Francesca Simonelli

List of Publications by Year
in descending order

131	9,421	87886 38	42393 92
papers	citations	h-index	g-index

133	133	133	7839
all docs	docs citations	times ranked	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.	27.0	1,941
2	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.	13.7	1,250
3	Age-dependent effects of RPE65 gene therapy for Leber's congenital amaurosis: a phase 1 dose-escalation trial. Lancet, The, 2009, 374, 1597-1605.	13.7	774
4	Gene Therapy for Leber's Congenital Amaurosis is Safe and Effective Through 1.5 Years After Vector Administration. Molecular Therapy, 2010, 18, 643-650.	8.2	503
5	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.	13.7	377
6	AAV2 Gene Therapy Readministration in Three Adults with Congenital Blindness. Science Translational Medicine, 2012, 4, 120ra15.	12.4	340
7	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.	5.2	301
8	Efficacy, Safety, and Durability of Voretigene Neparvovec-rzyl in RPE65 Mutationâ€“Associated Inherited Retinal Dystrophy. Ophthalmology, 2019, 126, 1273-1285.	5.2	239
9	Idebenone Treatment In Leber's Hereditary Optic Neuropathy. Brain, 2011, 134, e188-e188.	7.6	192
10	Clinical and Molecular Genetics of Leber's Congenital Amaurosis: A Multicenter Study of Italian Patients. , 2007, 48, 4284.		131
11	The human visual cortex responds to gene therapyâ€“mediated recovery of retinal function. Journal of Clinical Investigation, 2011, 121, 2160-2168.	8.2	121
12	Analysis of the ABCA4 genomic locus in Stargardt disease. Human Molecular Genetics, 2014, 23, 6797-6806.	2.9	117
13	Intein-mediated protein trans-splicing expands adeno-associated virus transfer capacity in the retina. Science Translational Medicine, 2019, 11, .	12.4	109
14	Mutations in IMPG2, Encoding Interphotoreceptor Matrix Proteoglycan 2, Cause Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2010, 87, 199-208.	6.2	98
15	Development of a genotyping microarray for Usher syndrome. Journal of Medical Genetics, 2006, 44, 153-160.	3.2	94
16	Triple Vectors Expand AAV Transfer Capacity in the Retina. Molecular Therapy, 2018, 26, 524-541.	8.2	94
17	Resolving the dark matter of ABCA4 for 1054 Stargardt disease probands through integrated genomics and transcriptomics. Genetics in Medicine, 2020, 22, 1235-1246.	2.4	92
18	Vitreous Substitutes: The Present and the Future. BioMed Research International, 2014, 2014, 1-12.	1.9	86

#	Article	IF	Citations
19	Durability of Voretigene Neparvovec for Biallelic RPE65-Mediated Inherited Retinal Disease. Ophthalmology, 2021, 128, 1460-1468.	5.2	82
20	Apolipoprotein E Polymorphisms in Age-Related Macular Degeneration in an Italian Population. Ophthalmic Research, 2001, 33, 325-328.	1.9	78
21	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.	2.8	77
22	Macular abnormalities in Italian patients with retinitis pigmentosa. British Journal of Ophthalmology, 2014, 98, 946-950.	3.9	76
23	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.	2.9	73
24	Mutations in IMPG1 Cause Vitelliform Macular Dystrophies. American Journal of Human Genetics, 2013, 93, 571-578.	6.2	71
25	Correlation between Photoreceptor Layer Integrity and Visual Function in Patients with Stargardt Disease: Implications for Gene Therapy. , 2012, 53, 4409.		62
26	Idebenone treatment in patients with OPA1-mutant dominant optic atrophy. Brain, 2013, 136, e231-e231.	7.6	62
27	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
28	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
29	Molecular and Clinical Characterization of Albinism in a Large Cohort of Italian Patients. , 2011, 52, 1281.		58
30	MicroRNA-Restricted Transgene Expression in the Retina. PLoS ONE, 2011, 6, e22166.	2.5	55
31	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.	2.5	55
32	Autosomal recessive familial exudative vitreoretinopathy: evidence for genetic heterogeneity. Clinical Genetics, 1998, 54, 315-320.	2.0	54
33	Macular Function and Morphologic Features in Juvenile Stargardt Disease. Ophthalmology, 2014, 121, 2399-2405.	5.2	54
34	Standard versus trans-epithelial collagen cross-linking in keratoconus patients suitable for standard collagen cross-linking. Clinical Ophthalmology, 2015, 9, 503.	1.8	52
35	Mutations in CTNNA1 cause butterfly-shaped pigment dystrophy and perturbed retinal pigment epithelium integrity. Nature Genetics, 2016, 48, 144-151.	21.4	50
36	Clinical and genetic features in Italian Bietti crystalline dystrophy patients. British Journal of Ophthalmology, 2013, 97, 174-179.	3.9	48

