Xavier Estivill

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/2741146/publications.pdf

Version: 2024-02-01

560 papers	57,476 citations	1457 107 h-index	1561 217 g-index
586 all docs	586 docs citations	586 times ranked	64497 citing authors

#	Article	IF	Citations
1	Common Genetic Variation and Age of Onset of Anorexia Nervosa. Biological Psychiatry Global Open Science, 2022, 2, 368-378.	1.0	10
2	Genome sequencing data analysis for rare disease gene discovery. Briefings in Bioinformatics, 2022, 23, .	3.2	6
3	Analysis of incidental findings in Qatar genome participants reveals novel functional variants in LMNA and DSP. Human Molecular Genetics, 2022, , .	1.4	2
4	Identification of autosomal cis expression quantitative trait methylation (cis eQTMs) in childrenâ€™s blood. ELife, 2022, 11, .	2.8	28
5	Functional Characterization of the MYO6 Variant p.E60Q in Non-Syndromic Hearing Loss Patients. International Journal of Molecular Sciences, 2022, 23, 3369.	1.8	1
6	Cell type-specific novel long non-coding RNA and circular RNA in the BLUEPRINT hematopoietic transcriptomes atlas. Haematologica, 2021, 106, 2613-2623.	1.7	12
7	Shared genetic risk between eating disorderâ€•and substanceâ€•useâ€•related phenotypes: Evidence from genomeâ€•wide association studies. Addiction Biology, 2021, 26, e12880.	1.4	28
8	Efficient and flexible Integration of variant characteristics in rare variant association studies using integrated nested Laplace approximation. PLoS Computational Biology, 2021, 17, e1007784.	1.5	4
9	Genetic evaluation of cardiomyopathies in Qatar identifies enrichment of pathogenic sarcomere gene variants and possible founder disease mutations in the Arabs. Molecular Genetics & Genomic Medicine, 2021, 9, e1709.	0.6	2
10	The alternative serotonin transporter promoter P2 impacts gene function in females with irritable bowel syndrome. Journal of Cellular and Molecular Medicine, 2021, 25, 8047-8061.	1.6	5
11	Variability of multi-omics profiles in a population-based child cohort. BMC Medicine, 2021, 19, 166.	2.3	23
12	Actionable genomic variants in 6045 participants from the Qatar Genome Program. Human Mutation, 2021, 42, 1584-1601.	1.1	13
13	In utero and childhood exposure to tobacco smoke and multi-layer molecular signatures in children. BMC Medicine, 2020, 18, 243.	2.3	22
14	Genome-wide association study identifies eight risk loci and implicates metabo-psychiatric origins for anorexia nervosa. Nature Genetics, 2019, 51, 1207-1214.	9.4	641
15	GWAS on longitudinal growth traits reveals different genetic factors influencing infant, child, and adult BMI. Science Advances, 2019, 5, eaaw3095.	4.7	86
16	Associations Between Attention-Deficit/Hyperactivity Disorder and Various Eating Disorders: A Swedish Nationwide Population Study Using Multiple Genetically Informative Approaches. Biological Psychiatry, 2019, 86, 577-586.	0.7	43
17	Maternal and fetal genetic effects on birth weight and their relevance to cardio-metabolic risk factors. Nature Genetics, 2019, 51, 804-814.	9.4	402
18	eDiVAâ€”Classification and prioritization of pathogenic variants for clinical diagnostics. Human Mutation, 2019, 40, 865-878.	1.1	19

#	Article	IF	Citations
19	Biallelic loss-of-function LACC1/FAMIN Mutations Presenting as Rheumatoid Factor-Negative Polyarticular Juvenile Idiopathic Arthritis. Scientific Reports, 2019, 9, 4579.	1.6	20
20	Genome-wide association study in frontal fibrosing alopecia identifies four susceptibility loci including HLA-B*07:02. Nature Communications, 2019, 10, 1150.	5.8	82
21	Allele balance bias identifies systematic genotyping errors and false disease associations. Human Mutation, 2019, 40, 115-126.	1.1	23
22	Genome-wide association study of offspring birth weight in 86â€‰577 women identifies five novel loci and highlights maternal genetic effects that are independent of fetal genetics. Human Molecular Genetics, 2018, 27, 742-756.	1.4	156
23	Geolocalisation of athletes for out-of-competition drug testing: ethical considerations. Position statement by the WADA Ethics Panel. British Journal of Sports Medicine, 2018, 52, 456-459.	3.1	6
24	miRTrace reveals the organismal origins of microRNA sequencing data. Genome Biology, 2018, 19, 213.	3.8	44
25	Human Early Life Exposome (HELIX) study: a European population-based exposome cohort. BMJ Open, 2018, 8, e021311.	0.8	161
26	Circulating miRNAs, isomiRs and small RNA clusters in human plasma and breast milk. PLoS ONE, 2018, 13, e0193527.	1.1	51
27	Detailed analysis of inversions predicted between two human genomes: errors, real polymorphisms, and their origin and population distribution. Human Molecular Genetics, 2017, 26, ddw415.	1.4	12
28	Contribution of theTTC21Bgene to glomerular and cystic kidney diseases. Nephrology Dialysis Transplantation, 2017, 32, gfv453.	0.4	26
29	Survey of 800+ data sets from human tissue and body fluid reveals xenomiRs are likely artifacts. Rna, 2017, 23, 433-445.	1.6	65
30	Significant Locus and Metabolic Genetic Correlations Revealed in Genome-Wide Association Study of Anorexia Nervosa. American Journal of Psychiatry, 2017, 174, 850-858.	4.0	410
31	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American Journal of Human Genetics, 2017, 100, 695-705.	2.6	305
32	Genetic architecture distinguishes systemic juvenile idiopathic arthritis from other forms of juvenile idiopathic arthritis: clinical and therapeutic implications. Annals of the Rheumatic Diseases, 2017, 76, 906-913.	0.5	123
33	Signatures of positive selection reveal a universal role of chromatin modifiers as cancer driver genes. Scientific Reports, 2017, 7, 13124.	1.6	20
34	The acute effects of ultraviolet radiation on the blood transcriptome are independent of plasma 25OHD3. Environmental Research, 2017, 159, 239-248.	3.7	13
35	Exploration of large, rare copy number variants associated with psychiatric and neurodevelopmental disorders in individuals with anorexia nervosa. Psychiatric Genetics, 2017, 27, 152-158.	0.6	18
36	NGS-Based Assay for the Identification of Individuals Carrying Recessive Genetic Mutations in Reproductive Medicine. Human Mutation, 2016, 37, 516-523.	1.1	43

#	Article	IF	Citations
37	Identification of Gene Mutations and Fusion Genes in Patients with SÃ©zary Syndrome. Journal of Investigative Dermatology, 2016, 136, 1490-1499.	0.3	77
38	Tying malaria and microRNAs: from the biology to future diagnostic perspectives. Malaria Journal, 2016, 15, 167.	0.8	19
39	Genome-wide DNA methylation study in human placenta identifies novel loci associated with maternal smoking during pregnancy. International Journal of Epidemiology, 2016, 45, 1644-1655.	0.9	85
40	A genome-wide association meta-analysis of diarrhoeal disease in young children identifiesFUT2locus and provides plausible biological pathways. Human Molecular Genetics, 2016, 25, 4127-4142.	1.4	35
41	Heritability and Genome-Wide Association Analyses of Sleep Duration in Children: The EAGLE Consortium. Sleep, 2016, 39, 1859-1869.	0.6	34
42	Prenatal exposure to mixtures of xenoestrogens and genome-wide DNA methylation in human placenta. Epigenomics, 2016, 8, 43-54.	1.0	15
43	Specific small-RNA signatures in the amygdala at premotor and motor stages of Parkinsonâ€™s disease revealed by deep sequencing analysis. Bioinformatics, 2016, 32, 673-681.	1.8	29
44	Smellâ€“taste dysfunctions in extreme weight/eating conditions: analysis of hormonal and psychological interactions. Endocrine, 2016, 51, 256-267.	1.1	82
45	Targeting CAG repeat RNAs reduces Huntingtonâ€™s disease phenotype independently of huntingtin levels. Journal of Clinical Investigation, 2016, 126, 4319-4330.	3.9	59
46	Genome-wide meta-analysis identifies multiple novel associations and ethnic heterogeneity of psoriasis susceptibility. Nature Communications, 2015, 6, 6916.	5.8	154
47	Towards a European consensus for reporting incidental findings during clinical NGS testing. European Journal of Human Genetics, 2015, 23, 1601-1606.	1.4	85
48	HLA-DRB1*11and variants of the MHC class II locus are strong risk factors for systemic juvenile idiopathic arthritis. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 15970-15975.	3.3	139
49	CaracterÃsticas genÃ©ticas de pacientes reumatolÃ³gicos que desarrollan lesiones cutÃ¡neas inflamatorias inducidas por fÃ¡rmacos biolÃ³gicos. ReumatologÃa ClÃnica, 2015, 11, 126-127.	0.2	0
50	Rare variants in Î²-Amyloid precursor protein (APP) and Parkinsonâ€™s disease. European Journal of Human Genetics, 2015, 23, 1328-1333.	1.4	50
51	Genetic Characteristics of Rheumatic Patients Developing Inflammatory Skin Lesions Induced by Biologic Therapy. ReumatologÃa ClÃnica (English Edition), 2015, 11, 126-127.	0.2	0
52	Missense mutations inTENM4, a regulator of axon guidance and central myelination, cause essential tremor. Human Molecular Genetics, 2015, 24, 5677-5686.	1.4	134
53	Deregulation of key signaling pathways involved in oocyte maturation in FMR1 premutation carriers with Fragile X-associated primary ovarian insufficiency. Gene, 2015, 571, 52-57.	1.0	17
54	Non-coding recurrent mutations in chronic lymphocytic leukaemia. Nature, 2015, 526, 519-524.	13.7	749

