Soumeya Bekri

List of Publications by Year
in descending order

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92 papers	6,653 citations	172457 29 h-index	64796 79 g-index
102 all docs	102 docs citations	102 times ranked	14145 citing authors

#	Article	IF	Citations
1	Disentangling molecular and clinical stratification patterns in beta-galactosidase deficiency. Journal of Medical Genetics, 2022, 59, 377-384.	3.2	2
2	Sphingosine-1-Phosphate Levels Are Higher in Male Patients with Non-Classic Fabry Disease. Journal of Clinical Medicine, 2022, 11, 1233.	2.4	0
3	Cystinuria: clinical practice recommendation. Kidney International, 2021, 99, 48-58.	5.2	58
4	Muscle metabolic remodelling patterns in Duchenne muscular dystrophy revealed by ultra-high-resolution mass spectrometry imaging. Scientific Reports, 2021, 11, 1906.	3.3	19
5	Next-Generation Molecular Investigations in Lysosomal Diseases: Clinical Integration of a Comprehensive Targeted Panel. Diagnostics, 2021, 11, 294.	2.6	3
6	Mucopolysaccharidosis type VII as a cause of recurrent Non-Immune Hydrops Fetalis: The first Tunisian case confirmed by Next-Generation Sequencing. Clinica Chimica Acta, 2021, 513, 68-70.	1.1	2
7	NGLY1 Deficiency: A Rare Newly Described Condition with a Typical Presentation. Life, 2021, 11, 187.	2.4	12
8	Diagnosis and Management of Glioblastoma: A Comprehensive Perspective. Journal of Personalized Medicine, 2021, 11, 258.	2.5	23
9	Hepcidin and Iron Deficiency in Women One Year after Sleeve Gastrectomy: A Prospective Cohort Study. Nutrients, 2021, 13, 2516.	4.1	4
10	An Atypical Case of Head Tremor and Extensive White Matter in an Adult Female Caused by 3-Hydroxy-3-methylglutaryl-CoA Lyase Deficiency. Diagnostics, 2021, 11, 1561.	2.6	3
11	Large-scale screening of lipase acid deficiency in at risk population. Clinica Chimica Acta, 2021, 519, 64-69.	1.1	7
12	Parsing Fabry Disease Metabolic Plasticity Using Metabolomics. Journal of Personalized Medicine, 2021, 11, 898.	2.5	3
13	Precision Neurosurgery: A Path Forward. Journal of Personalized Medicine, 2021, 11, 1019.	2.5	2
14	Integrative Metabolomics Reveals Deep Tissue and Systemic Metabolic Remodeling in Glioblastoma. Cancers, 2021, 13, 5157.	3.7	9
15	Heterogenous Clinical Landscape in a Consanguineous Malonic Aciduria Family. International Journal of Molecular Sciences, 2021, 22, 12633.	4.1	4
16	An Atypical Case of Congenital Erythropoietic Porphyria. Genes, 2021, 12, 1828.	2.4	2
17	Sebelipase alfa enzyme replacement therapy in Wolman disease: a nationwide cohort with up to ten years of follow-up. Orphanet Journal of Rare Diseases, 2021, 16, 507.	2.7	8
18	Hemophagocytic Lymphohistiocytosis: A Rare Complication of an Ultrarare Lysosomal Storage Disease. Journal of Pediatric Hematology/Oncology, 2020, 42, 310-312.	0.6	6

