Alya Qari

List of Publications by Year
in descending order

12	701	1163117 8	1281871 11
papers	citations	h-index	g-index

12	12	12	1870
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Novel pathogenic MAPKBP1 variant in a family with nephronophthisis. CKJ: Clinical Kidney Journal, 2021, 14, 728-730.	2.9	0
2	Familial/inherited cancer syndrome: a focus on the highly consanguineous Arab population. Npj Genomic Medicine, 2020, 5, 3.	3.8	24
3	The morbid genome of ciliopathies: an update. Genetics in Medicine, 2020, 22, 1051-1060.	2.4	68
4	Lessons Learned from Large-Scale, First-Tier Clinical Exome Sequencing in a Highly Consanguineous Population. American Journal of Human Genetics, 2019, 104, 1182-1201.	6.2	184
5	The many faces of peroxisomal disorders: Lessons from a large Arab cohort. Clinical Genetics, 2019, 95, 310-319.	2.0	12
6	Autozygome and high throughput confirmation of disease genes candidacy. Genetics in Medicine, 2019, 21, 736-742.	2.4	81
7	Molecular autopsy in maternalâ€“fetal medicine. Genetics in Medicine, 2018, 20, 420-427.	2.4	84
8	Juvenile idiopathic arthritis in multiplex families: longitudinal follow-up. International Journal of Rheumatic Diseases, 2017, 20, 898-902.	1.9	5
9	The landscape of genetic diseases in Saudi Arabia based on the first 1000 diagnostic panels and exomes. Human Genetics, 2017, 136, 921-939.	3.8	209
10	Clinical, Endocrine, and Molecular Genetic Analysis of a Large Cohort of Saudi Arabian Patients with Laron Syndrome. Hormone Research in Paediatrics, 2017, 88, 119-126.	1.8	4
11	KCNA4 deficiency leads to a syndrome of abnormal striatum, congenital cataract and intellectual disability. Journal of Medical Genetics, 2016, 53, 786-792.	3.2	24
12	Spectrum of Mutations in 60 Saudi Patients with Mut Methylmalonic Acidemia. JIMD Reports, 2014, 29, 39-46.	1.5	6