Francesco Testa

List of Publications by Year in descending order

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Version: 2024-02-01

78 papers	5,532 citations	257101 24 h-index	102304 66 g-index
79 all docs	79 docs citations	79 times ranked	5112 citing authors

#	Article	IF	Citations
1	Safety and Efficacy of Gene Transfer for Leber's Congenital Amaurosis. New England Journal of Medicine, 2008, 358, 2240-2248.	13.9	1,941
2	Age-dependent effects of RPE65 gene therapy for Leber's congenital amaurosis: a phase 1 dose-escalation trial. Lancet, The, 2009, 374, 1597-1605.	6.3	774
3	Gene Therapy for Leber's Congenital Amaurosis is Safe and Effective Through 1.5 Years After Vector Administration. Molecular Therapy, 2010, 18, 643-650.	3.7	503
4	Three-Year Follow-up after Unilateral Subretinal Delivery of Adeno-Associated Virus in Patients with Leber Congenital Amaurosis Type 2. Ophthalmology, 2013, 120, 1283-1291.	2.5	301
5	Clinical and Molecular Genetics of Leber's Congenital Amaurosis: A Multicenter Study of Italian Patients. , 2007, 48, 4284.		131
6	Analysis of the ABCA4 genomic locus in Stargardt disease. Human Molecular Genetics, 2014, 23, 6797-6806.	1.4	117
7	Mutations in IMPG2, Encoding Interphotoreceptor Matrix Proteoglycan 2, Cause Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2010, 87, 199-208.	2.6	98
8	Vitreous Substitutes: The Present and the Future. BioMed Research International, 2014, 2014, 1-12.	0.9	86
9	Apolipoprotein E Polymorphisms in Age-Related Macular Degeneration in an Italian Population. Ophthalmic Research, 2001, 33, 325-328.	1.0	78
10	An innovative strategy for the molecular diagnosis of Usher syndrome identifies causal biallelic mutations in 93% of European patients. European Journal of Human Genetics, 2016, 24, 1730-1738.	1.4	77
11	Macular abnormalities in Italian patients with retinitis pigmentosa. British Journal of Ophthalmology, 2014, 98, 946-950.	2.1	76
12	Correlation between Photoreceptor Layer Integrity and Visual Function in Patients with Stargardt Disease: Implications for Gene Therapy. , 2012, 53, 4409.		62
13	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
14	Molecular and Clinical Characterization of Albinism in a Large Cohort of Italian Patients. , 2011, 52, 1281.		58
15	Comprehensive mutation analysis (20 families) of the choroideremia gene reveals a missense variant that prevents the binding of REP1 with rab geranylgeranyl transferase. Human Mutation, 2011, 32, 1460-1469.	1.1	55
16	Autosomal recessive familial exudative vitreoretinopathy: evidence for genetic heterogeneity. Clinical Genetics, 1998, 54, 315-320.	1.0	54
17	Macular Function and Morphologic Features in Juvenile Stargardt Disease. Ophthalmology, 2014, 121, 2399-2405.	2.5	54
18	Clinical and genetic features in Italian Bietti crystalline dystrophy patients. British Journal of Ophthalmology, 2013, 97, 174-179.	2.1	48

