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	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
2	Acquired erythropoietic uroporphyria secondary to myeloid malignancy: A case report and literature review. Photodermatology Photoimmunology and Photomedicine, 2022, 38, 86-91.	0.7	5
3	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
4	The Contribution of QF-PCR and Pathology Studies in the Diagnosis of Diandric Triploidy/Partial Mole. Diagnostics, 2021, 11, 1811.	1.3	2
5	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
6	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
7	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
8	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
9	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
10	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
11	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
12	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
13	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
14	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
15	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
16	<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
17	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
18	Genomic Microarray in Fetuses With Early Growth Restriction: A Multicenter Study. Obstetrical and Gynecological Survey, 2018, 73, 73-74.	0.2	1

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19	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
20	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
21	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
22	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
23	IRF4 rs12203592 functional variant and melanoma survival. International Journal of Cancer, 2017, 140, 1845-1849.	2.3	11
24	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
25	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
26	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
27	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
28	Genomic Microarray in Fetuses with Early Growth Restriction: A Multicenter Study. Fetal Diagnosis and Therapy, 2017, 42, 174-180.	0.6	33
29	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
30	Amelanotic melanoma in oculocutaneous albinism: a genetic, dermoscopic and reflectance confocal microscopy study. British Journal of Dermatology, 2017, 177, e333-e335.	1.4	9
31	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
32	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
33	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
34	Association Between Confocal Morphologic Classification and Clinical Phenotypes of Multiple Primary and Familial Melanomas. JAMA Dermatology, 2016, 152, 1099.	2.0	13
35	Genetic susceptibility to cutaneous melanoma in southern Switzerland: role ofCDKN2A,MC1RandMITF. British Journal of Dermatology, 2016, 175, 1030-1037.	1.4	17
36	A 92,XXXY Miscarriage Consecutive to a Digynic Triploid Pregnancy. Cytogenetic and Genome Research, 2016, 149, 258-261.	0.6	1

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37	Reply. Annals of Neurology, 2016, 79, 161-163.	2.8	3
38	Reply. Annals of Neurology, 2016, 79, 868-868.	2.8	0
39	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
40	Prevalence ofMITFp.E318K in Patients With Melanoma Independent of the Presence ofCDKN2ACausative Mutations. JAMA Dermatology, 2016, 152, 405.	2.0	41
41	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
42	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
43	Mutational status of naevus-associated melanomas. British Journal of Dermatology, 2015, 173, 671-680.	1.4	42
44	Reply. Annals of Neurology, 2015, 78, 153-154.	2.8	1
45	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
46	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
47	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
48	MultipleBRAFWild-Type Melanomas During Dabrafenib Treatment for MetastaticBRAF-Mutant Melanoma. JAMA Dermatology, 2015, 151, 544.	2.0	15
49	Update in genetic susceptibility in melanoma. Annals of Translational Medicine, 2015, 3, 210.	0.7	100
50	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
51	Clinical and Histopathological Characteristics between Familial and Sporadic Melanoma in Barcelona, Spain. Journal of Clinical & Experimental Dermatology Research, 2014, 05, 231.	0.1	7
52	Multiple Primary Acral Melanomas in Two Young Caucasian Patients. Dermatology, 2014, 228, 307-310.	0.9	10
53	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
54	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

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55	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
56	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
57	TERT Promoter Mutation Status as an Independent Prognostic Factor in Cutaneous Melanoma. Journal of the National Cancer Institute, 2014, 106, .	3.0	204
58	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
59	Successful Treatment of Congenital Erythropoietic Porphyria Using Matched Unrelated Hematopoietic Stem Cell Transplantation. Pediatric Dermatology, 2013, 30, 484-489.	0.5	24
60	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
61	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
62	Genetic alterations in RAS-regulated pathway in acral lentiginous melanoma. Experimental Dermatology, 2013, 22, 148-150.	1.4	49
63	Evaluation of <scp>PAX</scp>3 genetic variants and nevus number. Pigment Cell and Melanoma Research, 2013, 26, 666-676.	1.5	7
64	Multiple primary melanomas: do they look the same?. British Journal of Dermatology, 2013, 168, 1267-1272.	1.4	16
65	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
66	Benefits of total body photography and digital dermatoscopy (â€œtwo-step method of digital) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 30 American Academy of Dermatology, 2012, 67, e17-e27.	0.6	176
67	Genetic counseling in melanoma. Dermatologic Therapy, 2012, 25, 397-402.	0.8	28
68	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
69	Molecular characterization of human cutaneous melanoma-derived cell lines. Anticancer Research, 2012, 32, 1245-51.	0.5	7
70	Genome-wide association study identifies novel loci predisposing to cutaneous melanomaâ€. Human Molecular Genetics, 2011, 20, 5012-5023.	1.4	187
71	Genome-wide association study identifies three new melanoma susceptibility loci. Nature Genetics, 2011, 43, 1108-1113.	9.4	230
72	A SUMOylation-defective MITF germline mutation predisposes to melanoma and renal carcinoma. Nature, 2011, 480, 94-98.	13.7	466

