Carmen Ayuso

List of Publications by Year in descending order

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350 papers	24,308 citations	15504 65 h-index	9589 142 g-index
376 all docs	376 docs citations	376 times ranked	19571 citing authors

#	Article	IF	Citations
1	Arterial embolisation or chemoembolisation versus symptomatic treatment in patients with unresectable hepatocellular carcinoma: a randomised controlled trial. Lancet, The, 2002, 359, 1734-1739.	13.7	3,172
2	BCLC strategy for prognosis prediction and treatment recommendation: The 2022 update. Journal of Hepatology, 2022, 76, 681-693.	3.7	1,495
3	Diagnosis of hepatic nodules 20 mm or smaller in cirrhosis: Prospective validation of the noninvasive diagnostic criteria for hepatocellular carcinoma. Hepatology, 2008, 47, 97-104.	7.3	884
4	Chemoembolization of hepatocellular carcinoma with drug eluting beads: Efficacy and doxorubicin pharmacokinetics. Journal of Hepatology, 2007, 46, 474-481.	3.7	864
5	Increased risk of tumor seeding after percutaneous radiofrequency ablation for single hepatocellular carcinoma. Hepatology, 2001, 33, 1124-1129.	7.3	698
6	OPA1 mutations induce mitochondrial DNA instability and optic atrophy 'plus' phenotypes. Brain, 2008, 131, 338-351.	7.6	454
7	Survival of patients with hepatocellular carcinoma treated by transarterial chemoembolisation (TACE) using Drug Eluting Beads. Implications for clinical practice and trial design. Journal of Hepatology, 2012, 56, 1330-1335.	3.7	436
8	Initial response to percutaneous ablation predicts survival in patients with hepatocellular carcinoma. Hepatology, 2004, 40, 1352-1360.	7.3	409
9	Evaluation of tumor response after locoregional therapies in hepatocellular carcinoma. Cancer, 2009, 115, 616-623.	4.1	403
10	MRI angiography is superior to helical CT for detection of HCC prior to liver transplantation: An explant correlation. Hepatology, 2003, 38, 1034-1042.	7.3	401
11	Retinitis pigmentosa caused by a homozygous mutation in the Stargardt disease gene ABCR. Nature Genetics, 1998, 18, 11-12.	21.4	382
12	Mutation of a Gene Encoding a Protein with Extracellular Matrix Motifs in Usher Syndrome Type IIa. Science, 1998, 280, 1753-1757.	12.6	366
13	Highly conserved non-coding elements on either side of SOX9 associated with Pierre Robin sequence. Nature Genetics, 2009, 41, 359-364.	21.4	364
14	Treatment of small hepatocellular carcinoma in cirrhotic patients: A cohort study comparing surgical resection and percutaneous ethanol injection. Hepatology, 1993, 18, 1121-1126.	7.3	305
15	CAG repeat expansion in Huntington disease determines age at onset in a fully dominant fashion. Neurology, 2012, 78, 690-695.	1.1	303
16	Tumor size determines the efficacy of percutaneous ethanol injection for the treatment of small hepatocellular carcinoma. Hepatology, 1992, 16, 353-357.	7.3	273
17	Intrahepatic peripheral cholangiocarcinoma in cirrhosis patients may display a vascular pattern similar to hepatocellular carcinoma on contrast-enhanced ultrasound. Hepatology, 2010, 51, 2020-2029.	7.3	268
18	Diagnosis and staging of hepatocellular carcinoma (HCC): current guidelines. European Journal of Radiology, 2018, 101, 72-81.	2.6	263

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19	Cholangiocarcinoma in cirrhosis: Absence of contrast washout in delayed phases by magnetic resonance imaging avoids misdiagnosis of hepatocellular carcinoma. Hepatology, 2009, 50, 791-798.	7.3	253
20	Randomized controlled trial of interferon treatment for advanced hepatocellular carcinoma. Hepatology, 2000, 31, 54-58.	7.3	242
21	Postprogression survival of patients with advanced hepatocellular carcinoma: Rationale for second-line trial design. Hepatology, 2013, 58, 2023-2031.	7.3	217
