Carlo Rivolta

List of Publications by Year
in descending order

124	12,889	57758 44	24982 109
papers	citations	h-index	g-index

134	134	134	19777
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	c.-61G>A in OVOL2 is a Pathogenic 5â€² Untranslated Region Variant Causing Posterior Polymorphous Corneal Dystrophy 1. Cornea, 2022, 41, 89-94.	1.7	3
2	Mutations in the ribosome biogenesis factor gene LTV1 are linked to LIPHAK syndrome, a novel poikiloderma-like disorder. Human Molecular Genetics, 2022, 31, 1970-1978.	2.9	4
3	Analysis of missense variants in the human genome reveals widespread gene-specific clustering and improves prediction of pathogenicity. American Journal of Human Genetics, 2022, 109, 457-470.	6.2	29
4	Identification of New Vulnerabilities in Conjunctival Melanoma Using Image-Based High Content Drug Screening. Cancers, 2022, 14, 1575.	3.7	0
5	Genetic Analysis of Consanguineous Pakistani Families with Congenital Stationary Night Blindness. Ophthalmic Research, 2022, 65, 104-110.	1.9	1
6	A novel phenotype associated with the R162W variant in the KCNJ13 gene. Ophthalmic Genetics, 2022, , 1-8.	1.2	0
7	New clinical and molecular evidence linking mutations in ARSG to UsherÂsyndrome type IV. Human Mutation, 2021, 42, 261-271.	2.5	23
8	New variants and in silico analyses in GRK1 associated Oguchi disease. Human Mutation, 2021, 42, 164-176.	2.5	7
9	A hypomorphic variant in EYS detected by genome-wide association study contributes toward retinitis pigmentosa. Communications Biology, 2021, 4, 140.	4.4	6
10	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
11	Non-coding deletions identify Maenli lncRNA as a limb-specific En1 regulator. Nature, 2021, 592, 93-98.	27.8	53
12	Immune deficiency, autoimmune disease and intellectual disability: A pleiotropic disorder caused by biallelic variants in the <scp>TPP2</scp> gene. Clinical Genetics, 2021, 99, 780-788.	2.0	4
13	Transcriptomic Signature Differences BetweenÂSARS-CoV-2 and Influenza Virus Infected Patients. Frontiers in Immunology, 2021, 12, 666163.	4.8	27
14	Broadening INPP5E phenotypic spectrum: detection of rare variants in syndromic and non-syndromic IRD. Npj Genomic Medicine, 2021, 6, 53.	3.8	8
15	CEP78 functions downstream of CEP350 to control biogenesis of primary cilia by negatively regulating CP110 levels. ELife, 2021, 10, .	6.0	29
16	NGS and phenotypic ontology-based approaches increase the diagnostic yield in syndromic retinal diseases. Human Genetics, 2021, 140, 1665-1678.	3.8	9
17	Agenesis of the Corpus Callosum with Facial Dysmorphism and Intellectual Disability in Sibs Associated with Compound Heterozygous KDM5B Variants. Genes, 2021, 12, 1397.	2.4	1
18	Whole exome sequencing in 17 consanguineous Iranian pedigrees expands the mutational spectrum of inherited retinal dystrophies. Scientific Reports, 2021, 11, 19332.	3.3	2

