

Carlo Rivolta

List of Publications by Year in descending order

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Version: 2024-02-01

124
papers

12,889
citations

57758

44
h-index

24982

109
g-index

134
all docs

134
docs citations

134
times ranked

19777
citing authors

#	ARTICLE	IF	CITATIONS
1	The complete genome sequence of the Gram-positive bacterium <i>Bacillus subtilis</i> . <i>Nature</i> , 1997, 390, 249-256.	27.8	3,519
2	Hundreds of variants clustered in genomic loci and biological pathways affect human height. <i>Nature</i> , 2010, 467, 832-838.	27.8	1,789
3	Essential <i>Bacillus subtilis</i> genes. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2003, 100, 4678-4683.	7.1	1,261
4	Sex-stratified Genome-wide Association Studies Including 270,000 Individuals Show Sexual Dimorphism in Genetic Loci for Anthropometric Traits. <i>PLoS Genetics</i> , 2013, 9, e1003500.	3.5	371
5	The Influence of Age and Sex on Genetic Associations with Adult Body Size and Shape: A Large-Scale Genome-Wide Interaction Study. <i>PLoS Genetics</i> , 2015, 11, e1005378.	3.5	331
6	Missense Mutation in the <i>USH2A</i> Gene: Association with Recessive Retinitis Pigmentosa without Hearing Loss. <i>American Journal of Human Genetics</i> , 2000, 66, 1975-1978.	6.2	296
7	Retinitis pigmentosa and allied diseases: numerous diseases, genes, and inheritance patterns. <i>Human Molecular Genetics</i> , 2002, 11, 1219-1227.	2.9	251
8	The Zinc Transporter <i>SLC39A13/ZIP13</i> Is Required for Connective Tissue Development; Its Involvement in BMP/TGF- β^2 Signaling Pathways. <i>PLoS ONE</i> , 2008, 3, e3642.	2.5	240
9	A novel protein kinase that controls carbon catabolite repression in bacteria. <i>Molecular Microbiology</i> , 1998, 27, 1157-1169.	2.5	205
10	Worldwide carrier frequency and genetic prevalence of autosomal recessive inherited retinal diseases. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2020, 117, 2710-2716.	7.1	195
11	Genomewide Association Study Using a High-Density Single Nucleotide Polymorphism Array and Case-Control Design Identifies a Novel Essential Hypertension Susceptibility Locus in the Promoter Region of Endothelial NO Synthase. <i>Hypertension</i> , 2012, 59, 248-255.	2.7	144
12	Comprehensive Genetic Landscape of Uveal Melanoma by Whole-Genome Sequencing. <i>American Journal of Human Genetics</i> , 2016, 99, 1190-1198.	6.2	135
13	NANS-mediated synthesis of sialic acid is required for brain and skeletal development. <i>Nature Genetics</i> , 2016, 48, 777-784.	21.4	125
14	PRPF mutations are associated with generalized defects in spliceosome formation and pre-mRNA splicing in patients with retinitis pigmentosa. <i>Human Molecular Genetics</i> , 2011, 20, 2116-2130.	2.9	120
15	Cortical-Bone Fragility â€™ Insights from sFRP4 Deficiency in Pyleâ€™s Disease. <i>New England Journal of Medicine</i> , 2016, 374, 2553-2562.	27.0	119
16	Whole genome sequencing in patients with retinitis pigmentosa reveals pathogenic DNA structural changes and <i>NEK2</i> as a new disease gene. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 16139-16144.	7.1	115
17	Barrier to Autointegration Factor Interacts with the Cone-Rod Homeobox and Represses Its Transactivation Function. <i>Journal of Biological Chemistry</i> , 2002, 277, 43288-43300.	3.4	112
18	A Missense Mutation in <i>PRPF6</i> Causes Impairment of pre-mRNA Splicing and Autosomal-Dominant Retinitis Pigmentosa. <i>American Journal of Human Genetics</i> , 2011, 88, 643-649.	6.2	110

