Francesca Simonelli

List of Publications by Year
in descending order

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131 papers	9,421 citations	100601 38 h-index	48101 92 g-index
133 all docs	133 docs citations	133 times ranked	8487 citing authors

#	Article	IF	Citations
1	RPE65-Associated Retinopathies in the Italian Population: A Longitudinal Natural History Study. , 2022, 63, 13.		11
2	Combined photorefractive keratectomy and corneal collagen cross-linking for treatment of keratoconus: a 2-year follow-up study. Therapeutic Advances in Ophthalmology, 2022, 14, 251584142210833.	0.8	2
3	Serum Iba-1, GLUT5, and TSPO in Patients With Diabetic Retinopathy: New Biomarkers for Early Retinal Neurovascular Alterations? A Pilot Study. Translational Vision Science and Technology, 2022, 11, 16.	1.1	7
4	Resolution of Inflammation in Retinal Disorders: Briefly the State. International Journal of Molecular Sciences, 2022, 23, 4501.	1.8	9
5	Two-year macular volume assessment in multiple sclerosis patients treated with fingolimod. Neurological Sciences, 2021, 42, 731-733.	0.9	2
6	Clinical and Molecular Characterization of Achromatopsia Patients: A Longitudinal Study. International Journal of Molecular Sciences, 2021, 22, 1681.	1.8	19
7	Choroidal Vascularity Features in Patients with Choroideremia and Cystoid Spaces. Diagnostics, 2021, 11, 382.	1.3	4
8	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
9	Circulating miRNAs in diabetic retinopathy patients: Prognostic markers or pharmacological targets?. Biochemical Pharmacology, 2021, 186, 114473.	2.0	19
10	Evaluation of Donor and Recipient Characteristics Involved in Descemet Stripping Automated Endothelial Keratoplasty Outcomes. Frontiers in Medicine, 2021, 8, 605160.	1.2	2
11	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
12	RPE65-associated inherited retinal diseases: consensus recommendations for eligibility to gene therapy. Orphanet Journal of Rare Diseases, 2021, 16, 257.	1.2	24
13	Correlation Between Choriocapillaris Density and Retinal Sensitivity in Age-Related Macular Degeneration. Translational Vision Science and Technology, 2021, 10, 2.	1.1	1
14	Molecular Characterization of Choroideremia-Associated Deletions Reveals an Unexpected Regulation of CHM Gene Transcription. Genes, 2021, 12, 1111.	1.0	2
15	Spectrum of Disease Severity in Nonsyndromic Patients With Mutations in the CEP290 Gene: A Multicentric Longitudinal Study. , 2021, 62, 1.		3
16	Pupillometry via smartphone for low-resource settings. Biocybernetics and Biomedical Engineering, 2021, 41, 891-902.	3.3	9
17	Fingolimod and Diabetic Retinopathy: A Drug Repurposing Study. Frontiers in Pharmacology, 2021, 12, 718902.	1.6	13
18	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

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19	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
20	Durability of Voretigene Neparvovec for Biallelic RPE65-Mediated Inherited Retinal Disease. Ophthalmology, 2021, 128, 1460-1468.	2.5	82
21	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
22	Analysis of Corneal Distortion after Myopic PRK. Journal of Clinical Medicine, 2021, 10, 82.	1.0	1
23	Classification Tree to Analyze Factors Connected with Post Operative Complications of Cataract Surgery in a Teaching Hospital. Journal of Clinical Medicine, 2021, 10, 5399.	1.0	2
24	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
25	ORÃO: RESTful Cloud-Based Ophthalmologic Medical Record for Chromatic Pupillometry. IFMBE Proceedings, 2020, , 713-720.	0.2	5
26	A collaborative RESTful cloud-based tool for management of chromatic pupillometry in a clinical trial. Health and Technology, 2020, 10, 25-38.	2.1	8
27	Clinical and Genetic Analysis of a European Cohort with Pericentral Retinitis Pigmentosa. International Journal of Molecular Sciences, 2020, 21, 86.	1.8	25
28	Application of Artificial Intelligence in the Analysis of Features Affecting Cataract Surgery Complications in a Teaching Hospital. Frontiers in Medicine, 2020, 7, 607870.	1.2	8
29	Resolving the dark matter of ABCA4 for 1054 Stargardt disease probands through integrated genomics and transcriptomics. Genetics in Medicine, 2020, 22, 1235-1246.	1.1	92
30	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
31	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
32	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
33	Spectrum of Disease Severity in Patients With X-Linked Retinitis Pigmentosa Due to RPGR Mutations. , 2020, 61, 36.		17
34	Full-field electroretinography, visual acuity and visual fields in Usher syndrome: a multicentre European study. Documenta Ophthalmologica, 2019, 139, 151-160.	1.0	7
35	Efficacy, Safety, and Durability of Voretigene Neparvovec-rzyl in RPE65 Mutationâ€“Associated Inherited Retinal Dystrophy. Ophthalmology, 2019, 126, 1273-1285.	2.5	239
36	Carbonic anhydrase inhibitors in patients with X-linked retinoschisis: effects on macular morphology and function. Ophthalmic Genetics, 2019, 40, 207-212.	0.5	21