22	Characterization of Inflammation and Fibrosis in Crohnâ€™s Disease Lesions by Magnetic Resonance Imaging. American Journal of Gastroenterology, 2015, 110, 432-440.	0.4	215
23	CDH23 Mutation and Phenotype Heterogeneity: A Profile of 107 Diverse Families with Usher Syndrome and Nonsyndromic Deafness. American Journal of Human Genetics, 2002, 71, 262-275.	6.2	207
24	Early dermatologic adverse events predict better outcome in HCC patients treated with sorafenib. Journal of Hepatology, 2014, 61, 318-324.	3.7	203
25	Next-Generation Sequencing of a 40 Mb Linkage Interval Reveals TSPAN12 Mutations in Patients with Familial Exudative Vitreoretinopathy. American Journal of Human Genetics, 2010, 86, 240-247.	6.2	202
26	Treatment of hepatocellular carcinoma with tamoxifen: A double-blind placebo-controlled trial in 120 patients. Gastroenterology, 1995, 109, 917-922.	1.3	191
27	AHI1 is required for photoreceptor outer segment development and is a modifier for retinal degeneration in nephronophthisis. Nature Genetics, 2010, 42, 175-180.	21.4	171
28	Genomic Cloning and Characterization of the Human Homeobox Gene SIX6 Reveals a Cluster of SIX Genes in Chromosome 14 and Associates SIX6 Hemizygosity with Bilateral Anophthalmia and Pituitary Anomalies. Genomics, 1999, 61, 82-91.	2.9	163
29	An Update on the Genetics of Usher Syndrome. Journal of Ophthalmology, 2011, 2011, 1-8.	1.3	160
30	Non-invasive diagnosis of hepatocellular carcinoma â©½2cm in cirrhosis. Diagnostic accuracy assessing fat, capsule and signal intensity at dynamic MRI. Journal of Hepatology, 2012, 56, 1317-1323.	3.7	159
31	OPA1 R445H mutation in optic atrophy associated with sensorineural deafness. Annals of Neurology, 2005, 58, 958-963.	5.3	155
32	Prospective validation of an immunohistochemical panel (glypican 3, heat shock protein 70 and) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 2 61, 1481-1487.	12.1	154
33	Identification of an IMPDH1 mutation in autosomal dominant retinitis pigmentosa (RP10) revealed following comparative microarray analysis of transcripts derived from retinas of wild-type and Rho-/- mice. Human Molecular Genetics, 2002, 11, 547-558.	2.9	152
34	Localization of an autosomal dominant retinitis pigmentosa gene to chromosome 7q. Nature Genetics, 1993, 4, 54-58.	21.4	143
35	Molecular screening of 980 cases of suspected hereditary optic neuropathy with a report on 77 novelOPA1mutations. Human Mutation, 2009, 30, E692-E705.	2.5	140
36	2017 update on the relationship between diabetes and colorectal cancer: epidemiology, potential molecular mechanisms and therapeutic implications. Oncotarget, 2017, 8, 18456-18485.	1.8	134

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37	Prenatal detection of a cystic fibrosis mutation in fetal DNA from maternal plasma. Prenatal Diagnosis, 2002, 22, 946-948.	2.3	131
38	Overview of the mutation spectrum in familial exudative vitreoretinopathy and Norrie disease with identification of 21 novel variants in FZD4, LRP5, and NDP. Human Mutation, 2010, 31, 656-666.	2.5	126
39	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.	2.8	118
40	Analysis of the ABCA4 genomic locus in Stargardt disease. Human Molecular Genetics, 2014, 23, 6797-6806.	2.9	117
41	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.	7.3	115
42	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.	3.6	112
43	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
44	BBS1 Mutations in a Wide Spectrum of Phenotypes Ranging From Nonsyndromic Retinitis Pigmentosa to Bardet-Biedl Syndrome. JAMA Ophthalmology, 2012, 130, 1425.	2.4	106
45	Clinical decision making and research in hepatocellular carcinoma: Pivotal role of imaging techniques. Hepatology, 2011, 54, 2238-2244.	7.3	101
46	Retinitis pigmentosa and allied conditions today: a paradigm of translational research. Genome Medicine, 2010, 2, 34.	8.2	99