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19	CNOT3 Is a Modifier of PRPF31 Mutations in Retinitis Pigmentosa with Incomplete Penetrance. PLoS Genetics, 2012, 8, e1003040.	3.5	109
20	Comprehensive screening of the USH2A gene in Usher syndrome type II and non-syndromic recessive retinitis pigmentosa. Experimental Eye Research, 2004, 79, 167-173.	2.6	108
21	Variation in retinitis pigmentosa-11 (PRPF31 or RP11) gene expression between symptomatic and asymptomatic patients with dominant RP11 mutations. Human Mutation, 2006, 27, 644-653.	2.5	100
22	Premature termination codons in PRPF31 cause retinitis pigmentosa via haploinsufficiency due to nonsense-mediated mRNA decay. Journal of Clinical Investigation, 2008, 118, 1519-1531.	8.2	96
23	Dominant Leber congenital amaurosis, cone-rod degeneration, and retinitis pigmentosa caused by mutant versions of the transcription factor CRX. Human Mutation, 2001, 18, 488-498.	2.5	91
24	DOMINO: Using Machine Learning to Predict Genes Associated with Dominant Disorders. American Journal of Human Genetics, 2017, 101, 623-629.	6.2	90
25	Novel Approach Identifies SNPs in SLC2A10 and KCNK9 with Evidence for Parent-of-Origin Effect on Body Mass Index. PLoS Genetics, 2014, 10, e1004508.	3.5	80
26	Nonsense Mutations in FAM161A Cause RP28-Associated Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2010, 87, 376-381.	6.2	76
27	EXTL3 mutations cause skeletal dysplasia, immune deficiency, and developmental delay. Journal of Experimental Medicine, 2017, 214, 623-637.	8.5	76
28	Structural Variants Create New Topological-Associated Domains and Ectopic Retinal Enhancer-Gene Contact in Dominant Retinitis Pigmentosa. American Journal of Human Genetics, 2020, 107, 802-814.	6.2	75
29	Loss of function mutations in HARS cause a spectrum of inherited peripheral neuropathies. Brain, 2015, 138, 2161-2172.	7.6	71
30	Genetic landscape of 6089 inherited retinal dystrophies affected cases in Spain and their therapeutic and extended epidemiological implications. Scientific Reports, 2021, 11, 1526.	3.3	71
31	Exome Sequencing of Index Patients with Retinal Dystrophies as a Tool for Molecular Diagnosis. PLoS ONE, 2013, 8, e65574.	2.5	71
32	Analysis of the genetic basis of periodic fever with aphthous stomatitis, pharyngitis and cervical adenitis (PFAPA) syndrome. Scientific Reports, 2015, 5, 10200.	3.3	70
33	AutoMap is a high performance homozygosity mapping tool using next-generation sequencing data. Nature Communications, 2021, 12, 518.	12.8	68
34	NBAS mutations cause a multisystem disorder involving bone, connective tissue, liver, immune system, and retina. American Journal of Medical Genetics, Part A, 2015, 167, 2902-2912.	1.2	66
35	Mutations in the heat-shock protein A9 (HSPA9) gene cause the EVEN-PLUS syndrome of congenital malformations and skeletal dysplasia. Scientific Reports, 2015, 5, 17154.	3.3	65
36	Paternal Uniparental Heterodisomy With Partial Isodisomy of Chromosome 1 in a Patient With Retinitis Pigmentosa Without Hearing Loss and a Missense Mutation in the Usher Syndrome Type II Gene USH2A. JAMA Ophthalmology, 2002, 120, 1566.	2.4	62