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37	Intein-mediated protein trans-splicing expands adeno-associated virus transfer capacity in the retina. Science Translational Medicine, 2019, 11, .	5.8	109
38	CHM/REP1 Transcript Expression and Loss of Visual Function in Patients Affected by Choroideremia. , 2019, 60, 1547.		16
39	Retinal dystrophy in an individual carrying a de novo missense variant of SMARCA4. Molecular Genetics & Genomic Medicine, 2019, 7, e682.	0.6	8
40	Early posterior vitreous detachment is associated with LAMA5 dominant mutation. Ophthalmic Genetics, 2019, 40, 39-42.	0.5	10
41	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
42	ASSOCIATION BETWEEN GENOTYPE AND DISEASE PROGRESSION IN ITALIAN STARGARDT PATIENTS. Retina, 2019, 39, 1399-1409.	1.0	19
43	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
44	Triple Vectors Expand AAV Transfer Capacity in the Retina. Molecular Therapy, 2018, 26, 524-541.	3.7	94
45	Prevalence of macular abnormalities assessed by optical coherence tomography in patients with Usher syndrome. Ophthalmic Genetics, 2018, 39, 17-21.	0.5	10
46	Standard, transepithelial and iontophoresis corneal cross-linking: clinical analysis of three surgical techniques. International Ophthalmology, 2018, 38, 2585-2592.	0.6	13
47	Visual Cortex Activation in Patients With Stargardt Disease. , 2018, 59, 1503.		5
48	Usher Syndrome and Color Vision. Current Eye Research, 2018, 43, 1295-1301.	0.7	3
49	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
50	Intrafamilial heterogeneity of congenital optic disc pit maculopathy. Ophthalmic Genetics, 2017, 38, 267-272.	0.5	4
51	Identification and population history of CYP4V2 mutations in patients with Bietti crystalline corneoretinal dystrophy. European Journal of Human Genetics, 2017, 25, 461-471.	1.4	23
52	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
53	CLINICAL PRESENTATION AND DISEASE COURSE OF USHER SYNDROME BECAUSE OF MUTATIONS IN MYO7A OR USH2A. Retina, 2017, 37, 1581-1590.	1.0	36
54	Systematic Screening of Retinopathy in Diabetes (REaD Project): An Italian Implementation Campaign. European Journal of Ophthalmology, 2017, 27, 179-184.	0.7	9

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55	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
56	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
57	Efficacy and safety of voretigene neparvovec (AAV2-hRPE65v2) in patients with RPE65 -mediated inherited retinal dystrophy: a randomised, controlled, open-label, phase 3 trial. Lancet, The, 2017, 390, 849-860.	6.3	1,250
58	Morphological and Functional Retinal Assessment in Epiretinal Membrane Surgery. Seminars in Ophthalmology, 2017, 32, 751-758.	0.8	17
59	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
60	Clinical and Genetic Evaluation of a Cohort of Pediatric Patients with Severe Inherited Retinal Dystrophies. Genes, 2017, 8, 280.	1.0	23
61	Targeting and silencing of rhodopsin by ectopic expression of the transcription factor KLF15. JCI Insight, 2017, 2, .	2.3	12
62	Identifying fallers among ophthalmic patients using classification tree methodology. PLoS ONE, 2017, 12, e0174083.	1.1	7
63	En Face Spectral-Domain Optical Coherence Tomography for the Monitoring of Lesion Area Progression in Stargardt Disease. , 2016, 57, OCT247.		17
64	Mulibrey nanism: Two novel mutations in a child identified by Array CGH and DNA sequencing. American Journal of Medical Genetics, Part A, 2016, 170, 2196-2199.	0.7	14
65	Safety and durability of effect of contralateral-eye administration of AAV2 gene therapy in patients with childhood-onset blindness caused by RPE65 mutations: a follow-on phase 1 trial. Lancet, The, 2016, 388, 661-672.	6.3	377
66	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
67	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
68	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
69	Functional improvement assessed by multifocal electroretinogram after Ocriplasmin treatment for vitreomacular traction. BMC Ophthalmology, 2016, 16, 110.	0.6	7
70	Mutations in CTNNA1 cause butterfly-shaped pigment dystrophy and perturbed retinal pigment epithelium integrity. Nature Genetics, 2016, 48, 144-151.	9.4	50
71	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
72	Rhodopsin targeted transcriptional silencing by DNA-binding. ELife, 2016, 5, e12242.	2.8	33

