

AndrÃ© B P Van Kuilenburg

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/6218746/publications.pdf>

Version: 2024-02-01

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	Histone deacetylases (HDACs): characterization of the classical HDAC family. <i>Biochemical Journal</i> , 2003, 370, 737-749.	1.7	2,671
2	Dihydropyrimidine dehydrogenase and the efficacy and toxicity of 5-fluorouracil. <i>European Journal of Cancer</i> , 2004, 40, 939-950.	1.3	425
3	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. <i>Lancet Oncology</i> , The, 2015, 16, 1639-1650.	5.1	277
4	DPYD genotype-guided dose individualisation of fluoropyrimidine therapy in patients with cancer: a prospective safety analysis. <i>Lancet Oncology</i> , The, 2018, 19, 1459-1467.	5.1	238
5	High prevalence of the IVS14 + 1G>A mutation in the dihydropyrimidine dehydrogenase gene of patients with severe 5-fluorouracil-associated toxicity. <i>Pharmacogenetics and Genomics</i> , 2002, 12, 555-558.	5.7	166
6	Increased risk of grade IV neutropenia after administration of 5-fluorouracil due to a dihydropyrimidine dehydrogenase deficiency: High prevalence of the IVS14+1g>a mutation. <i>International Journal of Cancer</i> , 2002, 101, 253-258.	2.3	163
7	Dihydropyrimidine dehydrogenase pharmacogenetics in Caucasian subjects. <i>British Journal of Clinical Pharmacology</i> , 1998, 46, 151-156.	1.1	113
8	Novel disease-causing mutations in the dihydropyrimidine dehydrogenase gene interpreted by analysis of the three-dimensional protein structure. <i>Biochemical Journal</i> , 2002, 364, 157-163.	1.7	102
9	Intragenic deletions and a deep intronic mutation affecting pre-mRNA splicing in the dihydropyrimidine dehydrogenase gene as novel mechanisms causing 5-fluorouracil toxicity. <i>Human Genetics</i> , 2010, 128, 529-538.	1.8	101
10	Dihydropyrimidinase deficiency and severe 5-fluorouracil toxicity. <i>Clinical Cancer Research</i> , 2003, 9, 4363-7.	3.2	98
11	Role of Human Hypoxanthine Guanine Phosphoribosyltransferase in Activation of the Antiviral Agent T-705 (Favipiravir). <i>Molecular Pharmacology</i> , 2013, 84, 615-629.	1.0	94
12	Pharmacogenetic and clinical aspects of dihydropyrimidine dehydrogenase deficiency. <i>Annals of Clinical Biochemistry</i> , 2003, 40, 41-45.	0.8	93
13	Developments in the treatment of Fabry disease. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 908-921.	1.7	83
14	Retrospective study of long-term outcomes of enzyme replacement therapy in Fabry disease: Analysis of prognostic factors. <i>PLoS ONE</i> , 2017, 12, e0182379.	1.1	83
15	New advances in DPYD genotype and risk of severe toxicity under capecitabine. <i>PLoS ONE</i> , 2017, 12, e0175998.	1.1	82
16	Arts Syndrome Is Caused by Loss-of-Function Mutations in PRPS1. <i>American Journal of Human Genetics</i> , 2007, 81, 507-518.	2.6	80
17	Î²-Ureidopropionase deficiency: an inborn error of pyrimidine degradation associated with neurological abnormalities. <i>Human Molecular Genetics</i> , 2004, 13, 2793-2801.	1.4	78
18	Agalsidase alfa versus agalsidase beta for the treatment of Fabry disease: an international cohort study. <i>Journal of Medical Genetics</i> , 2018, 55, 351-358.	1.5	72

#	ARTICLE	IF	CITATIONS
19	Glutaminase Deficiency Caused by Short Tandem Repeat Expansion in <i>GLS</i> . <i>New England Journal of Medicine</i> , 2019, 380, 1433-1441.	13.9	71
20	Dihydropyrimidinase deficiency: Phenotype, genotype and structural consequences in 17 patients. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2010, 1802, 639-648.	1.8	67
21	Dihydropyrimidinase Deficiency: Structural Organization, Chromosomal Localization, and Mutation Analysis of the Human Dihydropyrimidinase Gene. <i>American Journal of Human Genetics</i> , 1998, 63, 717-726.	2.6	66
22	A cost analysis of upfront DPYD genotype‐guided dose individualisation in fluoropyrimidine-based anticancer therapy. <i>European Journal of Cancer</i> , 2019, 107, 60-67.	1.3	65