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37	A New CRB1 Rat Mutation Links Müller Glial Cells to Retinal Telangiectasia. <i>Journal of Neuroscience</i> , 2015, 35, 6093-6106.	3.6	54
38	Non-coding deletions identify Maenli lncRNA as a limb-specific En1 regulator. <i>Nature</i> , 2021, 592, 93-98.	27.8	53
39	Comparative genome analysis of <i>Pseudomonas knackmussii</i> , the first bacterium known to degrade chloroaromatic compounds. <i>Environmental Microbiology</i> , 2015, 17, 91-104.	3.8	52
40	Network-Guided Analysis of Genes with Altered Somatic Copy Number and Gene Expression Reveals Pathways Commonly Perturbed in Metastatic Melanoma. <i>PLoS ONE</i> , 2011, 6, e18369.	2.5	51
41	FAM161A, associated with retinitis pigmentosa, is a component of the cilia-basal body complex and interacts with proteins involved in ciliopathies. <i>Human Molecular Genetics</i> , 2012, 21, 5174-5184.	2.9	51
42	Genes Associated with Retinitis Pigmentosa and Allied Diseases Are Frequently Mutated in the General Population. <i>PLoS ONE</i> , 2012, 7, e41902.	2.5	50
43	Reciprocal modulation of mesenchymal stem cells and tumor cells promotes lung cancer metastasis. <i>EBioMedicine</i> , 2018, 29, 128-145.	6.1	50
44	Target Sequencing, Cell Experiments, and a Population Study Establish Endothelial Nitric Oxide Synthase (eNOS) Gene as Hypertension Susceptibility Gene. <i>Hypertension</i> , 2013, 62, 844-852.	2.7	48
45	A homozygous founder missense variant in arylsulfatase G abolishes its enzymatic activity causing atypical Usher syndrome in humans. <i>Genetics in Medicine</i> , 2018, 20, 1004-1012.	2.4	48
46	Two trans-acting eQTLs modulate the penetrance of PRPF31 mutations. <i>Human Molecular Genetics</i> , 2008, 17, 3154-3165.	2.9	47
47	Mineralocorticoid receptor antagonism limits experimental choroidal neovascularization and structural changes associated with neovascular age-related macular degeneration. <i>Nature Communications</i> , 2019, 10, 369.	12.8	47
48	UV light signature in conjunctival melanoma; not only skin should be protected from solar radiation. <i>Journal of Human Genetics</i> , 2016, 61, 361-362.	2.3	46
49	Identification of an RP1 Prevalent Founder Mutation and Related Phenotype in Spanish Patients with Early-Onset Autosomal Recessive Retinitis. <i>Ophthalmology</i> , 2012, 119, 2616-2621.	5.2	45
50	A single-base substitution within an intronic repetitive element causes dominant retinitis pigmentosa with reduced penetrance. <i>Human Mutation</i> , 2009, 30, 1340-1347.	2.5	44
51	Molecular Genetics of Charcot-Marie-Tooth Disease: From Genes to Genomes. <i>Molecular Syndromology</i> , 2012, 3, 204-214.	0.8	44
52	Exome Sequencing Extends the Phenotypic Spectrum for ABHD12 Mutations. <i>Ophthalmology</i> , 2014, 121, 1620-1627.	5.2	44
53	Mutations in CEP78 Cause Cone-Rod Dystrophy and Hearing Loss Associated with Primary-Cilia Defects. <i>American Journal of Human Genetics</i> , 2016, 99, 770-776.	6.2	44
54	Next generation sequencing of pooled samples reveals new SNRNP200 mutations associated with retinitis pigmentosa. <i>Human Mutation</i> , 2011, 32, E2246-E2258.	2.5	42

