AndrÃ© B P Van Kuilenburg

List of Publications by Year
in descending order

165 papers	8,085 citations	76196 40 h-index	53109 85 g-index
170 all docs	170 docs citations	170 times ranked	9787 citing authors

#	Article	IF	Citations
1	Children with atopic dermatitis show increased activity of Î²â€glucocerebrosidase and stratum corneum levels of glucosylcholesterol that are strongly related to the local cytokine milieu. British Journal of Dermatology, 2022, 186, 988-996.	1.4	9
2	Urinary 3-Methoxytyramine Is a Biomarker for MYC Activity in Patients With Neuroblastoma. JCO Precision Oncology, 2022, 6, e2000447.	1.5	4
3	Î²-Ureidopropionase deficiency due to novel and rare UPB1 mutations affecting pre-mRNA splicing and protein structural integrity and catalytic activity. Molecular Genetics and Metabolism, 2022, 136, 177-185.	0.5	3
4	Purine and Pyrimidine Metabolism. , 2021, , 183-234.		3
5	Preemptive screening of DPYD as part of clinical practice: high prevalence of a novel exon 4 deletion in the Finnish population. Cancer Chemotherapy and Pharmacology, 2021, 87, 657-663.	1.1	6
6	Co-therapy with S-adenosylmethionine and nicotinamide riboside improves t-cell survival and function in Arts Syndrome (PRPS1 deficiency). Molecular Genetics and Metabolism Reports, 2021, 26, 100709.	0.4	6
7	Thrombocytopenia after meta-iodobenzylguanidine (MIBG) therapy in neuroblastoma patients may be caused by selective MIBG uptake via the serotonin transporter located on megakaryocytes. EJNMMI Research, 2021, 11, 81.	1.1	1
8	Metachromatic leukodystrophy genotypes in The Netherlands reveal novel pathogenic ARSA variants in non-Caucasian patients. Neurogenetics, 2020, 21, 289-299.	0.7	9
9	Predicting the Development of Anti-Drug Antibodies against Recombinant alpha-Galactosidase A in Male Patients with Classical Fabry Disease. International Journal of Molecular Sciences, 2020, 21, 5784.	1.8	9
10	Atypical presentation of Arts syndrome due to a novel hemizygous loss-of-function variant in the PRPS1 gene. Molecular Genetics and Metabolism Reports, 2020, 25, 100677.	0.4	6
11	Developments in the treatment of Fabry disease. Journal of Inherited Metabolic Disease, 2020, 43, 908-921.	1.7	83
12	Biochemical and imaging parameters in acid sphingomyelinase deficiency: Potential utility as biomarkers. Molecular Genetics and Metabolism, 2020, 130, 16-26.	0.5	15
13	The Effects of Mercaptopurine on Pulmonary Vascular Resistance and BMPR2 Expression in Pulmonary Arterial Hypertension. American Journal of Respiratory and Critical Care Medicine, 2020, 202, 296-299.	2.5	10
14	Selective serotonin reuptake inhibitors (SSRIs) prevent meta-iodobenzylguanidine (MIBG) uptake in platelets without affecting neuroblastoma tumor uptake. EJNMMI Research, 2020, 10, 78.	1.1	5
15	DNA damage and transcription stress cause ATP-mediated redesign of metabolism and potentiation of anti-oxidant buffering. Nature Communications, 2019, 10, 4887.	5.8	43
16	Deficiency of perforin and hCNT1, a novel inborn error of pyrimidine metabolism, associated with a rapidly developing lethal phenotype due to multi-organ failure. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2019, 1865, 1182-1191.	1.8	8
17	Glutaminase Deficiency Caused by Short Tandem Repeat Expansion in GLS. New England Journal of Medicine, 2019, 380, 1433-1441.	13.9	71
18	Liposome-targeted recombinant human acid sphingomyelinase: Production, formulation, and in vitro evaluation. European Journal of Pharmaceutics and Biopharmaceutics, 2019, 137, 185-195.	2.0	12

