

# Anwar Baban

## List of Publications by Year in descending order

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Version: 2024-02-01

82  
papers

1,967  
citations

279798

23  
h-index

289244

40  
g-index

85  
all docs

85  
docs citations

85  
times ranked

3457  
citing authors

#	ARTICLE	IF	CITATIONS
1	Coffin-Siris Syndrome and the BAF Complex: Genotype-Phenotype Study in 63 Patients. <i>Human Mutation</i> , 2013, 34, 1519-1528.	2.5	178
2	Development of a Novel Risk Prediction Model for Sudden Cardiac Death in Childhood Hypertrophic Cardiomyopathy (HCM Risk-Kids). <i>JAMA Cardiology</i> , 2019, 4, 918.	6.1	147
3	Recurrent microdeletion at 17q12 as a cause of Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome: two case reports. <i>Orphanet Journal of Rare Diseases</i> , 2009, 4, 25.	2.7	105
4	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffin-Siris syndrome. <i>Genetics in Medicine</i> , 2019, 21, 1295-1307.	2.4	80
5	Cardiac defects, morbidity and mortality in patients affected by RASopathies. CARNET study results. <i>International Journal of Cardiology</i> , 2017, 245, 92-98.	1.7	75
6	RASopathies: Clinical Diagnosis in the First Year of Life. <i>Molecular Syndromology</i> , 2010, 1, 282-289.	0.8	73
7	Poland syndrome with bilateral features: Case description with review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 1597-1602.	1.2	62
8	Human B-cell memory is shaped by age- and tissue-specific T-independent and GC-dependent events. <i>European Journal of Immunology</i> , 2017, 47, 327-344.	2.9	62
9	Congenital heart defects in molecularly proven Kabuki syndrome patients. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2912-2922.	1.2	60
10	Diagnosis of Noonan syndrome and related disorders using target next generation sequencing. <i>BMC Medical Genetics</i> , 2014, 15, 14.	2.1	59
11	Identification of <i>TBX5</i> mutations in a series of 94 patients with Tetralogy of Fallot. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 3100-3107.	1.2	47
12	Clinical Presentation and Natural History of Hypertrophic Cardiomyopathy in RASopathies. <i>Heart Failure Clinics</i> , 2018, 14, 225-235.	2.1	44
13	Dextrocardia in patients with Poland syndrome: Phenotypic characterization provides insight into the pathogenesis. <i>Journal of Thoracic and Cardiovascular Surgery</i> , 2010, 139, 1177-1182.	0.8	43
14	Congenital heart disease and genetic syndromes: new insights into molecular mechanisms. <i>Expert Review of Molecular Diagnostics</i> , 2017, 17, 861-870.	3.1	39
15	An International Multicenter Evaluation of Type 5 Long QT Syndrome. <i>Circulation</i> , 2020, 141, 429-439.	1.6	39
16	Ebstein anomaly: Genetic heterogeneity and association with microdeletions 1p36 and 8p23.1. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2196-2202.	1.2	38
17	Exome sequencing identifies a novel mutation in PIK3R1 as the cause of SHORT syndrome. <i>BMC Medical Genetics</i> , 2014, 15, 51.	2.1	34
18	Congenital heart defects in recurrent reciprocal 1q21.1 deletion and duplication syndromes: Rare association with pulmonary valve stenosis. <i>European Journal of Medical Genetics</i> , 2013, 56, 144-149.	1.3	33

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19	Syndromic nonâ€œcompaction of the left ventricle: associated chromosomal anomalies. <i>Clinical Genetics</i> , 2013, 84, 362-367.	2.0	30
20	Hand and Upper Limb Anomalies in Poland Syndrome. <i>Journal of Pediatric Orthopaedics</i> , 2012, 32, 722-726.	1.2	29
21	Familial Poland anomaly revisited. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 140-149.	1.2	29
22	Clinical Profile of Cardiac Involvement in Danon Disease. <i>Circulation Genomic and Precision Medicine</i> , 2020, 13, e003117.	3.6	29
23	Targeted panel sequencing in pediatric primary cardiomyopathy supports a critical role of <i>TNNI3</i> . <i>Clinical Genetics</i> , 2019, 96, 549-559.	2.0	28
24	KBG syndrome: Common and uncommon clinical features based on 31 new patients. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1073-1083.	1.2	27
25	A spectrum of LMX1B mutations in Nail-Patella syndrome: New point mutations, deletion, and evidence of mosaicism in unaffected parents. <i>Genetics in Medicine</i> , 2010, 12, 431-439.	2.4	26
26	Long-term survival and phenotypic spectrum in heterotaxy syndrome: A 25-year follow-up experience. <i>International Journal of Cardiology</i> , 2018, 268, 100-105.	1.7	24
27	Atrioventricular canal defect in patients with RASopathies. <i>European Journal of Human Genetics</i> , 2013, 21, 200-204.	2.8	22
28	Clinical presentation and longâ€œterm outcomes of infantile hypertrophic cardiomyopathy: a European multicentre study. <i>ESC Heart Failure</i> , 2021, 8, 5057-5067.	3.1	22
29	Holtâ€œOram syndrome with intermediate atrioventricular canal defect, and aortic coarctation: Functional characterization of a de novo <i>TBX5</i> mutation. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1419-1424.	1.2	21
30	Assessment of copy number variations in 120 patients with Poland syndrome. <i>BMC Medical Genetics</i> , 2016, 17, 89.	2.1	20
31	Atrioventricular canal defect and genetic syndromes: The unifying role of sonic hedgehog. <i>Clinical Genetics</i> , 2019, 95, 268-276.	2.0	20
32	The role of the electrocardiographic phenotype in risk stratification for sudden cardiac death in childhood hypertrophic cardiomyopathy. <i>European Journal of Preventive Cardiology</i> , 2022, 29, 645-653.	1.8	20
33	Clinical Features and Natural History of Preadolescent Nonsyndromic Hypertrophic Cardiomyopathy. <i>Journal of the American College of Cardiology</i> , 2022, 79, 1986-1997.	2.8	20
34	Differences in morbidity and mortality in Down syndrome are related to the type of congenital heart defect. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1342-1350.	1.2	19
35	Heart rate reduction strategy using ivabradine in end-stage Duchenne cardiomyopathy. <i>International Journal of Cardiology</i> , 2019, 280, 99-103.	1.7	17
36	Atypical cardiac defects in patients with RASopathies: Updated data on CARNET study. <i>Birth Defects Research</i> , 2020, 112, 725-731.	1.5	17

