And Naif Am Almontashiri

List of Publications by Year in Descending Order

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	SARS-CoV-2 genomes from Saudi Arabia implicate nucleocapsid mutations in host response and increased viral load.. *Nature Communications*, **2022**, 13, 601	17.4	4
40	Commentary on Multiple Copy Number Variants Detected by Noninvasive Prenatal Screening.. *Clinical Chemistry*, **2022**, 68, 633	5.5	
39	Hyperammonemia, Lactic Acidosis, and Arrhythmia in a Newborn. *Clinical Chemistry*, **2021**, 67, 327-330	5.5	
38	Clinical, Biochemical, and Molecular Characterization of Neonatal-Onset Dubin-Johnson Syndrome in a Large Case Series From the Arabs. *Frontiers in Pediatrics*, **2021**, 9, 741835	3.4	1
37	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	3.9	2
36	Progressive Ataxia and Neurologic Regression in -Associated Bare Lymphocyte Syndrome. *Neurology: Genetics*, **2021**, 7, e586	3.8	0
35	A Child with Progressive Hypertrophic Cardiomyopathy and Lactic Acidosis.. *Clinical Chemistry*, **2021**, 67, 912-914	5.5	
34	The Leukodystrophy Spectrum in Saudi Arabia: Epidemiological, Clinical, Radiological, and Genetic Data. *Frontiers in Pediatrics*, **2021**, 9, 633385	3.4	2
33	Simultaneous detection and mutation surveillance of SARS-CoV-2 and multiple respiratory viruses by rapid field-deployable sequencing. *Med*, **2021**, 2, 689-700.e4	31.7	7
32	A discarded synonymous variant in NPHP3 explains nephronophthisis and congenital hepatic fibrosis in several families. *Human Mutation*, **2021**, 42, 1221-1228	4.7	3
31	New paradigms of USP53 disease: normal GGT cholestasis, BRIC, cholangiopathy, and responsiveness to rifampicin. *Journal of Human Genetics*, **2021**, 66, 151-159	4.3	12
30	Biallelic loss of function variant in the unfolded protein response gene PDIA6 is associated with asphyxiating thoracic dystrophy and neonatal-onset diabetes. *Clinical Genetics*, **2021**, 99, 694-703	4	3
29	A Robust, Safe, and Scalable Magnetic Nanoparticle Workflow for RNA Extraction of Pathogens from Clinical and Wastewater Samples. *Global Challenges*, **2021**, 5, 2000068	4.3	6
28	Clinical characterization and further confirmation of the autosomal recessive SLC12A2 disease. *Journal of Human Genetics*, **2021**, 66, 689-695	4.3	1
27	Early Humoral Response Correlates with Disease Severity and Outcomes in COVID-19 Patients. *Viruses*, **2020**, 12,	6.2	21
26	Clinical Validation of Targeted and Untargeted Metabolomics Testing for Genetic Disorders: A 3 Year Comparative Study. *Scientific Reports*, **2020**, 10, 9382	4.9	10
25	An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. *Human Mutation*, **2020**, 41, 1577-1587	4.7	4

24	Metabolic Acidosis and Hypoglycemia in a Child with Leigh-Like Phenotype. *Clinical Chemistry*, **2020**, 66, 739-741	5.5	
23	Phenotypic delineation of the retinal arterial macroaneurysms with supravalvular pulmonic stenosis syndrome. *Clinical Genetics*, **2020**, 97, 447-456	4	4
22	iSCAN: An RT-LAMP-coupled CRISPR-Cas12 module for rapid, sensitive detection of SARS-CoV-2. *Virus Research*, **2020**, 288, 198129	6.4	102
21	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	4.9	51
20	Clinical, molecular, and biochemical delineation of asparagine synthetase deficiency in Saudi cohort. *Genetics in Medicine*, **2020**, 22, 2071-2080	8.1	0
19	NEMF mutations that impair ribosome-associated quality control are associated with neuromuscular disease. *Nature Communications*, **2020**, 11, 4625	17.4	21
18	Performance of Commercially Available Rapid Serological Assays for the Detection of SARS-CoV-2 Antibodies. *Pathogens*, **2020**, 9,	4.5	3
17	Lessons Learned from Large-Scale, First-Tier Clinical Exome Sequencing in a Highly Consanguineous Population. *American Journal of Human Genetics*, **2019**, 104, 1182-1201	11	95
16	Serine Deficiency in a Child with Neurological Presentation, Hearing Loss, and Multiple Congenital Anomalies. *Clinical Chemistry*, **2018**, 64, 870-872	5.5	1
15	Recurrent variants in OTOF are significant contributors to prelingual nonsydromic hearing loss in Saudi patients. *Genetics in Medicine*, **2018**, 20, 536-544	8.1	14
14	Hyperammonemia in a Child Presenting with Growth Delay, Short Stature, and Diarrhea. *Clinical Chemistry*, **2018**, 64, 1260-1262	5.5	1
13	Abnormal Glycerol Metabolism in a Child with Global Developmental Delay, Adrenal Insufficiency, and Intellectual Disability. *Clinical Chemistry*, **2018**, 64, 1785-1787	5.5	1
12	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	1.7	5
11	Multiplexed Reference Materials as Controls for Diagnostic Next-Generation Sequencing: A Pilot Investigating Applications for Hypertrophic Cardiomyopathy. *Journal of Molecular Diagnostics*, **2016**, 18, 882-889	5.1	10
10	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	1.7	1
9	IRF2BP2 Reduces Macrophage Inflammation and Susceptibility to Atherosclerosis. *Circulation Research*, **2015**, 117, 671-83	15.7	46
8	9p21.3 Coronary Artery Disease Risk Variants Disrupt TEAD Transcription Factor-Dependent Transforming Growth Factor Regulation of p16 Expression in Human Aortic Smooth Muscle Cells. *Circulation*, **2015**, 132, 1969-78	16.7	31
7	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	

6	SPG7 variant escapes phosphorylation-regulated processing by AFG3L2, elevates mitochondrial ROS, and is associated with multiple clinical phenotypes. *Cell Reports*, **2014**, 7, 834-47	10.6	29
5	Functional genomics of the 9p21.3 locus for atherosclerosis: clarity or confusion?. *Current Cardiology Reports*, **2014**, 16, 502	4.2	36
4	Plasma PCSK9 levels are elevated with acute myocardial infarction in two independent retrospective angiographic studies. *PLoS ONE*, **2014**, 9, e106294	3.7	57
3	Interferon- activates expression of p15 and p16 regardless of 9p21.3 coronary artery disease risk genotype. *Journal of the American College of Cardiology*, **2013**, 61, 143-7	15.1	32
2	Identification of a phosphorylation-dependent nuclear localization motif in interferon regulatory factor 2 binding protein 2. *PLoS ONE*, **2011**, 6, e24100	3.7	17
1	Early Humoral Response Correlates with Disease Severity and Outcomes in COVID-19 Patients		2