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19	Plasma free metanephrines for diagnosis of neuroblastoma patients. Clinical Biochemistry, 2019, 66, 57-62.	0.8	14
20	Acute Aerobic Exercise Leads to Increased Plasma Levels of R- and S-Î²-Aminoisobutyric Acid in Humans. Frontiers in Physiology, 2019, 10, 1240.	1.3	51
21	A cost analysis of upfront DPYD genotypeâ€“guided dose individualisation in fluoropyrimidine-based anticancer therapy. European Journal of Cancer, 2019, 107, 60-67.	1.3	65
22	Translational Metabolism: A multidisciplinary approach towards precision diagnosis of inborn errors of metabolism in the omics era. Journal of Inherited Metabolic Disease, 2019, 42, 197-208.	1.7	20
23	Genome sequencing reveals a novel genetic mechanism underlying dihydropyrimidine dehydrogenase deficiency: A novel missense variant c.1700G>A and a large intragenic inversion in DPYD spanning intron 8 to intron 12. Human Mutation, 2018, 39, 947-953.	1.1	6
24	Agalsidase alfa versus agalsidase beta for the treatment of Fabry disease: an international cohort study. Journal of Medical Genetics, 2018, 55, 351-358.	1.5	72
25	Rapid screening for lipid storage disorders using biochemical markers. Expert center data and review of the literature. Molecular Genetics and Metabolism, 2018, 123, 76-84.	0.5	33
26	Capecitabineâ€based treatment of a patient with a novel DPYD genotype and complete dihydropyrimidine dehydrogenase deficiency. International Journal of Cancer, 2018, 142, 424-430.	2.3	15
27	The pathophysiology of human obstructive cholestasis is mimicked in cholestatic Gold Syrian hamsters. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 942-951.	1.8	11
28	Diagnostic and Therapeutic Strategies for Fluoropyrimidine Treatment of Patients Carrying Multiple DPYD Variants. Genes, 2018, 9, 585.	1.0	10
29	Dihydropyrimidine Dehydrogenase Deficiency: Homozygosity for an Extremely Rare Variant in DPYD due to Uniparental Isodisomy of Chromosome 1. JIMD Reports, 2018, 45, 65-69.	0.7	3
30	DPYD genotype-guided dose individualisation of fluoropyrimidine therapy in patients with cancer: a prospective safety analysis. Lancet Oncology, The, 2018, 19, 1459-1467.	5.1	238
31	Preliminary Evidence for Enhanced Thymine Absorption: A Putative New Phenotype Associated With Fluoropyrimidine Toxicity in Cancer Patients. Therapeutic Drug Monitoring, 2018, 40, 495-502.	1.0	4
32	Catecholamines profiles at diagnosis: Increased diagnostic sensitivity and correlation with biological and clinical features in neuroblastoma patients. European Journal of Cancer, 2017, 72, 235-243.	1.3	57
33	Favourable effect of early versus late start of enzyme replacement therapy on plasma globotriaosylsphingosine levels in men with classical Fabry disease. Molecular Genetics and Metabolism, 2017, 121, 157-161.	0.5	64
34	Long-Term Dose-Dependent Agalsidase Effects on Kidney Histology in Fabry Disease. Clinical Journal of the American Society of Nephrology: CJASN, 2017, 12, 1470-1479.	2.2	42
35	Yunis-VarÃ³n syndrome caused by biallelic VAC14 mutations. European Journal of Human Genetics, 2017, 25, 1049-1054.	1.4	21
36	Hypothermic perfusion with retrograde outflow during right hepatectomy is safe and feasible. Surgery, 2017, 162, 48-58.	1.0	5

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37	Severe fluoropyrimidine toxicity due to novel and rare DPYD missense mutations, deletion and genomic amplification affecting DPD activity and mRNA splicing. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2017, 1863, 721-730.	1.8	32
