

# And Naif Am Almontashiri

## List of Publications by Citations

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41  
papers

654  
citations

14  
h-index

25  
g-index

47  
ext. papers

956  
ext. citations

6.5  
avg, IF

3.62  
L-index

#	Paper	IF	Citations
41	iSCAN: An RT-LAMP-coupled CRISPR-Cas12 module for rapid, sensitive detection of SARS-CoV-2. <i>Virus Research</i> , <b>2020</b> , 288, 198129	6.4	102
40	Lessons Learned from Large-Scale, First-Tier Clinical Exome Sequencing in a Highly Consanguineous Population. <i>American Journal of Human Genetics</i> , <b>2019</b> , 104, 1182-1201	11	95
39	Plasma PCSK9 levels are elevated with acute myocardial infarction in two independent retrospective angiographic studies. <i>PLoS ONE</i> , <b>2014</b> , 9, e106294	3.7	57
38	SARS-CoV-2 S1 and N-based serological assays reveal rapid seroconversion and induction of specific antibody response in COVID-19 patients. <i>Scientific Reports</i> , <b>2020</b> , 10, 16561	4.9	51
37	IRF2BP2 Reduces Macrophage Inflammation and Susceptibility to Atherosclerosis. <i>Circulation Research</i> , <b>2015</b> , 117, 671-83	15.7	46
36	Functional genomics of the 9p21.3 locus for atherosclerosis: clarity or confusion?. <i>Current Cardiology Reports</i> , <b>2014</b> , 16, 502	4.2	36
35	Interferon- $\beta$ activates expression of p15 and p16 regardless of 9p21.3 coronary artery disease risk genotype. <i>Journal of the American College of Cardiology</i> , <b>2013</b> , 61, 143-7	15.1	32
34	9p21.3 Coronary Artery Disease Risk Variants Disrupt TEAD Transcription Factor-Dependent Transforming Growth Factor $\beta$ Regulation of p16 Expression in Human Aortic Smooth Muscle Cells. <i>Circulation</i> , <b>2015</b> , 132, 1969-78	16.7	31
33	SPG7 variant escapes phosphorylation-regulated processing by AFG3L2, elevates mitochondrial ROS, and is associated with multiple clinical phenotypes. <i>Cell Reports</i> , <b>2014</b> , 7, 834-47	10.6	29
32	Early Humoral Response Correlates with Disease Severity and Outcomes in COVID-19 Patients. <i>Viruses</i> , <b>2020</b> , 12,	6.2	21
31	NEMF mutations that impair ribosome-associated quality control are associated with neuromuscular disease. <i>Nature Communications</i> , <b>2020</b> , 11, 4625	17.4	21
30	Identification of a phosphorylation-dependent nuclear localization motif in interferon regulatory factor 2 binding protein 2. <i>PLoS ONE</i> , <b>2011</b> , 6, e24100	3.7	17
29	Recurrent variants in OTOF are significant contributors to prelingual nonsyndromic hearing loss in Saudi patients. <i>Genetics in Medicine</i> , <b>2018</b> , 20, 536-544	8.1	14
28	New paradigms of USP53 disease: normal GGT cholestasis, BRIC, cholangiopathy, and responsiveness to rifampicin. <i>Journal of Human Genetics</i> , <b>2021</b> , 66, 151-159	4.3	12
27	Clinical Validation of Targeted and Untargeted Metabolomics Testing for Genetic Disorders: A 3 Year Comparative Study. <i>Scientific Reports</i> , <b>2020</b> , 10, 9382	4.9	10
26	Multiplexed Reference Materials as Controls for Diagnostic Next-Generation Sequencing: A Pilot Investigating Applications for Hypertrophic Cardiomyopathy. <i>Journal of Molecular Diagnostics</i> , <b>2016</b> , 18, 882-889	5.1	10
25	Simultaneous detection and mutation surveillance of SARS-CoV-2 and multiple respiratory viruses by rapid field-deployable sequencing. <i>Med</i> , <b>2021</b> , 2, 689-700.e4	31.7	7