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73	Ocriplasmin use in a selected case with preserved visual acuity. BMC Ophthalmology, 2015, 15, 146.	0.6	8
74	Cerebral Involvement in Stargardt's Disease: A VBM and TBSS Study. , 2015, 56, 7388.		16
75	Investigation of Aberrant Splicing Induced byAIPL1Variations as a Cause of Leber Congenital Amaurosis. , 2015, 56, 7784.		11
76	Standard versus trans-epithelial collagen cross-linking in keratoconus patients suitable for standard collagen cross-linking. Clinical Ophthalmology, 2015, 9, 503.	0.9	52
77	Gene Therapy of Inherited Retinal Degenerations: Prospects and Challenges. Human Gene Therapy, 2015, 26, 193-200.	1.4	39
78	Improved dual AAV vectors with reduced expression of truncated proteins are safe and effective in the retina of a mouse model of Stargardt disease. Human Molecular Genetics, 2015, 24, 6811-6825.	1.4	73
79	Photorefractive Keratectomy on Purely Refractive Accommodative Esotropia. Seminars in Ophthalmology, 2015, 30, 25-28.	0.8	4
80	Preserved Outer Retina in AIPL1 Leber's Congenital Amaurosis: Implications for Gene Therapy. Ophthalmology, 2015, 122, 862-864.	2.5	31
81	IMPG2-Associated Retinitis Pigmentosa Displays Relatively Early Macular Involvement. , 2014, 55, 3939.		37
82	Macular abnormalities in Italian patients with retinitis pigmentosa. British Journal of Ophthalmology, 2014, 98, 946-950.	2.1	76
83	Vitreous Substitutes: The Present and the Future. BioMed Research International, 2014, 2014, 1-12.	0.9	86
84	Analysis of the ABCA4 genomic locus in Stargardt disease. Human Molecular Genetics, 2014, 23, 6797-6806.	1.4	117
85	A novel CISD2 intragenic deletion, optic neuropathy and platelet aggregation defect in Wolfram syndrome type 2. BMC Medical Genetics, 2014, 15, 88.	2.1	59
86	Macular Function and Morphologic Features in Juvenile Stargardt Disease. Ophthalmology, 2014, 121, 2399-2405.	2.5	54
87	The ADAMTS18 gene is responsible for autosomal recessive early onset severe retinal dystrophy. Orphanet Journal of Rare Diseases, 2013, 8, 16.	1.2	41
88	Mutations in IMPG1 Cause Vitelliform Macular Dystrophies. American Journal of Human Genetics, 2013, 93, 571-578.	2.6	71
89	Combined Rod and Cone Transduction by Adeno-Associated Virus 2/8. Human Gene Therapy, 2013, 24, 982-992.	1.4	36
90	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