38	De Novo Mutations in SLC25A24 Cause a Disorder Characterized by Early Aging, Bone Dysplasia, Characteristic Face, and Early Demise. American Journal of Human Genetics, 2017, 101, 844-855.	2.6	51
39	Novel PRPS1 gain-of-function mutation in a patient with congenital hyperuricemia and facial anomalies. , 2017, 173, 2736-2742.		11
40	Dihydropyrimidinase deficiency in four East Asian patients due to novel and rare DPYS mutations affecting protein structural integrity and catalytic activity. Molecular Genetics and Metabolism, 2017, 122, 216-222.	0.5	16
41	Multi-OMIC profiling of survival and metabolic signaling networks in cells subjected to photodynamic therapy. Cellular and Molecular Life Sciences, 2017, 74, 1133-1151.	2.4	34
42	Genotypes Affecting the Pharmacokinetics of Anticancer Drugs. Clinical Pharmacokinetics, 2017, 56, 317-337.	1.6	50
43	Treatment Algorithm for Homozygous or Compound Heterozygous DPYD Variant Allele Carriers With Low-Dose Capecitabine. JCO Precision Oncology, 2017, 1, 1-10.	1.5	8
44	New advances in DPYD genotype and risk of severe toxicity under capecitabine. PLoS ONE, 2017, 12, e0175998.	1.1	82
45	Retrospective study of long-term outcomes of enzyme replacement therapy in Fabry disease: Analysis of prognostic factors. PLoS ONE, 2017, 12, e0182379.	1.1	83
46	Altered Pre-mRNA Splicing Caused by a Novel Intronic Mutation c.1443+5G>A in the Dihydropyrimidinase (DPYS) Gene. International Journal of Molecular Sciences, 2016, 17, 86.	1.8	10
47	The Cytidine Analog Fluorocyclopentenylcytosine (RX-3117) Is Activated by Uridine-Cytidine Kinase 2. PLoS ONE, 2016, 11, e0162901.	1.1	20
48	Evaluation of an oral uracil loading test to identify DPDâ€deficient patients using a limited sampling strategy. British Journal of Clinical Pharmacology, 2016, 81, 553-561.	1.1	26
49	Dopamine induces lipid accumulation, NADPH oxidase-related oxidative stress, and a proinflammatory status of the plasma membrane in H9c2 cells. American Journal of Physiology - Heart and Circulatory Physiology, 2016, 311, H1097-H1107.	1.5	11
50	Purification, activity, and expression levels of two uridine-cytidine kinase isoforms in neuroblastoma cell lines. Nucleosides, Nucleotides and Nucleic Acids, 2016, 35, 613-618.	0.4	6
51	Creatine kinase inhibition lowers systemic arterial blood pressure in spontaneously hypertensive rats. Journal of Hypertension, 2016, 34, 2418-2426.	0.3	19
52	Patients homozygous for DPYD c.1129-5923C>G/haplotype B3 have partial DPD deficiency and require a dose reduction when treated with fluoropyrimidines. Cancer Chemotherapy and Pharmacology, 2016, 78, 875-880.	1.1	17
53	Hyperferritinemia and iron metabolism in Gaucher disease: Potential pathophysiological implications. Blood Reviews, 2016, 30, 431-437.	2.8	22
54	The pivotal role of uridine-cytidine kinases in pyrimidine metabolism and activation of cytotoxic nucleoside analogues in neuroblastoma. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2016, 1862, 1504-1512.	1.8	21

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55	Phenotypic and clinical implications of variants in the dihydropyrimidine dehydrogenase gene. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2016, 1862, 754-762.	1.8	41
56	Towards a test to predict 5-fluorouracil toxicity: Pharmacokinetic data for thymine and two sequential metabolites following oral thymine administration to healthy adult males. European Journal of Pharmaceutical Sciences, 2016, 81, 36-41.	1.9	20