23	Late-onset MNGIE due to partial loss of thymidine phosphorylase activity. <i>Annals of Neurology</i> , 2005, 58, 649-652.	2.8	64
24	Favourable effect of early versus late start of enzyme replacement therapy on plasma globotriaosylsphingosine levels in men with classical Fabry disease. <i>Molecular Genetics and Metabolism</i> , 2017, 121, 157-161.	0.5	64
25	Evidence for transformation-related increase in CTP synthetase activity in situ in human lymphoblastic leukemia. <i>FEBS Journal</i> , 1993, 216, 161-167.	0.2	62
26	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. <i>Clinical Pharmacokinetics</i> , 2012, 51, 163-174.	1.6	61
27	Detection of VDR gene Apal and TaqI polymorphisms in patients with type 2 diabetes mellitus using PCR-RFLP method in a Turkish population. <i>Journal of Diabetes and Its Complications</i> , 2010, 24, 186-191.	1.2	59
28	Catecholamines profiles at diagnosis: Increased diagnostic sensitivity and correlation with biological and clinical features in neuroblastoma patients. <i>European Journal of Cancer</i> , 2017, 72, 235-243.	1.3	57
29	New insights in dihydropyrimidine dehydrogenase deficiency: a pivotal role for beta-aminoisobutyric acid?. <i>Biochemical Journal</i> , 2004, 379, 119-124.	1.7	55
30	Predicting 5-fluorouracil toxicity: DPD genotype and 5,6-dihydrouracil:uracil ratio. <i>Pharmacogenomics</i> , 2014, 15, 1653-1666.	0.6	55
31	Fenretinide induces mitochondrial ROS and inhibits the mitochondrial respiratory chain in neuroblastoma. <i>Cellular and Molecular Life Sciences</i> , 2010, 67, 807-816.	2.4	51
32	De Novo Mutations in SLC25A24 Cause a Disorder Characterized by Early Aging, Bone Dysplasia, Characteristic Face, and Early Demise. <i>American Journal of Human Genetics</i> , 2017, 101, 844-855.	2.6	51
33	Acute Aerobic Exercise Leads to Increased Plasma Levels of R- and S- β -Aminoisobutyric Acid in Humans. <i>Frontiers in Physiology</i> , 2019, 10, 1240.	1.3	51
34	Genotypes Affecting the Pharmacokinetics of Anticancer Drugs. <i>Clinical Pharmacokinetics</i> , 2017, 56, 317-337.	1.6	50
35	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). <i>Human Genetics</i> , 2009, 125, 581-590.	1.8	48
36	The novel histone deacetylase inhibitor BL1521 inhibits proliferation and induces apoptosis in neuroblastoma cells. <i>Biochemical Pharmacology</i> , 2004, 68, 1279-1288.	2.0	46

#	ARTICLE	IF	CITATIONS
37	Evaluation of 5-fluorouracil pharmacokinetic models and therapeutic drug monitoring in cancer patients. <i>Pharmacogenomics</i> , 2013, 14, 799-811.	0.6	43
38	DNA damage and transcription stress cause ATP-mediated redesign of metabolism and potentiation of anti-oxidant buffering. <i>Nature Communications</i> , 2019, 10, 4887.	5.8	43
39	Hydrogen Sulfide Donor NaHS Reduces Organ Injury in a Rat Model of Pneumococcal Pneumosepsis, Associated with Improved Bio-Energetic Status. <i>PLoS ONE</i> , 2013, 8, e63497.	1.1	42
40	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. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 24.	1.2	42
41	Long-Term Dose-Dependent Agalsidase Effects on Kidney Histology in Fabry Disease. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2017, 12, 1470-1479.	2.2	42
42	Phenotypic and clinical implications of variants in the dihydropyrimidine dehydrogenase gene. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2016, 1862, 754-762.	1.8	41
43	Cholestasis Is Associated with Hepatic Microvascular Dysfunction and Aberrant Energy Metabolism Before and During Ischemia-Reperfusion. <i>Antioxidants and Redox Signaling</i> , 2012, 17, 1109-1123.	2.5	40
44	Gene expression profiling in response to the histone deacetylase inhibitor BL1521 in neuroblastoma. <i>Experimental Cell Research</i> , 2005, 309, 451-467.	1.2	38