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19	AutoMap is a high performance homozygosity mapping tool using next-generation sequencing data. Nature Communications, 2021, 12, 518.	12.8	68
20	Heterozygous deletions of noncoding parts of the gene cause retinitis pigmentosa via reduced gene expression. Molecular Vision, 2021, 27, 107-116.	1.1	3
21	Taurine treatment of retinal degeneration and cardiomyopathy in a consanguineous family with SLC6A6 taurine transporter deficiency. Human Molecular Genetics, 2020, 29, 618-623.	2.9	29
22	Whole-exome sequencing in a consanguineous Pakistani family identifies a mutational hotspot in the COL7A1 gene, causing recessive dystrophic epidermolysis bullosa. Clinical Dysmorphology, 2020, 29, 86-89.	0.3	3
23	Exploring the Genetic Landscape of Retinal Diseases in North-Western Pakistan Reveals a High Degree of Autozygosity and a Prevalent Founder Mutation in ABCA4. Genes, 2020, 11, 12.	2.4	13
24	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
25	Management of Full-Thickness Macular Hole in A Genetically Confirmed Case with Usher Syndrome. Ophthalmology and Therapy, 2020, 9, 677-684.	2.3	2
26	Clinical characteristics and high resolution retinal imaging of retinitis pigmentosa caused by RP1 gene variants. Japanese Journal of Ophthalmology, 2020, 64, 485-496.	1.9	13
27	Whole exome sequencing and homozygosity mapping reveals genetic defects in consanguineous Iranian families with inherited retinal dystrophies. Scientific Reports, 2020, 10, 19413.	3.3	9
28	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
29	Genetic predisposition and environmental factors associated with the development of atopic dermatitis in infancy: a prospective birth cohort study. European Journal of Pediatrics, 2020, 179, 1367-1377.	2.7	2
30	Improved Diagnosis of Rare Disease Patients through Systematic Detection of Runs of Homozygosity. Journal of Molecular Diagnostics, 2020, 22, 1205-1215.	2.8	14
31	Genotypeâ€“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
32	Worldwide carrier frequency and genetic prevalence of autosomal recessive inherited retinal diseases. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 2710-2716.	7.1	195
33	Functional characterization of the first missense variant in CEP78 , a founder allele associated with coneâ€rod dystrophy, hearing loss, and reduced male fertility. Human Mutation, 2020, 41, 998-1011.	2.5	15
34	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
35	Genomic and transcriptomic landscape of conjunctival melanoma. PLoS Genetics, 2020, 16, e1009201.	3.5	18
36	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

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37	Conjunctival Melanoma Targeted Therapy: MAPK and PI3K/mTOR Pathways Inhibition. , 2019, 60, 2764.		24
38	A frequent variant in the Japanese population determines quasi-Mendelian inheritance of rare retinal ciliopathy. Nature Communications, 2019, 10, 2884.	12.8	21
39	The Liberfarb syndrome, a multisystem disorder affecting eye, ear, bone, and brain development, is caused by a founder pathogenic variant in the PISD gene. Genetics in Medicine, 2019, 21, 2734-2743.	2.4	33
40	Mineralocorticoid receptor antagonism limits experimental choroidal neovascularization and structural changes associated with neovascular age-related macular degeneration. Nature Communications, 2019, 10, 369.	12.8	47
41	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
42	Peripheral neuropathy and cognitive impairment associated with a novel monoallelic <scp>HARS</scp> variant. Annals of Clinical and Translational Neurology, 2019, 6, 1072-1080.	3.7	15
43	A novel missense variant in IDH3A causes autosomal recessive retinitis pigmentosa. Ophthalmic Genetics, 2019, 40, 177-181.	1.2	10
44	Macular Dystrophy and Cone-Rod Dystrophy Caused by Mutations in theRP1Gene: Extending theRP1Disease Spectrum. , 2019, 60, 1192.		23
45	Herpes simplex encephalitis in adult patients with MASP-2 deficiency. PLoS Pathogens, 2019, 15, e1008168.	4.7	17
46	Reciprocal modulation of mesenchymal stem cells and tumor cells promotes lung cancer metastasis. EBioMedicine, 2018, 29, 128-145.	6.1	50
47	A homozygous founder missense variant in arylsulfatase G abolishes its enzymatic activity causing atypical Usher syndrome in humans. Genetics in Medicine, 2018, 20, 1004-1012.	2.4	48
48	EXTL3 mutations cause skeletal dysplasia, immune deficiency, and developmental delay. Journal of Experimental Medicine, 2017, 214, 623-637.	8.5	76
49	DOMINO: Using Machine Learning to Predict Genes Associated with Dominant Disorders. American Journal of Human Genetics, 2017, 101, 623-629.	6.2	90
50	A large multiexonic genomic deletion within the <scp>ALMS1</scp> gene causes AlstrÃ¶m syndrome in a consanguineous Pakistani family. Clinical Genetics, 2016, 89, 510-511.	2.0	5
51	NANS-mediated synthesis of sialic acid is required for brain and skeletal development. Nature Genetics, 2016, 48, 777-784.	21.4	125
52	UV light signature in conjunctival melanoma; not only skin should be protected from solar radiation. Journal of Human Genetics, 2016, 61, 361-362.	2.3	46
53	Isolated and Syndromic Retinal Dystrophy Caused by Biallelic Mutations in RCBTB1 , a Gene Implicated in Ubiquitination. American Journal of Human Genetics, 2016, 99, 470-480.	6.2	39
54	Mutations in the polyglutamylase geneTTLL5, expressed in photoreceptor cells and spermatozoa, are associated with cone-rod degeneration and reduced male fertility. Human Molecular Genetics, 2016, 25, ddw282.	2.9	27

