And Naif Am Almontashiri

List of Publications by Year
in descending order

41 papers	1,188 citations	516215 16 h-index	414034 32 g-index
47 all docs	47 docs citations	47 times ranked	2609 citing authors

#	Article	IF	Citations
1	SARS-CoV-2 genomes from Saudi Arabia implicate nucleocapsid mutations in host response and increased viral load. Nature Communications, 2022, 13, 601.	5.8	40
2	OUP accepted manuscript. Clinical Chemistry, 2022, 68, 633.	1.5	0
3	A Biallelic Variant in FRA10AC1 Is Associated With Neurodevelopmental Disorder and Growth Retardation. Neurology: Genetics, 2022, 8, e200010.	0.9	2
4	New paradigms of USP53 disease: normal GGT cholestasis, BRIC, cholangiopathy, and responsiveness to rifampicin. Journal of Human Genetics, 2021, 66, 151-159.	1.1	21
5	Biallelic loss of function variant in the unfolded protein response gene PDIA6 is associated with asphyxiating thoracic dystrophy and neonatalâ€onset diabetes. Clinical Genetics, 2021, 99, 694-703.	1.0	9
6	A Robust, Safe, and Scalable Magnetic Nanoparticle Workflow for RNA Extraction of Pathogens from Clinical and Wastewater Samples. Global Challenges, 2021, 5, 2000068.	1.8	10
7	Quick and Easy Assembly of a One-Step qRT-PCR Kit for COVID-19 Diagnostics Using In-House Enzymes. ACS Omega, 2021, 6, 7374-7386.	1.6	5
8	Progressive Ataxia and Neurologic Regression in RFXANK-Associated Bare Lymphocyte Syndrome. Neurology: Genetics, 2021, 7, e586.	0.9	4
9	A Child with Progressive Hypertrophic Cardiomyopathy and Lactic Acidosis. Clinical Chemistry, 2021, 67, 912-914.	1.5	0
10	The Leukodystrophy Spectrum in Saudi Arabia: Epidemiological, Clinical, Radiological, and Genetic Data. Frontiers in Pediatrics, 2021, 9, 633385.	0.9	15
11	Simultaneous detection and mutation surveillance of SARS-CoV-2 and multiple respiratory viruses by rapid field-deployable sequencing. Med, 2021, 2, 689-700.e4.	2.2	16
12	A discarded synonymous variant in NPHP3 explains nephronophthisis and congenital hepatic fibrosis in several families. Human Mutation, 2021, 42, 1221-1228.	1.1	12
13	Clinical characterization and further confirmation of the autosomal recessive SLC12A2 disease. Journal of Human Genetics, 2021, 66, 689-695.	1.1	5
14	Hyperammonemia, Lactic Acidosis, and Arrhythmia in a Newborn. Clinical Chemistry, 2021, 67, 327-330.	1.5	0
15	Clinical, Biochemical, and Molecular Characterization of Neonatal-Onset Dubinâ€"Johnson Syndrome in a Large Case Series From the Arabs. Frontiers in Pediatrics, 2021, 9, 741835.	0.9	3
16	Phenotypic delineation of the retinal arterial macroaneurysms with supravalvular pulmonic stenosis syndrome. Clinical Genetics, 2020, 97, 447-456.	1.0	7
17	iSCAN: An RT-LAMP-coupled CRISPR-Cas12 module for rapid, sensitive detection of SARS-CoV-2. Virus Research, 2020, 288, 198129.	1.1	226
18	SARS-CoV-2 S1 and N-based serological assays reveal rapid seroconversion and induction of specific antibody response in COVID-19 patients. Scientific Reports, 2020, 10, 16561.	1.6	84

