, 17, 721-723.	1.1	57
32	Cystathionine Î³-lyase: Clinical, metabolic, genetic, and structural studies. Molecular Genetics and Metabolism, 2009, 97, 250-259.	0.5	57
33	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
34	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
35	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
36	Cystathionine Î²-synthase mutations: effect of mutation topology on folding and activity. Human Mutation, 2010, 31, 809-819.	1.1	50

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37	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
38	Vascular presentation of cystathionine betaâ€synthase deficiency in adulthood. Journal of Inherited Metabolic Disease, 2011, 34, 33-37.	1.7	47
39	Biogenesis of Hydrogen Sulfide and Thioethers by Cystathionine Beta-Synthase. Antioxidants and Redox Signaling, 2018, 28, 311-323.	2.5	47
40	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
41	A proactive genotype-to-patient-phenotype map for cystathionine beta-synthase. Genome Medicine, 2020, 12, 13.	3.6	45
42	cblEType of homocystinuria due to methionine synthase reductase deficiency: Functional correction by minigene expression. Human Mutation, 2005, 25, 239-247.	1.1	44
43	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
44	Metabolic Syndrome, Alcohol Consumption and Genetic Factors Are Associated with Serum Uric Acid Concentration. PLoS ONE, 2014, 9, e97646.	1.1	41
45	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
46	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
47	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
48	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
49	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
50	Genetic variants of homocysteine metabolizing enzymes and the risk of coronary artery disease. Molecular Genetics and Metabolism, 2003, 79, 167-175.	0.5	38
51	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
52	Newborn screening for homocystinurias: Recent recommendations versus current practice. Journal of Inherited Metabolic Disease, 2019, 42, 128-139.	1.7	37
53	Enzyme replacement with PEGylated cystathionine Î²-synthase ameliorates homocystinuria in murine model. Journal of Clinical Investigation, 2016, 126, 2372-2384.	3.9	37
54	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

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55	Novel structural arrangement of nematode cystathionine Î²-synthases: characterization of Caenorhabditis elegans CBS-1. Biochemical Journal, 2012, 443, 535-547.	1.7	33
56	Shifting landscapes of human MTHFR missense-variant effects. American Journal of Human Genetics, 2021, 108, 1283-1300.	2.6	33
57	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
58	CDH13 promoter SNPs with pleiotropic effect on cardiometabolic parameters represent methylation QTLs. Human Genetics, 2015, 134, 291-303.	1.8	32
59	Clinical picture of S-adenosylhomocysteine hydrolase deficiency resembles phosphomannomutase 2 deficiency. Molecular Genetics and Metabolism, 2012, 107, 611-613.	0.5	30
60	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
61	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
62	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
63	Cystathionine Î²-synthase p.S466L mutation causes hyperhomocysteinemia in mice. Human Mutation, 2008, 29, 1048-1054.	1.1	28
64	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
65	Chaperone therapy for homocystinuria: the rescue of CBS mutations by heme arginate. Journal of Inherited Metabolic Disease, 2015, 38, 287-294.	1.7	28
66	Thioethers as markers of hydrogen sulfide production in homocystinurias. Biochimie, 2016, 126, 14-20.	1.3	28
67	Potential Pharmacological Chaperones for Cystathionine Beta-Synthase-Deficient Homocystinuria. Handbook of Experimental Pharmacology, 2017, 245, 345-383.	0.9	28
68	Enzyme Replacement Therapy Ameliorates Multiple Symptoms of Murine Homocystinuria. Molecular Therapy, 2018, 26, 834-844.	3.7	28
69	Cystathionine Î²-synthase mutations in homocystinuria. Human Mutation, 1999, 13, 362.	1.1	28
70	Molecular defect in a patient with pyridoxine-responsive homocystinuria. Human Molecular Genetics, 1993, 2, 815-816.	1.4	27
71	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
72	Metabolism of sulfur compounds in homocystinurias. British Journal of Pharmacology, 2019, 176, 594-606.	2.7	27