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55	Mutations in CEP78 Cause Cone-Rod Dystrophy and Hearing Loss Associated with Primary-Cilia Defects. American Journal of Human Genetics, 2016, 99, 770-776.	6.2	44
56	Comprehensive Genetic Landscape of Uveal Melanoma by Whole-Genome Sequencing. American Journal of Human Genetics, 2016, 99, 1190-1198.	6.2	135
57	Transcriptional regulation of PRPF31 gene expression by MSR1 repeat elements causes incomplete penetrance in retinitis pigmentosa. Scientific Reports, 2016, 6, 19450.	3.3	42
58	Cortical-Bone Fragility â€" Insights from sFRP4 Deficiency in Pyleâ€™s Disease. New England Journal of Medicine, 2016, 374, 2553-2562.	27.0	119
59	Brief Report: Peripheral Osteolysis in Adults Linked to ASAH1 (Acid Ceramidase) Mutations: A New Presentation of Farber's Disease. Arthritis and Rheumatology, 2016, 68, 2323-2327.	5.6	17
60	OR2W3 sequence variants are unlikely to cause inherited retinal diseases. Ophthalmic Genetics, 2016, 37, 366-368.	1.2	13
61	A Comprehensive Analysis of Choroideremia: From Genetic Characterization to Clinical Practice. PLoS ONE, 2016, 11, e0151943.	2.5	41
62	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
63	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
64	Identification of two novel mutations in CDHR1 in consanguineous Spanish families with autosomal recessive retinal dystrophy. Scientific Reports, 2015, 5, 13902.	3.3	30
65	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
66	The Influence of Age and Sex on Genetic Associations with Adult Body Size and Shape: A Large-Scale Genome-Wide Interaction Study. PLoS Genetics, 2015, 11, e1005378.	3.5	331
67	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
68	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
69	Loss of function mutations inHARScause a spectrum of inherited peripheral neuropathies. Brain, 2015, 138, 2161-2172.	7.6	71
70	Homozygosity mapping reveals novel and known mutations in Pakistani families with inherited retinal dystrophies. Scientific Reports, 2015, 5, 9965.	3.3	28
71	Sequencing and characterizing the genome of Estrella lausannensis as an undergraduate project: training students and biological insights. Frontiers in Microbiology, 2015, 6, 101.	3.5	32
72	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