#	Article	IF	Citations
55	Effect of predicted protein-truncating genetic variants on the human transcriptome. Science, 2015, 348, 666-669.	6.0	252
56	Switching to zebrafish neurobehavioral models: The obsessiveâ€“compulsive disorder paradigm. European Journal of Pharmacology, 2015, 759, 142-150.	1.7	12
57	Genetic variation and alternative splicing. Nature Biotechnology, 2015, 33, 357-359.	9.4	5
58	Mutations in DCHS1 cause mitral valve prolapse. Nature, 2015, 525, 109-113.	13.7	150
59	Circulating Betatrophin Levels Are Increased in Anorexia and Decreased in Morbidly Obese Women. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E1188-E1196.	1.8	42
60	A novel common variant in DCST2 is associated with length in early life and height in adulthood. Human Molecular Genetics, 2015, 24, 1155-1168.	1.4	109
61	Targeted next-generation sequencing in steroid-resistant nephrotic syndrome: mutations in multiple glomerular genes may influence disease severity. European Journal of Human Genetics, 2015, 23, 1192-1199.	1.4	72
62	Association of Irisin with Fat Mass, Resting Energy Expenditure, and Daily Activity in Conditions of Extreme Body Mass Index. International Journal of Endocrinology, 2014, 2014, 1-9.	0.6	151
63	The Human Early-Life Exposome (HELIX): Project Rationale and Design. Environmental Health Perspectives, 2014, 122, 535-544.	2.8	280
64	Validation and Genotyping of Multiple Human Polymorphic Inversions Mediated by Inverted Repeats Reveals a High Degree of Recurrence. PLoS Genetics, 2014, 10, e1004208.	1.5	28
65	Extensive sequence analysis of <scp>CFTR</scp>, <scp>SCNN1A</scp>, <scp>SCNN1B</scp>, <scp>SCNN1G</scp> and <scp>SERPINA1</scp> suggests an oligogenic basis for cystic fibrosisâ€like phenotypes. Clinical Genetics, 2014, 86, 91-95.	1.0	19
66	Activating Mutations Cluster in the â€œMolecular Brakeâ€•Regions of Protein Kinases and Do Not Associate with Conserved or Catalytic Residues. Human Mutation, 2014, 35, 318-328.	1.1	20
67	Accurate molecular diagnosis of phenylketonuria and tetrahydrobiopterin-deficient hyperphenylalaninemias using high-throughput targeted sequencing. European Journal of Human Genetics, 2014, 22, 528-534.	1.4	36
68	Blood expression profiles of fragile X premutation carriers identify candidate genes involved in neurodegenerative and infertility phenotypes. Neurobiology of Disease, 2014, 65, 43-54.	2.1	23
69	Diagnosis of autosomal dominant polycystic kidney disease using efficient PKD1 and PKD2 targeted nextâ€generation sequencing. Molecular Genetics & Genomic Medicine, 2014, 2, 412-421.	0.6	67
70	Prenatal exposure to mixtures of xenoestrogens and repetitive element DNA methylation changes in human placenta. Environment International, 2014, 71, 81-87.	4.8	52
71	Relationship between genome and epigenome - challenges and requirements for future research. BMC Genomics, 2014, 15, 487.	1.2	24
72	Evidence for the biogenesis of more than 1,000 novel human microRNAs. Genome Biology, 2014, 15, R57.	13.9	222

#	Article	IF	Citations
73	A genome-wide association study of anorexia nervosa. Molecular Psychiatry, 2014, 19, 1085-1094.	4.1	282
74	Genome-wide analysis of single nucleotide polymorphisms and copy number variants in fibromyalgia suggest a role for the central nervous system. Pain, 2014, 155, 1102-1109.	2.0	54
75	ALDH5A1 variability in opioid dependent patients could influence response to methadone treatment. European Neuropsychopharmacology, 2014, 24, 420-424.	0.3	10
76	<scp>MicroRNA</scp> expression profiling in blood from fragile Xâ€associated tremor/ataxia syndrome patients. Genes, Brain and Behavior, 2013, 12, 595-603.	1.1	25
77	Worldwide population distribution of the common LCE3C-LCE3B deletion associated with psoriasis and other autoimmune disorders. BMC Genomics, 2013, 14, 261.	1.2	9
78	Genetic characterization of northeastern Italian population isolates in the context of broader European genetic diversity. European Journal of Human Genetics, 2013, 21, 659-665.	1.4	64
79	A highly expressed miR-101 isomiR is a functional silencing small RNA. BMC Genomics, 2013, 14, 104.	1.2	93
80	A common 56-kilobase deletion in a primate-specific segmental duplication creates a novel butyrophilin-like protein. BMC Genetics, 2013, 14, 61.	2.7	27
81	Reproducibility of high-throughput mRNA and small RNA sequencing across laboratories. Nature Biotechnology, 2013, 31, 1015-1022.	9.4	251
82	Storage conditions and stability of global DNA methylation in placental tissue. Epigenomics, 2013, 5, 341-348.	1.0	34
83	Transcriptome and genome sequencing uncovers functional variation in humans. Nature, 2013, 501, 506-511.	13.7	1,857
84	Interaction of SLC1A1 gene variants and life stress on pharmacological resistance in obsessiveâ€“compulsive disorder. Pharmacogenomics Journal, 2013, 13, 470-475.	0.9	15
85	The interaction between Comt and Bdnf variants influences obsessiveâ€“compulsive-related dysfunctional beliefs. Journal of Anxiety Disorders, 2013, 27, 321-327.	1.5	17
86	Screening for the presence of FMR1 premutation alleles in women with fibromyalgia. Gene, 2013, 512, 305-308.	1.0	13
87	VAL66MET BDNF GENOTYPES IN MELANCHOLIC DEPRESSION: EFFECTS ON BRAIN STRUCTURE AND TREATMENT OUTCOME. Depression and Anxiety, 2013, 30, 225-233.	2.0	39
88	Upregulation of a small vault RNA (svtRNA2-1a) is an early event in Parkinson disease and induces neuronal dysfunction. RNA Biology, 2013, 10, 1093-1106.	1.5	44
89	Next generation diagnostics of cystic fibrosis andCFTR-related disorders by targeted multiplex high-coverage resequencing ofCFTR. Journal of Medical Genetics, 2013, 50, 455-462.	1.5	39
90	Sporadic and reversible chromothripsis in chronic lymphocytic leukemia revealed by longitudinal genomic analysis. Leukemia, 2013, 27, 2376-2379.	3.3	29