45	Activity of Pyrimidine Degradation Enzymes in Normal Tissues. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2006, 25, 1211-1214.	0.4	36
46	cDNA cloning, genomic structure and chromosomal localization of the human BUP-1 gene encoding β -ureidopropionase. <i>Biochimica Et Biophysica Acta Gene Regulatory Mechanisms</i> , 1999, 1447, 251-257.	2.4	35
47	Lack of Susceptibility of Bicyclic Nucleoside Analogs, Highly Potent Inhibitors of Varicella-Zoster Virus, to the Catabolic Action of Thymidine Phosphorylase and Dihydropyrimidine Dehydrogenase. <i>Molecular Pharmacology</i> , 2002, 61, 1140-1145.	1.0	35
48	Analysis of Pyrimidine Synthesis α -Novo α -Intermediates in Urine and Dried Urine Filter- Paper Strips with HPLC α -Electrospray Tandem Mass Spectrometry. <i>Clinical Chemistry</i> , 2004, 50, 2117-2124.	1.5	34
49	Multi-OMIC profiling of survival and metabolic signaling networks in cells subjected to photodynamic therapy. <i>Cellular and Molecular Life Sciences</i> , 2017, 74, 1133-1151.	2.4	34
50	Rapid screening for lipid storage disorders using biochemical markers. Expert center data and review of the literature. <i>Molecular Genetics and Metabolism</i> , 2018, 123, 76-84.	0.5	33
51	Severe fluoropyrimidine toxicity due to novel and rare DPYD missense mutations, deletion and genomic amplification affecting DPD activity and mRNA splicing. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2017, 1863, 721-730.	1.8	32
52	Pharmacokinetics of orally administered uracil in healthy volunteers and in DPD-deficient patients, a possible tool for screening of DPD deficiency. <i>Cancer Chemotherapy and Pharmacology</i> , 2011, 68, 1611-1617.	1.1	31
53	Induced hypothermia is protective in a rat model of pneumococcal pneumonia associated with increased adenosine triphosphate availability and turnover*. <i>Critical Care Medicine</i> , 2012, 40, 919-926.	0.4	31
54	Synergistic interaction between cisplatin and gemcitabine in neuroblastoma cell lines and multicellular tumor spheroids. <i>Cancer Letters</i> , 2012, 319, 23-30.	3.2	31

#	ARTICLE	IF	CITATIONS
55	Prenatal growth restriction, retinal dystrophy, diabetes insipidus and white matter disease: expanding the spectrum of PRPS1-related disorders. <i>European Journal of Human Genetics</i> , 2015, 23, 310-316.	1.4	30
56	Clinical, biochemical and genetic findings in two siblings with a dihydropyrimidinase deficiency. <i>Molecular Genetics and Metabolism</i> , 2007, 91, 157-164.	0.5	29
57	6-Mercaptopurine Inhibits Atherosclerosis in Apolipoprotein E*3-Leiden Transgenic Mice Through Atheroprotective Actions on Monocytes and Macrophages. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 2010, 30, 1591-1597.	1.1	29
58	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?. <i>JIMD Reports</i> , 2014, 15, 39-45.	0.7	29
59	Cyclopentenyl cytosine increases the phosphorylation and incorporation into DNA of 1- ¹² -D-arabinofuranosyl cytosine in a human T-lymphoblastic cell line. <i>International Journal of Cancer</i> , 2002, 98, 616-623.	2.3	27
60	Screening for Dihydropyrimidine Dehydrogenase Deficiency: To Do or Not To Do, That's The Question. <i>Cancer Investigation</i> , 2006, 24, 215-217.	0.6	27
61	ÄUreidopropionase deficiency: Phenotype, genotype and protein structural consequences in 16 patients. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2012, 1822, 1096-1108.	1.8	27
62	Evaluation of an oral uracil loading test to identify DPDÄdeficient patients using a limited sampling strategy. <i>British Journal of Clinical Pharmacology</i> , 2016, 81, 553-561.	1.1	26
63	Rapid gas chromatographicÄmass spectrometric diagnosis of dihydropyrimidine dehydrogenase deficiency and dihydropyrimidinase deficiency. <i>Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences</i> , 2003, 792, 107-115.	1.2	25