57	Novel Genetic Mutations in the First Swedish Patient with Purine Nucleoside Phosphorylase Deficiency and Clinical Outcome After Hematopoietic Stem Cell Transplantation with HLA-Matched Unrelated Donor. JIMD Reports, 2015, 24, 83-89.	0.7	12
58	Prenatal growth restriction, retinal dystrophy, diabetes insipidus and white matter disease: expanding the spectrum of PRPS1-related disorders. European Journal of Human Genetics, 2015, 23, 310-316.	1.4	30
59	Influence of metastatic disease on the usefulness of uracil pharmacokinetics as a screening tool for DPD activity in colorectal cancer patients. Cancer Chemotherapy and Pharmacology, 2015, 76, 47-52.	1.1	7
60	Clinical relevance of DPYD variants c.1679T>G, c.1236G>A/HapB3, and c.1601G>A as predictors of severe fluoropyrimidine-associated toxicity: a systematic review and meta-analysis of individual patient data. Lancet Oncology, The, 2015, 16, 1639-1650.	5.1	277
61	Report of Two Never Treated Adult Sisters with Aromatic l-Amino Acid Decarboxylase Deficiency: A Portrait of the Natural History of the Disease or an Expanding Phenotype?. JIMD Reports, 2014, 15, 39-45.	0.7	29
62	A Korean Case of Î²-Ureidopropionase Deficiency Presenting with Intractable Seizure, Global Developmental Delay, and Microcephaly. JIMD Reports, 2014, 19, 117-121.	0.7	9
63	Predicting 5-fluorouracil toxicity: DPD genotype and 5,6-dihydrouracil:uracil ratio. Pharmacogenomics, 2014, 15, 1653-1666.	0.6	55
64	X-linked Charcot-Marie-Tooth disease, Arts syndrome, and prelingual non-syndromic deafness form a disease continuum: evidence from a family with a novel PRPS1 mutation. Orphanet Journal of Rare Diseases, 2014, 9, 24.	1.2	42
65	Clinical, biochemical and molecular analysis of 13 Japanese patients with Î²â€ureidopropionase deficiency demonstrates high prevalence of the c.977G > A (p.R326Q) mutation. Journal of Inherited Metabolic Disease, 2014, 37, 801-812.	1.7	22
66	Dihydropyrimidine Dehydrogenase Deficiency in Two Malaysian Siblings with Abnormal MRI Findings. Molecular Syndromology, 2014, 5, 299-303.	0.3	8
67	Severe phenotype of severe combined immunodeficiency caused by adenosine deaminase deficiency in a patient with a homozygous mutation due to uniparental disomy. Journal of Allergy and Clinical Immunology, 2013, 132, 222-223.	1.5	5
68	Î²-Ureidopropionase deficiency: phenotype, genotype and protein structural consequences in 16 patients. Tijdschrift Voor Kindergeneeskunde, 2013, 81, 73-74.	0.0	1
69	Evaluation of 5-fluorouracil pharmacokinetic models and therapeutic drug monitoring in cancer patients. Pharmacogenomics, 2013, 14, 799-811.	0.6	43
70	Purine and Pyrimidine Metabolism. , 2013, , 1-38.		1
71	Role of Human Hypoxanthine Guanine Phosphoribosyltransferase in Activation of the Antiviral Agent T-705 (Favipiravir). Molecular Pharmacology, 2013, 84, 615-629.	1.0	94
72	Hydrogen Sulfide Donor NaHS Reduces Organ Injury in a Rat Model of Pneumococcal Pneumosepsis, Associated with Improved Bio-Energetic Status. PLoS ONE, 2013, 8, e63497.	1.1	42

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73	Dihydropyrimidine Dehydrogenase Deficiency and 5-Fluorouracil Toxicity. , 2013, , 337-351.		0
74	Cholestasis Is Associated with Hepatic Microvascular Dysfunction and Aberrant Energy Metabolism Before and During Ischemia-Reperfusion. Antioxidants and Redox Signaling, 2012, 17, 1109-1123.	2.5	40