#	Article	IF	Citations
91	New loci associated with birth weight identify genetic links between intrauterine growth and adult height and metabolism. Nature Genetics, 2013, 45, 76-82.	9.4	293
92	The complex SNP and CNV genetic architecture of the increased risk of congenital heart defects in Down syndrome. Genome Research, 2013, 23, 1410-1421.	2.4	65
93	PeSV-Fisher: Identification of Somatic and Non-Somatic Structural Variants Using Next Generation Sequencing Data. PLoS ONE, 2013, 8, e63377.	1.1	17
94	Cluster Analysis of Clinical Data Identifies Fibromyalgia Subgroups. PLoS ONE, 2013, 8, e74873.	1.1	49
95	Small non-coding RNAs add complexity to the RNA pathogenic mechanisms in trinucleotide repeat expansion diseases. Frontiers in Molecular Neuroscience, 2013, 6, 45.	1.4	10
96	A Pathogenic Mechanism in Huntington's Disease Involves Small CAG-Repeated RNAs with Neurotoxic Activity. PLoS Genetics, 2012, 8, e1002481.	1.5	161
97	Association between the NMDA glutamate receptor GRIN2B gene and obsessiveâ€“compulsive disorder. Journal of Psychiatry and Neuroscience, 2012, 37, 273-281.	1.4	46
98	DNA Hypomethylation atALOX12Is Associated with Persistent Wheezing in Childhood. American Journal of Respiratory and Critical Care Medicine, 2012, 185, 937-943.	2.5	97
99	CYP2D6 polymorphism in patients with eating disorders. Pharmacogenomics Journal, 2012, 12, 173-175.	0.9	25
100	Common variants at 12q15 and 12q24 are associated with infant head circumference. Nature Genetics, 2012, 44, 532-538.	9.4	130
101	KLHL3 mutations cause familial hyperkalemic hypertension by impairing ion transport in the distal nephron. Nature Genetics, 2012, 44, 456-460.	9.4	281
102	Variation in the BDNF Val66Met polymorphism and response to cognitive-behavior therapy in obsessive-compulsive disorder. European Psychiatry, 2012, 27, 386-390.	0.1	36
103	Genetic epistasis in female suicide attempters. Progress in Neuro-Psychopharmacology and Biological Psychiatry, 2012, 38, 294-301.	2.5	9
104	Influence of fetal glutathione S-transferase copy number variants on adverse reproductive outcomes. BJOG: an International Journal of Obstetrics and Gynaecology, 2012, 119, 1141-1146.	1.1	11
105	Aberrant brain microRNA target and miRISC gene expression in the anx/anx anorexia mouse model. Gene, 2012, 497, 181-190.	1.0	12
106	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. Nature Genetics, 2012, 44, 1341-1348.	9.4	848
107	Exome sequencing identifies recurrent mutations of the splicing factor SF3B1 gene in chronic lymphocytic leukemia. Nature Genetics, 2012, 44, 47-52.	9.4	893
108	Fat Mass and Obesity-Associated Gene (<i>FTO</i>) in Eating Disorders: Evidence for Association of the rs9939609 Obesity Risk Allele with Bulimia nervosa and Anorexia nervosa. Obesity Facts, 2012, 5, 408-419.	1.6	46

#	Article	IF	Citations
109	BLUEPRINT to decode the epigenetic signature written in blood. Nature Biotechnology, 2012, 30, 224-226.	9.4	323
110	Overexpression of the CHRNA5/A3/B4 genomic cluster in mice increases the sensitivity to nicotine and modifies its reinforcing effects. Amino Acids, 2012, 43, 897-909.	1.2	36
111	Association of Neurexin 3 polymorphisms with smoking behavior. Genes, Brain and Behavior, 2012, 11, 704-711.	1.1	29
112	Whole-genome sequencing identifies recurrent mutations in chronic lymphocytic leukaemia. Nature, 2011, 475, 101-105.	13.7	1,364
113	MicroRNA profiling of Parkinson's disease brains identifies early downregulation of miR-34b/c which modulate mitochondrial function. Human Molecular Genetics, 2011, 20, 3067-3078.	1.4	433
114	Human microRNAs miR-22, miR-138-2, miR-148a, and miR-488 Are Associated with Panic Disorder and Regulate Several Anxiety Candidate Genes and Related Pathways. Biological Psychiatry, 2011, 69, 526-533.	0.7	167
115	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. Nature, 2011, 478, 97-102.	13.7	394
116	D184E mutation in aquaporin-4 gene impairs water permeability and links to deafness. Neuroscience, 2011, 197, 80-88.	1.1	31
117	Genetic Variants of the FADS Gene Cluster and ELOVL Gene Family, Colostrums LC-PUFA Levels, Breastfeeding, and Child Cognition. PLoS ONE, 2011, 6, e17181.	1.1	111
118	DNA methylation in neurodegenerative disorders: a missing link between genome and environment?. Clinical Genetics, 2011, 80, 1-14.	1.0	53
119	Geneâ€“environment interaction in anorexia nervosa: relevance of non-shared environment and the serotonin transporter gene. Molecular Psychiatry, 2011, 16, 590-592.	4.1	60
120	High risk of lifetime history of suicide attempts among CYP2D6 ultrarapid metabolizers with eating disorders. Molecular Psychiatry, 2011, 16, 691-692.	4.1	45
121	When catastrophe strikes a cell. Nature, 2011, 470, 476-477.	13.7	77
122	Variants in estrogen receptor alpha gene are associated with phenotypical expression of obsessive-compulsive disorder. Psychoneuroendocrinology, 2011, 36, 473-483.	1.3	38
123	Maternal C-reactive protein levels in pregnancy are associated with wheezing and lower respiratory tract infections in the offspring. American Journal of Obstetrics and Gynecology, 2011, 204, 164.e1-164.e9.	0.7	29
124	Deletion of LCE3C and LCE3B is a susceptibility factor for psoriatic arthritis: A study in Spanish and Italian populations and meta-analysis. Arthritis and Rheumatism, 2011, 63, 1860-1865.	6.7	31
125	Meta-Analysis Confirms the LCE3C_LCE3B Deletion as a Risk Factor for Psoriasis in Several Ethnic Groups and Finds Interaction with HLA-Cw6. Journal of Investigative Dermatology, 2011, 131, 1105-1109.	0.3	89
126	ADRB2 Gly16Arg polymorphism, asthma control and lung function decline. European Respiratory Journal, 2011, 38, 1029-1035.	3.1	24

#	Article	IF	Citations
127	A non-biased framework for the annotation and classification of the non-miRNA small RNA transcriptome. Bioinformatics, 2011, 27, 3202-3203.	1.8	49
128	TÃ©cnicas de ReflexiÃ³n EstratÃ©gica: Search Conference Momentum. Profesional De La Informacion, 2011, 20, 71-77.	2.7	0
129	Nucleotide variation in central nervous system genes among male suicide attempters. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 208-213.	1.1	13
130	Independent Contribution of Common CFTR Variants to Chronic Pancreatitis. Pancreas, 2010, 39, 209-215.	0.5	30
131	Correlation of BDNF blood levels with interoceptive awareness and maturity fears in anorexia and bulimia nervosa patients. Journal of Neural Transmission, 2010, 117, 505-512.	1.4	22
132	Association study of 44 candidate genes with depressive and anxiety symptoms in post-partum women. Journal of Psychiatric Research, 2010, 44, 717-724.	1.5	69
133	Role of the neurotrophin network in eating disordersâ€™ subphenotypes: Body mass index and age at onset of the disease. Journal of Psychiatric Research, 2010, 44, 834-840.	1.5	10
134	Comprehensive copy number variant (CNV) analysis of neuronal pathways genes in psychiatric disorders identifies rare variants within patients. Journal of Psychiatric Research, 2010, 44, 971-978.	1.5	65
135	Gene expression signatures in breast cancer distinguish phenotype characteristics, histologic subtypes, and tumor invasiveness. Cancer, 2010, 116, 486-496.	2.0	60
136	Variants at APOE influence risk of deep and lobar intracerebral hemorrhage. Annals of Neurology, 2010, 68, 934-943.	2.8	241
137	Deletion of the late cornified envelope genes, LCE3C and LCE3B, is associated with rheumatoid arthritis. Arthritis and Rheumatism, 2010, 62, 1246-1251.	6.7	26
138	Overexpression of miR-128 specifically inhibits the truncated isoform of NTRK3 and upregulates BCL2 in SH-SY5Y neuroblastoma cells. BMC Molecular Biology, 2010, 11, 95.	3.0	78
139	Resequencing and association analysis of arylalkylamine N-acetyltransferase (AANAT) gene and its contribution to major depression susceptibility. Journal of Pineal Research, 2010, 49, no-no.	3.4	35
140	Characterization of a mouse model overexpressing betaâ€site APPâ€cleaving enzyme 2 reveals a new role for BACE2. Genes, Brain and Behavior, 2010, 9, 160-172.	1.1	23
141	A haplotype of glycogen synthase kinase 3Î² is associated with early onset of unipolar major depression. Genes, Brain and Behavior, 2010, 9, 799-807.	1.1	48
142	Design and evaluation of a panel of single-nucleotide polymorphisms in microRNA genomic regions for association studies in human disease. European Journal of Human Genetics, 2010, 18, 218-226.	1.4	34
143	Additional support for the association of SLITRK1 var321 and Tourette syndrome. Molecular Psychiatry, 2010, 15, 447-450.	4.1	55
144	Association of common copy number variants at the glutathione S-transferase genes and rare novel genomic changes with schizophrenia. Molecular Psychiatry, 2010, 15, 1023-1033.	4.1	74