64	Cyclopentenyl cytosine-induced activation of deoxycytidine kinase increases gemcitabine anabolism and cytotoxicity in neuroblastoma. <i>Cancer Chemotherapy and Pharmacology</i> , 2006, 57, 105-113.	1.1	25
65	Identification of a cDNA encoding an isoform of human CTP synthetase. <i>Biochimica Et Biophysica Acta Gene Regulatory Mechanisms</i> , 2000, 1492, 548-552.	2.4	24
66	Quantification of 5,6-Dihydrouracil by HPLCÄElectrospray Tandem Mass Spectrometry. <i>Clinical Chemistry</i> , 2004, 50, 236-238.	1.5	24
67	Identification of three novel mutations in the dihydropyrimidine dehydrogenase gene associated with altered pre-mRNA splicing or protein function. <i>Biological Chemistry</i> , 2005, 386, 319-324.	1.2	22
68	Histone deacetylase inhibitor BL1521 induces a G1-phase arrest in neuroblastoma cells through altered expression of cell cycle proteins. <i>FEBS Letters</i> , 2005, 579, 1523-1528.	1.3	22
69	Increased dihydropyrimidine dehydrogenase activity associated with mild toxicity in patients treated with 5-fluorouracil and leucovorin. <i>European Journal of Cancer</i> , 2007, 43, 459-465.	1.3	22
70	Pharmacokinetics of 5-Fluorouracil in Patients Heterozygous for the IVS14+1G > A Mutation in the Dihydropyrimidine Dehydrogenase Gene. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2008, 27, 692-698.	0.4	22
71	Clinical, biochemical and molecular analysis of 13 Japanese patients with Î²Äureidopropionase deficiency demonstrates high prevalence of the c.977G > A (p.R326Q) mutation. <i>Journal of Inherited Metabolic Disease</i> , 2014, 37, 801-812.	1.7	22
72	Hyperferritinemia and iron metabolism in Gaucher disease: Potential pathophysiological implications. <i>Blood Reviews</i> , 2016, 30, 431-437.	2.8	22

#	ARTICLE	IF	CITATIONS
73	Cyclopentenyl cytosine primes SK-N-BE(2)c neuroblastoma cells for cytarabine toxicity. <i>International Journal of Cancer</i> , 2003, 103, 387-392.	2.3	21
74	The pivotal role of uridine-cytidine kinases in pyrimidine metabolism and activation of cytotoxic nucleoside analogues in neuroblastoma. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2016, 1862, 1504-1512.	1.8	21
75	Yunis-VarĂ±n syndrome caused by biallelic VAC14 mutations. <i>European Journal of Human Genetics</i> , 2017, 25, 1049-1054.	1.4	21
76	The cytostatic- and differentiation-inducing effects of cyclopentenyl cytosine on neuroblastoma cell lines11Abbreviations: CPEC, cyclopentenyl cytosine; and CPECTP, cyclopentenyl cytosine-5â€™-triphosphate.. <i>Biochemical Pharmacology</i> , 2001, 62, 1099-1105.	2.0	20
77	The Cytidine Analog Fluorocyclopentenylcytosine (RX-3117) Is Activated by Uridine-Cytidine Kinase 2. <i>PLoS ONE</i> , 2016, 11, e0162901.	1.1	20
78	Towards a test to predict 5-fluorouracil toxicity: Pharmacokinetic data for thymine and two sequential metabolites following oral thymine administration to healthy adult males. <i>European Journal of Pharmaceutical Sciences</i> , 2016, 81, 36-41.	1.9	20
79	Translational Metabolism: A multidisciplinary approach towards precision diagnosis of inborn errors of metabolism in the omics era. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 197-208.	1.7	20
80	Detection of Î²-ureidopropionase deficiency with HPLC-electrospray tandem mass spectrometry and confirmation of the defect at the enzyme level. <i>Journal of Inherited Metabolic Disease</i> , 2001, 24, 725-732.	1.7	19
81	Paradoxical Elevated Thiopurine S-Methyltransferase Activity After Pancytopenia During Azathioprine Therapy: Potential Influence of Red Blood Cell Age. <i>Therapeutic Drug Monitoring</i> , 2008, 30, 390-393.	1.0	19
82	Creatine kinase inhibition lowers systemic arterial blood pressure in spontaneously hypertensive rats. <i>Journal of Hypertension</i> , 2016, 34, 2418-2426.	0.3	19
83	beta-Ureidopropionase Deficiency Presenting with Febrile Status Epilepticus. <i>Epilepsia</i> , 2006, 47, 215-217.	2.6	18