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19	Peculiar combinations of individually non-pathogenic missense mitochondrial DNA variants cause low penetrance Leberâ€™s hereditary optic neuropathy. PLoS Genetics, 2018, 14, e1007210.	1.5	47
20	Serum oxidative and antioxidant parameters in a group of Italian patients with age-related maculopathy. Clinica Chimica Acta, 2002, 320, 111-115.	0.5	43
21	Clinical features of X linked juvenile retinoschisis associated with new mutations in the XLRS1 gene in Italian families. British Journal of Ophthalmology, 2003, 87, 1130-1134.	2.1	43
22	Genotype-Phenotype Correlation in Italian Families with Stargardt Disease. Ophthalmic Research, 2005, 37, 159-167.	1.0	42
23	Evaluation of Italian Patients with Leber Congenital Amaurosis due to AIPL1 Mutations Highlights the Potential Applicability of Gene Therapy. , 2011, 52, 5618.		41
24	The ADAMTS18 gene is responsible for autosomal recessive early onset severe retinal dystrophy. Orphanet Journal of Rare Diseases, 2013, 8, 16.	1.2	41
25	Identification of novel RP2 mutations in a subset of X-linked retinitis pigmentosa families and prediction of new domains. Human Mutation, 2001, 18, 109-119.	1.1	39
26	CLINICAL PRESENTATION AND DISEASE COURSE OF USHER SYNDROME BECAUSE OF MUTATIONS IN MYO7A OR USH2A. Retina, 2017, 37, 1581-1590.	1.0	36
27	Mutation analysis of the RPGR gene reveals novel mutations in south European patients with X-linked retinitis pigmentosa. European Journal of Human Genetics, 1999, 7, 687-694.	1.4	30
28	Pupillometric analysis for assessment of gene therapy in Leber Congenital Amaurosis patients. BioMedical Engineering OnLine, 2012, 11, 40.	1.3	27
29	Wearable Improved Vision System for Color Vision Deficiency Correction. IEEE Journal of Translational Engineering in Health and Medicine, 2017, 5, 1-7.	2.2	26
30	Clinical and Genetic Analysis of a European Cohort with Pericentral Retinitis Pigmentosa. International Journal of Molecular Sciences, 2020, 21, 86.	1.8	25
31	Melanocortin receptor agonists <scp>MCR</scp>_{1â€5} protect photoreceptors from highâ€glucose damage and restore antioxidant enzymes in primary retinal cell culture. Journal of Cellular and Molecular Medicine, 2017, 21, 968-974.	1.6	24
32	Macular Hole Surgery: The Healing Process of Outer Retinal Layers to Visual Acuity Recovery. European Journal of Ophthalmology, 2017, 27, 235-239.	0.7	24
33	RPE65-associated inherited retinal diseases: consensus recommendations for eligibility to gene therapy. Orphanet Journal of Rare Diseases, 2021, 16, 257.	1.2	24
34	Clinical and Genetic Evaluation of a Cohort of Pediatric Patients with Severe Inherited Retinal Dystrophies. Genes, 2017, 8, 280.	1.0	23
35	Association of a Homozygous Nonsense Mutation in the ABCA4 (ABCR) Gene with Cone-Rod Dystrophy Phenotype in an Italian Family. Ophthalmic Research, 2004, 36, 82-88.	1.0	22
36	An Atypical Form of Bietti Crystalline Dystrophy. Ophthalmic Genetics, 2011, 32, 118-121.	0.5	22

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37	Carbonic anhydrase inhibitors in patients with X-linked retinoschisis: effects on macular morphology and function. Ophthalmic Genetics, 2019, 40, 207-212.	0.5	21
38	ASSOCIATION BETWEEN GENOTYPE AND DISEASE PROGRESSION IN ITALIAN STARGARDT PATIENTS. Retina, 2019, 39, 1399-1409.	1.0	19
39	Clinical and Molecular Characterization of Achromatopsia Patients: A Longitudinal Study. International Journal of Molecular Sciences, 2021, 22, 1681.	1.8	19
40	Subretinal Fibrosis in StargardtÂ's Disease with Fundus Flavimaculatus and ABCA4 Gene Mutation. Case Reports in Ophthalmology, 2012, 3, 410-417.	0.3	17
41	En Face Spectral-Domain Optical Coherence Tomography for the Monitoring of Lesion Area Progression in Stargardt Disease. , 2016, 57, OCT247.		17
42	Morphological and Functional Retinal Assessment in Epiretinal Membrane Surgery. Seminars in Ophthalmology, 2017, 32, 751-758.	0.8	17
43	Spectrum of Disease Severity in Patients With X-Linked Retinitis Pigmentosa Due to RPGR Mutations. , 2020, 61, 36.		17
44	Cerebral Involvement in Stargardt's Disease: A VBM and TBSS Study. , 2015, 56, 7388.		16
45	CHM/REP1 Transcript Expression and Loss of Visual Function in Patients Affected by Choroideremia. , 2019, 60, 1547.		16
46	Union Makes Strength: A Worldwide Collaborative Genetic and Clinical Study to Provide a Comprehensive Survey of RD3 Mutations and Delineate the Associated Phenotype. PLoS ONE, 2013, 8, e51622.	1.1	16
47	Biallelic sequence and structural variants in RAX2 are a novel cause for autosomal recessive inherited retinal disease. Genetics in Medicine, 2019, 21, 1319-1329.	1.1	15
48	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
49	Characterization of MPP4, a gene highly expressed in photoreceptor cells, and mutation analysis in retinitis pigmentosa. Gene, 2002, 297, 33-38.	1.0	14
50	An Alu-mediated duplication in NMNAT1, involved in NAD biosynthesis, causes a novel syndrome, SHILCA, affecting multiple tissues and organs. Human Molecular Genetics, 2020, 29, 2250-2260.	1.4	14
51	Identification and characterization of C1orf36, a transcript highly expressed in photoreceptor cells, and mutation analysis in retinitis pigmentosa. Biochemical and Biophysical Research Communications, 2003, 308, 414-421.	1.0	13
52	RPE65-Associated Retinopathies in the Italian Population: A Longitudinal Natural History Study. , 2022, 63, 13.		11
53	Prevalence of macular abnormalities assessed by optical coherence tomography in patients with Usher syndrome. Ophthalmic Genetics, 2018, 39, 17-21.	0.5	10
54	Early posterior vitreous detachment is associated with LAMA5 dominant mutation. Ophthalmic Genetics, 2019, 40, 39-42.	0.5	10