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73	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
74	Conformational Properties of Nine Purified Cystathionine Î²-Synthase Mutants. Biochemistry, 2012, 51, 4755-4763.	1.2	24
75	Enzyme replacement prevents neonatal death, liver damage, and osteoporosis in murine homocystinuria. FASEB Journal, 2017, 31, 5495-5506.	0.2	24
76	Quality of analytical performance in inherited metabolic disorders: the role of ERNDIM. Journal of Inherited Metabolic Disease, 2008, 31, 680-689.	1.7	23
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	Effect of folic acid on fenofibrate-induced elevation of homocysteine and cysteine. American Heart Journal, 2003, 146, 110A-115A.	1.2	22
79	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
80	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
81	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
82	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
83	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
84	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
85	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
86	Lessons Learned from Inherited Metabolic Disorders of Sulfur-Containing Amino Acids Metabolism. Journal of Nutrition, 2020, 150, 2506S-2517S.	1.3	16
87	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
88	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
89	U-IMD: the first Unified European registry for inherited metabolic diseases. Orphanet Journal of Rare Diseases, 2021, 16, 95.	1.2	15
90	Mevalonic aciduria. Journal of Inherited Metabolic Disease, 1991, 14, 265-266.	1.7	14

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91	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
92	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
93	Towards Achieving Equity and Innovation in Newborn Screening across Europe. International Journal of Neonatal Screening, 2022, 8, 31.	1.2	14
94	A homozygous deletion in the SLC19A1 gene as a cause of folate-dependent recurrent megaloblastic anemia. Blood, 2020, 135, 2427-2431.	0.6	13
95	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
96	Methionine-loading test: evaluation of adverse effects and safety in an epidemiological study. Clinical Nutrition, 2002, 21, 151-156.	2.3	12
97	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
98	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
99	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
100	Regulatory landscape of providing information on newborn screening to parents across Europe. European Journal of Human Genetics, 2021, 29, 67-78.	1.4	11
101	Informing Parents about Newborn Screening: A European Comparison Study. International Journal of Neonatal Screening, 2021, 7, 13.	1.2	11
102	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
103	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
104	Rare Allelic Variants Determine Folate Status in an Unsupplemented European Population. Journal of Nutrition, 2012, 142, 1403-1409.	1.3	9
105	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
106	Homocystinuria patient and caregiver survey: experiences of diagnosis and patient satisfaction. Orphanet Journal of Rare Diseases, 2021, 16, 124.	1.2	9
107	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
108	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

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109	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
110	Factors Influencing Parental Awareness about Newborn Screening. International Journal of Neonatal Screening, 2019, 5, 35.	1.2	8
111	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
112	Disorders of Sulfur Amino Acid Metabolism. , 2016, , 309-320.		8
113	Di- and trihydroxycholestanaemia in twin sisters. Journal of Inherited Metabolic Disease, 1991, 14, 357-360.	1.7	7
114	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
115	Longâ€term uninterrupted enzyme replacement therapy prevents liver disease in murine model of severe homocystinuria. Human Mutation, 2020, 41, 1662-1670.	1.1	7
116	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
117	Accumulation and impaired in vivo metabolism of di- and trihydroxycholestanoic acid in two patients. Clinica Chimica Acta, 1991, 202, 123-132.	0.5	6
118	Unusual clinical presentation in two boys with cytochromec oxidase deficiency. Journal of Inherited Metabolic Disease, 1992, 15, 320-322.	1.7	5
119	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
120	Postauthorization safety study of betaine anhydrous. Journal of Inherited Metabolic Disease, 2022, 45, 719-733.	1.7	5
121	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
122	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
123	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
124	Laboratory newborn screening. Cesko-Slovenska Pediatrie, 2022, 77, 12-18.	0.0	3
125	Pyridoxine Responsive and Unresponsive Homocystinuria. Journal of Nutritional Science and Vitaminology, 1992, 38, 589-592.	0.2	2
126	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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127	Research activity and capability in the European reference network MetabERN. Orphanet Journal of Rare Diseases, 2019, 14, 119.	1.2	2
128	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
129	High Cysteine Diet Reduces Insulin Resistance in SHR-CRP Rats. Physiological Research, 2021, 70, 687-700.	0.4	2
130	Reply to Sajantila and Budowle. European Journal of Human Genetics, 2016, 24, 330-330.	1.4	1
131	A key leader in homocystinuria research: Jan P. Kraus (1942â€“2019). Human Mutation, 2019, 40, 1909-1909.	1.1	1
132	Newborn screening for homocystinurias: recent recommendations versus current practice. Journal of Inherited Metabolic Disease, 0, , .	1.7	1
133	Foreword to special issue on homocysteine disorders. Journal of Inherited Metabolic Disease, 2011, 34, 1-2.	1.7	0
134	Folate-Dependent Normocytic Anemia Caused By a Hypomorphic Mutation in SLC19A1 gene. Blood, 2018, 132, 502-502.	0.6	0