Carmen Ayuso

List of Publications by Year
in descending order

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Version: 2024-02-01

350 papers	24,308 citations	17776 65 h-index	10955 142 g-index
376 all docs	376 docs citations	376 times ranked	20923 citing authors

#	Article	IF	Citations
1	Liver Imaging Reporting and Data System: Review of Pros and Cons. Seminars in Liver Disease, 2022, 42, 104-111.	1.8	2
2	Liver cancer risk after HCV cure in patients with advanced liver disease without non-characterized nodules. Journal of Hepatology, 2022, 76, 874-882.	1.8	17
3	BCLC strategy for prognosis prediction and treatment recommendation: The 2022 update. Journal of Hepatology, 2022, 76, 681-693.	1.8	1,495
4	An evaluation of pipelines for DNA variant detection can guide a reanalysis protocol to increase the diagnostic ratio of genetic diseases. Npj Genomic Medicine, 2022, 7, 7.	1.7	8
5	Impact of Next Generation Sequencing in Unraveling the Genetics of 1036 Spanish Families With Inherited Macular Dystrophies. , 2022, 63, 11.		11
6	First evidence of <scp>SOX2</scp> mutations in Peters' anomaly: Lessons from molecular screening of 95 patients. Clinical Genetics, 2022, 101, 494-506.	1.0	9
7	SARS-CoV-2 Point Mutation and Deletion Spectra and Their Association with Different Disease Outcomes. Microbiology Spectrum, 2022, 10, e0022122.	1.2	10
8	Vaccine breakthrough infections with SARS-CoV-2 Alpha mirror mutations in Delta Plus, Iota, and Omicron. Journal of Clinical Investigation, 2022, 132, .	3.9	10
9	Comprehensive variant spectrum of the CNGA3 gene in patients affected by achromatopsia. Human Mutation, 2022, 43, 832-858.	1.1	8
10	Reliability of extracellular contrast versus gadoxetic acid in assessing small liver lesions using liver imaging reporting and data system v.2018 and European association for the study of the liver criteria. Hepatology, 2022, 76, 1318-1328.	3.6	10
11	Prevalence, multimodal imaging and genotype-phenotype assessment of trauma related subretinal fibrosis in stargardt disease. European Journal of Ophthalmology, 2022, , 112067212210939.	0.7	0
12	Presence of rare potential pathogenic variants in subjects under 65Âyears old with very severe or fatal COVID-19. Scientific Reports, 2022, 12, .	1.6	6
13	Novel genes and sex differences in COVID-19 severity. Human Molecular Genetics, 2022, 31, 3789-3806.	1.4	38
14	SARS-CoV-2 Mutant Spectra at Different Depth Levels Reveal an Overwhelming Abundance of Low Frequency Mutations. Pathogens, 2022, 11, 662.	1.2	16
15	Portal hypertension may influence the registration of hypointensity of small hepatocellular carcinoma in the hepatobiliary phase in gadoxetic acid MR. Radiology and Oncology, 2022, 56, 292-302.	0.6	0
16	Allelic overload and its clinical modifier effect in Bardet-Biedl syndrome. Npj Genomic Medicine, 2022, 7, .	1.7	7
17	For how long and with what relevance do genetics articles retracted due to research misconduct remain active in the scientific literature. Accountability in Research, 2021, 28, 280-296.	1.6	11
18	Pathogenic variants in IMPG1 cause autosomal dominant and autosomal recessive retinitis pigmentosa. Journal of Medical Genetics, 2021, 58, 570-578.	1.5	10

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19	CSVS, a crowdsourcing database of the Spanish population genetic variability. Nucleic Acids Research, 2021, 49, D1130-D1137.	6.5	34
