Jörgen Bierau

List of Publications by Year in descending order

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516710 454955 31 930 16 30 citations g-index h-index papers 32 32 32 2096 docs citations times ranked citing authors all docs

#	Article	IF	CITATIONS
1	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
2	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
3	<i>CAD</i> mutations and uridine-responsive epileptic encephalopathy. Brain, 2017, 140, 279-286.	7.6	106
4	Post-Prandial Protein Handling: You Are What You Just Ate. PLoS ONE, 2015, 10, e0141582.	2.5	96
5	Recessive <scp><i>ITPA</i></scp> mutations cause an early infantile encephalopathy. Annals of Neurology, 2015, 78, 649-658.	5.3	45
6	Persistent metabolic changes in HIV-infected patients during the first year of combination antiretroviral therapy. Scientific Reports, 2018, 8, 16947.	3.3	43
7	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
8	Defective postreperfusion metabolic recovery directly associates with incident delayed graft function. Kidney International, 2016, 90, 181-191.	5.2	28
9	Prolyl hydroxylase substrate adenylosuccinate lyase is an oncogenic driver in triple negative breast cancer. Nature Communications, 2019, 10, 5177.	12.8	27
10	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
11	Metabolic Profiling Associates with Disease Severity in Nonischemic Dilated Cardiomyopathy. Journal of Cardiac Failure, 2020, 26, 212-222.	1.7	22
12	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
13	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
14	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
15	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
16	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
17	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
18	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

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19	Impaired fertility and motor function in a zebrafish model for classic galactosemia. Journal of Inherited Metabolic Disease, 2018, 41, 117-127.	3.6	16
20	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
21	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
22	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
23	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
24	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
25	Kidney and vascular function in adult patients with hereditary fructose intolerance. Molecular Genetics and Metabolism Reports, 2020, 23, 100600.	1.1	7
26	Erythrocyte Inosine Triphosphatase Activity Is Decreased in HIV-Seropositive Individuals. PLoS ONE, 2012, 7, e30175.	2.5	5
27	Erythrocyte Inosine triphosphatase activity: A potential biomarker for adverse events during combination antiretroviral therapy for HIV. PLoS ONE, 2018, 13, e0191069.	2.5	5
28	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
29	An HPLC-Based Assay of Adenylosuccinate Lyase in Erythrocytes. Nucleosides, Nucleotides and Nucleic Acids, 2011, 30, 908-917.	1.1	3
30	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
31	Î ² -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