#	Article	IF	Citations
145	International network of cancer genome projects. Nature, 2010, 464, 993-998.	13.7	2,114
146	A genome-wide association study identifies new psoriasis susceptibility loci and an interaction between HLA-C and ERAP1. Nature Genetics, 2010, 42, 985-990.	9.4	918
147	Positionally cloned genes and age-specific effects in asthma and atopy: an international population-based cohort study (ECRHS). Thorax, 2010, 65, 124-131.	2.7	25
148	Genetic variants and abnormal processing of pre-miR-182, a circadian clock modulator, in major depression patients with late insomnia. Human Molecular Genetics, 2010, 19, 4017-4025.	1.4	150
149	SeqBuster, a bioinformatic tool for the processing and analysis of small RNAs datasets, reveals ubiquitous miRNA modifications in human embryonic cells. Nucleic Acids Research, 2010, 38, e34-e34.	6.5	168
150	A myriad of miRNA variants in control and Huntingtonâ€™s disease brain regions detected by massively parallel sequencing. Nucleic Acids Research, 2010, 38, 7219-7235.	6.5	270
151	Deletion of Late Cornified Envelope 3B and 3C Genes Is Not Associated with Atopic Dermatitis. Journal of Investigative Dermatology, 2010, 130, 2057-2061.	0.3	25
152	Replication of LCE3Câ€“LCE3B CNV as a Risk Factor for Psoriasis and Analysis of Interaction with Other Genetic Risk Factors. Journal of Investigative Dermatology, 2010, 130, 979-984.	0.3	61
153	Deletion of LCE3C and LCE3B genes at PSORS4 does not contribute to susceptibility to psoriatic arthritis in German patients. Annals of the Rheumatic Diseases, 2010, 69, 876-878.	0.5	34
154	Assessment of the Neuropeptide S System in Anxiety Disorders. Biological Psychiatry, 2010, 68, 474-483.	0.7	79
155	Differential Association of Circadian Genes with Mood Disorders: CRY1 and NPAS2 are Associated with Unipolar Major Depression and CLOCK and VIP with Bipolar Disorder. Neuropsychopharmacology, 2010, 35, 1279-1289.	2.8	310
156	Response to Methadone Maintenance Treatment is Associated with the MYOCD and GRM6 Genes. Molecular Diagnosis and Therapy, 2010, 14, 171-178.	1.6	28
157	Genetic Structure of Europeans: A View from the Northâ€“East. PLoS ONE, 2009, 4, e5472.	1.1	279
158	Aneuploidy: From a Physiological Mechanism of Variance to Down Syndrome. Physiological Reviews, 2009, 89, 887-920.	13.1	106
159	GSTM1 polymorphisms modify the effect of maternal smoking during pregnancy on cognitive functioning in preschoolers. International Journal of Epidemiology, 2009, 38, 690-697.	0.9	26
160	Positive Selection and Gene Conversion Drive the Evolution of a Brain-Expressed snoRNAs Cluster. Molecular Biology and Evolution, 2009, 26, 2563-2571.	3.5	4
161	Traffic-Related Air Pollution, Oxidative Stress Genes, and Asthma (ECHRS). Environmental Health Perspectives, 2009, 117, 1919-1924.	2.8	78
162	Association between leptin receptor (LEPR) and brain-derived neurotrophic factor (BDNF) gene variants and obesity: a case-control study. Nutritional Neuroscience, 2009, 12, 183-188.	1.5	12

#	Article	IF	Citations
163	Joint effect of obesity and TNFA variability on asthma: two international cohort studies. European Respiratory Journal, 2009, 33, 1003-1009.	3.1	43
164	Absence of cytogenetic effects in children and adults with attention-deficit/hyperactivity disorder treated with methylphenidate. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2009, 666, 44-49.	0.4	18
165	Accounting for uncertainty when assessing association between copy number and disease: a latent class model. BMC Bioinformatics, 2009, 10, 172.	1.2	22
166	Identification of new putative susceptibility genes for several psychiatric disorders by association analysis of regulatory and nonâ€synonymous SNPs of 306 genes involved in neurotransmission and neurodevelopment. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2009, 150B, 808-816.	1.1	98
167	Allele variants in functional MicroRNA target sites of the neurotrophin-3 receptor gene (NTRK3) as susceptibility factors for anxiety disorders. Human Mutation, 2009, 30, 1062-1071.	1.1	86
168	A pooling-based genome-wide analysis identifies new potential candidate genes for atopy in the European Community Respiratory Health Survey (ECRHS). BMC Medical Genetics, 2009, 10, 128.	2.1	43
169	BAC array CGH in patients with Velocardiofacial syndrome-like features reveals genomic aberrations on chromosome region 1q21.1. BMC Medical Genetics, 2009, 10, 144.	2.1	33
170	Exploration of 19 serotoninergic candidate genes in adults and children with attention-deficit/hyperactivity disorder identifies association for 5HT2A, DDC and MAOB. Molecular Psychiatry, 2009, 14, 71-85.	4.1	141
171	Deletion of the late cornified envelope LCE3B and LCE3C genes as a susceptibility factor for psoriasis. Nature Genetics, 2009, 41, 211-215.	9.4	482
172	Are MYO1C and MYO1F associated with hearing loss?. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2009, 1792, 27-32.	1.8	28
173	A Brain-Derived Neurotrophic Factor Haplotype Is Associated with Therapeutic Response in Obsessive-Compulsive Disorder. Biological Psychiatry, 2009, 66, 674-680.	0.7	34
174	Case-Control Study of Six Genes Asymmetrically Expressed in the Two Cerebral Hemispheres: Association of BAIAP2 with Attention-Deficit/Hyperactivity Disorder. Biological Psychiatry, 2009, 66, 926-934.	0.7	59
175	Suicide attempts in bulimia nervosa: Personality and psychopathological correlates. European Psychiatry, 2009, 24, 91-97.	0.1	59
176	Functional variants of the serotonin receptor type 3A and B gene are associated with eating disorders. Pharmacogenetics and Genomics, 2009, 19, 790-799.	0.7	35
177	Uniparental disomies, homozygous deletions, amplifications, and target genes in mantle cell lymphoma revealed by integrative high-resolution whole-genome profiling. Blood, 2009, 113, 3059-3069.	0.6	162
178	Identification of Copy Number Variants Defining Genomic Differences among Major Human Groups. PLoS ONE, 2009, 4, e7230.	1.1	32
179	Nucleotide, Cytogenetic and Expression Impact of the Human Chromosome 8p23.1 Inversion Polymorphism. PLoS ONE, 2009, 4, e8269.	1.1	18
180	Developmental analysis of Lingoâ€1/Lern1 protein expression in the mouse brain: Interaction of its intracellular domain with Myt1l. Developmental Neurobiology, 2008, 68, 521-541.	1.5	44

#	Article	IF	Citations
181	An integrated genetic and functional analysis of the role of type II transmembrane serine proteases (TMPRSSs) in hearing loss. Human Mutation, 2008, 29, 130-141.	1.1	70
182	Detection of unrecognized low-level mtDNA heteroplasmy may explain the variable phenotypic expressivity of apparently homoplasmic mtDNA mutations. Human Mutation, 2008, 29, 248-257.	1.1	44
183	Maximizing association statistics over genetic models. Genetic Epidemiology, 2008, 32, 246-254.	0.6	101
184	DYRK1A-Dosage Imbalance Perturbs NRSF/REST Levels, Deregulating Pluripotency and Embryonic Stem Cell Fate in Down Syndrome. American Journal of Human Genetics, 2008, 83, 388-400.	2.6	139
185	Targeting Dyrk1A with AAVshRNA Attenuates Motor Alterations in TgDyrk1A, a Mouse Model of Down Syndrome. American Journal of Human Genetics, 2008, 83, 479-488.	2.6	60
186	BDNF variability in opioid addicts and response to methadone treatment: preliminary findings. Genes, Brain and Behavior, 2008, 7, 515-522.	1.1	53
187	Genetic susceptibility to obsessiveâ€compulsive hoarding: the contribution of neurotrophic tyrosine kinase receptor type 3 gene¹. Genes, Brain and Behavior, 2008, 7, 778-785.	1.1	43
188	Probe-specific mixed-model approach to detect copy number differences using multiplex ligation-dependent probe amplification (MLPA). BMC Bioinformatics, 2008, 9, 261.	1.2	16
189	Genetic and genomic analysis modeling of germline c-MYC overexpression and cancer susceptibility. BMC Genomics, 2008, 9, 12.	1.2	27
190	ProSeeK: A web server for MLPA probe design. BMC Genomics, 2008, 9, 573.	1.2	6
191	Contribution of the serotoninergic system to anxious and depressive traits that may be partially responsible for the phenotypical variability of bulimia nervosa. Journal of Psychiatric Research, 2008, 42, 50-57.	1.5	38
192	Failure to detect the 22q11.2 duplication syndrome rearrangement among patients with schizophrenia. Behavioral and Brain Functions, 2008, 4, 10.	1.4	20
193	Copy number variants: a common mechanism in complex diseases. European Journal of Cancer, Supplement, 2008, 6, 187.	2.2	0
194	Extensive Genotyping of the BDNF and NTRK2 Genes Define Protective Haplotypes Against Obsessive-Compulsive Disorder. Biological Psychiatry, 2008, 63, 619-628.	0.7	66
195	Association Study of 10 Genes Encoding Neurotrophic Factors and Their Receptors in Adult and Child Attention-Deficit/Hyperactivity Disorder. Biological Psychiatry, 2008, 63, 935-945.	0.7	93
196	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. New England Journal of Medicine, 2008, 359, 1685-1699.	13.9	663
197	Efficient and specific transduction of cochlear supporting cells by adeno-associated virus serotype 5. Neuroscience Letters, 2008, 442, 134-139.	1.0	31
198	Analysis of the multi-copy gene family FAM90A as a copy number variant in different ethnic backgrounds. Gene, 2008, 420, 113-117.	1.0	3

