Celia Badenas

List of Publications by Year
in descending order

153 papers	6,309 citations	94381 37 h-index	76872 74 g-index
159 all docs	159 docs citations	159 times ranked	8169 citing authors

#	Article	IF	Citations
1	Familial Progressive Sensorineural Deafness Is Mainly Due to the mtDNA A1555G Mutation and Is Enhanced by Treatment with Aminoglycosides. American Journal of Human Genetics, 1998, 62, 27-35.	2.6	504
2	A SUMOylation-defective MITF germline mutation predisposes to melanoma and renal carcinoma. Nature, 2011, 480, 94-98.	13.7	466
3	High-risk Melanoma Susceptibility Genes and Pancreatic Cancer, Neural System Tumors, and Uveal Melanoma across GenoMEL. Cancer Research, 2006, 66, 9818-9828.	0.4	373
4	Features associated with germline CDKN2A mutations: a GenoMEL study of melanoma-prone families from three continents. Journal of Medical Genetics, 2006, 44, 99-106.	1.5	350
5	Penetrance of FMR1 premutation associated pathologies in fragile X syndrome families. European Journal of Human Genetics, 2009, 17, 1359-1362.	1.4	254
6	Genome-wide association study identifies three new melanoma susceptibility loci. Nature Genetics, 2011, 43, 1108-1113.	9.4	230
7	TERT Promoter Mutation Status as an Independent Prognostic Factor in Cutaneous Melanoma. Journal of the National Cancer Institute, 2014, 106, .	3.0	204
8	Genome-wide association study identifies novel loci predisposing to cutaneous melanomaâ€. Human Molecular Genetics, 2011, 20, 5012-5023.	1.4	187
9	A melanoma-associated germline mutation in exon 1Î² inactivates p14ARF. Oncogene, 2001, 20, 5543-5547.	2.6	178
10	Benefits of total body photography and digital dermatoscopy (â€œtwo-step method of digital) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 38 American Academy of Dermatology, 2012, 67, e17-e27.	0.6	176
11	Role of the CDKN2A Locus in Patients With Multiple Primary Melanomas. Journal of Clinical Oncology, 2005, 23, 3043-3051.	0.8	138
12	Association of MC1R Variants and Host Phenotypes With Melanoma Risk in CDKN2A Mutation Carriers: A GenoMEL Study. Journal of the National Cancer Institute, 2010, 102, 1568-1583.	3.0	108
13	Mutations in theCOL4A4 and COL4A3 Genes Cause Familial Benign Hematuria. Journal of the American Society of Nephrology: JASN, 2002, 13, 1248-1254.	3.0	106
14	Update in genetic susceptibility in melanoma. Annals of Translational Medicine, 2015, 3, 210.	0.7	100
15	Autosomal Dominant Polycystic Kidney Disease Types 1 and 2: Assessment of US Sensitivity for Diagnosis. Radiology, 1999, 213, 273-276.	3.6	89
16	ALAS2 acts as a modifier gene in patients with congenital erythropoietic porphyria. Blood, 2011, 118, 1443-1451.	0.6	80
17	A novel mutation in JARID1C gene associated with mental retardation. European Journal of Human Genetics, 2006, 14, 583-586.	1.4	78
18	A Novel Elastin Gene Mutation Resulting in an Autosomal Dominant Form of Cutis Laxa. Archives of Dermatology, 2004, 140, 1135-9.	1.7	73

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19	Autosomal dominant polycystic kidney disease with anticipation and Caroli's disease associated with a PKD1 mutation Rapid Communication. Kidney International, 1997, 52, 33-38.	2.6	59
20	Influence of the ACE gene polymorphism in the progression of renal failure in autosomal dominant polycystic kidney disease. American Journal of Kidney Diseases, 1999, 34, 273-278.	2.1	57
21	X-chromosome tiling path array detection of copy number variants in patients with chromosome X-linked mental retardation. BMC Genomics, 2007, 8, 443.	1.2	57
22	Incidence of Fragile X in 5,000 Consecutive Newborn Males. Genetic Testing and Molecular Biomarkers, 2003, 7, 339-343.	1.7	54