75	Introduction of a Fluorine Atom at C3 of 3-Deazauridine Shifts Its Antimetabolic Activity from Inhibition of CTP Synthetase to Inhibition of Orotidylate Decarboxylase, an Early Event in the de Novo Pyrimidine Nucleotide Biosynthesis Pathway*. Journal of Biological Chemistry, 2012, 287, 30444-30454.	1.6	7
76	Induced hypothermia is protective in a rat model of pneumococcal pneumonia associated with increased adenosine triphosphate availability and turnover*. Critical Care Medicine, 2012, 40, 919-926.	0.4	31
77	Evaluation of 5-Fluorouracil Pharmacokinetics in Cancer Patients with a C.1905+1G>A Mutation in DPYD by Means of a Bayesian Limited Sampling Strategy. Clinical Pharmacokinetics, 2012, 51, 163-174.	1.6	61
78	ÃŸ-Ureidopropionase deficiency: Phenotype, genotype and protein structural consequences in 16 patients. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 1096-1108.	1.8	27
79	Synergistic interaction between cisplatin and gemcitabine in neuroblastoma cell lines and multicellular tumor spheroids. Cancer Letters, 2012, 319, 23-30.	3.2	31
80	Promising effects of the 4HPRâ€“BSO combination in neuroblastoma monolayers and spheroids. Free Radical Biology and Medicine, 2011, 51, 1213-1220.	1.3	9
81	Pharmacokinetics of orally administered uracil in healthy volunteers and in DPD-deficient patients, a possible tool for screening of DPD deficiency. Cancer Chemotherapy and Pharmacology, 2011, 68, 1611-1617.	1.1	31
82	A mild phenotype of dihydropyrimidine dehydrogenase deficiency and developmental retardation associated with a missense mutation affecting cofactor binding. Clinical Biochemistry, 2011, 44, 722-724.	0.8	4
83	SNPs and Haplotypes in DPYD and Outcome of Capecitabineâ€“Letter. Clinical Cancer Research, 2011, 17, 5837-5837.	3.2	0
84	Fenretinide induces mitochondrial ROS and inhibits the mitochondrial respiratory chain in neuroblastoma. Cellular and Molecular Life Sciences, 2010, 67, 807-816.	2.4	51
85	Intragenic deletions and a deep intronic mutation affecting pre-mRNA splicing in the dihydropyrimidine dehydrogenase gene as novel mechanisms causing 5-fluorouracil toxicity. Human Genetics, 2010, 128, 529-538.	1.8	101
86	Detection of VDR gene ApaI and TaqI polymorphisms in patients with type 2 diabetes mellitus using PCR-RFLP method in a Turkish population. Journal of Diabetes and Its Complications, 2010, 24, 186-191.	1.2	59
87	Cyclopentenylcytosine does not enhance cisplatin-induced radiosensitization in human lung tumour cells. Oncology Letters, 2010, 1, 537-540.	0.8	2
88	6-Mercaptopurine Inhibits Atherosclerosis in Apolipoprotein E*3-Leiden Transgenic Mice Through Atheroprotective Actions on Monocytes and Macrophages. Arteriosclerosis, Thrombosis, and Vascular Biology, 2010, 30, 1591-1597.	1.1	29
89	Determination of Adenosine Deaminase Activity in Dried Blood Spots by a Nonradiochemical Assay Using Reversed-Phase High-Performance Liquid Chromatography. Nucleosides, Nucleotides and Nucleic Acids, 2010, 29, 461-465.	0.4	3
90	Identification of Purine Nucleoside Phosphorylase Deficiency in Dried Blood Spots by a Non-Radiochemical Assay Using Reversed-Phase High-Performance Liquid Chromatography. Nucleosides, Nucleotides and Nucleic Acids, 2010, 29, 466-470.	0.4	5

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91	Dihydropyrimidine Dehydrogenase Deficiency Caused by a Novel Genomic Deletion c.505_513del ofDPYD. Nucleosides, Nucleotides and Nucleic Acids, 2010, 29, 509-514.	0.4	4