#	Article	IF	Citations
199	Suicide attempts in bulimia nervosa: Personality, Psychopathological and Genetic correlates. European Psychiatry, 2008, 23, S181.	0.1	2
200	Association of NTRK3 and its interaction with NGF suggest an altered cross-regulation of the neurotrophin signaling pathway in eating disorders. European Psychiatry, 2008, 23, S182.	0.1	2
201	The emerging role of structural variations in common disorders: initial findings and discovery challenges. Cytogenetic and Genome Research, 2008, 123, 108-117.	0.6	5
202	Association of NTRK3 and its interaction with NGF suggest an altered cross-regulation of the neurotrophin signaling pathway in eating disorders. Human Molecular Genetics, 2008, 17, 1234-1244.	1.4	50
203	Origin of Primate Orphan Genes: A Comparative Genomics Approach. Molecular Biology and Evolution, 2008, 26, 603-612.	3.5	201
204	TNFA -308G>A in two international population-based cohorts and risk of asthma. European Respiratory Journal, 2008, 32, 350-361.	3.1	28
205	A brain-derived neurotrophic factor (BDNF) haplotype is associated with antidepressant treatment outcome in mood disorders. Pharmacogenomics Journal, 2008, 8, 101-112.	0.9	76
206	Hypothalamus transcriptome profile suggests an anorexia-cachexia syndrome in the anx/anx mouse model. Physiological Genomics, 2008, 35, 341-350.	1.0	22
207	Influence of Glutathione S -Transferase Polymorphisms on Cognitive Functioning Effects Induced by p,p â€²-DDT among Preschoolers. Environmental Health Perspectives, 2008, 116, 1581-1585.	2.8	32
208	SNPassoc: an R package to perform whole genome association studies. Bioinformatics, 2007, 23, 654-655.	1.8	607
209	Copy Number Variants and Common Disorders: Filling the Gaps and Exploring Complexity in Genome-Wide Association Studies. PLoS Genetics, 2007, 3, e190.	1.5	178
210	RCAN1 (DSCR1) increases neuronal susceptibility to oxidative stress: a potential pathogenic process in neurodegeneration. Human Molecular Genetics, 2007, 16, 1039-1050.	1.4	84
211	Age and origin of major Smith-Lemli-Opitz syndrome (SLOS) mutations in European populations. Journal of Medical Genetics, 2007, 45, 200-209.	1.5	47
212	Blood Levels of Brain-Derived Neurotrophic Factor Correlate with Several Psychopathological Symptoms in Anorexia Nervosa Patients. Neuropsychobiology, 2007, 56, 185-190.	0.9	28
213	Renaming the DSCR1 / Adapt78 gene family as RCAN : regulators of calcineurin. FASEB Journal, 2007, 21, 3023-3028.	0.2	157
214	Characterization and evolution of the novel gene family FAM90A in primates originated by multiple duplication and rearrangement events. Human Molecular Genetics, 2007, 16, 2572-2582.	1.4	24
215	Brain-Derived Neurotrophic Factor Val66Met and Psychiatric Disorders: Meta-Analysis of Case-Control Studies Confirm Association to Substance-Related Disorders, Eating Disorders, and Schizophrenia. Biological Psychiatry, 2007, 61, 911-922.	0.7	381
216	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

#	Article	IF	Citations
217	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
218	MRPS18CP2 alleles and DEFA3absence as putative chromosome 8p23.1 modifiers of hearing loss due to mtDNA mutation A1555G in the 12S rRNA gene. BMC Medical Genetics, 2007, 8, 81.	2.1	10
219	Copy number variants and genetic traits: closer to the resolution of phenotypic to genotypic variability. Nature Reviews Genetics, 2007, 8, 639-646.	7.7	391
220	Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. Nature, 2007, 447, 799-816.	13.7	4,709
221	Cystic fibrosis in a southern Brazilian population: characteristics of 90% of the alleles. Clinical Genetics, 2007, 72, 218-223.	1.0	13
222	Spectrum of Mutations in the CFTR Gene in Cystic Fibrosis Patients of Spanish Ancestry. Annals of Human Genetics, 2007, 71, 194-201.	0.3	53
223	Altered brain-derived neurotrophic factor blood levels and gene variability are associated with anorexia and bulimia. Genes, Brain and Behavior, 2007, 6, 706-716.	1.1	73
224	Candidate genes for panic disorder: insight from human and mouse genetic studies. Genes, Brain and Behavior, 2007, 6, 2-23.	1.1	25
225	Inter-population variability of DEFA3 gene absence: correlation with haplotype structure and population variability. BMC Genomics, 2007, 8, 14.	1.2	20
226	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
227	Differential responses to anxiogenic drugs in a mouse model of panic disorder as revealed by Fos immunocytochemistry in specific areas of the fear circuitry. Amino Acids, 2007, 33, 677-688.	1.2	13
228	Mutation in TRMU Related to Transfer RNA Modification Modulates the Phenotypic Expression of the Deafness-Associated Mitochondrial 12S Ribosomal RNA Mutations. American Journal of Human Genetics, 2006, 79, 291-302.	2.6	212
229	Mitochondrial 12S rRNA gene mutations affect RNA secondary structure and lead to variable penetrance in hearing impairment. Biochemical and Biophysical Research Communications, 2006, 341, 950-957.	1.0	61
230	Human TRMU encoding the mitochondrial 5-methylaminomethyl-2-thiouridylate-methyltransferase is a putative nuclear modifier gene for the phenotypic expression of the deafness-associated 12S rRNA mutations. Biochemical and Biophysical Research Communications, 2006, 342, 1130-1136.	1.0	31
231	Cochlear alterations in deaf and unaffected subjects carrying the deafness-associated A1555G mutation in the mitochondrial 12S rRNA gene. Biochemical and Biophysical Research Communications, 2006, 344, 511-516.	1.0	36
232	Reply to correspondence by Abreu-Silva et al. regarding Ballana et al.: Mutation T1291C in the mitochondrial 12S rRNA gene involved in deafness in a Cuban family belongs to the macrohaplogroup L1 of African origin. Biochemical and Biophysical Research Communications, 2006, 346, 619-620.	1.0	1
233	Cooperation to amplify gene-dosage-imbalance effects. Trends in Molecular Medicine, 2006, 12, 451-454.	3.5	17
234	A novel G21R mutation of the GJB2 gene causes autosomal dominant non-syndromic congenital deafness in a Cuban family. Genetics and Molecular Biology, 2006, 29, 443-445.	0.6	4

#	Article	IF	Citations
235	Caseâ€“control and combined family trios analysis of three polymorphisms in the ghrelin gene in European patients with anorexia and bulimia nervosa. Psychiatric Genetics, 2006, 16, 51-52.	0.6	40
236	Genome assembly comparison identifies structural variants in the human genome. Nature Genetics, 2006, 38, 1413-1418.	9.4	150
237	Global variation in copy number in the human genome. Nature, 2006, 444, 444-454.	13.7	3,831
238	Identification of large-scale human-specific copy number differences by inter-species array comparative genomic hybridization. Human Genetics, 2006, 119, 185-198.	1.8	35
239	Complex patterns of copy number variation at sites of segmental duplications: an important category of structural variation in the human genome. Human Genetics, 2006, 120, 270-284.	1.8	68
240	Transgenic mice overexpressing the full-length neurotrophin receptor TrkC exhibit increased catecholaminergic neuron density in specific brain areas and increased anxiety-like behavior and panic reaction. Neurobiology of Disease, 2006, 24, 403-418.	2.1	50
241	A recessive Mendelian model to predict carrier probabilities of DFNB1 for nonsyndromic deafness. Human Mutation, 2006, 27, 1135-1142.	1.1	4
242	Association of BDNF with restricting anorexia nervosa and minimum body mass index: a family-based association study of eight European populations. European Journal of Human Genetics, 2005, 13, 428-434.	1.4	131
243	Contribution of NTRK2 to the genetic susceptibility to anorexia nervosa, Harm avoidance and minimum body mass index. Molecular Psychiatry, 2005, 10, 851-860.	4.1	48
244	Different mechanisms preclude mutant CLDN14 proteins from forming tight junctions in vitro. Human Mutation, 2005, 25, 543-549.	1.1	45
245	Espin gene (ESPN) mutations associated with autosomal dominant hearing loss cause defects in microvillar elongation or organisation. Journal of Medical Genetics, 2005, 43, 157-161.	1.5	59
246	Murine segmental duplications are hot spots for chromosome and gene evolution. Genomics, 2005, 86, 692-700.	1.3	28
247	Genome Variation Influencing Gene Copy Number and Disease. Drugs and the Pharmaceutical Sciences, 2005, , 611-637.	0.1	0
248	Association of BDNF with anorexia, bulimia and age of onset of weight loss in six European populations. Human Molecular Genetics, 2004, 13, 1205-1212.	1.4	193
249	Bronchiectasis in adult patients: an expression of heterozygosity for CFTR gene mutations?. Clinical Genetics, 2004, 65, 490-495.	1.0	81
250	Small marker chromosomes in two patients with segmental aneusomy for proximal 17p. Human Genetics, 2004, 115, 1-7.	1.8	24
251	Combined family trio and case-control analysis of the COMT Val158Met polymorphism in European patients with anorexia nervosa. American Journal of Medical Genetics Part A, 2004, 124B, 68-72.	2.4	43
252	Characterization of the segmental duplication LCR7-20 in the human genome. Genomics, 2004, 83, 262-269.	1.3	7