84	Patients homozygous for DPYD c.1129-5923C>G/haplotype B3 have partial DPD deficiency and require a dose reduction when treated with fluoropyrimidines. <i>Cancer Chemotherapy and Pharmacology</i> , 2016, 78, 875-880.	1.1	17
85	Dihydropyrimidinase deficiency in four East Asian patients due to novel and rare DPYS mutations affecting protein structural integrity and catalytic activity. <i>Molecular Genetics and Metabolism</i> , 2017, 122, 216-222.	0.5	16
86	Determination of 5-Fluorouracil in Plasma with HPLC-Tandem Mass Spectrometry. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2006, 25, 1257-1260.	0.4	15
87	Capecitabineâ€based treatment of a patient with a novel <i>DPYD</i> genotype and complete dihydropyrimidine dehydrogenase deficiency. <i>International Journal of Cancer</i> , 2018, 142, 424-430.	2.3	15
88	Biochemical and imaging parameters in acid sphingomyelinase deficiency: Potential utility as biomarkers. <i>Molecular Genetics and Metabolism</i> , 2020, 130, 16-26.	0.5	15
89	Defects of Pyrimidine Degradation: Clinical, Molecular and Diagnostic Aspects. <i>Advances in Experimental Medicine and Biology</i> , 2002, 486, 233-241.	0.8	14
90	5-fluorocytosine-related bone-marrow depression and conversion to fluorouracil: a pilot study. <i>Fundamental and Clinical Pharmacology</i> , 2002, 16, 39-47.	1.0	14

#	ARTICLE	IF	CITATIONS
91	Plasma free metanephrines for diagnosis of neuroblastoma patients. <i>Clinical Biochemistry</i> , 2019, 66, 57-62.	0.8	14
92	Ganciclovir nucleotides accumulate in mitochondria of rat liver cells expressing the herpes simplex virus thymidine kinase gene. <i>Journal of Gene Medicine</i> , 2003, 5, 1018-1027.	1.4	13
93	Determination of the deoxycytidine kinase activity in cell homogenates with a non-radiochemical assay using reversed-phase high performance liquid chromatography. <i>Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences</i> , 2004, 805, 339-346.	1.2	13
94	Absence of cardiotoxicity of the experimental cytotoxic drug cyclopentenyl cytosine (CPEC) in rats. <i>Archives of Toxicology</i> , 2005, 79, 268-276.	1.9	13
95	Quantitative Analysis of the Pyrimidine Metabolism in Pheochromocytoma PC-12 Cells. <i>FEBS Journal</i> , 1995, 233, 538-543.	0.2	12
96	MIBG causes oxidative stress and up-regulation of anti-oxidant enzymes in the human neuroblastoma cell line SK-N-BE(2C)., 1997, 72, 486-490.		12
97	HPLC-Electrospray Tandem Mass Spectrometry for Rapid Determination of Dihydropyrimidine Dehydrogenase Activity. <i>Clinical Chemistry</i> , 2007, 53, 528-530.	1.5	12
98	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. <i>JIMD Reports</i> , 2015, 24, 83-89.	0.7	12
99	Liposome-targeted recombinant human acid sphingomyelinase: Production, formulation, and in vitro evaluation. <i>European Journal of Pharmaceutics and Biopharmaceutics</i> , 2019, 137, 185-195.	2.0	12
100	Subcellular Localization of Dihydropyrimidine Dehydrogenase. <i>Biological Chemistry</i> , 1997, 378, 1047-53.	1.2	11
101	Simultaneous determination of F- $\hat{1}^2$ -alanine and $\hat{1}^2$ -alanine in plasma and urine with dual-column reversed-phase high-performance liquid chromatography. <i>Biomedical Applications</i> , 2001, 759, 51-61.	1.7	11
102	Determination of thymidine phosphorylase activity by a non-radiochemical assay using reversed-phase high-performance liquid chromatography. <i>Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences</i> , 2005, 820, 271-275.	1.2	11
103	Dopamine induces lipid accumulation, NADPH oxidase-related oxidative stress, and a proinflammatory status of the plasma membrane in H9c2 cells. <i>American Journal of Physiology - Heart and Circulatory Physiology</i> , 2016, 311, H1097-H1107.	1.5	11
104	Novel PRPS1 gain-of-function mutation in a patient with congenital hyperuricemia and facial anomalies. , 2017, 173, 2736-2742.		11