24	A Robust, Safe, and Scalable Magnetic Nanoparticle Workflow for RNA Extraction of Pathogens from Clinical and Wastewater Samples. <i>Global Challenges</i> , <b>2021</b> , 5, 2000068	4.3	6
23	The 9p21.3 risk locus for coronary artery disease: A 10-year search for its mechanism. <i>Journal of Taibah University Medical Sciences</i> , <b>2017</b> , 12, 199-204	1.7	5
22	An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. <i>Human Mutation</i> , <b>2020</b> , 41, 1577-1587	4.7	4
21	SARS-CoV-2 genomes from Saudi Arabia implicate nucleocapsid mutations in host response and increased viral load.. <i>Nature Communications</i> , <b>2022</b> , 13, 601	17.4	4
20	Phenotypic delineation of the retinal arterial macroaneurysms with supra-avalvular pulmonic stenosis syndrome. <i>Clinical Genetics</i> , <b>2020</b> , 97, 447-456	4	4
19	Performance of Commercially Available Rapid Serological Assays for the Detection of SARS-CoV-2 Antibodies. <i>Pathogens</i> , <b>2020</b> , 9,	4.5	3
18	A discarded synonymous variant in NPHP3 explains nephronophthisis and congenital hepatic fibrosis in several families. <i>Human Mutation</i> , <b>2021</b> , 42, 1221-1228	4.7	3
17	Biallelic loss of function variant in the unfolded protein response gene PDIA6 is associated with asphyxiating thoracic dystrophy and neonatal-onset diabetes. <i>Clinical Genetics</i> , <b>2021</b> , 99, 694-703	4	3
16	Early Humoral Response Correlates with Disease Severity and Outcomes in COVID-19 Patients		2
15	Quick and Easy Assembly of a One-Step qRT-PCR Kit for COVID-19 Diagnostics Using In-House Enzymes. <i>ACS Omega</i> , <b>2021</b> , 6, 7374-7386	3.9	2
14	The Leukodystrophy Spectrum in Saudi Arabia: Epidemiological, Clinical, Radiological, and Genetic Data. <i>Frontiers in Pediatrics</i> , <b>2021</b> , 9, 633385	3.4	2
13	Usefulness of genome-wide association studies to identify novel genetic variants underlying the plasma lipoprotein metabolism as risk factors for CAD. <i>Journal of Taibah University Medical Sciences</i> , <b>2015</b> , 10, 266-270	1.7	1
12	Serine Deficiency in a Child with Neurological Presentation, Hearing Loss, and Multiple Congenital Anomalies. <i>Clinical Chemistry</i> , <b>2018</b> , 64, 870-872	5.5	1
11	Hyperammonemia in a Child Presenting with Growth Delay, Short Stature, and Diarrhea. <i>Clinical Chemistry</i> , <b>2018</b> , 64, 1260-1262	5.5	1
10	Clinical, Biochemical, and Molecular Characterization of Neonatal-Onset Dubin-Johnson Syndrome in a Large Case Series From the Arabs. <i>Frontiers in Pediatrics</i> , <b>2021</b> , 9, 741835	3.4	1
9	Abnormal Glycerol Metabolism in a Child with Global Developmental Delay, Adrenal Insufficiency, and Intellectual Disability. <i>Clinical Chemistry</i> , <b>2018</b> , 64, 1785-1787	5.5	1
8	Clinical characterization and further confirmation of the autosomal recessive SLC12A2 disease. <i>Journal of Human Genetics</i> , <b>2021</b> , 66, 689-695	4.3	1
7	Clinical, molecular, and biochemical delineation of asparagine synthetase deficiency in Saudi cohort. <i>Genetics in Medicine</i> , <b>2020</b> , 22, 2071-2080	8.1	0

- 6 Progressive Ataxia and Neurologic Regression in -Associated Bare Lymphocyte Syndrome. *Neurology: Genetics*, **2021**, 7, e586 3.8 0
- 5 Metabolic Acidosis and Hypoglycemia in a Child with Leigh-Like Phenotype. *Clinical Chemistry*, **2020**, 66, 739-741 5.5
- 4 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 1.7
- 3 Hyperammonemia, Lactic Acidosis, and Arrhythmia in a Newborn. *Clinical Chemistry*, **2021**, 67, 327-330 5.5
- 2 A Child with Progressive Hypertrophic Cardiomyopathy and Lactic Acidosis.. *Clinical Chemistry*, **2021**, 67, 912-914 5.5
- 1 Commentary on Multiple Copy Number Variants Detected by Noninvasive Prenatal Screening.. *Clinical Chemistry*, **2022**, 68, 633 5.5