#	Article	IF	Citations
37	Peculiar combinations of individually non-pathogenic missense mitochondrial DNA variants cause low penetrance Leberâ€™s hereditary optic neuropathy. PLoS Genetics, 2018, 14, e1007210.	3.5	47
38	Serum oxidative and antioxidant parameters in a group of Italian patients with age-related maculopathy. Clinica Chimica Acta, 2002, 320, 111-115.	1.1	43
39	Genotype-Phenotype Correlation in Italian Families with Stargardt Disease. Ophthalmic Research, 2005, 37, 159-167.	1.9	42
40	Evaluation of Italian Patients with Leber Congenital Amaurosis due to AIPL1 Mutations Highlights the Potential Applicability of Gene Therapy. , 2011, 52, 5618.		41
41	The ADAMTS18 gene is responsible for autosomal recessive early onset severe retinal dystrophy. Orphanet Journal of Rare Diseases, 2013, 8, 16.	2.7	41
42	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.	2.5	39
43	Gene Therapy of Inherited Retinal Degenerations: Prospects and Challenges. Human Gene Therapy, 2015, 26, 193-200.	2.7	39
44	IMPG2-Associated Retinitis Pigmentosa Displays Relatively Early Macular Involvement. , 2014, 55, 3939.		37
45	Combined Rod and Cone Transduction by Adeno-Associated Virus 2/8. Human Gene Therapy, 2013, 24, 982-992.	2.7	36
46	CLINICAL PRESENTATION AND DISEASE COURSE OF USHER SYNDROME BECAUSE OF MUTATIONS IN MYO7A OR USH2A. Retina, 2017, 37, 1581-1590.	1.7	36
47	Rhodopsin targeted transcriptional silencing by DNA-binding. ELife, 2016, 5, e12242.	6.0	33
48	Preserved Outer Retina in AIPL1 Leber's Congenital Amaurosis: Implications for Gene Therapy. Ophthalmology, 2015, 122, 862-864.	5.2	31
49	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.	2.8	30
50	Molecular Diagnosis of Usher Syndrome: Application of Two Different Next Generation Sequencing-Based Procedures. PLoS ONE, 2012, 7, e43799.	2.5	29
51	Mitochondrial Diabetes in Children: Seek and You Will Find It. PLoS ONE, 2012, 7, e34956.	2.5	28
52	Pupillometric analysis for assessment of gene therapy in Leber Congenital Amaurosis patients. BioMedical Engineering OnLine, 2012, 11, 40.	2.7	27
53	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.	2.7	27
54	Molecular epidemiology of Usher syndrome in Italy. Molecular Vision, 2011, 17, 1662-8.	1.1	27

#	Article	IF	Citations
55	Wearable Improved Vision System for Color Vision Deficiency Correction. IEEE Journal of Translational Engineering in Health and Medicine, 2017, 5, 1-7.	3.7	26
56	Clinical and Genetic Analysis of a European Cohort with Pericentral Retinitis Pigmentosa. International Journal of Molecular Sciences, 2020, 21, 86.	4.1	25
57	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.	3.6	24
58	RPE65-associated inherited retinal diseases: consensus recommendations for eligibility to gene therapy. Orphanet Journal of Rare Diseases, 2021, 16, 257.	2.7	24
59	Systemic human diseases as oxidative risk factors in cataractogenesis. II. Chronic renal failure. Experimental Eye Research, 1990, 51, 631-635.	2.6	23
60	Expression of VEGF-A, Otx Homeobox and p53 Family Genes in Proliferative Vitreoretinopathy. Mediators of Inflammation, 2013, 2013, 1-8.	3.0	23
61	Identification and population history of CYP4V2 mutations in patients with Bietti crystalline corneoretinal dystrophy. European Journal of Human Genetics, 2017, 25, 461-471.	2.8	23
62	Clinical and Genetic Evaluation of a Cohort of Pediatric Patients with Severe Inherited Retinal Dystrophies. Genes, 2017, 8, 280.	2.4	23
63	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.	1.9	22
64	An Atypical Form of Bietti Crystalline Dystrophy. Ophthalmic Genetics, 2011, 32, 118-121.	1.2	22
65	Carbonic anhydrase inhibitors in patients with X-linked retinoschisis: effects on macular morphology and function. Ophthalmic Genetics, 2019, 40, 207-212.	1.2	21
66	ASSOCIATION BETWEEN GENOTYPE AND DISEASE PROGRESSION IN ITALIAN STARGARDT PATIENTS. Retina, 2019, 39, 1399-1409.	1.7	19
67	Clinical and Molecular Characterization of Achromatopsia Patients: A Longitudinal Study. International Journal of Molecular Sciences, 2021, 22, 1681.	4.1	19
68	Circulating miRNAs in diabetic retinopathy patients: Prognostic markers or pharmacological targets?. Biochemical Pharmacology, 2021, 186, 114473.	4.4	19
69	Subretinal Fibrosis in StargardtÂ's Disease with Fundus Flavimaculatus and ABCA4 Gene Mutation. Case Reports in Ophthalmology, 2012, 3, 410-417.	0.7	17
70	En Face Spectral-Domain Optical Coherence Tomography for the Monitoring of Lesion Area Progression in Stargardt Disease. , 2016, 57, OCT247.		17
71	Morphological and Functional Retinal Assessment in Epiretinal Membrane Surgery. Seminars in Ophthalmology, 2017, 32, 751-758.	1.6	17
72	Spectrum of Disease Severity in Patients With X-Linked Retinitis Pigmentosa Due to RPGR Mutations. , 2020, 61, 36.		17

