R Rozen

List of Publications by Citations

94 papers	15,987 citations	50 h-index	96 g-index
96 ext. papers	16,871 ext. citations	8.2 avg, IF	5.58 L-index

#	Paper	IF	Citations
94	A candidate genetic risk factor for vascular disease: a common mutation in methylenetetrahydrofolate reductase. *Nature Genetics*, **1995**, 10, 111-3	36.3	4669
93	A second genetic polymorphism in methylenetetrahydrofolate reductase (MTHFR) associated with decreased enzyme activity. *Molecular Genetics and Metabolism*, **1998**, 64, 169-72	3.7	1021
92	Relation between folate status, a common mutation in methylenetetrahydrofolate reductase, and plasma homocysteine concentrations. *Circulation*, **1996**, 93, 7-9	16.7	941
91	Mutated methylenetetrahydrofolate reductase as a risk factor for spina bifida. *Lancet, The*, **1995**, 346, 1070-1	40	703
90	Human methylenetetrahydrofolate reductase: isolation of cDNA, mapping and mutation identification. *Nature Genetics*, **1994**, 7, 195-200	36.3	657
89	Methylenetetrahydrofolate reductase polymorphism, dietary interactions, and risk of colorectal cancer. *Cancer Research*, **1997**, 57, 1098-102	10.1	470
88	Mice deficient in methylenetetrahydrofolate reductase exhibit hyperhomocysteinemia and decreased methylation capacity, with neuropathology and aortic lipid deposition. *Human Molecular Genetics*, **2001**, 10, 433-43	5.6	458
87	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	11	421
86	A common variant in methionine synthase reductase combined with low cobalamin (vitamin B12) increases risk for spina bifida. *Molecular Genetics and Metabolism*, **1999**, 67, 317-23	3.7	350
85	The structure and properties of methylenetetrahydrofolate reductase from Escherichia coli suggest how folate ameliorates human hyperhomocysteinemia. *Nature Structural Biology*, **1999**, 6, 359-65		315
84	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, 14853-8	11.5	308
83	The 1298A-->C polymorphism in methylenetetrahydrofolate reductase (MTHFR): in vitro expression and association with homocysteine. *Atherosclerosis*, **2001**, 156, 409-15	3.1	304
82	Methylenetetrahydrofolate reductase polymorphism, plasma folate, homocysteine, and risk of myocardial infarction in US physicians. *Circulation*, **1996**, 94, 2410-6	16.7	304
81	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, 1867-74	5.6	286
80	Polymorphisms in genes involved in folate metabolism as maternal risk factors for Down syndrome. *American Journal of Human Genetics*, **2000**, 67, 623-30	11	285
79	Socio-economic disparities in preterm birth: causal pathways and mechanisms. *Paediatric and Perinatal Epidemiology*, **2001**, 15 Suppl 2, 104-23	2.7	251
78	A rapid procedure for extracting genomic DNA from leukocytes. *Nucleic Acids Research*, **1991**, 19, 408	20.1	250

77	Gene structure of human and mouse methylenetetrahydrofolate reductase (MTHFR). *Mammalian Genome*, **1998**, 9, 652-6	3.2	245
76	Genetic polymorphisms in methylenetetrahydrofolate reductase and methionine synthase, folate levels in red blood cells, and risk of neural tube defects. *American Journal of Medical Genetics Part A* , **1999**, 84, 151-7		210
75	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	11	174
74	5,10 Methylenetetrahydrofolate reductase genetic polymorphism as a risk factor for neural tube defects. *American Journal of Medical Genetics Part A*, **1996**, 63, 610-4		164
73	Polymorphisms in the methylenetetrahydrofolate reductase gene: clinical consequences. *Molecular Diagnosis and Therapy*, **2001**, 1, 189-201		151
72	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, 569-73	9.4	150
71	Homocysteine and risk of premature coronary heart disease. Evidence for a common gene mutation. *Circulation*, **1996**, 94, 2154-8	16.7	144
70	Maternal vitamin use, genetic variation of infant methylenetetrahydrofolate reductase, and risk for spina bifida. *American Journal of Epidemiology*, **1998**, 148, 30-7	3.8	137
69	Gene deletion and restriction fragment length polymorphisms at the human ornithine transcarbamylase locus. *Nature*, **1985**, 313, 815-7	50.4	124
68	Glycine N-methyltransferase deficiency: a novel inborn error causing persistent isolated hypermethioninaemia. *Journal of Inherited Metabolic Disease*, **2001**, 24, 448-64	5.4	116
67	Molecular genetics of methylenetetrahydrofolate reductase deficiency. *Journal of Inherited Metabolic Disease*, **1996**, 19, 589-94	5.4	105
66	Folate status is the major determinant of fasting total plasma homocysteine levels in maintenance dialysis patients. *Atherosclerosis*, **1996**, 123, 193-202	3.1	99
65	Genetic predisposition to hyperhomocysteinemia: deficiency of methylenetetrahydrofolate reductase (MTHFR). *Thrombosis and Haemostasis*, **1997**, 78, 523-6	7	98
64	Infant C677T mutation in MTHFR, maternal periconceptional vitamin use, and cleft lip. *American Journal of Medical Genetics Part A*, **1998**, 80, 196-8		96
63	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	11	92
62	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	5.4	84
61	Association between the methylenetetrahydrofolate reductase 677C-->T missense mutation and schizophrenia. *Molecular Psychiatry*, **2000**, 5, 323-6	15.1	76
60	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, 360-4		74