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19	Clinical, molecular, and biochemical delineation of asparagine synthetase deficiency in Saudi cohort. Genetics in Medicine, 2020, 22, 2071-2080.	1.1	7
20	NEMF mutations that impair ribosome-associated quality control are associated with neuromuscular disease. Nature Communications, 2020, 11, 4625.	5.8	47
21	Performance of Commercially Available Rapid Serological Assays for the Detection of SARS-CoV-2 Antibodies. Pathogens, 2020, 9, 1067.	1.2	7
22	Early Humoral Response Correlates with Disease Severity and Outcomes in COVID-19 Patients. Viruses, 2020, 12, 1390.	1.5	42
23	Clinical Validation of Targeted and Untargeted Metabolomics Testing for Genetic Disorders: A 3 Year Comparative Study. Scientific Reports, 2020, 10, 9382.	1.6	24
24	An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. Human Mutation, 2020, 41, 1577-1587.	1.1	10
25	Metabolic Acidosis and Hypoglycemia in a Child with Leigh-Like Phenotype. Clinical Chemistry, 2020, 66, 739-741.	1.5	0
26	Lessons Learned from Large-Scale, First-Tier Clinical Exome Sequencing in a Highly Consanguineous Population. American Journal of Human Genetics, 2019, 104, 1182-1201.	2.6	184
27	Serine Deficiency in a Child with Neurological Presentation, Hearing Loss, and Multiple Congenital Anomalies. Clinical Chemistry, 2018, 64, 870-872.	1.5	1
28	Recurrent variants in OTOF are significant contributors to prelingual nonsydromic hearing loss in Saudi patients. Genetics in Medicine, 2018, 20, 536-544.	1.1	18
29	Abnormal Glycerol Metabolism in a Child with Global Developmental Delay, Adrenal Insufficiency, and Intellectual Disability. Clinical Chemistry, 2018, 64, 1785-1787.	1.5	1
30	Hyperammonemia in a Child Presenting with Growth Delay, Short Stature, and Diarrhea. Clinical Chemistry, 2018, 64, 1260-1262.	1.5	1
31	The 9p21.3 risk locus for coronary artery disease: A 10-year search for its mechanism. Journal of Taibah University Medical Sciences, 2017, 12, 199-204.	0.5	8
32	Multiplexed Reference Materials as Controls for Diagnostic Next-Generation Sequencing. Journal of Molecular Diagnostics, 2016, 18, 882-889.	1.2	13
33	The need for population-based studies to estimate the rate of consanguinity in Almadinah Almunawwarah. Journal of Taibah University Medical Sciences, 2015, 10, 509-511.	0.5	0
34	Usefulness of genome-wide association studies to identify novel genetic variants underlying the plasma lipoprotein metabolism as risk factors for CAD. Journal of Taibah University Medical Sciences, 2015, 10, 266-270.	0.5	1
35	IRF2BP2 Reduces Macrophage Inflammation and Susceptibility to Atherosclerosis. Circulation Research, 2015, 117, 671-683.	2.0	64
36	9p21.3 Coronary Artery Disease Risk Variants Disrupt TEAD Transcription Factorâ€“Dependent Transforming Growth Factor Î² Regulation of p16 Expression in Human Aortic Smooth Muscle Cells. Circulation, 2015, 132, 1969-1978.	1.6	47

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37	Plasma PCSK9 Levels Are Elevated with Acute Myocardial Infarction in Two Independent Retrospective Angiographic Studies. PLoS ONE, 2014, 9, e106294.	1.1	75
38	SPG7 Variant Escapes Phosphorylation-Regulated Processing by AFG3L2, Elevates Mitochondrial ROS, and Is Associated with Multiple Clinical Phenotypes. Cell Reports, 2014, 7, 834-847.	2.9	39
39	Functional Genomics of the 9p21.3 Locus for Atherosclerosis: Clarity or Confusion?. Current Cardiology Reports, 2014, 16, 502.	1.3	39
40	Interferon-Î³ Activates Expression of p15 and p16 Regardless of 9p21.3 Coronary Artery Disease Risk Genotype. Journal of the American College of Cardiology, 2013, 61, 143-147.	1.2	37
41	Identification of a Phosphorylation-Dependent Nuclear Localization Motif in Interferon Regulatory Factor 2 Binding Protein 2. PLoS ONE, 2011, 6, e24100.	1.1	21