#	Article	IF	Citations
73	Cerebral Involvement in Stargardt's Disease: A VBM and TBSS Study. , 2015, 56, 7388.		16
74	CHM/REP1 Transcript Expression and Loss of Visual Function in Patients Affected by Choroideremia. , 2019, 60, 1547.		16
75	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.	2.5	16
76	Biallelic sequence and structural variants in RAX2 are a novel cause for autosomal recessive inherited retinal disease. Genetics in Medicine, 2019, 21, 1319-1329.	2.4	15
77	Voretigene Neparvovec Gene Therapy in Clinical Practice: Treatment of the First Two Italian Pediatric Patients. Translational Vision Science and Technology, 2021, 10, 11.	2.2	15
78	Characterization of MPP4, a gene highly expressed in photoreceptor cells, and mutation analysis in retinitis pigmentosa. Gene, 2002, 297, 33-38.	2.2	14
79	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.	1.2	14
80	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.	2.9	14
81	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.	2.1	13
82	Recombinant Vectors Based on Porcine Adeno-Associated Viral Serotypes Transduce the Murine and Pig Retina. PLoS ONE, 2013, 8, e59025.	2.5	13
83	Standard, transepithelial and iontophoresis corneal cross-linking: clinical analysis of three surgical techniques. International Ophthalmology, 2018, 38, 2585-2592.	1.4	13
84	Fingolimod and Diabetic Retinopathy: A Drug Repurposing Study. Frontiers in Pharmacology, 2021, 12, 718902.	3.5	13
85	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.	1.1	12
86	Targeting and silencing of rhodopsin by ectopic expression of the transcription factor KLF15. JCI Insight, 2017, 2, .	5.0	12
87	Investigation of Aberrant Splicing Induced byAIPL1Variations as a Cause of Leber Congenital Amaurosis. , 2015, 56, 7784.		11
88	RPE65-Associated Retinopathies in the Italian Population: A Longitudinal Natural History Study. , 2022, 63, 13.		11
89	Prevalence of macular abnormalities assessed by optical coherence tomography in patients with Usher syndrome. Ophthalmic Genetics, 2018, 39, 17-21.	1.2	10
90	Early posterior vitreous detachment is associated with LAMA5 dominant mutation. Ophthalmic Genetics, 2019, 40, 39-42.	1.2	10

#	Article	IF	Citations
91	Systematic Screening of Retinopathy in Diabetes (REaD Project): An Italian Implementation Campaign. European Journal of Ophthalmology, 2017, 27, 179-184.	1.3	9
92	Pupillometry via smartphone for low-resource settings. Biocybernetics and Biomedical Engineering, 2021, 41, 891-902.	5.9	9
93	Resolution of Inflammation in Retinal Disorders: Briefly the State. International Journal of Molecular Sciences, 2022, 23, 4501.	4.1	9
94	Ocriplasmin use in a selected case with preserved visual acuity. BMC Ophthalmology, 2015, 15, 146.	1.4	8
95	Retinal dystrophy in an individual carrying a de novo missense variant of SMARCA4. Molecular Genetics & Genomic Medicine, 2019, 7, e682.	1.2	8
96	A collaborative RESTful cloud-based tool for management of chromatic pupillometry in a clinical trial. Health and Technology, 2020, 10, 25-38.	3.6	8
97	Application of Artificial Intelligence in the Analysis of Features Affecting Cataract Surgery Complications in a Teaching Hospital. Frontiers in Medicine, 2020, 7, 607870.	2.6	8
98	Urine concentrating defect as presenting sign of progressive renal failure in Bardetâ€“Biedl syndrome patients. CKJ: Clinical Kidney Journal, 2021, 14, 1545-1551.	2.9	8
99	Functional improvement assessed by multifocal electroretinogram after Ocriplasmin treatment for vitreomacular traction. BMC Ophthalmology, 2016, 16, 110.	1.4	7
100	Full-field electroretinography, visual acuity and visual fields in Usher syndrome: a multicentre European study. Documenta Ophthalmologica, 2019, 139, 151-160.	2.2	7
101	Identifying fallers among ophthalmic patients using classification tree methodology. PLoS ONE, 2017, 12, e0174083.	2.5	7
102	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.	2.2	7
103	Intrafamilial Clinical Heterogeneity Associated with a Novel Mutation of the Retinal Degeneration Slow/Peripherin Gene. Ophthalmic Research, 2007, 39, 255-259.	1.9	6
104	The Role of Optical Coherence Tomography in an Atypical Case of Oculocutaneous Albinism: A Case Report. Case Reports in Ophthalmology, 2012, 3, 113-117.	0.7	6
105	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.	1.6	6
106	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.	2.0	6
107	Visual Cortex Activation in Patients With Stargardt Disease. , 2018, 59, 1503.		5
108	ORÃO: RESTful Cloud-Based Ophthalmologic Medical Record for Chromatic Pupillometry. IFMBE Proceedings, 2020, , 713-720.	0.3	5