23	Increased prevalence of lung, breast, and pancreatic cancers in addition to melanoma risk in families bearing the cyclin-dependent kinase inhibitor 2A mutation: Implications for genetic counseling. Journal of the American Academy of Dermatology, 2014, 71, 888-895.	0.6	52
24	The <scp>MC1R</scp> melanoma risk variant p.<scp>R160W</scp> is associated with <scp>P</scp>arkinson disease. Annals of Neurology, 2015, 77, 889-894.	2.8	52
25	A Loss-of-Function Model for Cystogenesis in Human Autosomal Dominant Polycystic Kidney Disease Type 2. American Journal of Human Genetics, 1999, 65, 345-352.	2.6	51
26	Genetic alterations in RAS-regulated pathway in acral lentiginous melanoma. Experimental Dermatology, 2013, 22, 148-150.	1.4	49
27	Increased prevalence of polycystic kidney disease type 2 among elderly polycystic patients. American Journal of Kidney Diseases, 2000, 36, 728-734.	2.1	48
28	Dermoscopic features of melanomas associated withMC1Rvariants in SpanishCDKN2Amutation carriers. British Journal of Dermatology, 2009, 160, 48-53.	1.4	48
29	Autosomal recessive Alportâ€™s syndrome and benign familial hematuria are collagen type IV diseases. American Journal of Kidney Diseases, 2003, 42, 952-959.	2.1	47
30	Cutaneous phenotype andMC1R variants as modifying factors for the development of melanoma inCDKN2A G101W mutation carriers from 4 countries. International Journal of Cancer, 2007, 121, 825-831.	2.3	45
31	Prevalence and predictors of germline CDKN2A mutations for melanoma cases from Australia, Spain and the United Kingdom. Hereditary Cancer in Clinical Practice, 2014, 12, 20.	0.6	45
32	MC1R gene variants and non-melanoma skin cancer: a pooled-analysis from the M-SKIP project. British Journal of Cancer, 2015, 113, 354-363.	2.9	43
33	Mutational status of naevus-associated melanomas. British Journal of Dermatology, 2015, 173, 671-680.	1.4	42
34	Mutational analysis within the 3â€² region of the PKD1 gene. Kidney International, 1999, 55, 1225-1233.	2.6	41
35	Prevalence ofMITFp.E318K in Patients With Melanoma Independent of the Presence ofCDKN2ACausative Mutations. JAMA Dermatology, 2016, 152, 405.	2.0	41
36	AURKA Overexpression Is Driven byÂFOXM1 and MAPK/ERK Activation inÂMelanoma Cells Harboring BRAF orÂNRASÂMutations: Impact on MelanomaÂPrognosis and Therapy. Journal of Investigative Dermatology, 2017, 137, 1297-1310.	0.3	40

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37	Functional characterization of a multi-cancer risk locus on chr5p15.33 reveals regulation of TERT by ZNF148. Nature Communications, 2017, 8, 15034.	5.8	40
38	Assessment of QF-PCR as the First Approach in Prenatal Diagnosis. Journal of Molecular Diagnostics, 2010, 12, 828-834.	1.2	39
39	Facilitated diagnosis of the contiguous gene syndrome: Tuberous sclerosis and polycystic kidneys by means of haplotype studies. American Journal of Kidney Diseases, 1998, 31, 1038-1043.	2.1	37
40	<scp>POT</scp> 1 germline mutations but not <scp>TERT</scp> promoter mutations are implicated in melanoma susceptibility in a large cohort of Spanish melanoma families. British Journal of Dermatology, 2019, 181, 105-113.	1.4	37
41	Association between BDNF Val66Met polymorphism and age at onset in Huntington disease. Neurology, 2005, 65, 964-965.	1.5	36
42	Capturing the biological impact of CDKN2A and MC1R genes as an early predisposing event in melanoma and non melanoma skin cancer. Oncotarget, 2014, 5, 1439-1451.	0.8	35
43	TERT gene amplification is associated with poor outcome in acral lentiginous melanoma. Journal of the American Academy of Dermatology, 2014, 71, 839-841.	0.6	35