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55	Transcriptional regulation of PRPF31 gene expression by MSR1 repeat elements causes incomplete penetrance in retinitis pigmentosa. <i>Scientific Reports</i> , 2016, 6, 19450.	3.3	42
56	A Comprehensive Analysis of Choroideremia: From Genetic Characterization to Clinical Practice. <i>PLoS ONE</i> , 2016, 11, e0151943.	2.5	41
57	Novel frameshift mutations in CRX associated with Leber congenital amaurosis. <i>Human Mutation</i> , 2001, 18, 550-551.	2.5	40
58	Isolated and Syndromic Retinal Dystrophy Caused by Biallelic Mutations in RCBTB1 , a Gene Implicated in Ubiquitination. <i>American Journal of Human Genetics</i> , 2016, 99, 470-480.	6.2	39
59	Subunit II of Bacillus subtilis Cytochrome c Oxidase Is a Lipoprotein. <i>Journal of Bacteriology</i> , 1999, 181, 685-688.	2.2	37
60	The Liberfarb syndrome, a multisystem disorder affecting eye, ear, bone, and brain development, is caused by a founder pathogenic variant in the PISD gene. <i>Genetics in Medicine</i> , 2019, 21, 2734-2743.	2.4	33
61	Sequencing and characterizing the genome of <i>Estrella lausannensis</i> as an undergraduate project: training students and biological insights. <i>Frontiers in Microbiology</i> , 2015, 6, 101.	3.5	32
62	Bacillus subtilis Contains Two Small c-Type Cytochromes with Homologous Heme Domains but Different Types of Membrane Anchors. <i>Journal of Biological Chemistry</i> , 1999, 274, 26179-26184.	3.4	31
63	PLEKHG5 deficiency leads to an intermediate form of autosomal-recessive Charcot-Marie-Tooth disease. <i>Human Molecular Genetics</i> , 2013, 22, 4224-4232.	2.9	31
64	Identification of two novel mutations in CDHR1 in consanguineous Spanish families with autosomal recessive retinal dystrophy. <i>Scientific Reports</i> , 2015, 5, 13902.	3.3	30
65	Taurine treatment of retinal degeneration and cardiomyopathy in a consanguineous family with SLC6A6 taurine transporter deficiency. <i>Human Molecular Genetics</i> , 2020, 29, 618-623.	2.9	29
66	CEP78 functions downstream of CEP350 to control biogenesis of primary cilia by negatively regulating CP110 levels. <i>ELife</i> , 2021, 10, .	6.0	29
67	Analysis of missense variants in the human genome reveals widespread gene-specific clustering and improves prediction of pathogenicity. <i>American Journal of Human Genetics</i> , 2022, 109, 457-470.	6.2	29
68	Dominant PRPF31 Mutations Are Hypostatic to a Recessive CNOT3 Polymorphism in Retinitis Pigmentosa: A Novel Phenomenon of "Linked Trans-Acting Epistasis". <i>Annals of Human Genetics</i> , 2014, 78, 62-71.	0.8	28
69	Homozygosity mapping reveals novel and known mutations in Pakistani families with inherited retinal dystrophies. <i>Scientific Reports</i> , 2015, 5, 9965.	3.3	28
70	Mutations in the polyglutamylase gene TLL5, expressed in photoreceptor cells and spermatozoa, are associated with cone-rod degeneration and reduced male fertility. <i>Human Molecular Genetics</i> , 2016, 25, ddw282.	2.9	27
71	Transcriptomic Signature Differences Between SARS-CoV-2 and Influenza Virus Infected Patients. <i>Frontiers in Immunology</i> , 2021, 12, 666163.	4.8	27
72	Conjunctival Melanoma Targeted Therapy: MAPK and PI3K/mTOR Pathways Inhibition. , 2019, 60, 2764.		24

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73	Macular Dystrophy and Cone-Rod Dystrophy Caused by Mutations in the <i>RP1</i> Gene: Extending the <i>RP1</i> Disease Spectrum. , 2019, 60, 1192.		23
74	New clinical and molecular evidence linking mutations in <i>ARSG</i> to Usher syndrome type IV. Human Mutation, 2021, 42, 261-271.	2.5	23
75	A frequent variant in the Japanese population determines quasi-Mendelian inheritance of rare retinal ciliopathy. Nature Communications, 2019, 10, 2884.	12.8	21
76	A 35.7 kb DNA fragment from the Bacillus subtilis chromosome containing a putative 12.3 kb operon involved in hexuronate catabolism and a perfectly symmetrical hypothetical catabolite-responsive element. Microbiology (United Kingdom), 1998, 144, 877-884.	1.8	20
77	Whole genome sequencing as a means to assess pathogenic mutations in medical genetics and cancer. Cellular and Molecular Life Sciences, 2015, 72, 1463-1471.	5.4	20
78	Genotype-Phenotype Correlations in a Spanish Cohort of 506 Families With Biallelic ABCA4 Pathogenic Variants. American Journal of Ophthalmology, 2020, 219, 195-204.	3.3	20
79	Interactome analysis reveals that FAM161A, deficient in recessive retinitis pigmentosa, is a component of the Golgi-centrosomal network. Human Molecular Genetics, 2015, 24, 3359-3371.	2.9	19
80	Two specific mutations are prevalent causes of recessive retinitis pigmentosa in North American patients of Jewish ancestry. Genetics in Medicine, 2015, 17, 285-290.	2.4	19
81	Mutations in ARL2BP, a protein required for ciliary microtubule structure, cause syndromic male infertility in humans and mice. PLoS Genetics, 2019, 15, e1008315.	3.5	19
82	Genomic and transcriptomic landscape of conjunctival melanoma. PLoS Genetics, 2020, 16, e1009201.	3.5	18
83	Brief Report: Peripheral Osteolysis in Adults Linked to <i>ASAH1</i> (Acid Ceramidase) Mutations: A New Presentation of Farber's Disease. Arthritis and Rheumatology, 2016, 68, 2323-2327.	5.6	17
84	Expanded Phenotypic Spectrum of Retinopathies Associated with Autosomal Recessive and Dominant Mutations in PROM1. American Journal of Ophthalmology, 2019, 207, 204-214.	3.3	17
85	Herpes simplex encephalitis in adult patients with MASP-2 deficiency. PLoS Pathogens, 2019, 15, e1008168.	4.7	17
86	Peripheral neuropathy and cognitive impairment associated with a novel monoallelic <i>HARS</i> variant. Annals of Clinical and Translational Neurology, 2019, 6, 1072-1080.	3.7	15
87	Functional characterization of the first missense variant in <i>CEP78</i> , a founder allele associated with cone-rod dystrophy, hearing loss, and reduced male fertility. Human Mutation, 2020, 41, 998-1011.	2.5	15
88	Childhood neurodegeneration associated with a specific UBTF variant: a new case report and review of the literature. BMC Neurology, 2020, 20, 17.	1.8	15
89	<i>PRPF31</i> Alternative Splicing and Expression in Human Retina. Ophthalmic Genetics, 2009, 30, 76-83.	1.2	14
90	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

