# R Rozen

## List of Publications by Year in Descending Order



| | | | |
|---|---|---|---|
| 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 | Oncogenic role of PDK4 in human colon cancer cells. *British Journal of Cancer*, **2017**, 116, 930-936 | 8.7 | 55 |
| 93 | 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 |
| 92 | 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 |
| 91 | Vasculopathic and thrombophilic risk factors for spontaneous preterm birth. *International Journal of Epidemiology*, **2009**, 38, 715-23 | 7.8 | 50 |
| 90 | Methylenetetrahydrofolate reductase deficiency and low dietary folate reduce tumorigenesis in Apc min/+ mice. *Gut*, **2009**, 58, 805-11 | 19.2 | 29 |
| 89 | Methylenetetrahydrofolate reductase (MTHFR) deficiency enhances resistance against cytomegalovirus infection. *Genes and Immunity*, **2009**, 10, 662-6 | 4.4 | 12 |
| 88 | 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 |
| 87 | 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 |
| 86 | Homocysteine modulates the effect of simvastatin on expression of ApoA-I and NF-kappaB/iNOS. *Cardiovascular Research*, **2008**, 80, 151-8 | 9.9 | 19 |
| 85 | Betaine supplementation improves the atherogenic risk factor profile in a transgenic mouse model of hyperhomocysteinemia. *Atherosclerosis*, **2007**, 195, e100-7 | 3.1 | 36 |
| 84 | 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 | |
| 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, 419-25 | 3.7 | 8 |
| 82 | Antisense inhibition of methylenetetrahydrofolate reductase reduces survival of methionine-dependent tumour lines. *British Journal of Cancer*, **2002**, 87, 225-30 | 8.7 | 9 |
| 81 | Renal function, homocysteine, and other plasma thiol concentrations during the postrenal transplant period. *Transplantation Proceedings*, **2002**, 34, 1159-60 | 1.1 | 0 |
| 80 | Plasma homocysteine concentration in children with chronic renal failure. *Pediatric Nephrology*, **2001**, 16, 805-11 | 3.2 | 43 |
| 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 | Glycine N-methyltransferase deficiency: a novel inborn error causing persistent isolated hypermethioninaemia. *Journal of Inherited Metabolic Disease*, **2001**, 24, 448-64 | 5.4 | 116 |

| | | | |
|---|---|---|---|
| 77 | 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 |
| 76 | Fenofibrate raises plasma homocysteine levels in the fasted and fed states. *Atherosclerosis*, **2001**, 155, 455-62 | 3.1 | 60 |
| 75 | The 1298A-->C polymorphism in methylenetetrahydrofolate reductase (MTHFR): in vitro expression and association with homocysteine. *Atherosclerosis*, **2001**, 156, 409-15 | 3.1 | 304 |
| 74 | Is the SLC7A10 gene on chromosome 19 a candidate locus for cystinuria?. *Molecular Genetics and Metabolism*, **2001**, 73, 333-9 | 3.7 | 14 |
| 73 | Polymorphisms in the methylenetetrahydrofolate reductase gene: clinical consequences. *Molecular Diagnosis and Therapy*, **2001**, 1, 189-201 | | 151 |
| 72 | 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 |
| 71 | 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 |
| 70 | Characterization of six novel mutations in the methylenetetrahydrofolate reductase (MTHFR) gene in patients with homocystinuria. *Human Mutation*, **2000**, 15, 280-7 | 4.7 | 70 |
| 69 | 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 |
| 68 | Association between the methylenetetrahydrofolate reductase 677C-->T missense mutation and schizophrenia. *Molecular Psychiatry*, **2000**, 5, 323-6 | 15.1 | 76 |
| 67 | The molecular basis of cystinuria: an update. *Nephron Experimental Nephrology*, **2000**, 8, 123-7 | | 26 |
| 66 | Genetic modulation of homocysteinemia. *Seminars in Thrombosis and Hemostasis*, **2000**, 26, 255-61 | 5.3 | 61 |
| 65 | 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 |
| 64 | 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 |
| 63 | The structure and properties of methylenetetrahydrofolate reductase from Escherichia coli suggest how folate ameliorates human hyperhomocysteinemia. *Nature Structural Biology*, **1999**, 6, 359-65 | | 315 |
| 62 | Cerebral vascular complication and hyperhomocysteinemia in a cystinotic uremic child. *Pediatric Nephrology*, **1999**, 13, 73-6 | 3.2 | 1 |
| 61 | 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 |
| 60 | 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 |