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55	Multilayer Microstructure of Idiopathic Epiretinal Macular Membranes. European Journal of Ophthalmology, 2017, 27, 762-768.	0.7	9
56	Ocriplasmin use in a selected case with preserved visual acuity. BMC Ophthalmology, 2015, 15, 146.	0.6	8
57	Functional improvement assessed by multifocal electroretinogram after Ocriplasmin treatment for vitreomacular traction. BMC Ophthalmology, 2016, 16, 110.	0.6	7
58	Full-field electroretinography, visual acuity and visual fields in Usher syndrome: a multicentre European study. Documenta Ophthalmologica, 2019, 139, 151-160.	1.0	7
59	Identifying fallers among ophthalmic patients using classification tree methodology. PLoS ONE, 2017, 12, e0174083.	1.1	7
60	Intrafamilial Clinical Heterogeneity Associated with a Novel Mutation of the Retinal Degeneration Slow/Peripherin Gene. Ophthalmic Research, 2007, 39, 255-259.	1.0	6
61	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
62	Evaluation of Ocular Gene Therapy in an Italian Patient Affected by Congenital Leber Amaurosis Type 2 Treated in Both Eyes. Advances in Experimental Medicine and Biology, 2016, 854, 533-539.	0.8	6
63	Visual Cortex Activation in Patients With Stargardt Disease. , 2018, 59, 1503.		5
64	Toward a Novel Medical Device Based on Chromatic Pupillometry for Screening and Monitoring of Inherited Ocular Disease: A Pilot Study. IFMBE Proceedings, 2019, , 387-390.	0.2	5
65	Genetics of diabetic retinopathy. Seminars in Ophthalmology, 2001, 16, 41-51.	0.8	4
66	Photorefractive Keratectomy on Purely Refractive Accommodative Esotropia. Seminars in Ophthalmology, 2015, 30, 25-28.	0.8	4
67	Intrafamilial heterogeneity of congenital optic disc pit maculopathy. Ophthalmic Genetics, 2017, 38, 267-272.	0.5	4
68	Choroidal Vascularity Features in Patients with Choroideremia and Cystoid Spaces. Diagnostics, 2021, 11, 382.	1.3	4
69	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
70	Usher Syndrome and Color Vision. Current Eye Research, 2018, 43, 1295-1301.	0.7	3
71	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
72	Spectrum of Disease Severity in Nonsyndromic Patients With Mutations in the CEP290 Gene: A Multicentric Longitudinal Study. , 2021, 62, 1.		3

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73	Characteristics of Retinitis Pigmentosa Associated with ADGRV1 and Comparison with USH2A in Patients from a Multicentric Usher Syndrome Study Treatrush. International Journal of Molecular Sciences, 2021, 22, 10352.	1.8	3
74	Molecular Characterization of Choroideremia-Associated Deletions Reveals an Unexpected Regulation of CHM Gene Transcription. Genes, 2021, 12, 1111.	1.0	2
75	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
76	Correlation Between Choriocapillaris Density and Retinal Sensitivity in Age-Related Macular Degeneration. Translational Vision Science and Technology, 2021, 10, 2.	1.1	1
77	The ABCA4 2588G>C Stargardt mutation: single origin and increasing frequency from South-West to North-East Europe. , 0, .		1
78	Expression of Otx Genes in MÃ¼ller Cells Using an In Vitro Experimental Model of Retinal Hypoxia. Journal of Ophthalmology, 2021, 2021, 1-10.	0.6	1