20	Evaluation of LI-RADS 3 category by magnetic resonance in US-detected nodules â‰¥â€‰2 cm in cirrhotic patients. European Radiology, 2021, 31, 4794-4803.	2.3	8
21	Genetic landscape of 6089 inherited retinal dystrophies affected cases in Spain and their therapeutic and extended epidemiological implications. Scientific Reports, 2021, 11, 1526.	1.6	71
22	Prevalent ALMS1 Pathogenic Variants in Spanish AlstrÃ¶m Patients. Genes, 2021, 12, 282.	1.0	4
23	Gene Correction Recovers Phagocytosis in Retinal Pigment Epithelium Derived from Retinitis Pigmentosa-Human-Induced Pluripotent Stem Cells. International Journal of Molecular Sciences, 2021, 22, 2092.	1.8	10
24	Prioritizing variants of uncertain significance for reclassification using a rule-based algorithm in inherited retinal dystrophies. Npj Genomic Medicine, 2021, 6, 18.	1.7	20
25	Genotypeâ€“phenotype correlation in patients with Usher syndrome and pathogenic variants in MYO7A: implications for future clinical trials. Acta Ophthalmologica, 2021, 99, 922-930.	0.6	8
26	Radiological response to nivolumab in patients with hepatocellular carcinoma: A multicenter analysis of real-life practice. European Journal of Radiology, 2021, 135, 109484.	1.2	20
27	Sanger sequencing is no longer always necessary based on a single-center validation of 1109 NGS variants in 825 clinical exomes. Scientific Reports, 2021, 11, 5697.	1.6	28
28	Comparison of the diagnostic yield of aCGH and genome-wide sequencing across different neurodevelopmental disorders. Npj Genomic Medicine, 2021, 6, 25.	1.7	27
29	Activation of cryptic donor splice sites by non-coding and coding PAX6 variants contributes to congenital aniridia. Journal of Medical Genetics, 2021, , jmedgenet-2020-106932.	1.5	8
30	KCNV2-Associated Retinopathy: Detailed Retinal Phenotype and Structural Endpointsâ€”KCNV2 Study Group Report 2. American Journal of Ophthalmology, 2021, 230, 1-11.	1.7	11
31	KCNV2-Associated Retinopathy: Genetics, Electrophysiology, and Clinical Courseâ€”KCNV2 Study Group Report 1. American Journal of Ophthalmology, 2021, 225, 95-107.	1.7	17
32	Diagnosis and treatment of hepatocellular carcinoma. Update of the consensus document of the AEEH, AEC, SEOM, SERAM, SERVEI, and SETH. Medicina ClÃ¡nica (English Edition), 2021, 156, 463.e1-463.e30.	0.1	16
33	Schuursâ€“Hoeijmakers Syndrome (PACS1 Neurodevelopmental Disorder): Seven Novel Patients and a Review. Genes, 2021, 12, 738.	1.0	13
34	Apparent but unconfirmed digenism in an Iranian consanguineous family with syndromic Retinal Disease. Experimental Eye Research, 2021, 207, 108533.	1.2	1
35	Limited tumour progression beyond Milan criteria while on the waiting list does not result in unacceptable impairment of survival. Journal of Hepatology, 2021, 75, 1154-1163.	1.8	9
36	EarlyÂdiarrhoea under sorafenib as a marker to consider the early migration to secondâ€line drugs. United European Gastroenterology Journal, 2021, 9, 655-661.	1.6	2

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37	GenÃ©tica y epidemiologÃa de la aniridia congÃ©nita: actualizaciÃ³n de buenas prÃ¡cticas para el diagnÃ³stico genÃ©tico. Archivos De La Sociedad Espanola De Oftalmologia, 2021, 96, 4-14.	0.1	6
38	NGS and phenotypic ontology-based approaches increase the diagnostic yield in syndromic retinal diseases. Human Genetics, 2021, 140, 1665-1678.	1.8	9