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73	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
74	ALAS2 acts as a modifier gene in patients with congenital erythropoietic porphyria. Blood, 2011, 118, 1443-1451.	0.6	80
75	Hepatoerythropoietic porphyria due to a novel mutation in the uroporphyrinogen decarboxylase gene. British Journal of Dermatology, 2011, 165, no-no.	1.4	4
76	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
77	Familial and Sporadic Porphyria Cutanea Tarda. Medicine (United States), 2010, 89, 69-74.	0.4	33
78	Chorionic villus sampling in the prenatal diagnosis of placental mesenchymal dysplasia. Ultrasound in Obstetrics and Gynecology, 2010, 36, 644-645.	0.9	7
79	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
80	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
81	Assessment of QF-PCR as the First Approach in Prenatal Diagnosis. Journal of Molecular Diagnostics, 2010, 12, 828-834.	1.2	39
82	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
83	Fragile X syndrome prenatal diagnosis: parental attitudes and reproductive responses. Reproductive BioMedicine Online, 2010, 21, 560-565.	1.1	7
84	Structure and Regulation of the Versican Promoter. Journal of Biological Chemistry, 2009, 284, 12306-12317.	1.6	29
85	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
86	Dermoscopic features of melanomas associated withMC1Rvariants in SpanishCDKN2Amutation carriers. British Journal of Dermatology, 2009, 160, 48-53.	1.4	48
87	CDKN2Amutations in melanoma families from Uruguay. British Journal of Dermatology, 2009, 161, 536-541.	1.4	20
88	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
89	Penetrance of FMR1 premutation associated pathologies in fragile X syndrome families. European Journal of Human Genetics, 2009, 17, 1359-1362.	1.4	254
90	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

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91	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
92	Premature ovarian failure and fragile X female premutation carriers. Menopause, 2009, 16, 944-949.	0.8	27
93	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
94	Deletion of the OPHN1 gene detected by aCGH. Journal of Intellectual Disability Research, 2008, 52, 190-194.	1.2	14
95	A comparison of CDKN2A mutation detection within the Melanoma Genetics Consortium (GenoMEL). European Journal of Cancer, 2008, 44, 1269-1274.	1.3	26
96	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
97	Screening for FXTAS in 95 Spanish Patients Negative for Huntington Disease. Genetic Testing and Molecular Biomarkers, 2008, 12, 135-138.	1.7	13
98	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
99	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
100	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
101	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
102	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
103	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
104	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
105	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
106	Novedades en genÃ©tica del melanoma. Piel, 2006, 21, 272-274.	0.0	7
107	Screening for MECP2 mutations in Spanish patients with an unexplained mental retardation. Clinical Genetics, 2006, 70, 140-144.	1.0	13
108	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

