R Rozen

List of Publications by Year
in descending order

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94 papers	18,432 citations	41258 49 h-index	49773 87 g-index
96 all docs	96 docs citations	96 times ranked	10070 citing authors

#	Article	IF	Citations
1	A candidate genetic risk factor for vascular disease: a common mutation in methylenetetrahydrofolate reductase. Nature Genetics, 1995, 10, 111-113.	9.4	5,180
2	Relation Between Folate Status, a Common Mutation in Methylenetetrahydrofolate Reductase, and Plasma Homocysteine Concentrations. Circulation, 1996, 93, 7-9.	1.6	1,173
3	A Second Genetic Polymorphism in Methylenetetrahydrofolate Reductase (MTHFR) Associated with Decreased Enzyme Activity. Molecular Genetics and Metabolism, 1998, 64, 169-172.	0.5	1,154
4	Mutated methylenetetrahydrofolate reductase as a risk factor for spina bifida. Lancet, The, 1995, 346, 1070-1071.	6.3	786
5	Human methylenetetrahydrofolate reductase: isolation of cDNA, mapping and mutation identification. Nature Genetics, 1994, 7, 195-200.	9.4	773
6	Mice deficient in methylenetetrahydrofolate reductase exhibit hyperhomocysteinemia and decreased methylation capacity, with neuropathology and aortic lipid deposition. Human Molecular Genetics, 2001, 10, 433-443.	1.4	539
7	Methylenetetrahydrofolate reductase polymorphism, dietary interactions, and risk of colorectal cancer. Cancer Research, 1997, 57, 1098-102.	0.4	528
8	Molecular genetic analysis in mild hyperhomocysteinemia: a common mutation in the methylenetetrahydrofolate reductase gene is a genetic risk factor for cardiovascular disease. American Journal of Human Genetics, 1996, 58, 35-41.	2.6	458
9	Methylenetetrahydrofolate Reductase Polymorphism, Plasma Folate, Homocysteine, and Risk of Myocardial Infarction in US Physicians. Circulation, 1996, 94, 2410-2416.	1.6	399
10	A Common Variant in Methionine Synthase Reductase Combined with Low Cobalamin (Vitamin B12) Increases Risk for Spina Bifida. Molecular Genetics and Metabolism, 1999, 67, 317-323.	0.5	398
11	The structure and properties of methylenetetrahydrofolate reductase from Escherichia coli suggest how folate ameliorates human hyperhomocysteinemia. Nature Structural Biology, 1999, 6, 359-365.	9.7	392
12	Effects of common polymorphisms on the properties of recombinant human methylenetetrahydrofolate reductase. Proceedings of the National Academy of Sciences of the United States of America, 2001, 98, 14853-14858.	3.3	358
13	Human methionine synthase: cDNA cloning and identification of mutations in patients of the cblG complementation group of folate/cobalamin disorders. Human Molecular Genetics, 1996, 5, 1867-1874.	1.4	344
14	The 1298Aât'C polymorphism in methylenetetrahydrofolate reductase (MTHFR): in vitro expression and association with homocysteine. Atherosclerosis, 2001, 156, 409-415.	0.4	339
15	Polymorphisms in Genes Involved in Folate Metabolism as Maternal Risk Factors for Down Syndrome. American Journal of Human Genetics, 2000, 67, 623-630.	2.6	333
16	A rapid procedure for extracting genomic DNA from leukocytes. Nucleic Acids Research, 1991, 19, 408-408.	6.5	313
17	Socio-economic disparities in preterm birth: causal pathways and mechanisms. Paediatric and Perinatal Epidemiology, 2001, 15, 104-123.	0.8	308
18	Gene structure of human and mouse methylenetetrahydrofolate reductase (MTHFR). Mammalian Genome, 1998, 9, 652-656.	1.0	287