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91	Expression of VEGF-A, Otx Homeobox and p53 Family Genes in Proliferative Vitreoretinopathy. Mediators of Inflammation, 2013, 2013, 1-8.	1.4	23
92	Clinical and genetic features in Italian Bietti crystalline dystrophy patients. British Journal of Ophthalmology, 2013, 97, 174-179.	2.1	48
93	Idebenone treatment in patients with OPA1-mutant dominant optic atrophy. Brain, 2013, 136, e231-e231.	3.7	62
94	Recombinant Vectors Based on Porcine Adeno-Associated Viral Serotypes Transduce the Murine and Pig Retina. PLoS ONE, 2013, 8, e59025.	1.1	13
95	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
96	Subretinal Fibrosis in StargardtÂ's Disease with Fundus Flavimaculatus and ABCA4 Gene Mutation. Case Reports in Ophthalmology, 2012, 3, 410-417.	0.3	17
97	Pupillometric analysis for assessment of gene therapy in Leber Congenital Amaurosis patients. BioMedical Engineering OnLine, 2012, 11, 40.	1.3	27
98	Mitochondrial Diabetes in Children: Seek and You Will Find It. PLoS ONE, 2012, 7, e34956.	1.1	28
99	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
100	AAV2 Gene Therapy Readministration in Three Adults with Congenital Blindness. Science Translational Medicine, 2012, 4, 120ra15.	5.8	340
101	Correlation between Photoreceptor Layer Integrity and Visual Function in Patients with Stargardt Disease: Implications for Gene Therapy. , 2012, 53, 4409.		62
102	Molecular Diagnosis of Usher Syndrome: Application of Two Different Next Generation Sequencing-Based Procedures. PLoS ONE, 2012, 7, e43799.	1.1	29
103	MicroRNA-Restricted Transgene Expression in the Retina. PLoS ONE, 2011, 6, e22166.	1.1	55
104	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
105	Evaluation of Italian Patients with Leber Congenital Amaurosis due to AIPL1 Mutations Highlights the Potential Applicability of Gene Therapy. , 2011, 52, 5618.		41
106	An Atypical Form of Bietti Crystalline Dystrophy. Ophthalmic Genetics, 2011, 32, 118-121.	0.5	22
107	Molecular and Clinical Characterization of Albinism in a Large Cohort of Italian Patients. , 2011, 52, 1281.		58
108	Idebenone Treatment In Leber's Hereditary Optic Neuropathy. Brain, 2011, 134, e188-e188.	3.7	192

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109	The human visual cortex responds to gene therapyâ€“mediated recovery of retinal function. Journal of Clinical Investigation, 2011, 121, 2160-2168.	3.9	121
110	The human visual cortex responds to gene therapyâ€“mediated recovery of retinal function. Journal of Clinical Investigation, 2011, 121, 2945-2945.	3.9	2
111	Molecular epidemiology of Usher syndrome in Italy. Molecular Vision, 2011, 17, 1662-8.	1.1	27
112	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
113	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
114	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
115	Safety and Efficacy of Gene Transfer for Leber's Congenital Amaurosis. New England Journal of Medicine, 2008, 358, 2240-2248.	13.9	1,941
116	Intrafamilial Clinical Heterogeneity Associated with a Novel Mutation of the Retinal Degeneration Slow/Peripherin Gene. Ophthalmic Research, 2007, 39, 255-259.	1.0	6
117	Clinical and Molecular Genetics of Leber's Congenital Amaurosis: A Multicenter Study of Italian Patients. , 2007, 48, 4284.		131
118	Development of a genotyping microarray for Usher syndrome. Journal of Medical Genetics, 2006, 44, 153-160.	1.5	94
119	Genotype-Phenotype Correlation in Italian Families with Stargardt Disease. Ophthalmic Research, 2005, 37, 159-167.	1.0	42
120	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
121	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
122	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
123	Characterization of MPP4, a gene highly expressed in photoreceptor cells, and mutation analysis in retinitis pigmentosa. Gene, 2002, 297, 33-38.	1.0	14
124	Apolipoprotein E Polymorphisms in Age-Related Macular Degeneration in an Italian Population. Ophthalmic Research, 2001, 33, 325-328.	1.0	78
125	Genetics of diabetic retinopathy. Seminars in Ophthalmology, 2001, 16, 41-51.	0.8	4
126	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

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127	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
128	Autosomal recessive familial exudative vitreoretinopathy: evidence for genetic heterogeneity. Clinical Genetics, 1998, 54, 315-320.	1.0	54
129	Senile cataractic lenses do not accumulate galactitol in either lactose tolerant or intolerant subjects. Clinica Chimica Acta, 1993, 220, 115-118.	0.5	0
130	Systemic human diseases as oxidative risk factors in cataractogenesis. II. Chronic renal failure. Experimental Eye Research, 1990, 51, 631-635.	1.2	23
131	The ABCA4 2588G>C Stargardt mutation: single origin and increasing frequency from South-West to North-East Europe. , 0, .		1