47	Transarterial embolization for hepatocellular carcinoma. Antibiotic prophylaxis and clinical meaning of postembolization fever. Journal of Hepatology, 1995, 22, 410-415.	3.7	95
48	Retinal degeneration associated with RDH12 mutations results from decreased 11- cis retinal synthesis due to disruption of the visual cycle. Human Molecular Genetics, 2005, 14, 3865-3875.	2.9	94
49	SOX2 anophthalmia syndrome: 12 new cases demonstrating broader phenotype and high frequency of large gene deletions. British Journal of Ophthalmology, 2007, 91, 1471-1476.	3.9	92
50	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
51	Mutations in the Pre-mRNA Splicing-Factor GenesPRPF3,PRPF8, andPRPF31in Spanish Families with Autosomal Dominant Retinitis Pigmentosa. , 2003, 44, 2171.		91
52	Mutation analysis of 272 Spanish families affected by autosomal recessive retinitis pigmentosa using a genotyping microarray. Molecular Vision, 2010, 16, 2550-8.	1.1	91
53	Novel heterozygousOTX2mutations and whole gene deletions in anophthalmia, microphthalmia and coloboma. Human Mutation, 2008, 29, E278-E283.	2.5	89
54	C9ORF72 hexanucleotide expansions of 20â€“22 repeats are associated with frontotemporal deterioration. Neurology, 2013, 80, 366-370.	1.1	89

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55	On the molecular pathology of neurodegeneration in IMPDH1-based retinitis pigmentosa. Human Molecular Genetics, 2004, 13, 641-650.	2.9	86
56	Comparison of three magnetic resonance enterography indices for grading activity in Crohnâ€™s disease. Journal of Gastroenterology, 2017, 52, 585-593.	5.1	83
57	Huntington diseaseâ€“unaffected fetus diagnosed from maternal plasma using QFâ€PCR. Prenatal Diagnosis, 2003, 23, 232-234.	2.3	80
58	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.	3.2	78
59	Identification of 14 novel mutations in the long isoform of USH2A in Spanish patients with Usher syndrome type II. Journal of Medical Genetics, 2006, 43, e55-e55.	3.2	75
60	Mutation Screening of 299 Spanish Families with Retinal Dystrophies by Leber Congenital Amaurosis Genotyping Microarray. , 2007, 48, 5653.		74
61	Foetal sex determination in maternal blood from the seventh week of gestation and its role in diagnosing haemophilia in the foetuses of female carriers. Haemophilia, 2008, 14, 593-598.	2.1	74
62	Outcome of ABCA4 Disease-Associated Alleles in Autosomal Recessive Retinal Dystrophies. Ophthalmology, 2013, 120, 2332-2337.	5.2	71
63	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
64	Exome Sequencing of Index Patients with Retinal Dystrophies as a Tool for Molecular Diagnosis. PLoS ONE, 2013, 8, e65574.	2.5	71
65	Discovery and Functional Analysis of a Retinitis Pigmentosa Gene, C2ORF71. American Journal of Human Genetics, 2010, 86, 686-695.	6.2	70
66	Mutations in USH2A in Spanish patients with autosomal recessive retinitis pigmentosa: high prevalence and phenotypic variation. Journal of Medical Genetics, 2003, 40, 8e-8.	3.2	69
67	Mutation spectrum of EYS in Spanish patients with autosomal recessive retinitis pigmentosa. Human Mutation, 2010, 31, E1772-E1800.	2.5	69
68	Homozygous tandem duplication within the gene encoding the Î²-subunit of rod phosphodiesterase as a cause for autosomal recessive retinitis pigmentosa. Human Mutation, 1995, 5, 228-234.	2.5	68
69	Imaging of HCC. Abdominal Imaging, 2012, 37, 215-230.	2.0	67
70	Time association between hepatitis C therapy and hepatocellular carcinoma emergence in cirrhosis: Relevance of non-characterized nodules. Journal of Hepatology, 2019, 70, 874-884.	3.7	67
71	Genetic Heterogeneity of Usher Syndrome: Analysis of 151 Families with Usher Type I. American Journal of Human Genetics, 2000, 67, 1569-1574.	6.2	63