39	Attention Deficit Hyperactivity and Autism Spectrum Disorders as the Core Symptoms of AUTS2 Syndrome: Description of Five New Patients and Update of the Frequency of Manifestations and Genotype-Phenotype Correlation. Genes, 2021, 12, 1360.	1.0	16
40	High SARS-CoV-2 viral load is associated with a worse clinical outcome of COVID-19 disease. Access Microbiology, 2021, 3, 000259.	0.2	13
41	Homozygous females for a X-linked RPGR-ORF15 mutation in an Iranian family with retinitis pigmentosa. Experimental Eye Research, 2021, 211, 108714.	1.2	2
42	RPE65-related retinal dystrophy: Mutational and phenotypic spectrum in 45 affected patients. Experimental Eye Research, 2021, 212, 108761.	1.2	11
43	Pancreatic Insufficiency in Patients Under Sorafenib Treatment for Hepatocellular Carcinoma. Journal of Clinical Gastroenterology, 2021, 55, 263-270.	1.1	3
44	Genetics and epidemiology of aniridia: Updated guidelines for genetic study. Archivos De La Sociedad Espanola De Oftalmologia, 2021, 96, 4-14.	0.1	6
45	Fine Breakpoint Mapping by Genome Sequencing Reveals the First Large X Inversion Disrupting the NHS Gene in a Patient with Syndromic Cataracts. International Journal of Molecular Sciences, 2021, 22, 12713.	1.8	2
46	Deepâ€intronic variants in CNGB3 cause achromatopsia by pseudoexon activation. Human Mutation, 2020, 41, 255-264.	1.1	26
47	Thermal Ablation for Intrahepatic Cholangiocarcinoma in Cirrhosis: Safety and Efficacy in Non-Surgical Patients. Journal of Vascular and Interventional Radiology, 2020, 31, 710-719.	0.2	25
48	Pharmacokinetics and pharmacogenetics of sorafenib in patients with hepatocellular carcinoma: Implications for combination trials. Liver International, 2020, 40, 2476-2488.	1.9	6
49	Clinical Phenotype and Course of PDE6A-Associated Retinitis Pigmentosa Disease, Characterized in Preparation for a Gene Supplementation Trial. JAMA Ophthalmology, 2020, 138, 1241.	1.4	9
50	Expanding the phenotype of CRYAA nucleotide variants to a complex presentation of anterior segment dysgenesis. Orphanet Journal of Rare Diseases, 2020, 15, 207.	1.2	9
51	Participant-funded clinical trials on rare diseases. Anales De PediatrÃa (English Edition), 2020, 93, 267.e1-267.e9.	0.1	2
52	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
53	Hepatic epithelioid hemangioendothelioma: An international multicenter study. Digestive and Liver Disease, 2020, 52, 1041-1046.	0.4	13
54	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

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55	Retinal Organoids derived from hiPSCs of an AIPL1-LCA Patient Maintain Cytoarchitecture despite Reduced levels of Mutant AIPL1. Scientific Reports, 2020, 10, 5426.	1.6	39
56	Does transient arterial-phase respiratory-motion-related artifact impact on diagnostic performance? An intra-patient comparison of extracellular gadolinium versus gadoxetic acid. European Radiology, 2020, 30, 6694-6701.	2.3	8
57	Genotypeâ€“Phenotype Correlations in a Spanish Cohort of 506 Families With Biallelic ABCA4 Pathogenic Variants. American Journal of Ophthalmology, 2020, 219, 195-204.	1.7	20
58	Novel PXDN biallelic variants in patients with microphthalmia and anterior segment dysgenesis. Journal of Human Genetics, 2020, 65, 487-491.	1.1	5
59	Posterior column ataxia with retinitis pigmentosa (PCARP) in an Iranian patient associated with the FLVCR1 gene. Ophthalmic Genetics, 2020, 41, 90-92.	0.5	3
60	Retinal Structure inRPE65-Associated Retinal Dystrophy. , 2020, 61, 47.		27