92	Dihydropyrimidinase deficiency: Phenotype, genotype and structural consequences in 17 patients. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2010, 1802, 639-648.	1.8	67
93	Cyclopentenyl cytosine has biological and anti-tumour activity, but does not enhance the efficacy of gemcitabine and radiation in two animal tumour models. International Journal of Oncology, 2009, 34, 813-9.	1.4	3
94	Analysis of severely affected patients with dihydropyrimidine dehydrogenase deficiency reveals large intragenic rearrangements of DPYD and a de novo interstitial deletion del(1)(p13.3p21.3). Human Genetics, 2009, 125, 581-590.	1.8	48
95	Quantitative analysis of the experimental cytotoxic drug cyclopentenyl cytosine and its metabolite in plasma with HPLC tandem mass spectrometry. Biomedical Chromatography, 2008, 22, 1368-1373.	0.8	1
96	Will cyclopentenyl cytosine (CPEC) ever have a future in the clinic?. Leukemia Research, 2008, 32, 201-202.	0.4	3
97	Mycophenolate mofetil inhibits T-cell proliferation in kidney transplant recipients without lowering intracellular dGTP and GTP. Transplant International, 2008, 21, ???-???.	0.8	10
98	ÃŸ-Alanine and ÃŸ-Aminoisobutyric Acid Levels in Two Siblings With Dihydropyrimidinase Deficiency. Nucleosides, Nucleotides and Nucleic Acids, 2008, 27, 825-829.	0.4	7
99	Identification of Two Novel Mutations C79X and R235Q in the Dihydropyrimidine Dehydrogenase Gene in a Patient Presenting With Hematuria. Nucleosides, Nucleotides and Nucleic Acids, 2008, 27, 809-815.	0.4	4
100	Pharmacokinetics of 5-Fluorouracil in Patients Heterozygous for the IVS14+1G > A Mutation in the Dihydropyrimidine Dehydrogenase Gene. Nucleosides, Nucleotides and Nucleic Acids, 2008, 27, 692-698.	0.4	22
101	Altered Dihydropyrimidine Dehydrogenase Activity Associated with Mild Toxicity in Patients Treated with 5-Fluorouracil Containing Chemotherapy. Nucleosides, Nucleotides and Nucleic Acids, 2008, 27, 726-732.	0.4	2
102	Paradoxical Elevated Thiopurine S-Methyltransferase Activity After Pancytopenia During Azathioprine Therapy: Potential Influence of Red Blood Cell Age. Therapeutic Drug Monitoring, 2008, 30, 390-393.	1.0	19
103	Dihydropyrimidine dehydrogenase deficiency presenting with psychomotor retardation in the first Polish patient.. Acta Biochimica Polonica, 2008, 55, 787-790.	0.3	3
104	Screening for Disorders of Purine and Pyrimidine Metabolism Using HPLC-Electrospray Tandem Mass Spectrometry. , 2008, , 725-737.		2
105	HPLC-Electrospray Tandem Mass Spectrometry for Rapid Determination of Dihydropyrimidine Dehydrogenase Activity. Clinical Chemistry, 2007, 53, 528-530.	1.5	12
106	Clinical, biochemical and genetic findings in two siblings with a dihydropyrimidinase deficiency. Molecular Genetics and Metabolism, 2007, 91, 157-164.	0.5	29
107	Increased dihydropyrimidine dehydrogenase activity associated with mild toxicity in patients treated with 5-fluorouracil and leucovorin. European Journal of Cancer, 2007, 43, 459-465.	1.3	22
108	Arts Syndrome Is Caused by Loss-of-Function Mutations in PRPS1. American Journal of Human Genetics, 2007, 81, 507-518.	2.6	80

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109	Determination of 5-Fluorouracil in Plasma with HPLC-Tandem Mass Spectrometry. Nucleosides, Nucleotides and Nucleic Acids, 2006, 25, 1257-1260.	0.4	15
110	Screening for Dihydropyrimidine Dehydrogenase Deficiency: To Do or Not To Do, That's The Question. Cancer Investigation, 2006, 24, 215-217.	0.6	27