72	Double trisomy in spontaneous miscarriages: cytogenetic and molecular approach. Human Reproduction, 2006, 21, 958-966.	0.9	61

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73	Targeted next generation sequencing for molecular diagnosis of Usher syndrome. Orphanet Journal of Rare Diseases, 2014, 9, 168.	2.7	61
74	Mutations in the Spliceosome Component CWC27 Cause Retinal Degeneration with or without Additional Developmental Anomalies. American Journal of Human Genetics, 2017, 100, 592-604.	6.2	61
75	Common SNP-Based Haplotype Analysis of the 4p16.3 Huntington Disease Gene Region. American Journal of Human Genetics, 2012, 90, 434-444.	6.2	60
76	New strategy for the prenatal detection/exclusion of paternal cystic fibrosis mutations in maternal plasma. Journal of Cystic Fibrosis, 2008, 7, 505-510.	0.7	59
77	High frequency of CRB1 mutations as cause of Early-Onset Retinal Dystrophies in the Spanish population. Orphanet Journal of Rare Diseases, 2013, 8, 20.	2.7	59
78	Clinical Aspects of Usher Syndrome and theUSH2AGene in a Cohort of 433 Patients. JAMA Ophthalmology, 2015, 133, 157.	2.5	59
79	Rab escort protein 1 (REP1) in intracellular traffic: a functional and pathophysiological overview. Ophthalmic Genetics, 2004, 25, 101-110.	1.2	58
80	Therapeutic benefit derived from RNAi-mediated ablation of IMPDH1 transcripts in a murine model of autosomal dominant retinitis pigmentosa (RP10). Human Molecular Genetics, 2008, 17, 2084-2100.	2.9	58
81	Toward the Mutational Landscape of Autosomal Dominant Retinitis Pigmentosa: A Comprehensive Analysis of 258 Spanish Families. , 2018, 59, 2345.		58
82	Fourteen novel OPA1 mutations in autosomal dominant optic atrophy including two de novo mutations in sporadic optic atrophy. Human Mutation, 2003, 21, 656-656.	2.5	57
83	Microarray-Based Mutation Analysis of 183 Spanish Families with Usher Syndrome. , 2010, 51, 1311.		57
84	A New Locus for Autosomal Recessive Retinitis Pigmentosa (RP19) Maps to 1p13â€“1p21. Genomics, 1997, 40, 142-146.	2.9	56
85	An excess of chromosome 1 breakpoints in male infertility. European Journal of Human Genetics, 2004, 12, 993-1000.	2.8	56
86	USH2A Gene Editing Using the CRISPR System. Molecular Therapy - Nucleic Acids, 2017, 8, 529-541.	5.1	56
87	CYP1B1 mutations in Spanish patients with primary congenital glaucoma: phenotypic and functional variability. Molecular Vision, 2009, 15, 417-31.	1.1	56
88	Frequency of constitutional chromosome alterations in patients with hematologic neoplasias. Cancer Genetics and Cytogenetics, 1987, 24, 345-354.	1.0	55
89	Complete response under sorafenib in patients with hepatocellular carcinoma: Relationship with dermatologic adverse events. Hepatology, 2018, 67, 612-622.	7.3	55
90	Diagnostic accuracy of fine-needle aspiration biopsy in patients with hepatocellular carcinoma. Digestive Diseases and Sciences, 1989, 34, 1765-1769.	2.3	54

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91	Abdominal Computed Tomography Predicts Progression in Patients With Rai Stage 0 Chronic Lymphocytic Leukemia. Journal of Clinical Oncology, 2007, 25, 1576-1580.	1.6	54
92	Retinitis pigmentosa in Spain. Clinical Genetics, 1995, 48, 120-122.	2.0	54
93	MYO7A mutation screening in Usher syndrome type I patients from diverse origins. Journal of Medical Genetics, 2006, 44, e71-e71.	3.2	53
94	The first COL7A1 mutation survey in a large Spanish dystrophic epidermolysis bullosa cohort: c.6527insC disclosed as an unusually recurrent mutation. British Journal of Dermatology, 2010, 163, 155-161.	1.5	53
95	Characterization of a Germline Mosaicism in Families with Lowe Syndrome, and Identification of Seven Novel Mutations in the OCRL1 Gene. American Journal of Human Genetics, 1999, 65, 68-76.	6.2	52
96	Analysis of the developmentalSIX6homeobox gene in patients with anophthalmia/microphthalmia. , 2004, 129A, 92-94.		52
