JÃ¶rgen Bierau

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/7817359/publications.pdf

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31	930	516710	454955
papers	citations	16	30
		h-index	g-index

32	32	32	2096
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	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.	1.1	3
2	Targeted urine metabolomics with a graphical reporting tool for rapid diagnosis of inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2021, 44, 1113-1123.	3.6	16
3	Follow-Up Study of Growth Hormone Therapy in Children with Kabuki Syndrome: Two-Year Treatment Results. Hormone Research in Paediatrics, 2021, 94, 285-296.	1.8	4
4	Metabolic Profiling Associates with Disease Severity in Nonischemic Dilated Cardiomyopathy. Journal of Cardiac Failure, 2020, 26, 212-222.	1.7	22
5	Dosage of 6-Mercaptopurine in Relation to Genetic TPMT and ITPA Variants: Toward Individualized Pediatric Acute Lymphoblastic Leukemia Maintenance Treatment. Journal of Pediatric Hematology/Oncology, 2020, 42, e94-e97.	0.6	9
6	Results of an explorative clinical evaluation suggest immediate and persistent post-reperfusion metabolic paralysis drives kidney ischemia reperfusion injury. Kidney International, 2020, 98, 1476-1488.	5.2	20
7	Expanding the clinical and genetic spectrum of CAD deficiency: an epileptic encephalopathy treatable with uridine supplementation. Genetics in Medicine, 2020, 22, 1589-1597.	2.4	19
8	Kidney and vascular function in adult patients with hereditary fructose intolerance. Molecular Genetics and Metabolism Reports, 2020, 23, 100600.	1.1	7
9	Specific Nutritional Biomarker Profiles in Mild Cognitive Impairment and Subjective Cognitive Decline Are Associated With Clinical Progression: The NUDAD Project. Journal of the American Medical Directors Association, 2020, 21, 1513.e1-1513.e17.	2.5	17
10	Metabolic events in HIV-infected patients using abacavir are associated with erythrocyte inosine triphosphatase activity. Journal of Antimicrobial Chemotherapy, 2019, 74, 157-164.	3.0	3
11	Extended diagnosis of purine and pyrimidine disorders from urine: LC MS/MS assay development and clinical validation. PLoS ONE, 2019, 14, e0212458.	2.5	25
12	Prolyl hydroxylase substrate adenylosuccinate lyase is an oncogenic driver in triple negative breast cancer. Nature Communications, 2019, 10, 5177.	12.8	27
13	Emotional and behavioral problems, quality of life and metabolic control in NTBC-treated Tyrosinemia type 1 patients. Orphanet Journal of Rare Diseases, 2019, 14, 285.	2.7	19
14	Evolution of Dihydropyrimidine Dehydrogenase Diagnostic Testing in a Single Center during an 8-Year Period of Time. Current Therapeutic Research, 2019, 90, 1-7.	1.2	8
15	Fast and accurate quantitative organic acid analysis with LCâ€QTOF/MS facilitates screening of patients for inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2018, 41, 415-424.	3.6	37
16	Titin cardiomyopathy leads to altered mitochondrial energetics, increased fibrosis and long-term life-threatening arrhythmias. European Heart Journal, 2018, 39, 864-873.	2.2	132
17	Impaired fertility and motor function in a zebrafish model for classic galactosemia. Journal of Inherited Metabolic Disease, 2018, 41, 117-127.	3.6	16
18	Persistent metabolic changes in HIV-infected patients during the first year of combination antiretroviral therapy. Scientific Reports, 2018, 8, 16947.	3.3	43

#	Article	IF	Citations
19	Erythrocyte Inosine triphosphatase activity: A potential biomarker for adverse events during combination antiretroviral therapy for HIV. PLoS ONE, 2018, 13, e0191069.	2.5	5
20	Nutrients required for phospholipid synthesis are lower in blood and cerebrospinal fluid in mild cognitive impairment and Alzheimer's disease dementia. Alzheimer's and Dementia: Diagnosis, Assessment and Disease Monitoring, 2017, 8, 139-146.	2.4	19
21	Classic Galactosemia: Study on the Late Prenatal Development of GALT Specific Activity in a Sheep Model. Anatomical Record, 2017, 300, 1570-1575.	1.4	7
22	The hypoxanthine-xanthine oxidase axis is not involved in the initial phase of clinical transplantation-related ischemia-reperfusion injury. American Journal of Physiology - Renal Physiology, 2017, 312, F457-F464.	2.7	7
23	CADmutations and uridine-responsive epileptic encephalopathy. Brain, 2017, 140, 279-286.	7.6	106
24	Increasing Insulin Availability Does Not Augment Postprandial Muscle Protein Synthesis Rates in Healthy Young and Older Men. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 3978-3988.	3.6	19
25	Defective postreperfusion metabolic recovery directly associates with incident delayed graft function. Kidney International, 2016, 90, 181-191.	5.2	28
26	Recessive <scp>ITPA</scp> mutations cause an early infantile encephalopathy. Annals of Neurology, 2015, 78, 649-658.	5.3	45
27	Post-Prandial Protein Handling: You Are What You Just Ate. PLoS ONE, 2015, 10, e0141582.	2.5	96
28	Serotonergic, noradrenergic and dopaminergic markers are related to cognitive function in adults with 22q11 deletion syndrome. International Journal of Neuropsychopharmacology, 2014, 17, 1159-1165.	2.1	17
29	Erythrocyte Inosine Triphosphatase Activity Is Decreased in HIV-Seropositive Individuals. PLoS ONE, 2012, 7, e30175.	2.5	5
30	An HPLC-Based Assay of Adenylosuccinate Lyase in Erythrocytes. Nucleosides, Nucleotides and Nucleic Acids, 2011, 30, 908-917.	1.1	3
31	Quantitative UPLC-MS/MS analysis of underivatised amino acids in body fluids is a reliable tool for the diagnosis and follow-up of patients with inborn errors of metabolism. Clinica Chimica Acta, 2009, 407, 36-42.	1.1	146