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19	Optimization of ion trajectories in a dynamically harmonized Fourierâ€transform ion cyclotron resonance cell using a design of experiments strategy. Rapid Communications in Mass Spectrometry, 2020, 34, e8659.	1.5	9
20	High-throughput omics in the precision medicine ecosystem. , 2020, , 19-31.		1
21	Intraventricular Hemorrhage in Very Preterm Infants: A Comprehensive Review. Journal of Clinical Medicine, 2020, 9, 2447.	2.4	29
22	A Proteomics-Based Analysis Reveals Predictive Biological Patterns in Fabry Disease. Journal of Clinical Medicine, 2020, 9, 1325.	2.4	18
23	Hypertonic sodium lactate improves microcirculation, cardiac function, and inflammation in a rat model of sepsis. Critical Care, 2020, 24, 354.	5.8	13
24	High diagnostic value of plasma Niemann-Pick type C biomarkers in adults with selected neurological and/or psychiatric disorders. Journal of Neurology, 2020, 267, 3371-3377.	3.6	8
25	Analysis of Mucopolysaccharidosis Type VI through Integrative Functional Metabolomics. International Journal of Molecular Sciences, 2019, 20, 446.	4.1	18
26	Hyperinsulinemic Hypoglycemia in a Neonate. Clinical Chemistry, 2019, 65, 351-353.	3.2	0
27	Integrative metabolic profiling in Sanfilippo syndrome. Molecular Genetics and Metabolism, 2019, 126, S30.	1.1	0
28	Next generation sequencing sheds light on inherited metabolic diseases in nonimmune hydrops fetalis investigations. Molecular Genetics and Metabolism, 2019, 126, S30-S31.	1.1	0
29	A new optimization strategy for MALDI FTICR MS tissue analysis for untargeted metabolomics using experimental design and data modeling. Analytical and Bioanalytical Chemistry, 2019, 411, 3891-3903.	3.7	14
30	Paving the Way to Precision Nutrition Through Metabolomics. Frontiers in Nutrition, 2019, 6, 41.	3.7	84
31	Fabry patientsâ€™ needs and expectations regarding their treatment in France: Development of a Patientsâ€™ Need Questionnaire (PNQ Fabry). Molecular Genetics and Metabolism, 2019, 126, S110.	1.1	0
32	Metabolomics Tools and Information Retrieval in Microbiome Hacking. , 2019, , 43-59.		0
33	Treatment needs and expectations for Fabry disease in France: development of a new Patient Needs Questionnaire. Orphanet Journal of Rare Diseases, 2019, 14, 284.	2.7	6
34	A Novel Mutation c.153 C>A in a Tunisian Girl With Wolman Disease and Unusual Presentation: Hemophagocytic Lymphohistiocytosis. Journal of Pediatric Hematology/Oncology, 2019, 41, e193-e196.	0.6	14
35	Metabolic causes of nonimmune hydrops fetalis: A next-generation sequencing panel as a first-line investigation. Clinica Chimica Acta, 2018, 481, 1-8.	1.1	32
36	Functional assessment of creatine transporter in control and X-linked SLC6A8-deficient fibroblasts. Molecular Genetics and Metabolism, 2018, 123, 463-471.	1.1	3

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37	Dilated Cardiomyopathy and Premature Ovarian Failure Unveiling Propionic Aciduria. Clinical Chemistry, 2018, 64, 752-754.	3.2	5
38	Advances in metabolome information retrieval: turning chemistry into biology. Part II: biological information recovery. Journal of Inherited Metabolic Disease, 2018, 41, 393-406.	3.6	16
39	Advances in metabolome information retrieval: turning chemistry into biology. Part I: analytical chemistry of the metabolome. Journal of Inherited Metabolic Disease, 2018, 41, 379-391.	3.6	29
40	Acute Respiratory Infection Unveiling CPT II Deficiency. International Journal of Molecular Sciences, 2018, 19, 2950.	4.1	2
41	Unveiling metabolic remodeling in mucopolysaccharidosis type III through integrative metabolomics and pathway analysis. Journal of Translational Medicine, 2018, 16, 248.	4.4	19
42	Major remodeling of brain microvessels during neonatal period in the mouse: A proteomic and transcriptomic study. Journal of Cerebral Blood Flow and Metabolism, 2017, 37, 495-513.	4.3	15
43	Prenatal alcohol exposure impairs autophagy in neonatal brain cortical microvessels. Cell Death and Disease, 2017, 8, e2610-e2610.	6.3	25
44	Enhancing the diagnosis of fabry disease in cardiology with a targeted information: a beforeâ€“after controlâ€“impact study. Open Heart, 2017, 4, e000567.	2.3	2
45	Urinary metabolic phenotyping of mucopolysaccharidosis type I combining untargeted and targeted strategies with data modeling. Clinica Chimica Acta, 2017, 475, 7-14.	1.1	19
46	Clinical and molecular characterization of cystinuria in a French cohort: relevance of assessing largeâ€scale rearrangements and splicing variants. Molecular Genetics & Genomic Medicine, 2017, 5, 373-389.	1.2	22
47	PLGF, a placental marker of fetal brain defects after in utero alcohol exposure. Acta Neuropathologica Communications, 2017, 5, 44.	5.2	42
48	Methylmalonyl-CoA Epimerase Deficiency Mimicking Propionic Aciduria. International Journal of Molecular Sciences, 2017, 18, 2294.	4.1	14
49	Proteomic and transcriptomic study of brain microvessels in neonatal and adult mice. PLoS ONE, 2017, 12, e0171048.	2.5	13
50	Clinical and Molecular Characterization of Patients with Mucopolysaccharidosis Type I in an Algerian Series. International Journal of Molecular Sciences, 2016, 17, 743.	4.1	19
51	Clinical Metabolomics: The New Metabolic Window for Inborn Errors of Metabolism Investigations in the Post-Genomic Era. International Journal of Molecular Sciences, 2016, 17, 1167.	4.1	92
52	Omics-Based Strategies in Precision Medicine: Toward a Paradigm Shift in Inborn Errors of Metabolism Investigations. International Journal of Molecular Sciences, 2016, 17, 1555.	4.1	135
53	The role of metabolomics in precision medicine. Expert Review of Precision Medicine and Drug Development, 2016, 1, 517-532.	0.7	13
54	Pyridoxine-dependent epilepsy: report on three families with neuropathology. Metabolic Brain Disease, 2016, 31, 1435-1443.	2.9	23