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109	Mutation of the tumour suppressor p33 ING1 b is rare in melanoma. British Journal of Dermatology, 2006, 155, 94-99.	1.4	12
110	A novel mutation in JARID1C gene associated with mental retardation. European Journal of Human Genetics, 2006, 14, 583-586.	1.4	78
111	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
112	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
113	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
114	Elastin Mutation Screening in a Group of Patients Affected by Vascular Abnormalities. Pediatric Cardiology, 2005, 26, 827-831.	0.6	22
115	46,XY,18q+/46,XY,18qâ˜ mosaicism in a fragile X prenatal diagnosis. Prenatal Diagnosis, 2005, 25, 448-450.	1.1	4
116	Association between BDNF Val66Met polymorphism and age at onset in Huntington disease. Neurology, 2005, 65, 964-965.	1.5	36
117	Role of the CDKN2A Locus in Patients With Multiple Primary Melanomas. Journal of Clinical Oncology, 2005, 23, 3043-3051.	0.8	138
118	Biochemical and genetic characterization of four cases of hereditary coproporphyria in Spain. Molecular Genetics and Metabolism, 2005, 85, 160-163.	0.5	8
119	A Novel Elastin Gene Mutation Resulting in an Autosomal Dominant Form of Cutis Laxa. Archives of Dermatology, 2004, 140, 1135-9.	1.7	73
120	Analysis of CGG variation through 642 meioses in Fragile X families. Molecular Human Reproduction, 2004, 10, 773-776.	1.3	22
121	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
122	Fetoplacental discrepancy involving structural abnormalities of chromosome 8 detected by prenatal diagnosis. Prenatal Diagnosis, 2003, 23, 319-322.	1.1	24
123	Cryptic chromosomal rearrangement screening in 30 patients with mental retardation and dysmorphic features. Clinical Genetics, 2003, 65, 17-23.	1.0	16
124	Incidence of Fragile X in 5,000 Consecutive Newborn Males. Genetic Testing and Molecular Biomarkers, 2003, 7, 339-343.	1.7	54
125	SCA8 in the Spanish population including one homozygous patient. Clinical Genetics, 2002, 62, 404-409.	1.0	16
126	Pilot study for the neonatal screening of fragile X syndrome. Prenatal Diagnosis, 2002, 22, 459-462.	1.1	8

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127	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
128	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
129	A melanoma-associated germline mutation in exon 1Î² inactivates p14ARF. Oncogene, 2001, 20, 5543-5547.	2.6	178
130	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
131	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
132	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
133	Rare variants in the promoter of the fragile X syndrome gene (FMR1). Molecular and Cellular Probes, 2000, 14, 115-119.	0.9	15
134	Increased prevalence of polycystic kidney disease type 2 among elderly polycystic patients. American Journal of Kidney Diseases, 2000, 36, 728-734.	2.1	48
135	Autosomal Dominant Polycystic Kidney Disease Types 1 and 2: Assessment of US Sensitivity for Diagnosis. Radiology, 1999, 213, 273-276.	3.6	89
136	Mutational analysis within the 3â€² region of the PKD1 gene. Kidney International, 1999, 55, 1225-1233.	2.6	41
137	Seven novel mutations of the PKD2 gene in families with autosomal dominant polycystic kidney disease. Kidney International, 1999, 56, 28-33.	2.6	29
138	Single-strand conformation polymorphism analysis in theFMR1. American Journal of Medical Genetics Part A, 1999, 84, 262-265.	2.4	8
139	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
140	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
141	Autosomal recessive Alport syndrome: linkage analysis and clinical features in two families. Nephrology Dialysis Transplantation, 1999, 14, 627-630.	0.4	16
142	Recurrence of the PKD1 nonsense mutation Q4041X in Spanish, Italian, and British families. Human Mutation, 1998, 11, S117-S120.	1.1	16
143	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
144	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

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145	Autosomal recessive polycystic kidney disease presenting in adulthood. Molecular diagnosis of the family. Nephrology Dialysis Transplantation, 1998, 13, 1273-1276.	0.4	8
146	Maternal transmission in sporadic Huntington's disease.. Journal of Neurology, Neurosurgery and Psychiatry, 1997, 62, 535-537.	0.9	14
147	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
148	Mutations and Intragenic Polymorphisms in the Diagnosis of Autosomal Dominant Polycystic Kidney Disease Type 1. , 1997, 122, 45-48.		2
149	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
150	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
151	A female compound heterozygote (pre- and full mutation) for the CGG FMR1 expansion. Human Genetics, 1996, 98, 419-421.	1.8	22
152	Isolation and characterization of a Tn5-induced tolQ mutant of Escherichia coli. Canadian Journal of Microbiology, 1994, 40, 503-507.	0.8	2
153	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