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73	A New CRB1 Rat Mutation Links MÃ¼ller Glial Cells to Retinal Telangiectasia. Journal of Neuroscience, 2015, 35, 6093-6106.	3.6	54
74	Comparative genome analysis of <scp>P</scp>seudomonas knackmussiiâ€...<scp>B</scp>13, the first bacterium known to degrade chloroaromatic compounds. Environmental Microbiology, 2015, 17, 91-104.	3.8	52
75	Molecular Genetics of FAM161A in North American Patients with Early-Onset Retinitis Pigmentosa. PLoS ONE, 2014, 9, e92479.	2.5	13
76	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
77	DominantPRPF31Mutations Are Hypostatic to a RecessiveCNOT3Polymorphism in Retinitis Pigmentosa: A Novel Phenomenon of â€œLinkedTrans-Acting Epistasisâ€• Annals of Human Genetics, 2014, 78, 62-71.	0.8	28
78	Exome Sequencing Extends the Phenotypic Spectrum for ABHD12 Mutations. Ophthalmology, 2014, 121, 1620-1627.	5.2	44
79	Mutational screening of splicing factor genes in cases with autosomal dominant retinitis pigmentosa. Molecular Vision, 2014, 20, 843-51.	1.1	11
80	Target Sequencing, Cell Experiments, and a Population Study Establish Endothelial Nitric Oxide Synthase (eNOS) Gene as Hypertension Susceptibility Gene. Hypertension, 2013, 62, 844-852.	2.7	48
81	Sex-stratified Genome-wide Association Studies Including 270,000 Individuals Show Sexual Dimorphism in Genetic Loci for Anthropometric Traits. PLoS Genetics, 2013, 9, e1003500.	3.5	371
82	PLEKHG5 deficiency leads to an intermediate form of autosomal-recessive Charcotâ€“Marieâ€“Tooth disease. Human Molecular Genetics, 2013, 22, 4224-4232.	2.9	31
83	Whole genome sequencing in patients with retinitis pigmentosa reveals pathogenic DNA structural changes and NEK2 as a new disease gene. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 16139-16144.	7.1	115
84	Exome Sequencing of Index Patients with Retinal Dystrophies as a Tool for Molecular Diagnosis. PLoS ONE, 2013, 8, e65574.	2.5	71
85	CNOT3 Is a Modifier of PRPF31 Mutations in Retinitis Pigmentosa with Incomplete Penetrance. PLoS Genetics, 2012, 8, e1003040.	3.5	109
86	FAM161A, associated with retinitis pigmentosa, is a component of the cilia-basal body complex and interacts with proteins involved in ciliopathies. Human Molecular Genetics, 2012, 21, 5174-5184.	2.9	51
87	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. Hypertension, 2012, 59, 248-255.	2.7	144
88	Molecular Genetics of Charcot-Marie-Tooth Disease: From Genes to Genomes. Molecular Syndromology, 2012, 3, 204-214.	0.8	44
89	Clinicopathologic and Molecular Analysis of a Choroidal Pigmented Schwannoma in the Context of a PTEN Hamartoma Tumor Syndrome. Ophthalmology, 2012, 119, 857-864.	5.2	13
90	Identification of an RP1 Prevalent Founder Mutation and Related Phenotype in Spanish Patients with Early-Onset Autosomal Recessive Retinitis. Ophthalmology, 2012, 119, 2616-2621.	5.2	45