44	Sonographic pattern of recessive polycystic kidney disease in young adults. Differences from the dominant form. Nephrology Dialysis Transplantation, 2000, 15, 1373-1378.	0.4	34
45	MLPA as first screening method for the detection of microduplications and microdeletions in patients with X-linked mental retardation. Genetics in Medicine, 2007, 9, 117-122.	1.1	34
46	Benefits of oral Polypodium Leucotomos extract in MM highâ€risk patients. Journal of the European Academy of Dermatology and Venereology, 2013, 27, 1095-1100.	1.3	34
47	Familial and Sporadic Porphyria Cutanea Tarda. Medicine (United States), 2010, 89, 69-74.	0.4	33
48	Genomic Microarray in Fetuses with Early Growth Restriction: A Multicenter Study. Fetal Diagnosis and Therapy, 2017, 42, 174-180.	0.6	33
49	Identification and characterization of novel uroporphyrinogen decarboxylase gene mutations in a large series of porphyria cutanea tarda patients and relatives. Clinical Genetics, 2009, 75, 346-353.	1.0	32
50	Loss of heterozygosity in renal and hepatic epithelial cystic cells from ADPKD1 patients. European Journal of Human Genetics, 2000, 8, 487-492.	1.4	31
51	Characterization of individuals at high risk of developing melanoma in Latin America: bases for genetic counseling in melanoma. Genetics in Medicine, 2016, 18, 727-736.	1.1	31
52	Seven novel mutations of the PKD2 gene in families with autosomal dominant polycystic kidney disease. Kidney International, 1999, 56, 28-33.	2.6	29
53	Structure and Regulation of the Versican Promoter. Journal of Biological Chemistry, 2009, 284, 12306-12317.	1.6	29
54	Screening for FMR1 and FMR2 mutations in 222 individuals from Spanish special schools: identification of a case of FRAXE-associated mental retardation. Human Genetics, 1997, 100, 503-507.	1.8	28

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55	Genetic counseling in melanoma. Dermatologic Therapy, 2012, 25, 397-402.	0.8	28
56	Melanocortin 1 receptor (<scp>MC</scp>1R) polymorphismsâ€™ influence on size and dermoscopic features of nevi. Pigment Cell and Melanoma Research, 2018, 31, 39-50.	1.5	28
57	Premature ovarian failure and fragile X female premutation carriers. Menopause, 2009, 16, 944-949.	0.8	27
58	Contiguous deletion of theNDP,MAOA,MAOB, andEFHC2 genes in a patient with Norrie disease, severe psychomotor retardation and myoclonic epilepsy. American Journal of Medical Genetics, Part A, 2007, 143A, 916-920.	0.7	26
59	A comparison of CDKN2A mutation detection within the Melanoma Genetics Consortium (GenoMEL). European Journal of Cancer, 2008, 44, 1269-1274.	1.3	26
60	Dermoscopic criteria associated with <scp>BRAF</scp> and <scp>NRAS</scp> mutation status in primary cutaneous melanoma. British Journal of Dermatology, 2014, 171, 754-759.	1.4	26
61	Clinical, Biochemical, and Genetic Study of 11 Patients With Erythropoietic Protoporphyria Including One With Homozygous Disease. Archives of Dermatology, 2007, 143, 1125-9.	1.7	25
62	Reproductive consequences of genome-wide paternal uniparental disomy mosaicism: description of two cases with different mechanisms of origin and pregnancy outcomes. Fertility and Sterility, 2009, 92, 393.e5-393.e9.	0.5	25
63	Distribution ofMC1Rvariants among melanoma subtypes: p.R163Q is associated with lentigo maligna melanoma in a Mediterranean population. British Journal of Dermatology, 2013, 169, 804-811.	1.4	25
64	Fetoplacental discrepancy involving structural abnormalities of chromosome 8 detected by prenatal diagnosis. Prenatal Diagnosis, 2003, 23, 319-322.	1.1	24
65	Genetic and biochemical characterization of 16 acute intermittent porphyria cases with a high prevalence of the R173W mutation. Journal of Inherited Metabolic Disease, 2006, 29, 580-585.	1.7	24
