Valentina Di iorio

List of Publications by Year
in descending order

41 papers	760 citations	623188 14 h-index	676716 22 g-index
41 all docs	41 docs citations	41 times ranked	1137 citing authors

#	Article	IF	Citations
1	Photodynamic therapy as a treatment option for peripapillary pachychoroid syndrome: a pilot study. Eye, 2022, 36, 716-723.	1.1	10
2	Applications of Optical Coherence Tomography in the Ocular Diagnosis: From the Tear Film to the Sclera. Diagnostics, 2022, 12, 673.	1.3	0
3	â€˜Target Signâ€™ - A near infrared feature and multimodal imaging in a pluri-ethnic cohort with RDH5-related fundus albipunctatus. Retina, 2022, Publish Ahead of Print, .	1.0	6
4	Clinical and Molecular Characterization of Achromatopsia Patients: A Longitudinal Study. International Journal of Molecular Sciences, 2021, 22, 1681.	1.8	19
5	Choroidal Vascularity Features in Patients with Choroideremia and Cystoid Spaces. Diagnostics, 2021, 11, 382.	1.3	4
6	Case Report: Ophthalmologic Evaluation Over a Long Follow-Up Time in a Patient With Wolfram Syndrome Type 2: Slowly Progressive Optic Neuropathy as a Possible Clinical Finding. Frontiers in Pediatrics, 2021, 9, 661434.	0.9	2
7	Nephroplex: a kidney-focused NGS panel highlights the challenges of PKD1 sequencing and identifies a founder BBS4 mutation. Journal of Nephrology, 2021, 34, 1855-1874.	0.9	6
8	Molecular Characterization of Choroideremia-Associated Deletions Reveals an Unexpected Regulation of CHM Gene Transcription. Genes, 2021, 12, 1111.	1.0	2
9	Spectrum of Disease Severity in Nonsyndromic Patients With Mutations in the CEP290 Gene: A Multicentric Longitudinal Study. , 2021, 62, 1.		3
10	Voretigene Neparvovec Gene Therapy in Clinical Practice: Treatment of the First Two Italian Pediatric Patients. Translational Vision Science and Technology, 2021, 10, 11.	1.1	15
11	Urine concentrating defect as presenting sign of progressive renal failure in Bardetâ€“Biedl syndrome patients. CKJ: Clinical Kidney Journal, 2021, 14, 1545-1551.	1.4	8
12	Care Pathway of RPE65-Related Inherited Retinal Disorders from Early Symptoms to Genetic Counseling: A Multicenter Narrative Medicine Project in Italy. Clinical Ophthalmology, 2021, Volume 15, 4591-4605.	0.9	4
13	Clinical and Genetic Analysis of a European Cohort with Pericentral Retinitis Pigmentosa. International Journal of Molecular Sciences, 2020, 21, 86.	1.8	25
14	Biofeedback Rehabilitation and Visual Cortex Response in Stargardt's Disease: A Randomized Controlled Trial. Translational Vision Science and Technology, 2020, 9, 6.	1.1	3
15	Mild form of Zellweger Spectrum Disorders (ZSD) due to variants in PEX1: Detailed clinical investigation in a 9-years-old female. Molecular Genetics and Metabolism Reports, 2020, 24, 100615.	0.4	12
16	Spectrum of Disease Severity in Patients With X-Linked Retinitis Pigmentosa Due to RPGR Mutations. , 2020, 61, 36.		17
17	Carbonic anhydrase inhibitors in patients with X-linked retinoschisis: effects on macular morphology and function. Ophthalmic Genetics, 2019, 40, 207-212.	0.5	21
18	CHM/REP1 Transcript Expression and Loss of Visual Function in Patients Affected by Choroideremia. , 2019, 60, 1547.		16