#	Article	IF	Citations
253	Human mitochondrial transcription factor B1 as a modifier gene for hearing loss associated with the mitochondrial A1555G mutation. Molecular Genetics and Metabolism, 2004, 82, 27-32.	0.5	75
254	Phenotype of non-syndromic deafness associated with the mitochondrial A1555G mutation is modulated by mitochondrial RNA modifying enzymes MTO1 and GTPBP3. Molecular Genetics and Metabolism, 2004, 83, 199-206.	0.5	64
255	On dendrites in Down syndrome and DS murine models: a spiny way to learn. Progress in Neurobiology, 2004, 74, 111-126.	2.8	124
256	Motor phenotypic alterations in TgDyrk1a transgenic mice implicate DYRK1A in Down syndrome motor dysfunction. Neurobiology of Disease, 2004, 15, 132-142.	2.1	75
257	Variation in a Repeat Sequence Determines Whether a Common Variant of the Cystic Fibrosis Transmembrane Conductance Regulator Gene Is Pathogenic or Benign. American Journal of Human Genetics, 2004, 74, 176-179.	2.6	227
258	Nonmuscle Myosin Heavy-Chain Gene MYH14 Is Expressed in Cochlea and Mutated in Patients Affected by Autosomal Dominant Hearing Impairment (DFNA4). American Journal of Human Genetics, 2004, 74, 770-776.	2.6	150
259	Different CFTR Mutational Spectrum in Alcoholic and Idiopathic Chronic Pancreatitis?. Pancreas, 2004, 28, 374-379.	0.5	34
260	Haploinsufficiency of Dyrk1A in Mice Leads to Specific Alterations in the Development and Regulation of Motor Activity.. Behavioral Neuroscience, 2004, 118, 815-821.	0.6	61
261	Identification and characterization of a novel splice-site mutation in a patient with Wiskott-Aldrich syndrome. Journal of Human Genetics, 2003, 48, 590-593.	1.1	4
262	Dyrk1A expression pattern supports specific roles of this kinase in the adult central nervous system. Brain Research, 2003, 964, 250-263.	1.1	125
263	The molecular basis of glutamate formiminotransferase deficiency. Human Mutation, 2003, 22, 67-73.	1.1	57
264	Suitability of oligonucleotide-mediated cystic fibrosis gene repair in airway epithelial cells. Journal of Gene Medicine, 2003, 5, 625-639.	1.4	5
265	Human nuclear transcription factor geneCREM: Genomic organization, mutation screening, and association analysis in panic disorder. , 2003, 117B, 70-78.		23
266	Met66 in the brain-derived neurotrophic factor (BDNF) precursor is associated with anorexia nervosa restrictive type. Molecular Psychiatry, 2003, 8, 745-751.	4.1	176
267	LRRN6A/LERN1 (leucine-rich repeat neuronal protein 1), a novel gene with enriched expression in limbic system and neocortex. European Journal of Neuroscience, 2003, 18, 3167-3182.	1.2	76
268	Prevalence and Evolutionary Origins of the del(GJB6-D13S1830) Mutation in the DFNB1 Locus in Hearing-Impaired Subjects: a Multicenter Study. American Journal of Human Genetics, 2003, 73, 1452-1458.	2.6	269
269	Wiskottâ€“Aldrich syndrome in a female with skewed X-chromosome inactivation. Blood Cells, Molecules, and Diseases, 2003, 31, 332-337.	0.6	30
270	Enrichment of segmental duplications in regions of breaks of synteny between the human and mouse genomes suggest their involvement in evolutionary rearrangements. Human Molecular Genetics, 2003, 12, 2201-2208.	1.4	121

#	Article	IF	Citations
271	Human Chromosome 7: DNA Sequence and Biology. Science, 2003, 300, 767-772.	6.0	185
272	Genome-wide detection of segmental duplications and potential assembly errors in the human genome sequence. Genome Biology, 2003, 4, R25.	13.9	207
273	DYRK1A accumulates in splicing speckles through a novel targeting signal and induces speckle disassembly. Journal of Cell Science, 2003, 116, 3099-3107.	1.2	137
274	Alterations of Neocortical Pyramidal Cell Phenotype in the Ts65Dn Mouse Model of Down Syndrome: Effects of Environmental Enrichment. Cerebral Cortex, 2003, 13, 758-764.	1.6	136
275	Phosphorylation of calcipressin 1 increases its ability to inhibit calcineurin and decreases calcipressin half-life. Biochemical Journal, 2003, 374, 567-575.	1.7	94
276	Recurrent mutations in the NF1 gene are common among neurofibromatosis type 1 patients. Journal of Medical Genetics, 2003, 40, 82e-82.	1.5	154
277	Genomic inversions of human chromosome 15q11-q13 in mothers of Angelman syndrome patients with class II (BP2/3) deletions. Human Molecular Genetics, 2003, 12, 849-858.	1.4	131
278	Coding haplotype analysis supports HCR as the putative susceptibility gene for psoriasis at the MHC PSORS1 locus. Human Molecular Genetics, 2002, 11, 589-597.	1.4	131
279	Human chromosome 15q11-q14 regions of rearrangements contain clusters of LCR15 duplicons. European Journal of Human Genetics, 2002, 10, 26-35.	1.4	92
280	Dyrk1A Haploinsufficiency Affects Viability and Causes Developmental Delay and Abnormal Brain Morphology in Mice. Molecular and Cellular Biology, 2002, 22, 6636-6647.	1.1	306
281	Chromosomal regions containing high-density and ambiguously mapped putative single nucleotide polymorphisms (SNPs) correlate with segmental duplications in the human genome. Human Molecular Genetics, 2002, 11, 1987-1995.	1.4	80
282	Expression profiles of the connexin genes, Gjb1 and Gjb3, in the developing mouse cochlea. Mechanisms of Development, 2002, 119, S111-S115.	1.7	18
283	Connexin mutations in hearing loss, dermatological and neurological disorders. Trends in Molecular Medicine, 2002, 8, 205-212.	3.5	75
284	Neurobehavioral development of two mouse lines commonly used in transgenic studies. Pharmacology Biochemistry and Behavior, 2002, 73, 19-25.	1.3	46
285	Intratumoral activation of cyclophosphamide by retroviral transfer of the cytochrome P450 2B1 in a pancreatic tumor model. Combination with the HSVtk/GCV system. Journal of Gene Medicine, 2002, 4, 141-149.	1.4	19
286	Non-viral vector-mediated uptake, distribution, and stability of chimeraplasts in human airway epithelial cells. Journal of Gene Medicine, 2002, 4, 308-322.	1.4	15
287	A common frameshift mutation and other variants in GJB4 (connexin 30.3): Analysis of hearing impairment families. Human Mutation, 2002, 19, 458-458.	1.1	16
288	Mutations in the TMPRSS3 gene are a rare cause of childhood nonsyndromic deafness in Caucasian patients. Journal of Molecular Medicine, 2002, 80, 124-131.	1.7	65

