Ammar Husami

List of Publications by Year
in descending order

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Version: 2024-02-01

35 papers	1,911 citations	430754 18 h-index	477173 29 g-index
38 all docs	38 docs citations	38 times ranked	3936 citing authors

#	Article	IF	Citations
1	vcf2fhir: a utility to convert VCF files into HL7 FHIR format for genomics-EHR integration. BMC Bioinformatics, 2021, 22, 104.	1.2	11
2	Congenital dyserythropoietic anemia type I: First report from the Congenital Dyserythropoietic Anemia Registry of North America (CDAR). Blood Cells, Molecules, and Diseases, 2021, 87, 102534.	0.6	3
3	Deleterious Variants in ABCC12 are Detected in Idiopathic Chronic Cholestasis and Cause Intrahepatic Bile Duct Loss in Model Organisms. Gastroenterology, 2021, 161, 287-300.e16.	0.6	12
4	GA4GH: International policies and standards for data sharing across genomic research and healthcare. Cell Genomics, 2021, 1, 100029.	3.0	94
5	Clinical utility of whole genome sequencing for the detection of mitochondrial genome mutations. Journal of Genetics and Genomics, 2020, 47, 167-169.	1.7	8
6	Systemic Juvenile Idiopathic Arthritisâ€“Associated Lung Disease: Characterization and Risk Factors. Arthritis and Rheumatology, 2019, 71, 1943-1954.	2.9	124
7	Partial growth hormone insensitivity and dysregulatory immune disease associated with de novo germline activating STAT3 mutations. Molecular and Cellular Endocrinology, 2018, 473, 166-177.	1.6	38
8	Brief Report: Novel UNC13D Intronic Variant Disrupting an NFâ€ºB Enhancer in a Patient With Recurrent Macrophage Activation Syndrome and Systemic Juvenile Idiopathic Arthritis. Arthritis and Rheumatology, 2018, 70, 963-970.	2.9	30
9	A comprehensive nextâ€generation sequencing assay for the diagnosis of epidermolysis bullosa. Pediatric Dermatology, 2018, 35, 188-197.	0.5	29
10	Prevalence of abnormal glucose metabolism in pediatric acute, acute recurrent and chronic pancreatitis. PLoS ONE, 2018, 13, e0204979.	1.1	12
11	Screening for Wiskott-Aldrich syndrome by flow cytometry. Journal of Allergy and Clinical Immunology, 2018, 142, 333-335.e8.	1.5	20
12	Dominant-negative IKZF1 mutations cause a T, B, and myeloid cell combined immunodeficiency. Journal of Clinical Investigation, 2018, 128, 3071-3087.	3.9	133
13	Peroxiredoxin II (PRDX2) Is a Novel Candidate Gene for Congenital Dyserythropoietic Anemia. Blood, 2018, 132, 3605-3605.	0.6	4
14	Concordance between Research Sequencing and Clinical Pharmacogenetic Genotyping in the eMERGE-PGx Study. Journal of Molecular Diagnostics, 2017, 19, 561-566.	1.2	18
15	Clinical delineation of the PACS1 â€related syndromeâ€”Report on 19 patients. American Journal of Medical Genetics, Part A, 2016, 170, 670-675.	0.7	44
16	Whole-Exome Sequencing Reveals Mutations in Genes Linked to Hemophagocytic Lymphohistiocytosis and Macrophage Activation Syndrome in Fatal Cases of H1N1 Influenza. Journal of Infectious Diseases, 2016, 213, 1180-1188.	1.9	133
17	Deep Sequencing Reveals Novel Genetic Variants in Children with Acute Liver Failure and Tissue Evidence of Impaired Energy Metabolism. PLoS ONE, 2016, 11, e0156738.	1.1	11
18	Clinical Impact and Cost-Effectiveness of Whole Exome Sequencing as a Diagnostic Tool: A Pediatric Centerâ€™s Experience. Frontiers in Pediatrics, 2015, 3, 67.	0.9	159

#	Article	IF	Citations
19	Patients with LRBA deficiency show CTLA4 loss and immune dysregulation responsive to abatacept therapy. Science, 2015, 349, 436-440.	6.0	580
20	The struggle to find reliable results in exome sequencing data: filtering out Mendelian errors. Frontiers in Genetics, 2014, 5, 16.	1.1	51
21	Wholeâ€Exome Sequencing Reveals Overlap Between Macrophage Activation Syndrome in Systemic Juvenile Idiopathic Arthritis and Familial Hemophagocytic Lymphohistiocytosis. Arthritis and Rheumatology, 2014, 66, 3486-3495.	2.9	158
22	The 253â€kb inversion and deep intronic mutations in UNC13D are present in North American patients with familial hemophagocytic lymphohistiocytosis 3. Pediatric Blood and Cancer, 2014, 61, 1034-1040.	0.8	33
23	Synergistic defects of different molecules in the cytotoxic pathway lead to clinical familial hemophagocytic lymphohistiocytosis. Blood, 2014, 124, 1331-1334.	0.6	115
24	Performance Evaluation of the Next-Generation Sequencing Approach for Molecular Diagnosis of Hereditary Hearing Loss. Otolaryngology - Head and Neck Surgery, 2013, 148, 1007-1016.	1.1	36
25	Next Generation Sequencing Technologies in Medical Genetics. SpringerBriefs in Genetics, 2013, , .	0.1	3
26	A Review of DNA Enrichment Technologies. SpringerBriefs in Genetics, 2013, , 25-32.	0.1	1
27	Development Of a Comprehensive Rapid Next-Generation Sequencing Assay For The Diagnosis Of Inherited Hemolytic Anemia. Blood, 2013, 122, 949-949.	0.6	1
28	Application of Next-Generationâ€“Sequencing to the Diagnosis of Genetic Disorders: A Brief Overview. SpringerBriefs in Genetics, 2013, , 35-43.	0.1	0
29	Exome Sequencing as a Discovery and Diagnostic Tool. SpringerBriefs in Genetics, 2013, , 75-86.	0.1	0
30	Diagnosis of Inherited Neuromuscular Disorders by Next-Generationâ€“Sequencing. SpringerBriefs in Genetics, 2013, , 57-65.	0.1	0
31	A Survey of Next-Generationâ€“Sequencing Technologies. SpringerBriefs in Genetics, 2013, , 13-24.	0.1	0
32	Application of Next-Generationâ€“Sequencing in Hearing Loss Diagnosis. SpringerBriefs in Genetics, 2013, , 67-74.	0.1	0
33	Sanger Sequencing Principles, History, and Landmarks. SpringerBriefs in Genetics, 2013, , 3-11.	0.1	3
34	Next Generation Sequencing for Diagnostic Testing of Erythrocyte Cytoskeleton Disorders. Blood, 2012, 120, 976-976.	0.6	0
35	High-throughput detection of mutations responsible for childhood hearing loss using resequencing microarrays. BMC Biotechnology, 2010, 10, 10.	1.7	47