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19	Genetic polymorphisms in methylenetetrahydrofolate reductase and methionine synthase, folate levels in red blood cells, and risk of neural tube defects. , 1999, 84, 151-157.		252
20	5,10 Methylenetetrahydrofolate reductase genetic polymorphism as a risk factor for neural tube defects. , 1996, 63, 610-614.		199
21	Homocysteine and Risk of Premature Coronary Heart Disease. Circulation, 1996, 94, 2154-2158.	1.6	196
22	Seven novel mutations in the methylenetetrahydrofolate reductase gene and genotype/phenotype correlations in severe methylenetetrahydrofolate reductase deficiency. American Journal of Human Genetics, 1995, 56, 1052-9.	2.6	190
23	Correlation of a Common Mutation in the Methylenetetrahydrofolate Reductase Gene With Plasma Homocysteine in Patients With Premature Coronary Artery Disease. Arteriosclerosis, Thrombosis, and Vascular Biology, 1997, 17, 569-573.	1.1	186
24	Polymorphisms in the Methylenetetrahydrofolate Reductase Gene. Molecular Diagnosis and Therapy, 2001, 1, 189-201.	3.3	177
25	Maternal Vitamin Use, Genetic Variation of Infant Methylenetetrahydrofolate Reducatase, and Risk for spina Bifida. American Journal of Epidemiology, 1998, 148, 30-37.	1.6	149
26	GlycineN-methyltransferase deficiency: A novel inborn error causing persistent isolated hypermethioninaemia. Journal of Inherited Metabolic Disease, 2001, 24, 448-464.	1.7	137
27	Gene deletion and restriction fragment length polymorphisms at the human ornithine transcarbamylase locus. Nature, 1985, 313, 815-817.	13.7	129
28	Annotation Molecular genetics of methylenetetrahydrofolate reductase deficiency. Journal of Inherited Metabolic Disease, 1996, 19, 589-594.	1.7	129
29	Genetic predisposition to hyperhomocysteinemia: deficiency of methylenetetrahydrofolate reductase (MTHFR). Thrombosis and Haemostasis, 1997, 78, 523-6.	1.8	120
30	Folate status is the major determinant of fasting total plasma homocysteine levels in maintenance dialysis patients. Atherosclerosis, 1996, 123, 193-202.	0.4	112
31	Infant C677T mutation in MTHFR, maternal periconceptional vitamin use, and cleft lip. , 1998, 80, 196-198.		108
32	Severe and mild mutations in cis for the methylenetetrahydrofolate reductase (MTHFR) gene, and description of five novel mutations in MTHFR. American Journal of Human Genetics, 1996, 59, 1268-75.	2.6	102
33	Primary structure of a human trifunctional enzyme. Isolation of a cDNA encoding methylenetetrahydrofolate dehydrogenase-methenyltetrahydrofolate cyclohydrolase-formyltetrahydrofolate synthetase. Journal of Biological Chemistry, 1988, 263, 15946-50.	1.6	89
34	MTHFR deficiency or reduced intake of folate or choline in pregnant mice results in impaired short-term memory and increased apoptosis in the hippocampus of wild-type offspring. Neuroscience, 2015, 300, 1-9.	1.1	84
35	Cystinuria subtype and the risk of nephrolithiasis11See Editorial by Chesney, p. 279. Kidney International, 1998, 54, 56-61.	2.6	83
36	Association between the methylenetetrahydrofolate reductase 677Câ†'T missense mutation and schizophrenia. Molecular Psychiatry, 2000, 5, 323-326.	4.1	81

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37	Cystic fibrosis mutations in French Canadians: Three CFTR mutations are relatively frequent in a Quebec population with an elevated incidence of cystic fibrosis. American Journal of Medical Genetics Part A, 1992, 42, 360-364.	2.4	80
38	Characterization of six novel mutations in the methylenetetrahydrofolate reductase (MTHFR) gene in patients with homocystinuria. Human Mutation, 2000, 15, 280-287.	1.1	80
39	Oncogenic role of PDK4 in human colon cancer cells. British Journal of Cancer, 2017, 116, 930-936.	2.9	80
40	Dietary and genetic compromise in folate availability reduces acetylcholine, cognitive performance and increases aggression: Critical role of S-adenosyl methionine. Journal of Nutrition, Health and Aging, 2008, 12, 252-261.	1.5	73
41	Molecular genetics of cystinuria: Mutation analysis of SLC3A1 and evidence for another gene in the Type I (silent) phenotype. Kidney International, 1998, 54, 48-55.	2.6	70
42	Incidence and expression of the N1303K mutation of the cystic fibrosis (CFTR) gene. Human Genetics, 1992, 89, 653-658.	1.8	69
43	Genetic Modulation of Homocysteinemia. Seminars in Thrombosis and Hemostasis, 2000, Volume 26, 255-262.	1.5	69
44	Isolation and Expression of a cDNA Encoding the Precursor for a Novel Member (ACADSB) of the Acyl-CoA Dehydrogenase Gene Family. Genomics, 1994, 24, 280-287.	1.3	65
45	Influence of methylenetetrahydrofolate reductase genotype, age, vitamin B-12, and folate status on plasma homocysteine in children. American Journal of Clinical Nutrition, 2000, 72, 1469-1473.	2.2	65
46	Fenofibrate raises plasma homocysteine levels in the fasted and fed states. Atherosclerosis, 2001, 155, 455-462.	0.4	65
47	Renal transport of taurine adapts to perturbed taurine homeostasis.. Proceedings of the National Academy of Sciences of the United States of America, 1982, 79, 2101-2105.	3.3	62
48	Vasculopathic and thrombophilic risk factors for spontaneous preterm birth. International Journal of Epidemiology, 2009, 38, 715-723.	0.9	58
49	Translational control of ornithine aminotransferase. Modulation by initiation factor eIF-4E. Journal of Biological Chemistry, 1991, 266, 16518-23.	1.6	55
50	Taurine transport in renal brush-border-membrane vesicles. Biochemical Journal, 1979, 180, 245-248.	1.7	54
51	Methylenetetrahydrofolate Reductase 677 C â†' T Polymorphism, Plasma Folate, Vitamin B12 Concentrations, and Risk of Preeclampsia among Black African Women from Zimbabwe. Molecular Genetics and Metabolism, 2000, 69, 33-39.	0.5	53
52	Recurrent mutation, gene conversion, or recombination at the human phenylalanine hydroxylase locus: evidence in French-Canadians and a catalog of mutations. American Journal of Human Genetics, 1990, 46, 970-4.	2.6	51
53	Plasma homocysteine concentration in children with chronic renal failure. Pediatric Nephrology, 2001, 16, 805-811.	0.9	49
54	Molecular cloning, expression and physical mapping of the human methionine synthase reductase gene. Gene, 1999, 240, 75-88.	1.0	45