| | | | |
|---|---|---|---|
| 59 | Molecular cloning, expression and physical mapping of the human methionine synthase reductase gene. *Gene*, **1999**, 240, 75-88 | 3.8 | 41 |
| 58 | 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 |
| 57 | Homocystinuria (methylenetetrahydrofolate reductase deficiency) and mutation of factor V gene. *Journal of Inherited Metabolic Disease*, **1998**, 21, 690-1 | 5.4 | 4 |
| 56 | Infant C677T mutation in MTHFR, maternal periconceptional vitamin use, and cleft lip. *American Journal of Medical Genetics Part A*, **1998**, 80, 196-8 | | 96 |
| 55 | 16th All Ireland social medicine meeting. *Irish Journal of Medical Science*, **1998**, 167, 27-32 | 1.9 | |
| 54 | Gene structure of human and mouse methylenetetrahydrofolate reductase (MTHFR). *Mammalian Genome*, **1998**, 9, 652-6 | 3.2 | 245 |
| 53 | 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 |
| 52 | Cystinuria subtype and the risk of nephrolithiasis. *Kidney International*, **1998**, 54, 56-61 | 9.9 | 66 |
| 51 | A second genetic polymorphism in methylenetetrahydrofolate reductase (MTHFR) associated with decreased enzyme activity. *Molecular Genetics and Metabolism*, **1998**, 64, 169-72 | 3.7 | 1021 |
| 50 | 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 |
| 49 | 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 |
| 48 | Methylenetetrahydrofolate reductase polymorphism, dietary interactions, and risk of colorectal cancer. *Cancer Research*, **1997**, 57, 1098-102 | 10.1 | 470 |
| 47 | Genetic predisposition to hyperhomocysteinemia: deficiency of methylenetetrahydrofolate reductase (MTHFR). *Thrombosis and Haemostasis*, **1997**, 78, 523-6 | 7 | 98 |
| 46 | Folate status is the major determinant of fasting total plasma homocysteine levels in maintenance dialysis patients. *Atherosclerosis*, **1996**, 123, 193-202 | 3.1 | 99 |
| 45 | 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 |
| 44 | Molecular genetics of methylenetetrahydrofolate reductase deficiency. *Journal of Inherited Metabolic Disease*, **1996**, 19, 589-94 | 5.4 | 105 |
| 43 | The methylenetetrahydrofolate reductase (Mthfr) gene maps to distal mouse chromosome 4. *Mammalian Genome*, **1996**, 7, 864-5 | 3.2 | 13 |
| 42 | 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 |