66	Successful Treatment of Congenital Erythropoietic Porphyria Using Matched Unrelated Hematopoietic Stem Cell Transplantation. Pediatric Dermatology, 2013, 30, 484-489.	0.5	24
67	Serum 25-hydroxyvitamin D3 levels and vitamin D receptor variants in melanoma patients from the Mediterranean area of Barcelona. BMC Medical Genetics, 2013, 14, 26.	2.1	24
68	Correlation among Dermoscopy, Confocal Reflectance Microscopy, and Histologic Features of Melanoma and Basal Cell Carcinoma Collision Tumor. Dermatologic Surgery, 2011, 37, 275-279.	0.4	23
69	A female compound heterozygote (pre- and full mutation) for the CGG FMR1 expansion. Human Genetics, 1996, 98, 419-421.	1.8	22
70	Analysis of CGG variation through 642 meioses in Fragile X families. Molecular Human Reproduction, 2004, 10, 773-776.	1.3	22
71	Elastin Mutation Screening in a Group of Patients Affected by Vascular Abnormalities. Pediatric Cardiology, 2005, 26, 827-831.	0.6	22
72	Should cellâ€free DNA testing be used in pregnancy with increased fetal nuchal translucency?. Ultrasound in Obstetrics and Gynecology, 2020, 55, 645-651.	0.9	21

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73	CDKN2Amutations in melanoma families from Uruguay. British Journal of Dermatology, 2009, 161, 536-541.	1.4	20
74	Maternal plasma genome-wide cell-free DNA can detect fetal aneuploidy in early and recurrent pregnancy loss and can be used to direct further workup. Human Reproduction, 2020, 35, 1222-1229.	0.4	19
75	Pathway-Based Analysis of a Melanoma Genome-Wide Association Study: Analysis of Genes Related to Tumour-Immunosuppression. PLoS ONE, 2011, 6, e29451.	1.1	18
76	Genetic susceptibility to cutaneous melanoma in southern Switzerland: role ofCDKN2A,MC1RandMITF. British Journal of Dermatology, 2016, 175, 1030-1037.	1.4	17
77	Recurrence of the PKD1 nonsense mutation Q4041X in Spanish, Italian, and British families. Human Mutation, 1998, 11, S117-S120.	1.1	16
78	Autosomal recessive Alport syndrome: linkage analysis and clinical features in two families. Nephrology Dialysis Transplantation, 1999, 14, 627-630.	0.4	16
79	SCA8 in the Spanish population including one homozygous patient. Clinical Genetics, 2002, 62, 404-409.	1.0	16
80	Cryptic chromosomal rearrangement screening in 30 patients with mental retardation and dysmorphic features. Clinical Genetics, 2003, 65, 17-23.	1.0	16
81	Multiple primary melanomas: do they look the same?. British Journal of Dermatology, 2013, 168, 1267-1272.	1.4	16
82	Association of Melanocortin-1 Receptor Variants with Pigmentary Traits in Humans: AÂPooled Analysis from the M-Skip Project. Journal of Investigative Dermatology, 2016, 136, 1914-1917.	0.3	16
83	MC1R variants in childhood and adolescent melanoma: a retrospective pooled analysis of a multicentre cohort. The Lancet Child and Adolescent Health, 2019, 3, 332-342.	2.7	16
84	Rare variants in the promoter of the fragile X syndrome gene (FMR1). Molecular and Cellular Probes, 2000, 14, 115-119.	0.9	15
85	Study of the genotypeâ€“phenotype relationship in four cases of congenital erythropoietic porphyria. Blood Cells, Molecules, and Diseases, 2007, 38, 242-246.	0.6	15
86	MultipleBRAFWild-Type Melanomas During Dabrafenib Treatment for MetastaticBRAF-Mutant Melanoma. JAMA Dermatology, 2015, 151, 544.	2.0	15
87	Association between dermoscopic and reflectance confocal microscopy features of cutaneous melanoma with <scp>BRAF</scp> mutational status. Journal of the European Academy of Dermatology and Venereology, 2017, 31, 643-649.	1.3	15
88	Maternal transmission in sporadic Huntington's disease.. Journal of Neurology, Neurosurgery and Psychiatry, 1997, 62, 535-537.	0.9	14