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55	Molecular genetics of cystinuria in French Canadians: Identification of four novel mutations in Type I patients. Kidney International, 1996, 49, 1401-1406.	2.6	41
56	Betaine supplementation improves the atherogenic risk factor profile in a transgenic mouse model of hyperhomocysteinemia. Atherosclerosis, 2007, 195, e100-e107.	0.4	41
57	A novel mouse model for genetic variation in 10-formyltetrahydrofolate synthetase exhibits disturbed purine synthesis with impacts on pregnancy and embryonic development. Human Molecular Genetics, 2013, 22, 3705-3719.	1.4	41
58	Novel PKU mutation on haplotype 2 in French-Canadians. American Journal of Human Genetics, 1989, 45, 905-9.	2.6	41
59	Methylenetetrahydrofolate reductase deficiency and low dietary folate reduce tumorigenesis in Apcmin/+ mice. Gut, 2009, 58, 805-811.	6.1	38
60	The thermolabile variant 677C?T can further reduce activity when expressed in CIS with severe mutations for human methylenetetrahydrofolate reductase. Human Mutation, 2000, 16, 132-138.	1.1	36
61	Assignment of the Gene for Cystinuria (SLC3A1) to Human Chromosome 2p21 by Fluorescence in Situ Hybridization. Genomics, 1994, 24, 413-414.	1.3	33
62	In vitro and in vivo correlations for I65T and M1V mutations at the phenylalanine hydroxylase locus. Human Mutation, 1992, 1, 147-153.	1.1	32
63	The Molecular Basis of Cystinuria: An Update. Nephron Experimental Nephrology, 2000, 8, 123-127.	2.4	30
64	Deficiency of complex III of the mitochondrial respiratory chain in a patient with facioscapulohumeral disease. American Journal of Human Genetics, 1991, 48, 502-10.	2.6	26
65	DNA analysis for ornithine transcarbamylase deficiency. Journal of Inherited Metabolic Disease, 1986, 9, 49-57.	1.7	24
66	Decreased proportion of female newborn infants homozygous for the 677 C?T mutation in methylenetetrahydrofolate reductase. , 1999, 83, 142-143.		23
67	Homocysteine modulates the effect of simvastatin on expression of ApoA-I and NF-ÂB/iNOS. Cardiovascular Research, 2008, 80, 151-158.	1.8	23
68	L206W mutation of the cystic fibrosis gene, relatively frequent in French Canadians, is associated with atypical presentations of cystic fibrosis. American Journal of Medical Genetics Part A, 1995, 57, 437-439.	2.4	22
69	Cystic fibrosis mutations in North American populations of French ancestry: analysis of Quebec French-Canadian and Louisiana Acadian families. American Journal of Human Genetics, 1990, 47, 606-10.	2.6	21
70	Localization of short/branched chain Acyl-CoA dehydrogenase (ACADSB) to human chromosome 10. Genomics, 1995, 25, 743-745.	1.3	19
71	Is the SLC7A10 Gene on Chromosome 19 a Candidate Locus for Cystinuria?. Molecular Genetics and Metabolism, 2001, 73, 333-339.	0.5	17
72	Mefolinate (5â€methyltetrahydrofolate), but not folic acid, decreases mortality in an animal model of severe methylenetetrahydrofolate reductase deficiency. Journal of Inherited Metabolic Disease, 2008, 31, 403-411.	1.7	17