111	A Pivotal Role for Î²-Aminoisobutyric Acid and Oxidative Stress in Dihydropyrimidine Dehydrogenase Deficiency?. Nucleosides, Nucleotides and Nucleic Acids, 2006, 25, 1103-1106.	0.4	8
112	Determination of Thymidine Phosphorylase Activity in Human Blood Cells and Fibroblasts by a Nonradiochemical Assay Using Reversed-Phase High-Performance Liquid Chromatography. Nucleosides, Nucleotides and Nucleic Acids, 2006, 25, 1261-1264.	0.4	3
113	Activity of Pyrimidine Degradation Enzymes in Normal Tissues. Nucleosides, Nucleotides and Nucleic Acids, 2006, 25, 1211-1214.	0.4	36
114	Antagonistic effects of sequential administration of BL1521, a histone deacetylase inhibitor, and gemcitabine to neuroblastoma cells. Cancer Letters, 2006, 233, 240-246.	3.2	7
115	beta-Ureidopropionase Deficiency Presenting with Febrile Status Epilepticus. Epilepsia, 2006, 47, 215-217.	2.6	18
116	Cyclopentenyl cytosine-induced activation of deoxycytidine kinase increases gemcitabine anabolism and cytotoxicity in neuroblastoma. Cancer Chemotherapy and Pharmacology, 2006, 57, 105-113.	1.1	25
117	Analysis of Pyrimidine Synthesis De Novo Intermediates in Urine During Crisis of a Patient with Ornithine Transcarbamylase Deficiency. Nucleosides, Nucleotides and Nucleic Acids, 2006, 25, 1251-1255.	0.4	3
118	Genetic Analysis of the First 4 Patients with Î²-Ureidopropionase Deficiency. Nucleosides, Nucleotides and Nucleic Acids, 2006, 25, 1093-1098.	0.4	8
119	Determination of thymidine phosphorylase activity by a non-radiochemical assay using reversed-phase high-performance liquid chromatography. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2005, 820, 271-275.	1.2	11
120	Erratum to â€œDetermination of thymidine phosphorylase activity by a non-radiochemical assay using reversed-phase high-performance liquid chromatographyâ€•[J. Chromatogr. B 820 (2005) 271â€“275]. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2005, 821, 245.	1.2	0
121	Late-onset MNGIE due to partial loss of thymidine phosphorylase activity. Annals of Neurology, 2005, 58, 649-652.	2.8	64
122	Absence of cardiotoxicity of the experimental cytotoxic drug cyclopentenyl cytosine (CPEC) in rats. Archives of Toxicology, 2005, 79, 268-276.	1.9	13
123	Identification of three novel mutations in the dihydropyrimidine dehydrogenase gene associated with altered pre-mRNA splicing or protein function. Biological Chemistry, 2005, 386, 1075-1075.	1.2	0
124	Identification of three novel mutations in the dihydropyrimidine dehydrogenase gene associated with altered pre-mRNA splicing or protein function. Biological Chemistry, 2005, 386, 319-324.	1.2	22
125	Gene expression profiling in response to the histone deacetylase inhibitor BL1521 in neuroblastoma. Experimental Cell Research, 2005, 309, 451-467.	1.2	38
126	Histone deacetylase inhibitor BL1521 induces a G1-phase arrest in neuroblastoma cells through altered expression of cell cycle proteins. FEBS Letters, 2005, 579, 1523-1528.	1.3	22

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127	Î²-Ureidopropionase deficiency: an inborn error of pyrimidine degradation associated with neurological abnormalities. Human Molecular Genetics, 2004, 13, 2793-2801.	1.4	78
128	Analysis of Pyrimidine Synthesis â€œde Novoâ€•Intermediates in Urine and Dried Urine Filter- Paper Strips with HPLCâ€“Electrospray Tandem Mass Spectrometry. Clinical Chemistry, 2004, 50, 2117-2124.	1.5	34
129	Quantification of 5,6-Dihydrouracil by HPLCâ€“Electrospray Tandem Mass Spectrometry. Clinical Chemistry, 2004, 50, 236-238.	1.5	24