#	Article	IF	Citations
19	Early posterior vitreous detachment is associated with LAMA5 dominant mutation. Ophthalmic Genetics, 2019, 40, 39-42.	0.5	10
20	ASSOCIATION BETWEEN GENOTYPE AND DISEASE PROGRESSION IN ITALIAN STARGARDT PATIENTS. Retina, 2019, 39, 1399-1409.	1.0	19
21	Autosomalâ€dominant myopia associated to a novel P4HA2 missense variant and defective collagen hydroxylation. Clinical Genetics, 2018, 93, 982-991.	1.0	21
22	Automatic segmentation of pigment deposits in retinal fundus images of Retinitis Pigmentosa. Computerized Medical Imaging and Graphics, 2018, 66, 73-81.	3.5	8
23	Visual Cortex Activation in Patients With Stargardt Disease. , 2018, 59, 1503.		5
24	Learning-based approach to segment pigment signs in fundus images for Retinitis Pigmentosa analysis. Neurocomputing, 2018, 308, 159-171.	3.5	10
25	Genetic characterization of Italian patients with Bardet-Biedl syndrome and correlation to ocular, renal and audio-vestibular phenotype: identification of eleven novel pathogenic sequence variants. BMC Medical Genetics, 2017, 18, 10.	2.1	59
26	Mutations in the PCYT1A gene are responsible for isolated forms of retinal dystrophy. European Journal of Human Genetics, 2017, 25, 651-655.	1.4	19
27	The Kidney in Bardet-Biedl Syndrome: Possible Pathogenesis of Urine Concentrating Defect. Kidney Diseases (Basel, Switzerland), 2017, 3, 57-65.	1.2	14
28	High Levels of Serum Ubiquitin and Proteasome in a Case of HLA-B27 Uveitis. International Journal of Molecular Sciences, 2017, 18, 505.	1.8	3
29	Clinical and Genetic Evaluation of a Cohort of Pediatric Patients with Severe Inherited Retinal Dystrophies. Genes, 2017, 8, 280.	1.0	23
30	MP864THE HISTORY OF THE RENAL LESIONS IN BARDET-BIEDL SYNDROME. Nephrology Dialysis Transplantation, 2017, 32, iii37-iii37.	0.4	0
31	En Face Spectral-Domain Optical Coherence Tomography for the Monitoring of Lesion Area Progression in Stargardt Disease. , 2016, 57, OCT247.		17
32	Reproducibility of en-face Optical Coherence Tomography Imaging for Macular Atrophy Area Evaluation in Juvenile Macular Degeneration. IFMBE Proceedings, 2016, , 250-253.	0.2	1
33	Renal phenotype in Bardet-Biedl syndrome: a combined defect of urinary concentration and dilution is associated with defective urinary AQP2 and UMOD excretion. American Journal of Physiology - Renal Physiology, 2016, 311, F686-F694.	1.3	27
34	Macular abnormalities in Italian patients with retinitis pigmentosa. British Journal of Ophthalmology, 2014, 98, 946-950.	2.1	76
35	Macular Function and Morphologic Features in Juvenile Stargardt Disease. Ophthalmology, 2014, 121, 2399-2405.	2.5	54
36	The Role of Optical Coherence Tomography in an Atypical Case of Oculocutaneous Albinism: A Case Report. Case Reports in Ophthalmology, 2012, 3, 113-117.	0.3	6

#	ARTICLE	IF	CITATIONS
37	Correlation between Photoreceptor Layer Integrity and Visual Function in Patients with Stargardt Disease: Implications for Gene Therapy. , 2012, 53, 4409.		62
38	Evaluation of Italian Patients with Leber Congenital Amaurosis due to AIPL1 Mutations Highlights the Potential Applicability of Gene Therapy. , 2011, 52, 5618.		41
39	An Atypical Form of Bietti Crystalline Dystrophy. Ophthalmic Genetics, 2011, 32, 118-121.	0.5	22
40	Molecular and Clinical Characterization of Albinism in a Large Cohort of Italian Patients. , 2011, 52, 1281.		58
41	A normal electro-oculography in a family affected by best disease with a novel spontaneous mutation of the BEST1 gene. British Journal of Ophthalmology, 2008, 92, 1467-1470.	2.1	32