#	Article	IF	Citations
289	Cloning of S4D-SRCRB, a new soluble member of the group B scavenger receptor cysteine-rich family (SRCR-SF) mapping to human Chromosome 7q11.23. Immunogenetics, 2002, 54, 621-634.	1.2	13
290	Human connexin26 (GJB2) deafness mutations affect the function of gap junction channels at different levels of protein expression.. Human Genetics, 2002, 111, 190-197.	1.8	86
291	5â€² UTR-region SNP in the NTRK3 gene is associated with panic disorder. Molecular Psychiatry, 2002, 7, 928-930.	4.1	28
292	Quantitative assessment of chimeraplast stability in biological fluids by polyacrylamide gel electrophoresis and laser-assisted fluorescence analysis. Pharmaceutical Research, 2002, 19, 914-918.	1.7	1
293	The 5-HT2A â^'1438G/A polymorphism in anorexia nervosa: a combined analysis of 316 trios from six European centres. Molecular Psychiatry, 2002, 7, 90-94.	4.1	6
294	Presence of a Major WFS1 Mutation in Spanish Wolfram Syndrome Pedigrees. Molecular Genetics and Metabolism, 2001, 72, 72-81.	0.5	67
295	Dscr1, a novel endogenous inhibitor of calcineurin signaling, is expressed in the primitive ventricle of the heart and during neurogenesis. Mechanisms of Development, 2001, 101, 289-292.	1.7	58
296	MYO6, the Human Homologue of the Gene Responsible for Deafness in Snellâ€™s Waltzer Mice, Is Mutated in Autosomal Dominant Nonsyndromic Hearing Loss. American Journal of Human Genetics, 2001, 69, 635-640.	2.6	212
297	Identification and characterization of BTBD1 , a novel BTB domain containing gene on human chromosome 15q24. Gene, 2001, 262, 275-281.	1.0	15
298	PACSIN 3 is a novel SH3 domain cytoplasmic adapter protein of the pacsin-syndapin-FAP52 gene family. Gene, 2001, 262, 199-205.	1.0	25
299	PALML, a novel paralemmin-related gene mapping on human chromosome 1p21. Gene, 2001, 278, 33-40.	1.0	10
300	Murine models for Down syndrome. Physiology and Behavior, 2001, 73, 859-871.	1.0	62
301	A Polymorphic Genomic Duplication on Human Chromosome 15 Is a Susceptibility Factor for Panic and Phobic Disorders. Cell, 2001, 106, 367-379.	13.5	219
302	Connexin 31 (GJB3) is expressed in the peripheral and auditory nerves and causes neuropathy and hearing impairment. Human Molecular Genetics, 2001, 10, 947-952.	1.4	109
303	Cloning, Mapping and Expression Analysis of C15 or f4, a Novel Human Gene with Homology to the Yeast Mitochondrial Ribosomal Protein Yml30 Gene. DNA Sequence, 2001, 12, 91-96.	0.7	2
304	Large de novo deletion in chromosome 12 affecting the PAH, IGF1, ASCL1, and TRA1 genes. Journal of Molecular Medicine, 2001, 78, 721-724.	1.7	6
305	Genomic organization, chromosomal localization, alternative splicing, and isoforms of the human synaptosome-associated protein-23 gene implicated in vesicle-membrane fusion processes. Human Genetics, 2001, 108, 211-215.	1.8	4
306	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

#	Article	IF	Citations
307	Retrovirus-mediated transfer of the herpes simplex virus thymidine kinase and connexin26 genes in pancreatic cells results in variable efficiency on the bystander killing: Implications for gene therapy. International Journal of Cancer, 2001, 94, 81-88.	2.3	32
308	Identification of WASP mutations in 14 Spanish families with Wiskott-Aldrich syndrome. American Journal of Medical Genetics Part A, 2001, 100, 116-121.	2.4	22
309	Somatic NF1 mutational spectrum in benign neurofibromas: mRNA splice defects are common among point mutations. Human Genetics, 2001, 108, 416-429.	1.8	72
310	Mitotic recombination effects homozygosity for NF1 germline mutations in neurofibromas. Nature Genetics, 2001, 28, 294-296.	9.4	72
311	ATB0/SLC1A5 gene. Fine localisation and exclusion of association with the intestinal phenotype of cystic fibrosis. European Journal of Human Genetics, 2001, 9, 860-866.	1.4	10
312	PTOV1, a novel protein overexpressed in prostate cancer containing a new class of protein homology blocks. Oncogene, 2001, 20, 1455-1464.	2.6	61
313	Identification and characterization of UBXD1, a novel UBX domain-containing gene on human chromosome 19p13, and its mouse ortholog. Biochimica Et Biophysica Acta Gene Regulatory Mechanisms, 2001, 1517, 298-301.	2.4	14
314	Characterization of human FSD1, a novel brain specific gene on chromosome 19 with paralogy to 9q31. Biochimica Et Biophysica Acta Gene Regulatory Mechanisms, 2001, 1518, 200-203.	2.4	8
315	The human intersectin genes and their spliced variants are differentially expressed. Biochimica Et Biophysica Acta Gene Regulatory Mechanisms, 2001, 1521, 1-11.	2.4	56
316	Functional analysis of mutations in SLC7A9, and genotypeâ€“phenotype correlation in non-Type I cystinuria. Human Molecular Genetics, 2001, 10, 305-316.	1.4	125
317	Adenosine Triphosphate-Binding Cassette Superfamily Transporter Gene Expression in Severe Male Infertility1. Biology of Reproduction, 2001, 65, 394-400.	1.2	20
318	Cystic fibrosis transmembrane regulator (CFTR) Delta F508 mutation and 5T allele in patients with chronic pancreatitis and exocrine pancreatic cancer. Gut, 2001, 48, 70-74.	6.1	107
319	Modifier locus for mitochondrial DNA disease: Linkage and linkage disequilibrium mapping of a nuclear modifier gene for maternally inherited deafness. Genetics in Medicine, 2001, 3, 177-180.	1.1	56
320	Identification of C15orf5, a Heart-Enriched Transcript on Chromosome 15q23-q24. DNA Sequence, 2001, 12, 67-69.	0.7	1
321	Neurodevelopmental delay, motor abnormalities and cognitive deficits in transgenic mice overexpressing Dyrk1A (minibrain), a murine model of Down's syndrome. Human Molecular Genetics, 2001, 10, 1915-1923.	1.4	357
322	Additional Complexity on Human Chromosome 15q: Identification of a Set of Newly Recognized Duplicons (LCR15) on 15q11-q13, 15q24, and 15q26. Genome Research, 2001, 11, 98-111.	2.4	60
323	Clinical and cytogenetic characterisation of a patient with Down syndrome resulting from a 21q22.1->qter duplication. Journal of Medical Genetics, 2001, 38, 73-76.	1.5	7
324	Cystic fibrosis patients with the 3272-26A>G splicing mutation have milder disease than F508del homozygotes: a large European study. Journal of Medical Genetics, 2001, 38, 777-783.	1.5	30

#	Article	IF	Citations
325	Optimization of a simple and rapid single-strand conformation analysis for detection of mutations in thePROS1 gene: Identification of seven novel mutations and three novel, apparently neutral, variants. , 2000, 15, 463-473.		22
326	Identification of seven novel SNPS (five nucleotide and two amino acid substitutions) in the connexin31 (GJB3) gene. , 2000, 15, 481-482.		17
327	Molecular genetics of hearing impairment due to mutations in gap junction genes encoding beta connexins. Human Mutation, 2000, 16, 190-202.	1.1	197
328	High carrier frequency of the 35delG deafness mutation in European populations. European Journal of Human Genetics, 2000, 8, 19-23.	1.4	363
329	Isolation and characterisation of a novel human gene (C9orf11) on chromosome 9p21, a region frequently deleted in human cancer. Biochimica Et Biophysica Acta Gene Regulatory Mechanisms, 2000, 1517, 128-134.	2.4	6
330	Association study of proposed candidate genes/regions in a population of Spanish asthmatics. European Journal of Epidemiology, 2000, 16, 745-750.	2.5	8
331	Characterization of a novel 21-kb deletion, CFTRdele2,3(21 kb), in the CFTR gene: a cystic fibrosis mutation of Slavic origin common in Central and East Europe. Human Genetics, 2000, 106, 259-268.	1.8	129
332	A new approach for identifying non-pathogenic mutations. An analysis of the cystic fibrosis transmembrane regulator gene in normal individuals. Human Genetics, 2000, 106, 172-178.	1.8	39
333	Homozygosity for the Protein S Heerlen Allele Is Associated with Type I PS Deficiency in a Thrombophilic Pedigree with Multiple Risk Factors. Thrombosis and Haemostasis, 2000, 83, 102-106.	1.8	21
334	DSCR1, overexpressed in Down syndrome, is an inhibitor of calcineurin-mediated signaling pathways. Human Molecular Genetics, 2000, 9, 1681-1690.	1.4	426
335	Cold shock induces the insertion of a cryptic exon in the neurofibromatosis type 1 (NF1) mRNA. Nucleic Acids Research, 2000, 28, 1307-1312.	6.5	48
336	Identification and Expression Analysis of C3orf1, a Novel Human Gene Homologous to the Drosophila RP140â€"Upstream Gene. DNA Sequence, 2000, 11, 335-338.	0.7	8
337	Cloning of the novel gene TM6SF1 reveals conservation of clusters of paralogous genes between human chromosomes 15q24â†'q26 and 19p13.3â†'p12. Cytogenetic and Genome Research, 2000, 90, 255-260.	0.6	26
338	Cloning, mapping and expression analysis of VPS33B, the human orthologue of rat Vps33b. Cytogenetic and Genome Research, 2000, 89, 92-95.	0.6	20
339	Mutations affecting mRNA splicing are the most common molecular defects in patients with neurofibromatosis type 1. Human Molecular Genetics, 2000, 9, 237-247.	1.4	319
340	Association Study of the Chromosomal Region Containing the FCER2 Gene Suggests It Has a Regulatory Role in Atopic Disorders. American Journal of Respiratory and Critical Care Medicine, 2000, 161, 700-706.	2.5	40
341	Hereditary neuropathy with liability to pressure palsies: two cases with a reciprocal translocation t(16;17)(q12;p11.2) interrupting the PMP22 gene. Journal of Medical Genetics, 2000, 37, 396-398.	1.5	8
342	Schwann cells harbor the somatic NF1 mutation in neurofibromas: evidence of two different Schwann cell subpopulations. Human Molecular Genetics, 2000, 9, 3055-3064.	1.4	232