89	Deletion of the OPHN1 gene detected by aCGH. Journal of Intellectual Disability Research, 2008, 52, 190-194.	1.2	14
90	Skewed X Inactivation in Women Carrying the <i>FMR1</i> Premutation and Its Relation with Fragile-X-Associated Tremor/Ataxia Syndrome. Neurodegenerative Diseases, 2016, 16, 290-292.	0.8	14

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91	Screening for MECP2 mutations in Spanish patients with an unexplained mental retardation. Clinical Genetics, 2006, 70, 140-144.	1.0	13
92	Screening for FXTAS in 95 Spanish Patients Negative for Huntington Disease. Genetic Testing and Molecular Biomarkers, 2008, 12, 135-138.	1.7	13
93	Association Between Confocal Morphologic Classification and Clinical Phenotypes of Multiple Primary and Familial Melanomas. JAMA Dermatology, 2016, 152, 1099.	2.0	13
94	Monitoring of Donorâ€Derived Cellâ€Free DNA by Short Tandem Repeats: Concentration of Total Cellâ€Free DNA and Fragment Size for Acute Rejection Risk Assessment in Liver Transplantation. Liver Transplantation, 2022, 28, 257-268.	1.3	13
95	Mutation of the tumour suppressor p33 ING1 b is rare in melanoma. British Journal of Dermatology, 2006, 155, 94-99.	1.4	12
96	Familial 4.8â€‰MB deletion on 18q23 associated with growth hormone insufficiency and phenotypic variability. American Journal of Medical Genetics, Part A, 2012, 158A, 611-616.	0.7	12
97	Lack of Mutations in POT1 Gene in Selected Families with Familial Non-Medullary Thyroid Cancer. Hormones and Cancer, 2020, 11, 111-116.	4.9	12
98	Childhood-onset mild cutaneous porphyria with compound heterozygotic mutations in the uroporphyrinogen decarboxylase gene. Clinical and Experimental Dermatology, 2008, 33, 602-605.	0.6	11
99	Characterization of a 5.8-Mb Interstitial Deletion of Chromosome 3p in a Girl with 46,XX,inv(7)dn Karyotype and Phenotypic Abnormalities. Cytogenetic and Genome Research, 2009, 125, 334-340.	0.6	11
100	Duplication of CXC chemokine genes on chromosome 4q13 in a melanomaâ€prone family. Pigment Cell and Melanoma Research, 2012, 25, 243-247.	1.5	11
101	IRF4 rs12203592 functional variant and melanoma survival. International Journal of Cancer, 2017, 140, 1845-1849.	2.3	11
102	Molecular characterization of a t(9;12)(p21;q13) balanced chromosome translocation in combination with integrative genomics analysis identifiesC9orf14as a candidate tumor-suppressor. Genes Chromosomes and Cancer, 2007, 46, 155-162.	1.5	10
103	Multiple Primary Acral Melanomas in Two Young Caucasian Patients. Dermatology, 2014, 228, 307-310.	0.9	10
104	Recombination in a male carrier of two reciprocal translocations involving chromosomes 14, 14â€², 15, and 21 leading to balanced and unbalanced rearrangements in offspring. , 2005, 134A, 309-314.		9
105	Genetic studies in variegate porphyria in Spain. Identification of gene mutations and family study for carrier detection. Journal of the European Academy of Dermatology and Venereology, 2006, 20, 060804053334005-???.	1.3	9
106	Amelanotic melanoma in oculocutaneous albinism: a genetic, dermoscopic and reflectance confocal microscopy study. British Journal of Dermatology, 2017, 177, e333-e335.	1.4	9
107	Autosomal recessive polycystic kidney disease presenting in adulthood. Molecular diagnosis of the family. Nephrology Dialysis Transplantation, 1998, 13, 1273-1276.	0.4	8
108	Single-strand conformation polymorphism analysis in theFMR1. American Journal of Medical Genetics Part A, 1999, 84, 262-265.	2.4	8

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109	Pilot study for the neonatal screening of fragile X syndrome. Prenatal Diagnosis, 2002, 22, 459-462.	1.1	8