97	Mutation profile of the MYO7A gene in Spanish patients with Usher syndrome type I. Human Mutation, 2006, 27, 290-291.	2.5	52
98	Study of the involvement of the RGR, CRPB1, and CRB1 genes in the pathogenesis of autosomal recessive retinitis pigmentosa. Journal of Medical Genetics, 2003, 40, 89e-89.	3.2	48
99	Panel-based NGS Reveals Novel Pathogenic Mutations in Autosomal Recessive Retinitis Pigmentosa. Scientific Reports, 2016, 6, 19531.	3.3	48
100	Genomic Landscape of Sporadic Retinitis Pigmentosa. Ophthalmology, 2019, 126, 1181-1188.	5.2	48
101	Identification of three novel mutations of the noggin gene in patients with fibrodysplasia ossificans progressiva. American Journal of Medical Genetics Part A, 2001, 102, 314-317.	2.4	47
102	Mutations in Myosin VIIA (MYO7A) and Usherin (USH2A) in Spanish patients with usher syndrome types I and II, respectively. Human Mutation, 2002, 20, 76-77.	2.5	47
103	Mutational screening of the USH2A gene in Spanish USH patients reveals 23 novel pathogenic mutations. Orphanet Journal of Rare Diseases, 2011, 6, 65.	2.7	47
104	Human iPSC derived disease model of MERTK-associated retinitis pigmentosa. Scientific Reports, 2015, 5, 12910.	3.3	47
105	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.	2.4	47
106	Clinical and genetic studies in Spanish patients with Usher syndrome type II: description of new mutations and evidence for a lack of genotype-phenotype correlation. Clinical Genetics, 2005, 68, 204-214.	2.0	46
107	Application of quantitative fluorescent PCR with short tandem repeat markers to the study of aneuploidies in spontaneous miscarriages. Human Reproduction, 2005, 20, 1235-1243.	0.9	46
108	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.	3.7	46

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109	Identification of splice defects due to noncanonical splice site or deepâ€intronic variants in ABCA4. Human Mutation, 2019, 40, 2365-2376.	2.5	46
110	CERKLMutations and Associated Phenotypes in Seven Spanish Families with Autosomal Recessive Retinitis Pigmentosa. , 2008, 49, 2709.		45
111	Fibrodysplasia ossificans progressiva in Spain: epidemiological, clinical, and genetic aspects. Bone, 2012, 51, 748-755.	2.9	45
112	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
113	Exome Sequencing Extends the Phenotypic Spectrum for ABHD12 Mutations. Ophthalmology, 2014, 121, 1620-1627.	5.2	44
114	Targeted Next-Generation Sequencing Improves the Diagnosis of Autosomal Dominant Retinitis Pigmentosa in Spanish Patients. , 2015, 56, 2173.		44
115	Microarray-based mutation analysis of the ABCA4 gene in Spanish patients with Stargardt disease: evidence of a prevalent mutated allele. Molecular Vision, 2006, 12, 902-8.	1.1	43
116	Frequency of ABCA4 mutations in 278 Spanish controls: an insight into the prevalence of autosomal recessive Stargardt disease. British Journal of Ophthalmology, 2009, 93, 1359-1364.	3.9	42
117	Exploring genotype-phenotype relationships in Bardet-Biedl syndrome families. Journal of Medical Genetics, 2015, 52, 503-513.	3.2	42
118	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.	2.9	41
119	A Comprehensive Analysis of Choroideremia: From Genetic Characterization to Clinical Practice. PLoS ONE, 2016, 11, e0151943.	2.5	41
120	Mutations P51U and G122E in retinal transcription factor NRL associated with autosomal dominant and sporadic retinitis pigmentosa. Human Mutation, 2001, 17, 520-520.	2.5	40
121	Prenatal diagnosis of Huntington disease in maternal plasma: direct and indirect study. European Journal of Neurology, 2008, 15, 1338-1344.	3.3	40
122	Choroideremia, sensorineural deafness, and primary ovarian failure in a woman with a balanced X-4 translocation. Ophthalmic Genetics, 2000, 21, 185-189.	1.2	39