| | | | |
|---|---|---|---|
| 41 | 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 |
| 40 | Relation between folate status, a common mutation in methylenetetrahydrofolate reductase, and plasma homocysteine concentrations. *Circulation*, **1996**, 93, 7-9 | 16.7 | 941 |
| 39 | Methylenetetrahydrofolate reductase polymorphism, plasma folate, homocysteine, and risk of myocardial infarction in US physicians. *Circulation*, **1996**, 94, 2410-6 | 16.7 | 304 |
| 38 | Homocysteine and risk of premature coronary heart disease. Evidence for a common gene mutation. *Circulation*, **1996**, 94, 2154-8 | 16.7 | 144 |
| 37 | 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 |
| 36 | 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 |
| 35 | 5,10 Methylenetetrahydrofolate reductase genetic polymorphism as a risk factor for neural tube defects **1996**, 63, 610 | | 3 |
| 34 | Molecular genetic aspects of hyperhomocysteinemia and its relation to folic acid. *Clinical and Investigative Medicine*, **1996**, 19, 171-8 | 0.9 | 12 |
| 33 | A candidate genetic risk factor for vascular disease: a common mutation in methylenetetrahydrofolate reductase. *Nature Genetics*, **1995**, 10, 111-3 | 36.3 | 4669 |
| 32 | Localization of short/branched chain acyl-CoA dehydrogenase (ACADSB) to human chromosome 10. *Genomics*, **1995**, 25, 743-5 | 4.3 | 8 |
| 31 | Mutated methylenetetrahydrofolate reductase as a risk factor for spina bifida. *Lancet, The*, **1995**, 346, 1070-1 | 40 | 703 |
| 30 | 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 |
| 29 | 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 |
| 28 | Human methylenetetrahydrofolate reductase: isolation of cDNA, mapping and mutation identification. *Nature Genetics*, **1994**, 7, 195-200 | 36.3 | 657 |
| 27 | 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 |
| 26 | 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 |
| 25 | Assignment of the gene for cystinuria (SLC3A1) to human chromosome 2p21 by fluorescence in situ hybridization. *Genomics*, **1994**, 24, 413-4 | 4.3 | 31 |
| 24 | Mutation profiles of phenylketonuria in Quebec populations: evidence of stratification and novel mutations. *American Journal of Human Genetics*, **1994**, 55, 321-6 | 11 | 12 |

| | | | |
|---|---|---|---|
| 23 | Translational control of ornithine-delta-aminotransferase (OAT) by estrogen. *Molecular and Cellular Endocrinology*, **1993**, 90, 171-7 | 4.4 | 10 |
| 22 | Incidence and expression of the N1303K mutation of the cystic fibrosis (CFTR) gene. *Human Genetics*, **1992**, 89, 653-8 | 6.3 | 62 |
| 21 | 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 |
| 20 | 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 |
| 19 | 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 |
| 18 | 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 |
| 17 | A rapid procedure for extracting genomic DNA from leukocytes. *Nucleic Acids Research*, **1991**, 19, 408 | 20.1 | 250 |
| 16 | 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 |
| 15 | Translational control of ornithine aminotransferase. Modulation by initiation factor eIF-4E. *Journal of Biological Chemistry*, **1991**, 266, 16518-23 | 5.4 | 51 |
| 14 | 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 |
| 13 | Congenital deficiency of a 20-kDa subunit of mitochondrial complex I in fibroblasts. *American Journal of Human Genetics*, **1991**, 48, 1121-6 | 11 | 6 |
| 12 | Purification and characterization of methylenetetrahydrofolate reductase from human cadaver liver. *Biochemical Medicine and Metabolic Biology*, **1990**, 43, 234-42 | | 12 |
| 11 | 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 |
| 10 | 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 |
| 9 | Novel PKU mutation on haplotype 2 in French-Canadians. *American Journal of Human Genetics*, **1989** , 45, 905-9 | 11 | 41 |
| 8 | 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 |
| 7 | 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 |
| 6 | Clinical application of DNA analysis in a family with OTC deficiency. *American Journal of Medical Genetics Part A*, **1986**, 25, 513-8 | | 14 |

| | | | |
|---|---|---|---|
| 5 | DNA analysis for ornithine transcarbamylase deficiency. *Journal of Inherited Metabolic Disease*, **1986** , 9 Suppl 1, 49-57 | 5.4 | 24 |
| 4 | DNA Analysis for Ornithine Transcarbamylase Deficiency **1986**, 49-57 | | |
| 3 | Gene deletion and restriction fragment length polymorphisms at the human ornithine transcarbamylase locus. *Nature*, **1985**, 313, 815-7 | 50.4 | 124 |
| 2 | 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 |
| 1 | Taurine transport in renal brush-border-membrane vesicles. *Biochemical Journal*, **1979**, 180, 245-8 | 3.8 | 49 |