Wadih M Zein

List of Publications by Year
in descending order

36	858	516710 16	526287 27
papers	citations	h-index	g-index

38	38	38	1654
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Prospective phenotyping of NGLY1-CDDG, the first congenital disorder of deglycosylation. Genetics in Medicine, 2017, 19, 160-168.	2.4	124
2	Molecular genetic findings and clinical correlations in 100 patients with Joubert syndrome and related disorders prospectively evaluated at a single center. Genetics in Medicine, 2017, 19, 875-882.	2.4	100
3	Healthcare recommendations for Joubert syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 229-249.	1.2	66
4	Lysosomal Storage and Albinism Due to Effects of a De Novo CLCN7 Variant on Lysosomal Acidification. American Journal of Human Genetics, 2019, 104, 1127-1138.	6.2	59
5	Mutations in human homologue of chickentalpid3gene (KIAA0586) cause a hybrid ciliopathy with overlapping features of Jeune and Joubert syndromes. Journal of Medical Genetics, 2015, 52, 830-839.	3.2	47
6	Expanding the clinical and molecular characteristics of PIGT-CDG, a disorder of glycosylphosphatidylinositol anchors. Molecular Genetics and Metabolism, 2015, 115, 128-140.	1.1	44
7	Joubert Syndrome: Ophthalmological Findings in Correlation with Genotype and Hepatorenal Disease in 99 Patients Prospectively Evaluated at a Single Center. Ophthalmology, 2018, 125, 1937-1952.	5.2	43
8	Ophthalmic Manifestations and Long-Term Visual Outcomes in Patients with Cobalamin C Deficiency. Ophthalmology, 2016, 123, 571-582.	5.2	34
9	Neuropsychological phenotypes of 76 individuals with Joubert syndrome evaluated at a single center. American Journal of Medical Genetics, Part A, 2017, 173, 1796-1812.	1.2	26
10	Cystic cerebellar dysplasia and biallelicLAMA1mutations: a lamininopathy associated with tics, obsessive compulsive traits and myopia due to cell adhesion and migration defects. Journal of Medical Genetics, 2016, 53, 318-329.	3.2	25
11	Longitudinal adaptive optics fluorescence microscopy reveals cellular mosaicism in patients. JCI Insight, 2019, 4, .	5.0	25
12	Defective ciliogenesis in INPP5Eâ€related Joubert syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 3231-3237.	1.2	22
13	DICER1 Syndrome. Ophthalmology, 2019, 126, 296-304.	5.2	20
14	Atypical and ultra-rare Usher syndrome: a review. Ophthalmic Genetics, 2020, 41, 401-412.	1.2	20
15	Genotypeâ€“phenotype associations in a large PRPH2 â€related retinopathy cohort. Human Mutation, 2020, 41, 1528-1539.	2.5	18
16	Vestibular <scp>phenotypeâ€genotype</scp> correlation in a cohort of 90 patients with Usher syndrome. Clinical Genetics, 2021, 99, 226-235.	2.0	18
17	In-depth investigations of adolescents and adults with holoprosencephaly identify unique characteristics. Genetics in Medicine, 2018, 20, 14-23.	2.4	15
18	The peroxisomal disorder spectrum and Heimler syndrome: Deep phenotyping and review of the literature. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 618-630.	1.6	14

#	Article	IF	Citations
19	Expanding the clinical phenotype in patients with disease causing variants associated with atypical Usher syndrome. Ophthalmic Genetics, 2021, 42, 664-673.	1.2	14
20	Severity modeling of propionic acidemia using clinical and laboratory biomarkers. Genetics in Medicine, 2021, 23, 1534-1542.	2.4	13
21	A genetic and clinical study of individuals with nonsyndromic retinopathy consequent upon sequence variants in <scp>HGSNAT</scp>, the gene associated with Sanfilippo C mucopolysaccharidosis. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 631-643.	1.6	12
22	Proposed therapy, developed in a Pcdh15-deficient mouse, for progressive loss of vision in human Usher syndrome. ELife, 2021, 10, .	6.0	12
23	Defining the clinical phenotype of Saulâ€“Wilson syndrome. Genetics in Medicine, 2020, 22, 857-866.	2.4	11
24	Characterization of the Spectrum of Ophthalmic Changes in Patients With Alagille Syndrome. , 2021, 62, 27.		11
25	Active Cell Appearance Model Induced Generative Adversarial Networks for Annotation-Efficient Cell Segmentation and Identification on Adaptive Optics Retinal Images. IEEE Transactions on Medical Imaging, 2021, 40, 2820-2831.	8.9	10
26	Photoreceptor degeneration in ABCA4-associated retinopathy and its genetic correlates. JCI Insight, 2022, 7, .	5.0	10
27	Identification of a novel mutation in HPS6 in a patient with hemophilia B and oculocutaneous albinism. Molecular Genetics and Metabolism, 2016, 119, 284-287.	1.1	9
28	DYRK1A pathogenic variants in two patients with syndromic intellectual disability and a review of the literature. Molecular Genetics & Genomic Medicine, 2020, 8, e1544.	1.2	8
29	Ocular and Systemic Findings in Adults with Uveal Coloboma. Ophthalmology, 2020, 127, 1772-1774.	5.2	8
30	Applying next generation sequencing with microdroplet PCR to determine the disease-causing mutations in retinal dystrophies. BMC Ophthalmology, 2017, 17, 157.	1.4	6
31	A sialidosis type I cohort and a quantitative approach to multimodal ophthalmic imaging of the macular cherry-red spot. British Journal of Ophthalmology, 2021, 105, 838-843.	3.9	4
32	ABCA4c.859-25A>G, a Frequent Palestinian Founder Mutation Affecting the Intron 7 Branchpoint, Is Associated With Early-Onset Stargardt Disease. , 2022, 63, 20.		3
33	Clinical Phenotypes of CDHR1-Associated Retinal Dystrophies. Genes, 2022, 13, 925.	2.4	3
34	Novel ophthalmic findings and deep phenotyping in Williams-Beuren syndrome. British Journal of Ophthalmology, 2023, 107, 1554-1559.	3.9	3
35	Systematic analysis of physical examination characteristics of 94 individuals with Joubert syndrome: Keys to suspecting the diagnosis. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2022, , .	1.6	1
36	Cover Image, Volume 173A, Number 12, December 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0