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37	Congenital heart defects in Noonan syndrome and RIT1 mutation. <i>Genetics in Medicine</i> , 2016, 18, 1320.	2.4	16
38	Pituitary hypoplasia and growth hormone deficiency in Coffinâ€“Siris syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 384-388.	1.2	15
39	Some Isolated Cardiac Malformations Can Be Related to Laterality Defects. <i>Journal of Cardiovascular Development and Disease</i> , 2018, 5, 24.	1.6	15
40	Clinical Manifestations of 22q11.2 Deletion Syndrome. <i>Heart Failure Clinics</i> , 2021, 18, 155-164.	2.1	15
41	Heterotaxy syndrome with and without spleen: Different infection risk and management. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 139, 1981-1984.e1.	2.9	14
42	Coronary artery ectasia in Noonan syndrome: Report of an individual with <i>SOS1</i> mutation and literature review. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 665-669.	1.2	13
43	Left pulmonary artery in 22q11.2 deletion syndrome. Echocardiographic evaluation in patients without cardiac defects and role of <i>Tbx1</i> in mice. <i>PLoS ONE</i> , 2019, 14, e0211170.	2.5	13
44	Bradyarrhythmias in Repaired Atrioventricular Septal Defects: Single-Center Experience Based on 34 Years of Follow-Up of 522 Patients. <i>Pediatric Cardiology</i> , 2018, 39, 1590-1597.	1.3	12
45	Cardiac Defects and Genetic Syndromes: Old Uncertainties and New Insights. <i>Genes</i> , 2021, 12, 1047.	2.4	12
46	Arrhythmogenic cardiomyopathy in children according to â€œPadua criteriaâ€“: Single pediatric center experience. <i>International Journal of Cardiology</i> , 2022, 350, 83-89.	1.7	12
47	Hypoplastic Left Heart Syndrome in Patients With Kabuki Syndrome. <i>Pediatric Cardiology</i> , 2010, 31, 1111-1113.	1.3	11
48	Cardiovascular malformations in Adamsâ€“Oliver syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1175-1177.	1.2	11
49	Medical conditions of children and young people with Down syndrome. <i>Journal of Intellectual Disability Research</i> , 2021, 65, 199-209.	2.0	11
50	<i>SOS1</i> mutations in Noonan syndrome: Cardiomyopathies and not only congenital heart defects! Report of six patients including two novel variants and literature review. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2083-2090.	1.2	10
51	A heterozygous, intragenic deletion of <i>CNOT2</i> recapitulates the phenotype of 12q15 deletion syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1615-1621.	1.2	10
52	<i>LTBP2</i> -related â€œMarfanâ€“like phenotype in two Roma/Gypsy subjects with the <i>LTBP2</i> homozygous p.R299X variant. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 104-112.	1.2	10
53	Cardiovascular Involvement in Pediatric Laminopathies. Report of Six Patients and Literature Revision. <i>Frontiers in Pediatrics</i> , 2020, 8, 374.	1.9	9
54	How wide is the ocular spectrum of Delleman syndrome?. <i>Clinical Dysmorphology</i> , 2004, 13, 33-34.	0.3	8