105	The pathophysiology of human obstructive cholestasis is mimicked in cholestatic Gold Syrian hamsters. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2018, 1864, 942-951.	1.8	11
106	Determination of CTP synthetase activity in crude cell homogenates by a fast and sensitive non-radiochemical assay using anion-exchange high-performance liquid chromatography. <i>Biomedical Applications</i> , 1997, 693, 287-295.	1.7	10
107	Mycophenolate mofetil inhibits T-cell proliferation in kidney transplant recipients without lowering intracellular dGTP and GTP. <i>Transplant International</i> , 2008, 21, ???-???	0.8	10
108	Altered Pre-mRNA Splicing Caused by a Novel Intronic Mutation c.1443+5G>A in the Dihydropyrimidinase (DPYS) Gene. <i>International Journal of Molecular Sciences</i> , 2016, 17, 86.	1.8	10

#	ARTICLE	IF	CITATIONS
109	Diagnostic and Therapeutic Strategies for Fluoropyrimidine Treatment of Patients Carrying Multiple DPYD Variants. <i>Genes</i> , 2018, 9, 585.	1.0	10
110	The Effects of Mercaptopurine on Pulmonary Vascular Resistance and BMPR2 Expression in Pulmonary Arterial Hypertension. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2020, 202, 296-299.	2.5	10
111	Combination therapy in childhood leukaemia: in vitro studies of thiopurines and inhibitors of purine metabolism on apoptosis. <i>Annals of Clinical Biochemistry</i> , 2003, 40, 70-74.	0.8	9
112	Promising effects of the 4HPR��BSO combination in neuroblastoma monolayers and spheroids. <i>Free Radical Biology and Medicine</i> , 2011, 51, 1213-1220.	1.3	9
113	A Korean Case of ��-Ureidopropionase Deficiency Presenting with Intractable Seizure, Global Developmental Delay, and Microcephaly. <i>JIMD Reports</i> , 2014, 19, 117-121.	0.7	9
114	Metachromatic leukodystrophy genotypes in The Netherlands reveal novel pathogenic ARSA variants in non-Caucasian patients. <i>Neurogenetics</i> , 2020, 21, 289-299.	0.7	9
115	Predicting the Development of Anti-Drug Antibodies against Recombinant alpha-Galactosidase A in Male Patients with Classical Fabry Disease. <i>International Journal of Molecular Sciences</i> , 2020, 21, 5784.	1.8	9
116	Children with atopic dermatitis show increased activity of ��-glucocerebrosidase and stratum corneum levels of glucosylcholesterol that are strongly related to the local cytokine milieu. <i>British Journal of Dermatology</i> , 2022, 186, 988-996.	1.4	9
117	A Pivotal Role for ��-Aminoisobutyric Acid and Oxidative Stress in Dihydropyrimidine Dehydrogenase Deficiency?. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2006, 25, 1103-1106.	0.4	8
118	Genetic Analysis of the First 4 Patients with ��-Ureidopropionase Deficiency. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2006, 25, 1093-1098.	0.4	8
119	Pleiotropic effects of fenretinide in neuroblastoma cell lines and multicellular tumor spheroids. <i>International Journal of Oncology</i> , 0, , .	1.4	8
120	Dihydropyrimidine Dehydrogenase Deficiency in Two Malaysian Siblings with Abnormal MRI Findings. <i>Molecular Syndromology</i> , 2014, 5, 299-303.	0.3	8
121	Treatment Algorithm for Homozygous or Compound Heterozygous DPYD Variant Allele Carriers With Low-Dose Capecitabine. <i>JCO Precision Oncology</i> , 2017, 1, 1-10.	1.5	8
122	Deficiency of perforin and hCNT1, a novel inborn error of pyrimidine metabolism, associated with a rapidly developing lethal phenotype due to multi-organ failure. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2019, 1865, 1182-1191.	1.8	8
123	Confirmation of the Enzyme Defect in the First Case of ��-Ureidopropionase Deficiency. <i>Advances in Experimental Medicine and Biology</i> , 2002, 486, 243-245.	0.8	7
124	Antagonistic effects of sequential administration of BL1521, a histone deacetylase inhibitor, and gemcitabine to neuroblastoma cells. <i>Cancer Letters</i> , 2006, 233, 240-246.	3.2	7