61	CPAMD8 loss-of-function underlies non-dominant congenital glaucoma with variable anterior segment dysgenesis and abnormal extracellular matrix. Human Genetics, 2020, 139, 1209-1231.	1.8	23
62	Exome sequencing identifies mutations in three cases diagnosed with Retinitis Pigmentosa and hearing impairment. Molecular Vision, 2020, 26, 216-225.	1.1	2
63	Expanding the phenotype of the X-linked BCOR microphthalmia syndromes. Human Genetics, 2019, 138, 1051-1069.	1.8	35
64	Identification of splice defects due to noncanonical splice site or deepâ€intronic variants in ABCA4. Human Mutation, 2019, 40, 2365-2376.	1.1	46
65	Molecular evidence of field cancerization initiated by diabetes in colon cancer patients. Molecular Oncology, 2019, 13, 857-872.	2.1	13
66	A Novel Chromosomal Translocation Identified due to Complex Genetic Instability in iPSC Generated for Choroideremia. Cells, 2019, 8, 1068.	1.8	4
67	Time association between hepatitis C therapy and hepatocellular carcinoma emergence in cirrhosis: Relevance of non-characterized nodules. Journal of Hepatology, 2019, 70, 874-884.	1.8	67
68	Expanded Phenotypic Spectrum of Retinopathies Associated with Autosomal Recessive and Dominant Mutations in PROM1. American Journal of Ophthalmology, 2019, 207, 204-214.	1.7	17
69	Diabetesâ€mediated promotion of colon mucosa carcinogenesis is associated with mitochondrial dysfunction. Molecular Oncology, 2019, 13, 1887-1897.	2.1	9
70	Mutation spectrum and clinical investigation of achromatopsia patients with mutations in the GNAT2 gene. Human Mutation, 2019, 40, 1145-1155.	1.1	15
71	Prospective evaluation of gadoxetic acid magnetic resonance for the diagnosis of hepatocellular carcinoma in newly detected nodules â‰¤2Âcm in cirrhosis. Liver International, 2019, 39, 1281-1291.	1.9	20
72	Genomic Landscape of Sporadic Retinitis Pigmentosa. Ophthalmology, 2019, 126, 1181-1188.	2.5	48

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73	Diagnosis of Hepatic Nodules in Patients at Risk for Hepatocellular Carcinoma: LI-RADS Probability Versus Certainty. Gastroenterology, 2019, 156, 860-862.	0.6	11
74	Reasons for and time to retraction of genetics articles published between 1970 and 2018. Journal of Medical Genetics, 2019, 56, 734-740.	1.5	24
75	Expanding the Genetic Landscape of Usher-Like Phenotypes. , 2019, 60, 4701.		9
76	Generation of gene-corrected human induced pluripotent stem cell lines derived from retinitis pigmentosa patient with Ser331Cysfs*5 mutation in MERTK. Stem Cell Research, 2019, 34, 101341.	0.3	10
77	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
78	New GJA8 variants and phenotypes highlight its critical role in a broad spectrum of eye anomalies. Human Genetics, 2019, 138, 1027-1042.	1.8	38
79	Genetic Diagnosis of Epidermolysis Bullosa: Recommendations From an Expert Spanish Research Group. Actas Dermo-sifiliogrÃ¡ficas, 2018, 109, 104-122.	0.2	4
80	Combining targeted panel-based resequencing and copy-number variation analysis for the diagnosis of inherited syndromic retinopathies and associated ciliopathies. Scientific Reports, 2018, 8, 5285.	1.6	28
81	Generation of a human iPSC line from a patient with congenital glaucoma caused by mutation in CYP1B1 gene. Stem Cell Research, 2018, 28, 96-99.	0.3	4
82	Diagnosis and staging of hepatocellular carcinoma (HCC): current guidelines. European Journal of Radiology, 2018, 101, 72-81.	1.2	263