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55	Hypoplastic left heart syndrome and 21q22.3 deletion. American Journal of Medical Genetics, Part A, 2015, 167, 579-586.	1.2	8
56	Genetics in Congenital Heart Diseases. Heart Failure Clinics, 2022, 18, 139-153.	2.1	8
57	Relationship Between Maximal Left Ventricular Wall Thickness and Sudden Cardiac Death in Childhood Onset Hypertrophic Cardiomyopathy. Circulation: Arrhythmia and Electrophysiology, 2022, 15, CIRCEP121010075.	4.8	8
58	CantÃ© syndrome versus Zimmermann-Laband syndrome: Report of nine individuals with ABCC9 variants. European Journal of Medical Genetics, 2020, 63, 103996.	1.3	7
59	Atrioventricular Canal Defect and Associated Genetic Disorders: New Insights into Polydactyly Syndromes. Neurology International, 2011, 1, e7.	0.5	6
60	Data on cardiac defects, morbidity and mortality in patients affected by RASopathies. CARNET study results. Data in Brief, 2018, 16, 649-654.	1.0	6
61	Pharmacological resources, diagnostic approach and coordination of care in joint hypermobility-related disorders. Expert Review of Clinical Pharmacology, 2018, 11, 689-703.	3.1	6
62	Discovering a new part of the phenotypic spectrum of Coffin-Siris syndrome in a fetal cohort. Genetics in Medicine, 2022, 24, 1753-1760.	2.4	6
63	Myocardial and Arrhythmic Spectrum of Neuromuscular Disorders in Children. Biomolecules, 2021, 11, 1578.	4.0	5
64	Cardiomyopathies in Children and Systemic Disorders When Is It Useful to Look beyond the Heart?. Journal of Cardiovascular Development and Disease, 2022, 9, 47.	1.6	5
65	Congenital heart defects in molecularly confirmed <scp>KBG</scp> syndrome patients. American Journal of Medical Genetics, Part A, 2022, 188, 1149-1159.	1.2	5
66	First evidence of maternally inherited mosaicism in TGFBR1 and subtle primary myocardial changes in Loeys-Dietz syndrome: a case report. BMC Medical Genetics, 2018, 19, 170.	2.1	4
67	Palmoplantar Keratoderma and Woolly Hair Revealing Asymptomatic Arrhythmogenic Cardiomyopathy. Acta Dermato-Venereologica, 2019, 99, 831-832.	1.3	4
68	Delayed appearance of 3â€methylglutaconic aciduria in neonates with early onset metabolic cardiomyopathies: A potential pitfall for the diagnosis. American Journal of Medical Genetics, Part A, 2020, 182, 64-70.	1.2	4
69	Troponin T Mutation as a Cause of Left Ventricular Systolic Dysfunction in a Young Patient with Previous Surgical Correction of Aortic Coarctation. Biomolecules, 2021, 11, 696.	4.0	4
70	8p23.1 deletion : Look out for left ventricular hypertrabeculation and not only congenital heart diseases . Single â€center experience and literature revision. American Journal of Medical Genetics, Part A, 2022, 188, 883-895.	1.2	4
71	ICD Outcome in Pediatric Cardiomyopathies. Journal of Cardiovascular Development and Disease, 2022, 9, 33.	1.6	3
72	Syndromic and Non-Syndromic Patients with Repaired Tetralogy of Fallot: Does It Affect the Long-Term Outcome?. Journal of Clinical Medicine, 2022, 11, 850.	2.4	3

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73	Response to Klinger and Merlob re: Case description with review of the literature. Am J Med Genet Part A 149A:1597-1602, 2009. American Journal of Medical Genetics, Part A, 2009, 149A, 2899-2899.	1.2	2
74	Deciphering Genetic Variants of Warfarin Metabolism in Children With Ventricular Assist Devices. Pediatric Cardiology, 2021, 42, 1082-1087.	1.3	2
75	The role of the electrocardiographic phenotype in risk stratification for sudden cardiac death in childhood hypertrophic cardiomyopathy. , 2021, , .		2
76	Unilateral radio-ulnar synostosis associated with hypotonia, developmental delay, and facial dysmorphism. American Journal of Medical Genetics, Part A, 2005, 137A, 106-108.	1.2	1
77	Further case of metaphyseal acroscyphodysplasia with cone-shaped epiphyses (Bellini disease or) Tj ETQq1 1 0.784314 rgBT /Overlock	0.3	1
78	A t(7;12) balanced translocation with breakpoints overlapping those of the Williams-Beuren and 12q14 microdeletion syndromes. American Journal of Medical Genetics, Part A, 2010, 152A, 1285-1294.	1.2	1
79	RDDR: a dysmorphology diagnostic network for newborns in central Italy. Journal of Maternal-Fetal and Neonatal Medicine, 2012, 25, 113-115.	1.5	1
80	Gershoni-Baruch syndrome: First report of a surviving child. American Journal of Medical Genetics, Part A, 2016, 170, 707-711.	1.2	1
81	Hidden Complexity in Routine Adult and Pediatric Arrhythmias Interpretation. Cardiac Electrophysiology Clinics, 2019, 11, 391-404.	1.7	0
82	Persistent myocardial atrophy despite LV reverse remodeling in Duchenne cardiomyopathy treated by LVAD. Pediatric Transplantation, 2021, 25, e13890.	1.0	0