Majed Dasouki

List of Publications by Year
in descending order

50 papers	1,891 citations	361413 20 h-index	265206 42 g-index
51 all docs	51 docs citations	51 times ranked	3644 citing authors

#	Article	IF	Citations
1	Metabolomics toward personalized medicine. Mass Spectrometry Reviews, 2019, 38, 221-238.	5.4	229
2	DOCK8 functions as an adaptor that links TLR-MyD88 signaling to B cell activation. Nature Immunology, 2012, 13, 612-620.	14.5	205
3	The extended clinical phenotype of 64 patients with dedicator of cytokinesis 8 deficiency. Journal of Allergy and Clinical Immunology, 2015, 136, 402-412.	2.9	163
4	Effect of Oral Eliglustat on Splenomegaly in Patients With Gaucher Disease Type 1. JAMA - Journal of the American Medical Association, 2015, 313, 695.	7.4	120
5	Unbiased targeted next-generation sequencing molecular approach for primary immunodeficiency diseases. Journal of Allergy and Clinical Immunology, 2016, 137, 1780-1787.	2.9	115
6	Pompe Disease. Neurologic Clinics, 2014, 32, 751-776.	1.8	104
7	Chromosomal microarray analysis of consecutive individuals with autism spectrum disorders or learning disability presenting for genetic services. Gene, 2014, 535, 70-78.	2.2	94
8	Dedicator of cytokinesis 8â€“deficient patients have aÂbreakdown in peripheral B-cell tolerance and defectiveÂregulatory T cells. Journal of Allergy and Clinical Immunology, 2014, 134, 1365-1374.	2.9	79
9	Metabolomics Based Profiling of Dexamethasone Side Effects in Rats. Frontiers in Pharmacology, 2018, 9, 46.	3.5	75
10	Deficient T Cell Receptor Excision Circles (TRECs) in autosomal recessive hyper IgE syndrome caused by DOCK8 mutation: Implications for pathogenesis and potential detection by newborn screening. Clinical Immunology, 2011, 141, 128-132.	3.2	57
11	High Incidence of Severe Combined Immunodeficiency Disease in Saudi Arabia Detected Through Combined T Cell Receptor Excision Circle and Next Generation Sequencing of Newborn Dried Blood Spots. Frontiers in Immunology, 2018, 9, 782.	4.8	57
12	Electrochemical immunosensors for the detection of survival motor neuron (SMN) protein using different carbon nanomaterials-modified electrodes. Biosensors and Bioelectronics, 2018, 101, 282-289.	10.1	55
13	Safety and efficacy of alternative alglucosidase alfa regimens in Pompe disease. Neuromuscular Disorders, 2015, 25, 321-332.	0.6	43
14	Flow cytometry biomarkers distinguish DOCK8 deficiency from severe atopic dermatitis. Clinical Immunology, 2014, 150, 220-224.	3.2	38
15	Genetic investigation of 93 families with microphthalmia or posterior microphthalmos. Clinical Genetics, 2018, 93, 1210-1222.	2.0	38
16	Duvoglustat HCl Increases Systemic and Tissue Exposure of Active Acid Î±-Glucosidase in Pompe Patients Co-administered with Alglucosidase Î±. Molecular Therapy, 2017, 25, 1199-1208.	8.2	36
17	Dexamethasone-Induced Perturbations in Tissue Metabolomics Revealed by Chemical Isotope Labeling LC-MS Analysis. Metabolites, 2020, 10, 42.	2.9	35
18	Recurrent agnathiaâ€“otocephaly caused by DNA replication slippage in PRRX1. American Journal of Medical Genetics, Part A, 2013, 161, 803-808.	1.2	27