125	��-Alanine and ��-Aminoisobutyric Acid Levels in Two Siblings With Dihydropyrimidinase Deficiency. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2008, 27, 825-829.	0.4	7
126	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*. <i>Journal of Biological Chemistry</i> , 2012, 287, 30444-30454.	1.6	7

#	ARTICLE	IF	CITATIONS
127	Influence of metastatic disease on the usefulness of uracil pharmacokinetics as a screening tool for DPD activity in colorectal cancer patients. <i>Cancer Chemotherapy and Pharmacology</i> , 2015, 76, 47-52.	1.1	7
128	Dihydropyrimidine Dehydrogenase (DPD) Deficiency: Novel Mutations in the DPD Gene. <i>Advances in Experimental Medicine and Biology</i> , 2002, 486, 247-250.	0.8	6
129	Purification, activity, and expression levels of two uridine-cytidine kinase isoforms in neuroblastoma cell lines. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2016, 35, 613-618.	0.4	6
130	Genome sequencing reveals a novel genetic mechanism underlying dihydropyrimidine dehydrogenase deficiency: A novel missense variant c.1700G>A and a large intragenic inversion in <i>DPYD</i> spanning intron 8 to intron 12. <i>Human Mutation</i> , 2018, 39, 947-953.	1.1	6
131	Atypical presentation of Arts syndrome due to a novel hemizygous loss-of-function variant in the PRPS1 gene. <i>Molecular Genetics and Metabolism Reports</i> , 2020, 25, 100677.	0.4	6
132	Preemptive screening of DPYD as part of clinical practice: high prevalence of a novel exon 4 deletion in the Finnish population. <i>Cancer Chemotherapy and Pharmacology</i> , 2021, 87, 657-663.	1.1	6
133	Co-therapy with S-adenosylmethionine and nicotinamide riboside improves t-cell survival and function in Arts Syndrome (PRPS1 deficiency). <i>Molecular Genetics and Metabolism Reports</i> , 2021, 26, 100709.	0.4	6
134	Identification of Purine Nucleoside Phosphorylase Deficiency in Dried Blood Spots by a Non-Radiochemical Assay Using Reversed-Phase High-Performance Liquid Chromatography. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2010, 29, 466-470.	0.4	5
135	Severe phenotype of severe combined immunodeficiency caused by adenosine deaminase deficiency in a patient with a homozygous mutation due to uniparental disomy. <i>Journal of Allergy and Clinical Immunology</i> , 2013, 132, 222-223.	1.5	5
136	Hypothermic perfusion with retrograde outflow during right hepatectomy is safe and feasible. <i>Surgery</i> , 2017, 162, 48-58.	1.0	5
137	Selective serotonin reuptake inhibitors (SSRIs) prevent meta-iodobenzylguanidine (MIBG) uptake in platelets without affecting neuroblastoma tumor uptake. <i>EJNMMI Research</i> , 2020, 10, 78.	1.1	5
138	Retinoic acid reduces the cytotoxicity of cyclopentenyl cytosine in neuroblastoma cells. <i>FEBS Letters</i> , 2002, 527, 229-233.	1.3	4
139	Identification of Two Novel Mutations C79X and R235Q in the Dihydropyrimidine Dehydrogenase Gene in a Patient Presenting With Hematuria. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2008, 27, 809-815.	0.4	4
140	Dihydropyrimidine Dehydrogenase Deficiency Caused by a Novel Genomic Deletion c.505_513del ofDPYD. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2010, 29, 509-514.	0.4	4
141	A mild phenotype of dihydropyrimidine dehydrogenase deficiency and developmental retardation associated with a missense mutation affecting cofactor binding. <i>Clinical Biochemistry</i> , 2011, 44, 722-724.	0.8	4
142	Preliminary Evidence for Enhanced Thymine Absorption: A Putative New Phenotype Associated With Fluoropyrimidine Toxicity in Cancer Patients. <i>Therapeutic Drug Monitoring</i> , 2018, 40, 495-502.	1.0	4
143	Urinary 3-Methoxytyramine Is a Biomarker for MYC Activity in Patients With Neuroblastoma. <i>JCO Precision Oncology</i> , 2022, 6, e2000447.	1.5	4
144	Determination of Thymidine Phosphorylase Activity in Human Blood Cells and Fibroblasts by a Nonradiochemical Assay Using Reversed-Phase High-Performance Liquid Chromatography. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2006, 25, 1261-1264.	0.4	3