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91	Improved Diagnosis of Rare Disease Patients through Systematic Detection of Runs of Homozygosity. <i>Journal of Molecular Diagnostics</i> , 2020, 22, 1205-1215.	2.8	14
92	Clinicopathologic and Molecular Analysis of a Choroidal Pigmented Schwannoma in the Context of a PTEN Hamartoma Tumor Syndrome. <i>Ophthalmology</i> , 2012, 119, 857-864.	5.2	13
93	Molecular Genetics of FAM161A in North American Patients with Early-Onset Retinitis Pigmentosa. <i>PLoS ONE</i> , 2014, 9, e92479.	2.5	13
94	<i>OR2W3</i> sequence variants are unlikely to cause inherited retinal diseases. <i>Ophthalmic Genetics</i> , 2016, 37, 366-368.	1.2	13
95	Exploring the Genetic Landscape of Retinal Diseases in North-Western Pakistan Reveals a High Degree of Autozygosity and a Prevalent Founder Mutation in ABCA4. <i>Genes</i> , 2020, 11, 12.	2.4	13
96	Clinical characteristics and high resolution retinal imaging of retinitis pigmentosa caused by RP1 gene variants. <i>Japanese Journal of Ophthalmology</i> , 2020, 64, 485-496.	1.9	13
97	The product of the <i>yvoC(geri)</i> gene of <i>Bacillus subtilis</i> is required for spore germination. <i>Microbiology (United Kingdom)</i> , 1998, 144, 3105-3109.	1.8	12
98	Ultra High Throughput Sequencing in Human DNA Variation Detection: A Comparative Study on the NDUFA3-PRPF31 Region. <i>PLoS ONE</i> , 2010, 5, e13071.	2.5	11
99	Mutational screening of splicing factor genes in cases with autosomal dominant retinitis pigmentosa. <i>Molecular Vision</i> , 2014, 20, 843-51.	1.1	11
100	Mutation screening of the glutamate cysteine ligase modifier (GCLM) gene in patients with schizophrenia. <i>Psychiatric Genetics</i> , 2009, 19, 201-208.	1.1	10
101	A novel missense variant in IDH3A causes autosomal recessive retinitis pigmentosa. <i>Ophthalmic Genetics</i> , 2019, 40, 177-181.	1.2	10
102	Whole exome sequencing and homozygosity mapping reveals genetic defects in consanguineous Iranian families with inherited retinal dystrophies. <i>Scientific Reports</i> , 2020, 10, 19413.	3.3	9
103	NGS and phenotypic ontology-based approaches increase the diagnostic yield in syndromic retinal diseases. <i>Human Genetics</i> , 2021, 140, 1665-1678.	3.8	9
104	Broadening INPP5E phenotypic spectrum: detection of rare variants in syndromic and non-syndromic IRD. <i>Npj Genomic Medicine</i> , 2021, 6, 53.	3.8	8
105	New variants and in silico analyses in GRK1 associated Oguchi disease. <i>Human Mutation</i> , 2021, 42, 164-176.	2.5	7
106	A hypomorphic variant in EYS detected by genome-wide association study contributes toward retinitis pigmentosa. <i>Communications Biology</i> , 2021, 4, 140.	4.4	6
107	A large multiexonic genomic deletion within the <i>ALMS1</i> gene causes Alström syndrome in a consanguineous Pakistani family. <i>Clinical Genetics</i> , 2016, 89, 510-511.	2.0	5
108	Evaluation of the ELOVL4 gene in patients with autosomal recessive retinitis pigmentosa and Leber congenital amaurosis. <i>Molecular Vision</i> , 2003, 9, 49-51.	1.1	5