#	Article	IF	Citations
19	Metabolomics Distinguishes DOCK8 Deficiency from Atopic Dermatitis: Towards a Biomarker Discovery. Metabolites, 2019, 9, 274.	2.9	23
20	Multiplexed detection of DOCK8, PGM3 and STAT3 proteins for the diagnosis of Hyper-Immunoglobulin E syndrome using gold nanoparticles-based immunosensor array platform. Biosensors and Bioelectronics, 2018, 117, 613-619.	10.1	20
21	Confirmation and further delineation of the 3q26.33â€“3q27.2 microdeletion syndrome. European Journal of Medical Genetics, 2014, 57, 76-80.	1.3	18
22	Carbon nanofiber-based multiplexed immunosensor for the detection of survival motor neuron 1, cystic fibrosis transmembrane conductance regulator and Duchenne Muscular Dystrophy proteins. Biosensors and Bioelectronics, 2018, 117, 84-90.	10.1	18
23	TREC and KREC profiling as a representative of thymus and bone marrow output in patients with various inborn errors of immunity. Clinical and Experimental Immunology, 2020, 202, 60-71.	2.6	18
24	Hematopoietic stem cell transplantation corrects WIP deficiency. Journal of Allergy and Clinical Immunology, 2017, 139, 1039-1040.e4.	2.9	17
25	Dried Blood Spot-Based Metabolomic Profiling in Adults with Cystic Fibrosis. Journal of Proteome Research, 2020, 19, 2346-2357.	3.7	17
26	Mutational profile and genotype/phenotype correlation of non-familial pheochromocytoma and paraganglioma. Oncotarget, 2019, 10, 5919-5931.	1.8	17
27	Lipidome Alterations Induced by Cystic Fibrosis, CFTR Mutation, and Lung Function. Journal of Proteome Research, 2021, 20, 549-564.	3.7	16
28	Distinctive metabolic profiles between Cystic Fibrosis mutational subclasses and lung function. Metabolomics, 2021, 17, 4.	3.0	15
29	Congenital Arthrogryposis: An Extension of the 15q11.2 BP1-BP2 Microdeletion Syndrome?. Case Reports in Genetics, 2014, 2014, 1-3.	0.2	13
30	Hypomorphic variants in AK2 reveal the contribution of mitochondrial function to B-cell activation. Journal of Allergy and Clinical Immunology, 2020, 146, 192-202.	2.9	13
31	Serum-Based Proteomics Profiling in Adult Patients with Cystic Fibrosis. International Journal of Molecular Sciences, 2020, 21, 7415.	4.1	12
32	Phenotypic variation of autosomal recessive pseudohypoaldosteronism type I: a case in point. Clinical Case Reports (discontinued), 2014, 2, 326-330.	0.5	11
33	Quantitative profiling of cytokines and chemokines in <scp>DOCK</scp>8â€deficient and atopic dermatitis patients. Allergy: European Journal of Allergy and Clinical Immunology, 2019, 74, 370-379.	5.7	11
34	A novel claudin-10 mutation with a unique mechanism in two unrelated families with HELIX syndrome. Kidney International, 2021, 100, 415-429.	5.2	11
35	Clinical Report of a 17q12 Microdeletion with Additionally Unreported Clinical Features. Case Reports in Genetics, 2014, 2014, 1-6.	0.2	10
36	Selection, characterization, and electrochemical biosensing application of DNA aptamers for sepiapterin. Talanta, 2020, 216, 120951.	5.5	9

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37	Electrochemical Immunosensors for the Rapid Screening of Cystic Fibrosis and Duchenne Muscular Dystrophy. Electroanalysis, 2017, 29, 1911-1917.	2.9	8
38	Comprehensive multi-omics analysis of G6PC3 deficiency-related congenital neutropenia with inflammatory bowel disease. IScience, 2021, 24, 102214.	4.1	7
39	A Clinical, Genomic and Proteomic Approach for the Characterization of Fanconi Anemia in Adolescent and Young Adult (AYA) Patients : A Single Center Study of 55 Patients from a National Bone Marrow Failure Referral Center. Blood, 2018, 132, 2594-2594.	1.4	7
40	Metabolomics Profiling of Cystic Renal Disease towards Biomarker Discovery. Biology, 2021, 10, 770.	2.8	6
41	THPOâ€“MPL pathway and bone marrow failure. Hematology/ Oncology and Stem Cell Therapy, 2015, 8, 6-9.	0.9	5
42	HSP and deafness. Neurology: Genetics, 2017, 3, e151.	1.9	5
43	Targeted Metabolomic Profiling of Total Fatty Acids in Human Plasma by Liquid Chromatography-Tandem Mass Spectrometry. Metabolites, 2020, 10, 400.	2.9	5
44	Development of Impedimetric Immunosensors for the Diagnosis of DOCK8 and STAT3 Related Hyperâ€Immunoglobulin E Syndrome. Electroanalysis, 2018, 30, 2021-2027.	2.9	2
45	Molecular classification of blood and bleeding disorder genes. Npj Genomic Medicine, 2021, 6, 62.	3.8	2
46	Proteomics Profiling to Distinguish DOCK8 Deficiency From Atopic Dermatitis. Frontiers in Allergy, 2021, 2, 774902.	2.8	2
47	Inborn Errors of Mitochondrial Fatty Acid Oxidation. , 0, , 767-802.		1
48	Novel human pathological mutations. Human Genetics, 2010, 127, 109-124.	3.8	1
49	Novel human pathological mutations. Gene symbol: DMD. Disease: Muscular Dystrophy, Duchenne. Human Genetics, 2010, 127, 109.	3.8	1
50	Familial emberger syndrome with autoimmunity, hyper-immunoglobulin E and lymphatic impairment caused by a novel GATA2 mutation. Hematology/ Oncology and Stem Cell Therapy, 2020, , .	0.9	0