110	Biochemical and genetic characterization of four cases of hereditary coproporphyria in Spain. Molecular Genetics and Metabolism, 2005, 85, 160-163.	0.5	8
111	Protocol proposal for Friedreich ataxia molecular diagnosis using fluorescent and triplet repeat primed polymerase chain reaction. Translational Research, 2010, 156, 309-314.	2.2	8
112	Dermoscopy comparative approach for early diagnosis in familial melanoma: influence of MC1R genotype. Journal of the European Academy of Dermatology and Venereology, 2021, 35, 403-410.	1.3	8
113	Novedades en genÃ©tica del melanoma. Piel, 2006, 21, 272-274.	0.0	7
114	Chorionic villus sampling in the prenatal diagnosis of placental mesenchymal dysplasia. Ultrasound in Obstetrics and Gynecology, 2010, 36, 644-645.	0.9	7
115	Fragile X syndrome prenatal diagnosis: parental attitudes and reproductive responses. Reproductive BioMedicine Online, 2010, 21, 560-565.	1.1	7
116	Evaluation of <scp>PAX</scp>3 genetic variants and nevus number. Pigment Cell and Melanoma Research, 2013, 26, 666-676.	1.5	7
117	Clinical and Histopathological Characteristics between Familial and Sporadic Melanoma in Barcelona, Spain. Journal of Clinical & Experimental Dermatology Research, 2014, 05, 231.	0.1	7
118	Late-onset cutaneous porphyria in a patient heterozygous for a uroporphyrinogen III synthase gene mutation. British Journal of Dermatology, 2016, 175, 1346-1350.	1.4	7
119	Acquired erythropoietic uroporphyria secondary to myelodysplastic syndrome with chromosome 3 alterations: a case report. British Journal of Dermatology, 2018, 179, 486-490.	1.4	7
120	Resolution of subclinical porphyria cutanea tarda after hepatitis C eradication with directâ€acting antiâ€virals. Alimentary Pharmacology and Therapeutics, 2020, 51, 968-973.	1.9	7
121	Molecular characterization of human cutaneous melanoma-derived cell lines. Anticancer Research, 2012, 32, 1245-51.	0.5	7
122	Parental Origin of the Retained X Chromosome in Monosomy X Miscarriages and Ongoing Pregnancies. Fetal Diagnosis and Therapy, 2019, 45, 118-124.	0.6	6
123	Excision and transposition of Tn5 upon insertion in the hha gene of Escherichia coli. Canadian Journal of Microbiology, 1994, 40, 597-601.	0.8	5
124	A Common Variant in the MC1R Gene (p.V92M) is associated with Alzheimerâ€™s Disease Risk. Journal of Alzheimer's Disease, 2017, 56, 1065-1074.	1.2	5
125	The p. R151C Polymorphism in MC1R Gene Modifies the Age of Onset in Spanish Huntingtonâ€™s Disease Patients. Molecular Neurobiology, 2017, 54, 3906-3910.	1.9	5
126	Novel clinical and molecular findings in Spanish patients with naevoid basal cell carcinoma syndrome. British Journal of Dermatology, 2018, 178, 198-206.	1.4	5

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127	Chromosome microarray analysis should be offered to all invasive prenatal diagnostic testing following a normal rapid aneuploidy test result. Clinical Genetics, 2020, 98, 379-383.	1.0	5
128	Acquired erythropoietic uroporphyria secondary to myeloid malignancy: A case report and literature review. Photodermatology Photoimmunology and Photomedicine, 2022, 38, 86-91.	0.7	5
129	46,XY,18q+/46,XY,18qâ^' mosaicism in a fragile X prenatal diagnosis. Prenatal Diagnosis, 2005, 25, 448-450.	1.1	4
130	A Retrospective and Theoretical Evaluation of Rapid Methods for Detecting Chromosome Abnormalities and Their Implications on Genetic Counseling Based on a Series of 3868 CVS Diagnoses. Fetal Diagnosis and Therapy, 2008, 23, 126-131.	0.6	4
131	Hepatoerythropoietic porphyria due to a novel mutation in the uroporphyrinogen decarboxylase gene. British Journal of Dermatology, 2011, 165, no-no.	1.4	4