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109	Immune deficiency, autoimmune disease and intellectual disability: A pleiotropic disorder caused by biallelic variants in the <i>TPP2</i> gene. <i>Clinical Genetics</i> , 2021, 99, 780-788.	2.0	4
110	Mutations in the ribosome biogenesis factor gene <i>LTV1</i> are linked to LIPHAK syndrome, a novel poikiloderma-like disorder. <i>Human Molecular Genetics</i> , 2022, 31, 1970-1978.	2.9	4
111	Whole-exome sequencing in a consanguineous Pakistani family identifies a mutational hotspot in the <i>COL7A1</i> gene, causing recessive dystrophic epidermolysis bullosa. <i>Clinical Dysmorphology</i> , 2020, 29, 86-89.	0.3	3
112	c.-61G>A in <i>OVOL2</i> is a Pathogenic 5' UTR Variant Causing Posterior Polymorphous Corneal Dystrophy 1. <i>Cornea</i> , 2022, 41, 89-94.	1.7	3
113	Genetic and Physical Maps of the <i>Bacillus subtilis</i> Chromosome. <i>Genetics</i> , 1999, 151, 1239-1244.	2.9	3
114	Ultra high throughput sequencing excludes <i>MDH1</i> as candidate gene for RP28-linked retinitis pigmentosa. <i>Molecular Vision</i> , 2009, 15, 2627-33.	1.1	3
115	Heterozygous deletions of noncoding parts of the gene cause retinitis pigmentosa via reduced gene expression. <i>Molecular Vision</i> , 2021, 27, 107-116.	1.1	3
116	Mutation screening of the peropsin gene, a retinal pigment epithelium specific rhodopsin homolog, in patients with retinitis pigmentosa and allied diseases. <i>Molecular Vision</i> , 2006, 12, 1511-5.	1.1	3
117	Retinitis pigmentosa and allied diseases: numerous diseases, genes, and inheritance patterns. <i>Human Molecular Genetics</i> , 2003, 12, 583-584.	2.9	2
118	Management of Full-Thickness Macular Hole in A Genetically Confirmed Case with Usher Syndrome. <i>Ophthalmology and Therapy</i> , 2020, 9, 677-684.	2.3	2
119	Genetic predisposition and environmental factors associated with the development of atopic dermatitis in infancy: a prospective birth cohort study. <i>European Journal of Pediatrics</i> , 2020, 179, 1367-1377.	2.7	2
120	Whole exome sequencing in 17 consanguineous Iranian pedigrees expands the mutational spectrum of inherited retinal dystrophies. <i>Scientific Reports</i> , 2021, 11, 19332.	3.3	2
121	Agenesis of the Corpus Callosum with Facial Dysmorphism and Intellectual Disability in Sibs Associated with Compound Heterozygous <i>KDM5B</i> Variants. <i>Genes</i> , 2021, 12, 1397.	2.4	1
122	Genetic Analysis of Consanguineous Pakistani Families with Congenital Stationary Night Blindness. <i>Ophthalmic Research</i> , 2022, 65, 104-110.	1.9	1
123	Identification of New Vulnerabilities in Conjunctival Melanoma Using Image-Based High Content Drug Screening. <i>Cancers</i> , 2022, 14, 1575.	3.7	0
124	A novel phenotype associated with the R162W variant in the <i>KCNJ13</i> gene. <i>Ophthalmic Genetics</i> , 2022, , 1-8.	1.2	0