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55	Clinical and pathologic features of Aicardiâ€“GoutiÃ¨res syndrome due to an IFIH1 mutation: A pediatric case report. American Journal of Medical Genetics, Part A, 2016, 170, 1317-1324.	1.2	14
56	Optimization of a liquid chromatography ion mobility-mass spectrometry method for untargeted metabolomics using experimental design and multivariate data analysis. Analytica Chimica Acta, 2016, 913, 55-62.	5.4	25
57	Phenotypic and Neuropathological Characterization of Fetal Pyruvate Dehydrogenase Deficiency. Journal of Neuropathology and Experimental Neurology, 2016, 75, 227-238.	1.7	145
58	Lethal Neonatal Progression of Fetal Cardiomegaly Associated to ACAD9 Deficiency. JIMD Reports, 2015, , 1-10.	1.5	11
59	A New Workflow for Proteomic Analysis of Urinary Exosomes and Assessment in Cystinuria Patients. Journal of Proteome Research, 2015, 14, 567-577.	3.7	39
60	From splitting GLUT1 deficiency syndromes to overlapping phenotypes. European Journal of Medical Genetics, 2015, 58, 443-454.	1.3	52
61	NMDA receptor blockade in the developing cortex induces autophagy-mediated death of immature cortical GABAergic interneurons: An ex vivo and in vivo study in Gad67-GFP mice. Experimental Neurology, 2015, 267, 177-193.	4.1	19
62	Adult-onset renal thrombotic microangiopathy and pulmonary arterial hypertension in cobalamin C deficiency. Lancet, The, 2015, 386, 1011-1012.	13.7	55
63	A 9-Year-Old Child with Methemoglobinemia. Clinical Chemistry, 2014, 60, 1126-1127.	3.2	1
64	3-MA Inhibits Autophagy and Favors Long-Term Integration of Grafted Gad67â€“GFP GABAergic Precursors in the Developing Neocortex by Preventing Apoptosis. Cell Transplantation, 2014, 23, 1425-1450.	2.5	9
65	Creatine and guanidinoacetate reference values in a French population. Molecular Genetics and Metabolism, 2013, 110, 263-267.	1.1	32
66	Study of LPIN1, LPIN2 and LPIN3 in rhabdomyolysis and exerciseâ€induced myalgia. Journal of Inherited Metabolic Disease, 2012, 35, 1119-1128.	3.6	75
67	Glutamine induces nuclear degradation of the NF-ÎºB p65 subunit in Caco-2/TC7 cells. Biochimie, 2012, 94, 806-815.	2.6	22
68	Screening for primary creatine deficiencies in French patients with unexplained neurological symptoms. Orphanet Journal of Rare Diseases, 2012, 7, 96.	2.7	33
69	Guidelines for the use and interpretation of assays for monitoring autophagy. Autophagy, 2012, 8, 445-544.	9.1	3,122
70	X-linked sideroblastic anemia and ataxia: A new family with identification of a fourth ABCB7 gene mutation. European Journal of Paediatric Neurology, 2012, 16, 730-735.	1.6	39
71	Fatal Rhabdomyolysis in 2 Children with LPIN1 Mutations. Journal of Pediatrics, 2012, 160, 1052-1054.	1.8	50
72	NO-dependent protective effect of VEGF against excitotoxicity on layer VI of the developing cerebral cortex. Neurobiology of Disease, 2012, 45, 871-886.	4.4	14