130	The novel histone deacetylase inhibitor BL1521 inhibits proliferation and induces apoptosis in neuroblastoma cells. Biochemical Pharmacology, 2004, 68, 1279-1288.	2.0	46
131	Determination of the deoxycytidine kinase activity in cell homogenates with a non-radiochemical assay using reversed-phase high performance liquid chromatography. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2004, 805, 339-346.	1.2	13
132	Dihydropyrimidine dehydrogenase and the efficacy and toxicity of 5-fluorouracil. European Journal of Cancer, 2004, 40, 939-950.	1.3	425
133	New insights in dihydropyrimidine dehydrogenase deficiency: a pivotal role for beta-aminoisobutyric acid?. Biochemical Journal, 2004, 379, 119-124.	1.7	55
134	Cyclopentenyl cytosine primes SK-N-BE(2)c neuroblastoma cells for cytarabine toxicity. International Journal of Cancer, 2003, 103, 387-392.	2.3	21
135	Ganciclovir nucleotides accumulate in mitochondria of rat liver cells expressing the herpes simplex virus thymidine kinase gene. Journal of Gene Medicine, 2003, 5, 1018-1027.	1.4	13
136	Rapid gas chromatographicâ€“mass spectrometric diagnosis of dihydropyrimidine dehydrogenase deficiency and dihydropyrimidinase deficiency. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2003, 792, 107-115.	1.2	25
137	Histone deacetylases (HDACs): characterization of the classical HDAC family. Biochemical Journal, 2003, 370, 737-749.	1.7	2,671
138	Pharmacogenetic and clinical aspects of dihydropyrimidine dehydrogenase deficiency. Annals of Clinical Biochemistry, 2003, 40, 41-45.	0.8	93
139	Combination therapy in childhood leukaemia: in vitro studies of thiopurines and inhibitors of purine metabolism on apoptosis. Annals of Clinical Biochemistry, 2003, 40, 70-74.	0.8	9
140	Dihydropyrimidinase deficiency and severe 5-fluorouracil toxicity. Clinical Cancer Research, 2003, 9, 4363-7.	3.2	98
141	Dihydropyrimidine Dehydrogenase (DPD) Deficiency: Novel Mutations in the DPD Gene. Advances in Experimental Medicine and Biology, 2002, 486, 247-250.	0.8	6
142	Lack of Susceptibility of Bicyclic Nucleoside Analogs, Highly Potent Inhibitors of Varicella-Zoster Virus, to the Catabolic Action of Thymidine Phosphorylase and Dihydropyrimidine Dehydrogenase. Molecular Pharmacology, 2002, 61, 1140-1145.	1.0	35
143	Defects of Pyrimidine Degradation: Clinical, Molecular and Diagnostic Aspects. Advances in Experimental Medicine and Biology, 2002, 486, 233-241.	0.8	14
144	High prevalence of the IVS14 + 1G>A mutation in the dihydropyrimidine dehydrogenase gene of patients with severe 5-fluorouracil-associated toxicity. Pharmacogenetics and Genomics, 2002, 12, 555-558.	5.7	166

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145	Novel disease-causing mutations in the dihydropyrimidine dehydrogenase gene interpreted by analysis of the three-dimensional protein structure. Biochemical Journal, 2002, 364, 157-163.	1.7	102
146	Confirmation of the Enzyme Defect in the First Case of Î²-Ureidopropionase Deficiency. Advances in Experimental Medicine and Biology, 2002, 486, 243-245.	0.8	7
147	Retinoic acid reduces the cytotoxicity of cyclopentenyl cytosine in neuroblastoma cells. FEBS Letters, 2002, 527, 229-233.	1.3	4
148	Cyclopentenyl cytosine increases the phosphorylation and incorporation into DNA of 1-Î²-D-arabinofuranosyl cytosine in a human T-lymphoblastic cell line. International Journal of Cancer, 2002, 98, 616-623.	2.3	27
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