132	Atypical Clinical Presentation of Xeroderma Pigmentosum in a Patient Harboring a Novel Missense Mutation in the XPC Gene: The Importance of Clinical Suspicion. Dermatology, 2015, 231, 217-221.	0.9	4
133	Genome-wide linkage analysis in Spanish melanoma-prone families identifies a new familial melanoma susceptibility locus at 11q. European Journal of Human Genetics, 2018, 26, 1188-1193.	1.4	4
134	Prognostic value of tyrosinase reverse transcriptase PCR analysis in melanoma sentinel lymph nodes: long-term follow-up analysis. Clinical and Experimental Dermatology, 2009, 34, 863-869.	0.6	3
135	Hepatoerythropoietic Porphyria and Familial Porphyria Cutanea Tarda in Spanish Patients: G281E Mutation in the Uroporphyrinogen Decarboxylase Gene. Archives of Dermatology, 2010, 146, 1313-4.	1.7	3
136	Reply. Annals of Neurology, 2016, 79, 161-163.	2.8	3
137	Genetic linkage analysis of a large family identifies FIGN as a candidate modulator of reduced penetrance in heritable pulmonary arterial hypertension. Journal of Medical Genetics, 2019, 56, 481-490.	1.5	3
138	Significance of Low Maternal Serum Î²-hCG Levels in the Assessment of the Risk of Atypical Chromosomal Abnormalities. Fetal Diagnosis and Therapy, 2021, 48, 849-856.	0.6	3
139	Isolation and characterization of a Tn5-induced tolQ mutant of Escherichia coli. Canadian Journal of Microbiology, 1994, 40, 503-507.	0.8	2
140	Mutations and Intragenic Polymorphisms in the Diagnosis of Autosomal Dominant Polycystic Kidney Disease Type 1. , 1997, 122, 45-48.		2
141	Molecular study of thePAK3 andGDI1 genes in nonsyndromic X-linked mental retardation Spanish patients. American Journal of Medical Genetics Part A, 2000, 94, 389-391.	2.4	2
142	Time and tumor type (primary or metastatic) do not influence the detection of BRAF/NRAS mutations in formalin fixed paraffin embedded samples from melanomas. Clinical Chemistry and Laboratory Medicine, 2016, 54, 1733-1738.	1.4	2
143	The Contribution of QF-PCR and Pathology Studies in the Diagnosis of Diandric Triploidy/Partial Mole. Diagnostics, 2021, 11, 1811.	1.3	2
144	DNA Repair and Immune Response Pathways Are Deregulated in Melanocyte-Keratinocyte Co-cultures Derived From the Healthy Skin of Familial Melanoma Patients. Frontiers in Medicine, 2021, 8, 692341.	1.2	2

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145	Hypertension in Polycystic Kidney Disease Types 1 and 2 and Its Effect on the Age of Onset of End-Stage Renal Disease. , 1997, 122, 28-30.		1
146	Linkage analysis in Spanish families with nonspecific X-linked mental retardation: Significant linkage at Xq13-q21. American Journal of Medical Genetics Part A, 2001, 98, 343-347.	2.4	1
147	Reply. Annals of Neurology, 2015, 78, 153-154.	2.8	1
148	A 92,XXXY Miscarriage Consecutive to a Digynic Triploid Pregnancy. Cytogenetic and Genome Research, 2016, 149, 258-261.	0.6	1
149	Genomic Microarray in Fetuses With Early Growth Restriction: A Multicenter Study. Obstetrical and Gynecological Survey, 2018, 73, 73-74.	0.2	1
150	A New Stepwise Molecular Work-Up After Chorionic Villi Sampling in Women With an Early Pregnancy Loss. Frontiers in Genetics, 2020, 11, 561720.	1.1	1
151	Discrepant mutational status between naevi and melanomas in naevus-associated melanomas: about mutation-specific immunohistochemistry: reply from the authors. British Journal of Dermatology, 2016, 175, 435-435.	1.4	0
152	Reply. Annals of Neurology, 2016, 79, 868-868.	2.8	0
153	FRI-435-Progression to cirrhosis is not infrequent in patients with Wilsonâ€™s disease despite treatment. Journal of Hepatology, 2019, 70, e585.	1.8	0