#	Article	IF	Citations
109	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.	0.3	5
110	Genetics of diabetic retinopathy. Seminars in Ophthalmology, 2001, 16, 41-51.	1.6	4
111	Photorefractive Keratectomy on Purely Refractive Accommodative Esotropia. Seminars in Ophthalmology, 2015, 30, 25-28.	1.6	4
112	Intrafamilial heterogeneity of congenital optic disc pit maculopathy. Ophthalmic Genetics, 2017, 38, 267-272.	1.2	4
113	Choroidal Vascularity Features in Patients with Choroideremia and Cystoid Spaces. Diagnostics, 2021, 11, 382.	2.6	4
114	High Levels of Serum Ubiquitin and Proteasome in a Case of HLA-B27 Uveitis. International Journal of Molecular Sciences, 2017, 18, 505.	4.1	3
115	Usher Syndrome and Color Vision. Current Eye Research, 2018, 43, 1295-1301.	1.5	3
116	Biofeedback Rehabilitation and Visual Cortex Response in Stargardt's Disease: A Randomized Controlled Trial. Translational Vision Science and Technology, 2020, 9, 6.	2.2	3
117	Spectrum of Disease Severity in Nonsyndromic Patients With Mutations in the CEP290 Gene: A Multicentric Longitudinal Study. , 2021, 62, 1.		3
118	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.	4.1	3
119	Two-year macular volume assessment in multiple sclerosis patients treated with fingolimod. Neurological Sciences, 2021, 42, 731-733.	1.9	2
120	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.	1.9	2
121	Evaluation of Donor and Recipient Characteristics Involved in Descemet Stripping Automated Endothelial Keratoplasty Outcomes. Frontiers in Medicine, 2021, 8, 605160.	2.6	2
122	Molecular Characterization of Choroideremia-Associated Deletions Reveals an Unexpected Regulation of CHM Gene Transcription. Genes, 2021, 12, 1111.	2.4	2
123	The human visual cortex responds to gene therapyâ€“mediated recovery of retinal function. Journal of Clinical Investigation, 2011, 121, 2945-2945.	8.2	2
124	Classification Tree to Analyze Factors Connected with Post Operative Complications of Cataract Surgery in a Teaching Hospital. Journal of Clinical Medicine, 2021, 10, 5399.	2.4	2
125	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.	1.4	2
126	Reproducibility of en-face Optical Coherence Tomography Imaging for Macular Atrophy Area Evaluation in Juvenile Macular Degeneration. IFMBE Proceedings, 2016, , 250-253.	0.3	1

#	Article	IF	Citations
127	Correlation Between Choriocapillaris Density and Retinal Sensitivity in Age-Related Macular Degeneration. Translational Vision Science and Technology, 2021, 10, 2.	2.2	1
128	The ABCA4 2588G>C Stargardt mutation: single origin and increasing frequency from South-West to North-East Europe. , 0, .		1
129	Analysis of Corneal Distortion after Myopic PRK. Journal of Clinical Medicine, 2021, 10, 82.	2.4	1
130	Expression of Otx Genes in MÃ¼ller Cells Using an In Vitro Experimental Model of Retinal Hypoxia. Journal of Ophthalmology, 2021, 2021, 1-10.	1.3	1
131	Senile cataractic lenses do not accumulate galactitol in either lactose tolerant or intolerant subjects. Clinica Chimica Acta, 1993, 220, 115-118.	1.1	0