#	ARTICLE	IF	CITATIONS
145	Analysis of Pyrimidine Synthesis De Novo Intermediates in Urine During Crisis of a Patient with Ornithine Transcarbamylase Deficiency. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2006, 25, 1251-1255.	0.4	3
146	Will cyclopentenyl cytosine (CPEC) ever have a future in the clinic?. <i>Leukemia Research</i> , 2008, 32, 201-202.	0.4	3
147	Cyclopentenyl cytosine has biological and anti-tumour activity, but does not enhance the efficacy of gemcitabine and radiation in two animal tumour models. <i>International Journal of Oncology</i> , 2009, 34, 813-9.	1.4	3
148	Determination of Adenosine Deaminase Activity in Dried Blood Spots by a Nonradiochemical Assay Using Reversed-Phase High-Performance Liquid Chromatography. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2010, 29, 461-465.	0.4	3
149	Dihydropyrimidine Dehydrogenase Deficiency: Homozygosity for an Extremely Rare Variant in DPYD due to Uniparental Isodisomy of Chromosome 1. <i>JIMD Reports</i> , 2018, 45, 65-69.	0.7	3
150	Purine and Pyrimidine Metabolism. , 2021, , 183-234.		3
151	Imbalance between the Pyrimidine Ribonucleotide Pools in Rat Rhabdomyosarcoma R1 Cells. <i>Advances in Experimental Medicine and Biology</i> , 1995, 370, 279-282.	0.8	3
152	Dihydropyrimidine dehydrogenase deficiency presenting with psychomotor retardation in the first Polish patient.. <i>Acta Biochimica Polonica</i> , 2008, 55, 787-790.	0.3	3
153	Î²-Ureidopropionase deficiency due to novel and rare UPB1 mutations affecting pre-mRNA splicing and protein structural integrity and catalytic activity. <i>Molecular Genetics and Metabolism</i> , 2022, 136, 177-185.	0.5	3
154	Altered Dihydropyrimidine Dehydrogenase Activity Associated with Mild Toxicity in Patients Treated with 5-Fluorouracil Containing Chemotherapy. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2008, 27, 726-732.	0.4	2
155	Cyclopentenylcytosine does not enhance cisplatin-induced radiosensitization in human lung tumour cells. <i>Oncology Letters</i> , 2010, 1, 537-540.	0.8	2
156	Screening for Disorders of Purine and Pyrimidine Metabolism Using HPLC-Electrospray Tandem Mass Spectrometry. , 2008, , 725-737.		2
157	Quantitative analysis of the experimental cytotoxic drug cyclopentenyl cytosine and its metabolite in plasma with HPLC tandem mass spectrometry. <i>Biomedical Chromatography</i> , 2008, 22, 1368-1373.	0.8	1
158	Î²-Ureidopropionase deficiency: phenotype, genotype and protein structural consequences in 16 patients. <i>Tijdschrift Voor Kindergeneeskunde</i> , 2013, 81, 73-74.	0.0	1
159	Purine and Pyrimidine Metabolism. , 2013, , 1-38.		1
160	Thrombocytopenia after meta-iodobenzylguanidine (MIBG) therapy in neuroblastoma patients may be caused by selective MIBG uptake via the serotonin transporter located on megakaryocytes. <i>EJNMMI Research</i> , 2021, 11, 81.	1.1	1
161	MIBG causes oxidative stress and up-regulation of anti-oxidant enzymes in the human neuroblastoma cell line SK-N-BE(2C). , 1997, 72, 486.		1
162	Erratum to "Determination of thymidine phosphorylase activity by a non-radiochemical assay using reversed-phase high-performance liquid chromatography" [J. Chromatogr. B 820 (2005) 271-275]. <i>Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences</i> , 2005, 821, 245.	1.2	0

#	ARTICLE	IF	CITATIONS
163	Identification of three novel mutations in the dihydropyrimidine dehydrogenase gene associated with altered pre-mRNA splicing or protein function. <i>Biological Chemistry</i> , 2005, 386, 1075-1075.	1.2	0
164	SNPs and Haplotypes in DPYD and Outcome of Capecitabine—Letter. <i>Clinical Cancer Research</i> , 2011, 17, 5837-5837.	3.2	0
165	Dihydropyrimidine Dehydrogenase Deficiency and 5-Fluorouracil Toxicity. , 2013, , 337-351.		0