59	Characterization of six novel mutations in the methylenetetrahydrofolate reductase (MTHFR) gene in patients with homocystinuria. *Human Mutation*, **2000**, 15, 280-7	4.7	70
58	Cystinuria subtype and the risk of nephrolithiasis. *Kidney International*, **1998**, 54, 56-61	9.9	66
57	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	3.9	63
56	Molecular genetics of cystinuria: mutation analysis of SLC3A1 and evidence for another gene in type I (silent) phenotype. *Kidney International*, **1998**, 54, 48-55	9.9	63
55	Incidence and expression of the N1303K mutation of the cystic fibrosis (CFTR) gene. *Human Genetics*, **1992**, 89, 653-8	6.3	62
54	Genetic modulation of homocysteinemia. *Seminars in Thrombosis and Hemostasis*, **2000**, 26, 255-61	5.3	61
53	Fenofibrate raises plasma homocysteine levels in the fasted and fed states. *Atherosclerosis*, **2001**, 155, 455-62	3.1	60
52	Influence of methylenetetrahydrofolate reductase genotype, age, vitamin B-12, and folate status on plasma homocysteine in children. *American Journal of Clinical Nutrition*, **2000**, 72, 1469-73	7	59
51	Renal transport of taurine adapts to perturbed taurine homeostasis. *Proceedings of the National Academy of Sciences of the United States of America*, **1982**, 79, 2101-5	11.5	57
50	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, 252-61	5.2	56
49	Oncogenic role of PDK4 in human colon cancer cells. *British Journal of Cancer*, **2017**, 116, 930-936	8.7	55
48	Isolation and expression of a cDNA encoding the precursor for a novel member (ACADSB) of the acyl-CoA dehydrogenase gene family. *Genomics*, **1994**, 24, 280-7	4.3	54
47	Translational control of ornithine aminotransferase. Modulation by initiation factor eIF-4E. *Journal of Biological Chemistry*, **1991**, 266, 16518-23	5.4	51
46	Vasculopathic and thrombophilic risk factors for spontaneous preterm birth. *International Journal of Epidemiology*, **2009**, 38, 715-23	7.8	50
45	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	11	50
44	Taurine transport in renal brush-border-membrane vesicles. *Biochemical Journal*, **1979**, 180, 245-8	3.8	49
43	Methylenetetrahydrofolate reductase 677 C --> T polymorphism, plasma folate, vitamin B(12) concentrations, and risk of preeclampsia among black African women from Zimbabwe. *Molecular Genetics and Metabolism*, **2000**, 69, 33-9	3.7	47
42	Plasma homocysteine concentration in children with chronic renal failure. *Pediatric Nephrology*, **2001**, 16, 805-11	3.2	43

41	Molecular cloning, expression and physical mapping of the human methionine synthase reductase gene. *Gene*, **1999**, 240, 75-88	3.8	41
40	Novel PKU mutation on haplotype 2 in French-Canadians. *American Journal of Human Genetics*, **1989**, 45, 905-9	11	41
39	Betaine supplementation improves the atherogenic risk factor profile in a transgenic mouse model of hyperhomocysteinemia. *Atherosclerosis*, **2007**, 195, e100-7	3.1	36
38	Molecular genetics of cystinuria in French Canadians: identification of four novel mutations in type I patients. *Kidney International*, **1996**, 49, 1401-6	9.9	36
37	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, 3705-19	5.6	31
36	The thermolabile variant 677C-->T can further reduce activity when expressed in cis with severe mutations for human methylenetetrahydrofolate reductase. *Human Mutation*, **2000**, 16, 132-8	4.7	31
35	Assignment of the gene for cystinuria (SLC3A1) to human chromosome 2p21 by fluorescence in situ hybridization. *Genomics*, **1994**, 24, 413-4	4.3	31
34	Methylenetetrahydrofolate reductase deficiency and low dietary folate reduce tumorigenesis in Apc min/+ mice. *Gut*, **2009**, 58, 805-11	19.2	29
33	In vitro and in vivo correlations for I65T and M1V mutations at the phenylalanine hydroxylase locus. *Human Mutation*, **1992**, 1, 147-53	4.7	29
32	The molecular basis of cystinuria: an update. *Nephron Experimental Nephrology*, **2000**, 8, 123-7		26
31	DNA analysis for ornithine transcarbamylase deficiency. *Journal of Inherited Metabolic Disease*, **1986**, 9 Suppl 1, 49-57	5.4	24
30	Deficiency of complex III of the mitochondrial respiratory chain in a patient with facioscapulohumeral disease. *American Journal of Human Genetics*, **1991**, 48, 502-10	11	23
29	Homocysteine modulates the effect of simvastatin on expression of ApoA-I and NF-kappaB/iNOS. *Cardiovascular Research*, **2008**, 80, 151-8	9.9	19
28	Decreased proportion of female newborn infants homozygous for the 677 C-->T mutation in methylenetetrahydrofolate reductase. *American Journal of Medical Genetics Part A*, **1999**, 83, 142-3		19
27	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, 437-9		17
26	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	11	16
25	Is the SLC7A10 gene on chromosome 19 a candidate locus for cystinuria?. *Molecular Genetics and Metabolism*, **2001**, 73, 333-9	3.7	14
24	Clinical application of DNA analysis in a family with OTC deficiency. *American Journal of Medical Genetics Part A*, **1986**, 25, 513-8		14