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73	Intestinal DMT1 Cotransporter Is Down-regulated by Hepcidin via Proteasome Internalization and Degradation. Gastroenterology, 2011, 140, 1261-1271.e1.	1.3	181
74	Hemojuvelin: A New Link Between Obesity and Iron Homeostasis. Obesity, 2011, 19, 1545-1551.	3.0	33
75	Clinical and biochemical heterogeneity associated with fumarase deficiency. Human Mutation, 2011, 32, 1046-1052.	2.5	28
76	Autophagosome maturation is impaired in Fabry disease. Autophagy, 2010, 6, 589-599.	9.1	88
77	Immunoassay for human serum hemojuvelin. Haematologica, 2010, 95, 2031-2037.	3.5	27
78	Evidence for a role of sphingosine-1 phosphate in cardiovascular remodelling in Fabry disease. European Heart Journal, 2010, 31, 67-76.	2.2	71
79	Bariatric Surgery Can Correct Iron Depletion in Morbidly Obese Women: A Link with Chronic Inflammation. Obesity Surgery, 2008, 18, 709-714.	2.1	63
80	Clinical results of enzyme replacement therapy in Fabry disease: a comprehensive review of literature. International Journal of Clinical Practice, 2007, 61, 293-302.	1.7	63
81	The Inflammatory C-Reactive Protein Is Increased in Both Liver and Adipose Tissue in Severely Obese Patients Independently from Metabolic Syndrome, Type 2 Diabetes, and NASH. American Journal of Gastroenterology, 2006, 101, 1824-1833.	0.4	162
82	Increased Adipose Tissue Expression of Hepcidin in Severe Obesity Is Independent From Diabetes and NASH. Gastroenterology, 2006, 131, 788-796.	1.3	416
83	The Role of Ceramide Trihexoside (Globotriaosylceramide) in the Diagnosis and Follow-Up of the Efficacy of Treatment of Fabry Disease: A Review of the Literature. Cardiovascular and Hematological Agents in Medicinal Chemistry, 2006, 4, 289-297.	1.0	51
84	Disruption of Autophagy at the Maturation Step by the Carcinogen Lindane Is Associated with the Sustained Mitogen-Activated Protein Kinase/Extracellular Signalâ€“Regulated Kinase Activity. Cancer Research, 2006, 66, 6861-6870.	0.9	172
85	Fabry Disease in Patients with End-Stage Renal Failure: The Potential Benefits of Screening. Nephron Clinical Practice, 2005, 101, c33-c38.	2.3	41
86	A promoter mutation in the erythroid-specific 5-aminolevulinate synthase (ALAS2) gene causes X-linked sideroblastic anemia. Blood, 2003, 102, 698-704.	1.4	51
87	Human ABC7 transporter: gene structure and mutation causing X-linked sideroblastic anemia with ataxia with disruption of cytosolic iron-sulfur protein maturation. Blood, 2000, 96, 3256-3264.	1.4	247
88	Detailed map of a region commonly amplified at 11q13â†'q14 in human breast carcinoma. Cytogenetic and Genome Research, 1997, 79, 125-131.	1.1	135
89	A 5.5-Mb High-Resolution Integrated Map of Distal 11q13. Genomics, 1997, 39, 340-347.	2.9	8
90	Increased urinary catecholamines in a hypertensive child with renal artery stenosis. Pediatric Nephrology, 1996, 10, 761-763.	1.7	1

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91	Urinary excretion of methylated catecholamine metabolites in a child with neuroblastoma maturing into ganglioneuroma. , 1996, 26, 57-60.		7
92	Expanded range of 11q13 breakpoints with differing patterns of cyclin D1 expression in B-cell malignancies. Genes Chromosomes and Cancer, 1993, 8, 80-87.	2.8	99