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73	The methylenetetrahydrofolate reductase (Mthfr) gene maps to distal mouse Chromosome 4. Mammalian Genome, 1996, 7, 864-865.	1.0	16
74	Purification and characterization of methylenetetrahydrofolate reductase from human cadaver liver. Biochemical Medicine and Metabolic Biology, 1990, 43, 234-242.	0.7	15
75	Clinical application of DNA analysis in a family with OTC deficiency. American Journal of Medical Genetics Part A, 1986, 25, 513-518.	2.4	14
76	Chromosomal localization of the gene for the human trifunctional enzyme, methylenetetrahydrofolate dehydrogenase-methenyltetrahydrofolate cyclohydrolase-formyltetrahydrofolate synthetase. American Journal of Human Genetics, 1989, 44, 781-6.	2.6	14
77	Mutation profiles of phenylketonuria in Quebec populations: evidence of stratification and novel mutations. American Journal of Human Genetics, 1994, 55, 321-6.	2.6	13
78	Methylenetetrahydrofolate reductase (MTHFR) deficiency enhances resistance against cytomegalovirus infection. Genes and Immunity, 2009, 10, 662-666.	2.2	12
79	Molecular genetic aspects of hyperhomocysteinemia and its relation to folic acid. Clinical and Investigative Medicine, 1996, 19, 171-8.	0.3	12
80	A pseudogene on the X chromosome for the human trifunctional enzyme MTHFD (methylenetetrahydrofolate dehydrogenase-methenyltetrahydrofolate) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 457 Td (cyclohydrolase-for	1.1	11
81	Translational control of ornithine-Î´-aminotransferase (OAT) by estrogen. Molecular and Cellular Endocrinology, 1993, 90, 171-177.	1.6	11
82	Antisense inhibition of methylenetetrahydrofolate reductase reduces survival of methionine-dependent tumour lines. British Journal of Cancer, 2002, 87, 225-230.	2.9	11
83	Studies of urinary cystine precipitation in vitro: ontogeny of cystine nephrolithiasis and identification of meso-2,3-dimercaptosuccinic acid as a potential therapy for cystinuria. Molecular Genetics and Metabolism, 2003, 80, 419-425.	0.5	10
84	Evidence for different clinical subtypes of Type 1 diabetes mellitus: a prospective study. Diabetes Research and Clinical Practice, 1994, 23, 95-102.	1.1	9
85	Five mutations at the PAH locus account for almost 90% of PKU mutations in French-Canadians from eastern Quebec. Human Mutation, 1992, 1, 72-74.	1.1	8
86	Congenital deficiency of a 20-kDa subunit of mitochondrial complex I in fibroblasts. American Journal of Human Genetics, 1991, 48, 1121-6.	2.6	7
87	Homocystinuria (methylenetetrahydrofolate reductase deficiency) and mutation of factor V gene. Journal of Inherited Metabolic Disease, 1998, 21, 690-691.	1.7	4
88	Cerebral vascular complication and hyperhomocysteinemia in a cystinotic uremic child. Pediatric Nephrology, 1999, 13, 73-76.	0.9	3
89	5,10 Methylenetetrahydrofolate reductase genetic polymorphism as a risk factor for neural tube defects. , 1996, 63, 610.		3
90	Deficient synthesis of MTHFD, a trifunctional folate-dependent enzyme, in the CHO Ade?E mutant. Somatic Cell and Molecular Genetics, 1991, 17, 391-398.	0.7	2

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91	Renal function, homocysteine, and other plasma thiol concentrations during the postrenal transplant period. Transplantation Proceedings, 2002, 34, 1159-1160.	0.3	1
92	16th All Ireland social medicine meeting. Irish Journal of Medical Science, 1998, 167, 27-32.	0.8	0
93	The Vasodilation at the Beginning of the Exercise in the Muscular Microcirculation is More Evident in the Repeated Exercise. High Blood Pressure and Cardiovascular Prevention, 2007, 14, 145-196.	1.0	0
94	DNA Analysis for Ornithine Transcarbamylase Deficiency. , 1986, , 49-57.		0