23	The methylenetetrahydrofolate reductase (Mthfr) gene maps to distal mouse chromosome 4. *Mammalian Genome*, **1996**, 7, 864-5	3.2	13
22	Methylenetetrahydrofolate reductase (MTHFR) deficiency enhances resistance against cytomegalovirus infection. *Genes and Immunity*, **2009**, 10, 662-6	4.4	12
21	Mefolinate (5-methyltetrahydrofolate), but not folic acid, decreases mortality in an animal model of severe methylenetetrahydrofolate reductase deficiency. *Journal of Inherited Metabolic Disease*, **2008**, 31, 403-11	5.4	12
20	Purification and characterization of methylenetetrahydrofolate reductase from human cadaver liver. *Biochemical Medicine and Metabolic Biology*, **1990**, 43, 234-42		12
19	Mutation profiles of phenylketonuria in Quebec populations: evidence of stratification and novel mutations. *American Journal of Human Genetics*, **1994**, 55, 321-6	11	12
18	Molecular genetic aspects of hyperhomocysteinemia and its relation to folic acid. *Clinical and Investigative Medicine*, **1996**, 19, 171-8	0.9	12
17	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	11	11
16	Translational control of ornithine-delta-aminotransferase (OAT) by estrogen. *Molecular and Cellular Endocrinology*, **1993**, 90, 171-7	4.4	10
15	A pseudogene on the X chromosome for the human trifunctional enzyme MTHFD (methylenetetrahydrofolate dehydrogenase-methenyltetrahydrofolate cyclohydrolase-formyltetrahydrofolate synthetase). *Genomics*, **1991**, 10, 1073-4	4.3	10
14	Antisense inhibition of methylenetetrahydrofolate reductase reduces survival of methionine-dependent tumour lines. *British Journal of Cancer*, **2002**, 87, 225-30	8.7	9
13	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, 419-25	3.7	8
12	Localization of short/branched chain acyl-CoA dehydrogenase (ACADSB) to human chromosome 10. *Genomics*, **1995**, 25, 743-5	4.3	8
11	Evidence for different clinical subtypes of type 1 diabetes mellitus: a prospective study. *Diabetes Research and Clinical Practice*, **1994**, 23, 95-102	7.4	8
10	Five mutations at the PAH locus account for almost 90% of PKU mutations in French-Canadians from eastern Quebec. *Human Mutation*, **1992**, 1, 72-4	4.7	6
9	Congenital deficiency of a 20-kDa subunit of mitochondrial complex I in fibroblasts. *American Journal of Human Genetics*, **1991**, 48, 1121-6	11	6
8	Homocystinuria (methylenetetrahydrofolate reductase deficiency) and mutation of factor V gene. *Journal of Inherited Metabolic Disease*, **1998**, 21, 690-1	5.4	4
7	5,10 Methylenetetrahydrofolate reductase genetic polymorphism as a risk factor for neural tube defects **1996**, 63, 610		3
6	Deficient synthesis of MTHFD, a trifunctional folate-dependent enzyme, in the CHO Ade E mutant. *Somatic Cell and Molecular Genetics*, **1991**, 17, 391-8		2

5	Cerebral vascular complication and hyperhomocysteinemia in a cystinotic uremic child. *Pediatric Nephrology*, **1999**, 13, 73-6	3.2	1
4	Renal function, homocysteine, and other plasma thiol concentrations during the postrenal transplant period. *Transplantation Proceedings*, **2002**, 34, 1159-60	1.1	0
3	16th All Ireland social medicine meeting. *Irish Journal of Medical Science*, **1998**, 167, 27-32	1.9	
2	10.14 Peroxisome Proliferator-Activated Receptors Activator, Rosiglitazone, Improves Vascular Structure and Reduces Lipid Deposition in Atherosclerotic Mthfr Knock-Out Mice. *High Blood Pressure and Cardiovascular Prevention*, **2007**, 14, 145-196	2.9	
1	DNA Analysis for Ornithine Transcarbamylase Deficiency **1986**, 49-57		