123	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
124	MLPA as a screening method of aneuploidy and unbalanced chromosomal rearrangements in spontaneous miscarriages. Prenatal Diagnosis, 2007, 27, 765-771.	2.3	39
125	Retinal Organoids derived from hiPSCs of an AIPL1-LCA Patient Maintain Cytoarchitecture despite Reduced levels of Mutant AIPL1. Scientific Reports, 2020, 10, 5426.	3.3	39
126	Analysis of the involvement of the NR2E3 gene in autosomal recessive retinal dystrophies. Clinical Genetics, 2008, 73, 360-366.	2.0	38

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127	New GJA8 variants and phenotypes highlight its critical role in a broad spectrum of eye anomalies. Human Genetics, 2019, 138, 1027-1042.	3.8	38
128	Novel genes and sex differences in COVID-19 severity. Human Molecular Genetics, 2022, 31, 3789-3806.	2.9	38
129	Genotype-phenotype variations in five Spanish families with Norrie disease or X-linked FEVR. Molecular Vision, 2005, 11, 705-12.	1.1	37
130	Correlation of Genetic and Clinical Findings in Spanish Patients with X-linked Juvenile Retinoschisis. , 2009, 50, 4342.		36
131	Genomic Cloning, Structure, Expression Pattern, and Chromosomal Location of the HumanSIX3Gene. Genomics, 1999, 55, 100-105.	2.9	35
132	Clinical presentation of a variant of Axenfeldâ€“Rieger syndrome associated with subtelomeric 6p deletion. European Journal of Medical Genetics, 2007, 50, 120-127.	1.3	35
133	Expanding the phenotype of the X-linked BCOR microphthalmia syndromes. Human Genetics, 2019, 138, 1051-1069.	3.8	35
134	Movement disorders in hereditary ataxias. Journal of the Neurological Sciences, 2002, 202, 59-64.	0.6	34
135	Implication of non-coding PAX6 mutations in aniridia. Human Genetics, 2018, 137, 831-846.	3.8	34
136	CSVS, a crowdsourcing database of the Spanish population genetic variability. Nucleic Acids Research, 2021, 49, D1130-D1137.	14.5	34
137	Effect of polymorphisms on the pharmacokinetics, pharmacodynamics, and safety of risperidone in healthy volunteers. Human Psychopharmacology, 2014, 29, 459-469.	1.5	33
138	NovelGUCA1AMutations Suggesting Possible Mechanisms of Pathogenesis in Cone, Cone-Rod, and Macular Dystrophy Patients. BioMed Research International, 2013, 2013, 1-15.	1.9	32
139	Expanding the phenotype of PRPS1 syndromes in females: neuropathy, hearing loss and retinopathy. Orphanet Journal of Rare Diseases, 2014, 9, 190.	2.7	31
140	Preserved Outer Retina in AIPL1 Leber's Congenital Amaurosis: Implications for Gene Therapy. Ophthalmology, 2015, 122, 862-864.	5.2	31
141	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
142	New mutations in BBS genes in small consanguineous families with Bardet-Biedl syndrome: detection of candidate regions by homozygosity mapping. Molecular Vision, 2010, 16, 137-43.	1.1	31
143	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
144	Prenatal diagnosis on fetal cells from maternal blood: practical comparative evaluation of the first and second trimesters. Prenatal Diagnosis, 2001, 21, 165-170.	2.3	30

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145	New syndrome with retinitis pigmentosa is caused by nonsense mutations in retinol dehydrogenase RDH11. Human Molecular Genetics, 2014, 23, 5774-5780.	2.9	30
146	Identification of two novel mutations in CDHR1 in consanguineous Spanish families with autosomal recessive retinal dystrophy. Scientific Reports, 2015, 5, 13902.	3.3	30
147	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.	3.3	30
148	Spectrum of theABCA4Gene Mutations Implicated in Severe Retinopathies in Spanish Patients. , 2007, 48, 985.		29
149	Prevalence of Rhodopsin mutations in autosomal dominant Retinitis Pigmentosa in Spain: clinical and analytical review in 200 families. Acta Ophthalmologica, 2015, 93, e38-44.	1.1	29