83	Identification ofPITX3mutations in individuals with various ocular developmental defects. Ophthalmic Genetics, 2018, 39, 314-320.	0.5	20
84	DiagnÃ³stico genÃ©tico de la epidermÃ³lisis bullosa: recomendaciones de un grupo espaÃ±ol de expertos. Actas Dermo-sifiliogrÃ¡ficas, 2018, 109, 104-122.	0.2	14
85	Complete response under sorafenib in patients with hepatocellular carcinoma: Relationship with dermatologic adverse events. Hepatology, 2018, 67, 612-622.	3.6	55
86	Pilot study of living donor liver transplantation for patients with hepatocellular carcinoma exceeding Milan Criteria (Barcelona Clinic Liver Cancer extended criteria). Liver Transplantation, 2018, 24, 369-379.	1.3	47
87	High-throughput sequencing for the molecular diagnosis of Usher syndrome reveals 42 novel mutations and consolidates CEP250 as Usher-like disease causative. Scientific Reports, 2018, 8, 17113.	1.6	30
88	Implication of non-coding PAX6 mutations in aniridia. Human Genetics, 2018, 137, 831-846.	1.8	34
89	Generation of a human iPSC line from a patient with Leber congenital amaurosis caused by mutation in AIPL1. Stem Cell Research, 2018, 33, 151-155.	0.3	4
90	Parental Mosaicism in PAX6 Causes Intra-Familial Variability: Implications for Genetic Counseling of Congenital Aniridia and Microphthalmia. Frontiers in Genetics, 2018, 9, 479.	1.1	21

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91	Toward the Mutational Landscape of Autosomal Dominant Retinitis Pigmentosa: A Comprehensive Analysis of 258 Spanish Families. , 2018, 59, 2345.		58
92	Unravelling the pathogenic role and genotype-phenotype correlation of the USH2A p.(Cys759Phe) variant among Spanish families. PLoS ONE, 2018, 13, e0199048.	1.1	17
93	Mutations in the Spliceosome Component CWC27 Cause Retinal Degeneration with or without Additional Developmental Anomalies. American Journal of Human Genetics, 2017, 100, 592-604.	2.6	61
94	Whole-Exome Sequencing of Congenital Glaucoma Patients Reveals Hypermorphic Variants in GPATCH3, a New Gene Involved in Ocular and Craniofacial Development. Scientific Reports, 2017, 7, 46175.	1.6	22
95	New CDH3 mutation in the first Spanish case of hypotrichosis with juvenile macular dystrophy, a case report. BMC Medical Genetics, 2017, 18, 1.	2.1	31
96	Establishment of a human DOA 'plus' iPSC line, IISHDOi003-A, with the mutation in the OPA1 gene: c.1635C > A; p.Ser545Arg. Stem Cell Research, 2017, 24, 81-84.	0.3	8
97	USH2A Gene Editing Using the CRISPR System. Molecular Therapy - Nucleic Acids, 2017, 8, 529-541.	2.3	56
98	Pharmacogenetics of methylphenidate in childhood attention-deficit/hyperactivity disorder: long-term effects. Scientific Reports, 2017, 7, 10391.	1.6	18
99	Mutations inSCAPERcause autosomal recessive retinitis pigmentosa with intellectual disability. Journal of Medical Genetics, 2017, 54, 698-704.	1.5	26
100	Comparison of three magnetic resonance enterography indices for grading activity in Crohnâ€™s disease. Journal of Gastroenterology, 2017, 52, 585-593.	2.3	83
101	2017 update on the relationship between diabetes and colorectal cancer: epidemiology, potential molecular mechanisms and therapeutic implications. Oncotarget, 2017, 8, 18456-18485.	0.8	134
102	Analysis of the PRPF31 Gene in Spanish Autosomal Dominant Retinitis Pigmentosa Patients: A Novel Genomic Rearrangement. , 2017, 58, 1045.		19