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91	Genes Associated with Retinitis Pigmentosa and Allied Diseases Are Frequently Mutated in the General Population. PLoS ONE, 2012, 7, e41902.	2.5	50
92	Network-Guided Analysis of Genes with Altered Somatic Copy Number and Gene Expression Reveals Pathways Commonly Perturbed in Metastatic Melanoma. PLoS ONE, 2011, 6, e18369.	2.5	51
93	A Missense Mutation in PRPF6 Causes Impairment of pre-mRNA Splicing and Autosomal-Dominant Retinitis Pigmentosa. American Journal of Human Genetics, 2011, 88, 643-649.	6.2	110
94	Next generation sequencing of pooled samples reveals new SNRNP200 mutations associated with retinitis pigmentosa. Human Mutation, 2011, 32, E2246-E2258.	2.5	42
95	PRPF mutations are associated with generalized defects in spliceosome formation and pre-mRNA splicing in patients with retinitis pigmentosa. Human Molecular Genetics, 2011, 20, 2116-2130.	2.9	120
96	Nonsense Mutations in FAM161A Cause RP28-Associated Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2010, 87, 376-381.	6.2	76
97	Hundreds of variants clustered in genomic loci and biological pathways affect human height. Nature, 2010, 467, 832-838.	27.8	1,789
98	Ultra High Throughput Sequencing in Human DNA Variation Detection: A Comparative Study on the NDUFA3-PRPF31 Region. PLoS ONE, 2010, 5, e13071.	2.5	11
99	PRPF31Alternative Splicing and Expression in Human Retina. Ophthalmic Genetics, 2009, 30, 76-83.	1.2	14
100	A single-base substitution within an intronic repetitive element causes dominant retinitis pigmentosa with reduced penetrance. Human Mutation, 2009, 30, 1340-1347.	2.5	44
101	Mutation screening of the glutamate cysteine ligase modifier (GCLM) gene in patients with schizophrenia. Psychiatric Genetics, 2009, 19, 201-208.	1.1	10
102	Ultra high throughput sequencing excludes MDH1 as candidate gene for RP28-linked retinitis pigmentosa. Molecular Vision, 2009, 15, 2627-33.	1.1	3
103	Two trans-acting eQTLs modulate the penetrance of PRPF31 mutations. Human Molecular Genetics, 2008, 17, 3154-3165.	2.9	47
104	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
105	The Zinc Transporter SLC39A13/ZIP13 Is Required for Connective Tissue Development; Its Involvement in BMP/TGF-Î² Signaling Pathways. PLoS ONE, 2008, 3, e3642.	2.5	240
106	Variation in retinitis pigmentosa-11 (PRPF31orRP11) gene expression between symptomatic and asymptomatic patients with dominantRP11mutations. Human Mutation, 2006, 27, 644-653.	2.5	100
107	Mutation screening of the peropsin gene, a retinal pigment epithelium specific rhodopsin homolog, in patients with retinitis pigmentosa and allied diseases. Molecular Vision, 2006, 12, 1511-5.	1.1	3
108	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

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109	Essential Bacillus subtilis genes. Proceedings of the National Academy of Sciences of the United States of America, 2003, 100, 4678-4683.	7.1	1,261
110	Retinitis pigmentosa and allied diseases: numerous diseases, genes, and inheritance patterns. Human Molecular Genetics, 2003, 12, 583-584.	2.9	2
111	Evaluation of the ELOVL4 gene in patients with autosomal recessive retinitis pigmentosa and Leber congenital amaurosis. Molecular Vision, 2003, 9, 49-51.	1.1	5
112	Retinitis pigmentosa and allied diseases: numerous diseases, genes, and inheritance patterns. Human Molecular Genetics, 2002, 11, 1219-1227.	2.9	251
113	Barrier to Autointegration Factor Interacts with the Cone-Rod Homeobox and Represses Its Transactivation Function. Journal of Biological Chemistry, 2002, 277, 43288-43300.	3.4	112
114	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
115	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
116	Novel frameshift mutations inCRX associated with Leber congenital amaurosis. Human Mutation, 2001, 18, 550-551.	2.5	40
117	Missense Mutation in the USH2A Gene: Association with Recessive Retinitis Pigmentosa without Hearing Loss. American Journal of Human Genetics, 2000, 66, 1975-1978.	6.2	296
118	Bacillus subtilis Contains Two Small c-Type Cytochromes with Homologous Heme Domains but Different Types of Membrane Anchors. Journal of Biological Chemistry, 1999, 274, 26179-26184.	3.4	31
119	Subunit II of Bacillus subtilis Cytochrome c Oxidase Is a Lipoprotein. Journal of Bacteriology, 1999, 181, 685-688.	2.2	37
120	Genetic and Physical Maps of the Bacillus subtilis Chromosome. Genetics, 1999, 151, 1239-1244.	2.9	3
121	A novel protein kinase that controls carbon catabolite repression in bacteria. Molecular Microbiology, 1998, 27, 1157-1169.	2.5	205
122	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
123	The product of the yvoC(geri)gene of Bacillus subtillis is required for spore germination. Microbiology (United Kingdom), 1998, 144, 3105-3109.	1.8	12
124	The complete genome sequence of the Gram-positive bacterium Bacillus subtilis. Nature, 1997, 390, 249-256.	27.8	3,519