150	Identification of Large Rearrangements of thePCDH15Gene by Combined MLPA and a CGH: Large Duplications Are Responsible for Usher Syndrome. , 2010, 51, 5480.		28
151	New Mutations in theRAB28Gene in 2 Spanish Families With Cone-Rod Dystrophy. JAMA Ophthalmology, 2015, 133, 133.	2.5	28
152	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.	3.3	28
153	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.	3.3	28
154	Genotyping microarray: mutation screening in Spanish families with autosomal dominant retinitis pigmentosa. Molecular Vision, 2012, 18, 1478-83.	1.1	28
155	Improvement in strategies for the non-invasive prenatal diagnosis of Huntington disease. Journal of Assisted Reproduction and Genetics, 2008, 25, 477-481.	2.5	27
156	Contribution of JAK2 mutations to T-cell lymphoblastic lymphoma development. Leukemia, 2016, 30, 94-103.	7.2	27
157	Retinal Structure inRPE65-Associated Retinal Dystrophy. , 2020, 61, 47.		27
158	Comparison of the diagnostic yield of aCGH and genome-wide sequencing across different neurodevelopmental disorders. Npj Genomic Medicine, 2021, 6, 25.	3.8	27
159	Calcifications in the portal venous system: comparison of plain films, sonography, and CT.. American Journal of Roentgenology, 1992, 159, 321-323.	2.2	26
160	Population stratification may bias analysis of PGC-1Î± as a modifier of age at Huntington disease motor onset. Human Genetics, 2012, 131, 1833-1840.	3.8	26
161	Attention deficit hyperactivity disorder: genetic association study in a cohort of Spanish children. Behavioral and Brain Functions, 2015, 12, 2.	3.3	26
162	Mutations inSCAPERcause autosomal recessive retinitis pigmentosa with intellectual disability. Journal of Medical Genetics, 2017, 54, 698-704.	3.2	26

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163	Improving molecular diagnosis of aniridia and WAGR syndrome using customized targeted array-based CGH. PLoS ONE, 2017, 12, e0172363.	2.5	26
164	Deepâ€intronic variants in CNGB3 cause achromatopsia by pseudoexon activation. Human Mutation, 2020, 41, 255-264.	2.5	26
165	Application of Fetal DNA Detection in Maternal Plasma: A Prenatal Diagnosis Unit Experience. Journal of Histochemistry and Cytochemistry, 2005, 53, 307-314.	2.5	25
166	Complexity of Phenotypeâ€“Genotype Correlations in Spanish Patients withRDH12Mutations. , 2009, 50, 1065.		25
167	Molecular analysis of the ABCA4 gene for reliable detection of allelic variations in Spanish patients: identification of 21 novel variants. British Journal of Ophthalmology, 2009, 93, 614-621.	3.9	25
168	Further Associations between Mutations and Polymorphisms in theABCA4Gene: Clinical Implication of Allelic Variants and Their Role as Protector/Risk Factors. , 2011, 52, 6206.		25
169	CYP2D6 poor metabolizer status might be associated with better response to risperidone treatment. Pharmacogenetics and Genomics, 2013, 23, 627-630.	1.5	25
170	Thermal Ablation for Intrahepatic Cholangiocarcinoma in Cirrhosis: Safety and Efficacy in Non-Surgical Patients. Journal of Vascular and Interventional Radiology, 2020, 31, 710-719.	0.5	25
171	ABCA4 mutations in Portuguese Stargardt patients: identification of new mutations and their phenotypic analysis. Molecular Vision, 2009, 15, 584-91.	1.1	25
172	Novel homozygous mutation in the alpha subunit of the rod cGMP gated channel (CNGA1) in two Spanish sibs affected with autosomal recessive retinitis pigmentosa. Journal of Medical Genetics, 2002, 39, 66e-66.	3.2	24
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174	Two novel recessive mutations in KRT14 identified in a cohort of 21 Spanish families with epidermolysis bullosa simplex. British Journal of Dermatology, 2011, 165, 683-692.	1.5	24
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