103	Improving molecular diagnosis of aniridia and WAGR syndrome using customized targeted array-based CGH. PLoS ONE, 2017, 12, e0172363.	1.1	26
104	Colon cancer modulation by a diabetic environment: A single institutional experience. PLoS ONE, 2017, 12, e0172300.	1.1	5
105	Functional Characterization of Three Concomitant MtDNA LHON Mutations Shows No Synergistic Effect on Mitochondrial Activity. PLoS ONE, 2016, 11, e0146816.	1.1	17
106	Dominant Retinitis Pigmentosa, p.Gly56Arg Mutation in NR2E3: Phenotype in a Large Cohort of 24 Cases. PLoS ONE, 2016, 11, e0149473.	1.1	21
107	Panel-based NGS Reveals Novel Pathogenic Mutations in Autosomal Recessive Retinitis Pigmentosa. Scientific Reports, 2016, 6, 19531.	1.6	48
108	Diagnosis and treatment of hepatocellular carcinoma. Update consensus document from the AEEH, SEOM, SERAM, SERVEI and SETH. Medicina ClÃnica (English Edition), 2016, 146, 511.e1-511.e22.	0.1	2

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109	Nine-year experience in Gaucher disease diagnosis at the Spanish reference center FundaciÃ³n JimÃ©nez DÃaz. Molecular Genetics and Metabolism Reports, 2016, 9, 79-85.	0.4	6
110	Identification of the Photoreceptor Transcriptional Co-Repressor SAMD11 as Novel Cause of Autosomal Recessive Retinitis Pigmentosa. Scientific Reports, 2016, 6, 35370.	1.6	13
111	Diversity of Cognitive Phenotypes Associated with C9ORF72 Hexanucleotide Expansion. Journal of Alzheimer's Disease, 2016, 52, 25-31.	1.2	0
112	Generation of a human iPSC line from a patient with a mitochondrial encephalopathy due to mutations in the GFM1 gene. Stem Cell Research, 2016, 16, 124-127.	0.3	8
113	Conclusive HCC diagnosis with hepatocyte-specific contrast-enhanced magnetic resonance imaging? Not yet. Journal of Hepatology, 2016, 65, 648-649.	1.8	2
114	Generation of a human iPSC line from a patient with an optic atrophy â€˜plusâ€™ phenotype due to a mutation in the OPA1 gene. Stem Cell Research, 2016, 16, 673-676.	0.3	12
115	Contribution of JAK2 mutations to T-cell lymphoblastic lymphoma development. Leukemia, 2016, 30, 94-103.	3.3	27
116	A Comprehensive Analysis of Choroideremia: From Genetic Characterization to Clinical Practice. PLoS ONE, 2016, 11, e0151943.	1.1	41
117	Identification of two novel mutations in CDHR1 in consanguineous Spanish families with autosomal recessive retinal dystrophy. Scientific Reports, 2015, 5, 13902.	1.6	30
118	Human iPSC derived disease model of MERTK-associated retinitis pigmentosa. Scientific Reports, 2015, 5, 12910.	1.6	47
119	Attention deficit hyperactivity disorder: genetic association study in a cohort of Spanish children. Behavioral and Brain Functions, 2015, 12, 2.	1.4	26
120	Characterization of Inflammation and Fibrosis in Crohnâ€™s Disease Lesions by Magnetic Resonance Imaging. American Journal of Gastroenterology, 2015, 110, 432-440.	0.2	215
121	Targeted Next-Generation Sequencing Improves the Diagnosis of Autosomal Dominant Retinitis Pigmentosa in Spanish Patients. , 2015, 56, 2173.		44
122	Management and return of incidental genomic findings in clinical trials. Pharmacogenomics Journal, 2015, 15, 1-5.	0.9	8
123	Exploring genotype-phenotype relationships in Bardet-Biedl syndrome families. Journal of Medical Genetics, 2015, 52, 503-513.	1.5	42
124	Whole-exome sequencing reveals ZNF408 as a new gene associated with autosomal recessive retinitis pigmentosa with vitreal alterations. Human Molecular Genetics, 2015, 24, 4037-4048.	1.4	41
125	New Mutations in theRAB28Gene in 2 Spanish Families With Cone-Rod Dystrophy. JAMA Ophthalmology, 2015, 133, 133.	1.4	28
126	Clinical Aspects of Usher Syndrome and theUSH2AGene in a Cohort of 433 Patients. JAMA Ophthalmology, 2015, 133, 157.	1.4	59

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127	Autosomal recessive retinitis pigmentosa withRP1mutations is associated with myopia. British Journal of Ophthalmology, 2015, 99, 1360-1365.	2.1	18
128	Liver Imaging Reporting and Data System with MR Imaging: Evaluation in Nodules 20 mm or Smaller Detected in Cirrhosis at Screening US. Radiology, 2015, 275, 698-707.	3.6	115
129	Patients with relapsed/refractory chronic lymphocytic leukaemia may benefit from inclusion in clinical trials irrespective of the therapy received: a case-control retrospective analsysis. Blood Cancer Journal, 2015, 5, e356-e356.	2.8	2
130	Preserved Outer Retina in AIPL1 Leber's Congenital Amaurosis: Implications for Gene Therapy. Ophthalmology, 2015, 122, 862-864.	2.5	31
131	Lack of arterial hypervascularity at contrast-enhanced ultrasound should not define the priority for diagnostic work-up of nodules <2cm. Journal of Hepatology, 2015, 62, 150-155.	1.8	46
132	Prevalence of Rhodopsin mutations in autosomal dominant Retinitis Pigmentosa in Spain: clinical and analytical review in 200 families. Acta Ophthalmologica, 2015, 93, e38-44.	0.6	29
133	Hypo- and Hypermorphic FOXC1 Mutations in Dominant Glaucoma: Transactivation and Phenotypic Variability. PLoS ONE, 2015, 10, e0119272.	1.1	24
134	Application of Whole Exome Sequencing in Six Families with an Initial Diagnosis of Autosomal Dominant Retinitis Pigmentosa: Lessons Learned. PLoS ONE, 2015, 10, e0133624.	1.1	19
135	New COL6A6 variant detected by whole-exome sequencing is linked to break points in intron 4 and 3'-UTR, deleting exon 5 of RHO, and causing adRP. Molecular Vision, 2015, 21, 857-70.	1.1	4
136	Reply to Townsend et al.. European Journal of Human Genetics, 2014, 22, 7-7.	1.4	1
137	Targeted next generation sequencing for molecular diagnosis of Usher syndrome. Orphanet Journal of Rare Diseases, 2014, 9, 168.	1.2	61
138	Managing Incidental Genomic Findings in Clinical Trials: Fulfillment of the Principle of Justice. PLoS Medicine, 2014, 11, e1001584.	3.9	14
139	Analysis of the ABCA4 genomic locus in Stargardt disease. Human Molecular Genetics, 2014, 23, 6797-6806.	1.4	117
140	Effect of polymorphisms on the pharmacokinetics, pharmacodynamics, and safety of risperidone in healthy volunteers. Human Psychopharmacology, 2014, 29, 459-469.	0.7	33
141	Expanding the phenotype of PRPS1 syndromes in females: neuropathy, hearing loss and retinopathy. Orphanet Journal of Rare Diseases, 2014, 9, 190.	1.2	31
142	Description of a new family with cryopyrin-associated periodic syndrome: risk of visual loss in patients bearing the R260W mutation. Rheumatology, 2014, 53, 1095-1099.	0.9	24
143	Systemic Therapy for Hepatocellular Carcinoma: The Issue of Treatment Stage Migration and Registration of Progression Using the BCLC-Refined RECIST. Seminars in Liver Disease, 2014, 34, 444-455.	1.8	112
144	Overview of Bardetâ€“Biedl syndrome in Spain: identification of novel mutations in <scp>BBS1,</scp><scp>BBS10</scp> and <scp>BBS12</scp> genes. Clinical Genetics, 2014, 86, 601-602.	1.0	20

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145	Molecular Testing for Fragile X: Analysis of 5062 Tests from 1105 Fragile X Familiesâ€"Performed in 12 Clinical Laboratories in Spain. BioMed Research International, 2014, 2014, 1-8.	0.9	13
146	The challenges of novel contrast agents for the imaging diagnosis of hepatocellular carcinoma. Hepatology International, 2014, 8, 4-6.	1.9	2
147	Contribution of Mutation Load to the Intrafamilial Genetic Heterogeneity in a Large Cohort of Spanish Retinal Dystrophies Families. , 2014, 55, 7562.		11
148	New syndrome with retinitis pigmentosa is caused by nonsense mutations in retinol dehydrogenase RDH11. Human Molecular Genetics, 2014, 23, 5774-5780.	1.4	30
149	Involvement of LCA5 in Leber Congenital Amaurosis and Retinitis Pigmentosa in the Spanish Population. Ophthalmology, 2014, 121, 399-407.	2.5	20
150	A homozygous nonsense CEP250 mutation combined with a heterozygous nonsense C2orf71 mutation is associated with atypical Usher syndrome. Journal of Medical Genetics, 2014, 51, 460-469.	1.5	78
151	Early dermatologic adverse events predict better outcome in HCC patients treated with sorafenib. Journal of Hepatology, 2014, 61, 318-324.	1.8	203
152	Exome Sequencing Extends the Phenotypic Spectrum for ABHD12 Mutations. Ophthalmology, 2014, 121, 1620-1627.	2.5	44
153	Mutational screening of splicing factor genes in cases with autosomal dominant retinitis pigmentosa. Molecular Vision, 2014, 20, 843-51.	1.1	11
154	Novel deletions involving the USH2A gene in patients with Usher syndrome and retinitis pigmentosa. Molecular Vision, 2014, 20, 1398-410.	1.1	12
155	High frequency of CRB1 mutations as cause of Early-Onset Retinal Dystrophies in the Spanish population. Orphanet Journal of Rare Diseases, 2013, 8, 20.	1.2	59
156	Postprogression survival of patients with advanced hepatocellular carcinoma: Rationale for second-line trial design. Hepatology, 2013, 58, 2023-2031.	3.6	217
157	Pharmacodynamic genetic variants related to antipsychotic adverse reactions in healthy volunteers. Pharmacogenomics, 2013, 14, 1203-1214.	0.6	9
158	GuÃa para el estudio genÃ©tico de la aniridia. Archivos De La Sociedad Espanola De Oftalmologia, 2013, 88, 145-152.	0.1	11
159	Candidate glutamatergic and dopaminergic pathway gene variants do not influence Huntingtonâ€™s disease motor onset. Neurogenetics, 2013, 14, 173-179.	0.7	10
160	C9ORF72 hexanucleotide expansions of 20â€"22 repeats are associated with frontotemporal deterioration. Neurology, 2013, 80, 366-370.	1.5	89
161	Recessive dystrophic epidermolysis bullosa: the origin of the c.6527insC mutation in the Spanish population. British Journal of Dermatology, 2013, 168, 226-229.	1.4	6
162	Outcome of ABCA4 Disease-Associated Alleles in Autosomal Recessive Retinal Dystrophies. Ophthalmology, 2013, 120, 2332-2337.	2.5	71

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163	Informed consent for whole-genome sequencing studies in the clinical setting. Proposed recommendations on essential content and process. European Journal of Human Genetics, 2013, 21, 1054-1059.	1.4	118
164	NovelGUCA1AMutations Suggesting Possible Mechanisms of Pathogenesis in Cone, Cone-Rod, and Macular Dystrophy Patients. BioMed Research International, 2013, 2013, 1-15.	0.9	32
165	CYP2D6 poor metabolizer status might be associated with better response to risperidone treatment. Pharmacogenetics and Genomics, 2013, 23, 627-630.	0.7	25
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