#	Article	IF	Citations
343	Identification and expression analysis of C15orf3, a novel gene on chromosome 15q21.1â†'q21.2. Cytogenetic and Genome Research, 2000, 88, 330-332.	0.6	0
344	Genes as causes: scientific fact or simplistic thinking?. Journal of Epidemiology and Community Health, 2000, 54, 559-559.	2.0	5
345	Genotype-phenotype correlation in three homozygotes and nine compound heterozygotes for the cystic fibrosis mutation 2183AAright-arrowG shows a severe phenotype. Journal of Medical Genetics, 2000, 37, 307-309.	1.5	6
346	HMG20A and HMG20B map to human chromosomes 15q24 and 19p13.3 and constitute a distinct class of HMG-box genes with ubiquitous expression. Cytogenetic and Genome Research, 2000, 88, 62-67.	0.6	36
347	Rare variants in the promoter of the fragile X syndrome gene (FMR1). Molecular and Cellular Probes, 2000, 14, 115-119.	0.9	15
348	Mutations in the Mitochondrial tRNA Ser(UCN) and in the GJB2 (Connexin 26) Gene Are Not Modifiers of the Age at Onset or Severity of Hearing Loss in Spanish Patients with the 12S rRNA A1555G Mutation. American Journal of Human Genetics, 2000, 66, 1465-1467.	2.6	16
349	Candidate Locus for a Nuclear Modifier Gene for Maternally Inherited Deafness. American Journal of Human Genetics, 2000, 66, 1905-1910.	2.6	103
350	Intersectin 2, a new multimodular protein involved in clathrin-mediated endocytosis. FEBS Letters, 2000, 478, 43-51.	1.3	83
351	A new aspartyl protease on 21q22.3, BACE2, is highly similar to Alzheimerâ€™s amyloid precursor protein Î²-secretase. Cytogenetic and Genome Research, 2000, 89, 177-184.	0.6	81
352	Heterogeneity for mutations in the CFTR gene and clinical correlations in patients with congenital absence of the vas deferens. Human Reproduction, 2000, 15, 1476-1483.	0.4	131
353	Cloning and characterization of human FTCD on 21q22.3, a candidate gene for glutamate formiminotransferase deficiency. Cytogenetic and Genome Research, 2000, 88, 43-49.	0.6	32
354	The Mouse Brain Transcriptome by SAGE: Differences in Gene Expression between P30 Brains of the Partial Trisomy 16 Mouse Model of Down Syndrome (Ts65Dn) and Normals. Genome Research, 2000, 10, 2006-2021.	2.4	13
355	A High-Resolution Physical Map of Human Chromosome 21p Using Yeast Artificial Chromosomes. Genome Research, 1999, 9, 1059-1073.	2.4	23
356	Cloning, expression, and mapping of PDCD9, the human homolog of Gallus gallus pro-apoptotic protein p52. Cytogenetic and Genome Research, 1999, 87, 85-88.	0.6	18
357	The human HERC3 gene maps<footref rid="foot01">¹</footref> to chromosome 4q21 by fluorescence in situ hybridization. Cytogenetic and Genome Research, 1999, 87, 263-264.	0.6	5
358	Mutational analysis within the 3â€² region of the PKD1 gene. Kidney International, 1999, 55, 1225-1233.	2.6	41
359	Mutations in GJB6 cause nonsyndromic autosomal dominant deafness at DFNA3 locus. Nature Genetics, 1999, 23, 16-18.	9.4	345
360	Non-type I cystinuria caused by mutations in SLC7A9, encoding a subunit (bo,+AT) of rBAT. Nature Genetics, 1999, 23, 52-57.	9.4	280

#	Article	IF	Citations
361	Detection of a cystic fibrosis modifier locus for meconium ileus on human chromosome 19q13. Nature Genetics, 1999, 22, 128-129.	9.4	216
362	Alu-splice cloning of human Intersectin (ITSN), a putative multivalent binding protein expressed in proliferating and differentiating neurons and overexpressed in Down syndrome. European Journal of Human Genetics, 1999, 7, 704-712.	1.4	74
363	Spinocerebellar ataxias in Spanish patients: genetic analysis of familial and sporadic cases. Human Genetics, 1999, 104, 516-522.	1.8	140
364	Protein S gene analysis reveals the presence of a cosegregating mutation in most pedigrees with type I but not type III PS deficiency. , 1999, 14, 30-39.		28
365	Molecular studies in 20 submicroscopic neurofibromatosis type 1 gene deletions. , 1999, 14, 387-393.		62
366	Missense mutations in the cystic fibrosis gene in adult patients with asthma. , 1999, 14, 510-519.		51
367	Splice-site mutation in thePDS gene may result in intrafamilial variability for deafness in Pendred syndrome. , 1999, 14, 520-526.		37
368	Prenatal diagnosis of sporadic neurofibromatosis type 1 (NF1) by RNA and DNA analysis of a splicing mutation. , 1999, 19, 739-742.		17
369	Anticipation is not associated with CAG repeat expansion in parent-offspring pairs of patients affected with schizophrenia. , 1999, 88, 50-56.		13
370	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
371	The A1555G Mutation in the 12S rRNA Gene of Human mtDNA: Recurrent Origins and Founder Events in Families Affected by Sensorineural Deafness. American Journal of Human Genetics, 1999, 65, 1349-1358.	2.6	111
372	Cloning, expression and chromosomal localization of a human testis 6-phosphofructo-2-kinase/fructose-2,6-bisphosphatase gene. Gene, 1999, 229, 83-89.	1.0	38
373	Cloning of the human phospholipase A2 activating protein (hPLAP) gene on the chromosome 9p21 melanoma deleted region. Gene, 1999, 239, 155-161.	1.0	6
374	Genetic engineering approaches for organ transplantation. Transplantation Proceedings, 1999, 31, 2228-2229.	0.3	0
375	Tools of gene transfer in organ transplantation. Transplantation Proceedings, 1999, 31, 2230-2231.	0.3	0
376	HumanMinibrainHomologue (MNBH/DYRK1): Characterization, Alternative Splicing, Differential Tissue Expression, and Overexpression in Down Syndrome. Genomics, 1999, 57, 407-418.	1.3	169
377	Recombinant Families Locate the Gene for Non-Type I Cystinuria between Markers C13 and D19S587 on Chromosome 19q13.1. Genomics, 1999, 60, 362-365.	1.3	9
378	Cloning and Characterization of DYRK1B, a Novel Member of the DYRK Family of Protein Kinases. Biochemical and Biophysical Research Communications, 1999, 254, 474-479.	1.0	70

#	Article	IF	Citations
379	Chapter 22: Molecular Basis of Deafness due to Mutations in the Connexin26 Gene (GJB2). Current Topics in Membranes, 1999, , 483-508.	0.5	0
380	Vestibular and hearing loss in genetic and metabolic disorders. Current Opinion in Neurology, 1999, 12, 35-39.	1.8	6
381	Paternal origin of a de novo novel CFTR mutation (L1065R) causing cystic fibrosis. Human Mutation, 1998, 11, S99-S102.	1.1	7
382	Recurrence of the PKD1 nonsense mutation Q4041X in Spanish, Italian, and British families. Human Mutation, 1998, 11, S117-S120.	1.1	16
383	Retention of theCDKN2A locus and low frequency of point mutations in primary and metastasic cutaneous malignant melanoma. , 1998, 76, 312-316.		35
384	Two novel mutations in exon 11 of thePAH gene (V1163 del TG and P362T) associated with classic phenylketonuria and mild phenylketonuria. , 1998, 11, 482-482.		24
385	Isolation and genomic characterization of the TUPLE1/HIRA gene of the pufferfish Fugu rubripes. Gene, 1998, 208, 279-283.	1.0	13
386	Connexin-26 mutations in sporadic and inherited sensorineural deafness. Lancet, The, 1998, 351, 394-398.	6.3	610
387	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
388	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
389	A Clinical Variant of Neurofibromatosis Type 1: Familial Spinal Neurofibromatosis with a Frameshift Mutation in the NF1 Gene. American Journal of Human Genetics, 1998, 62, 834-841.	2.6	61
390	Dating the Origin of the CCR5-Î”32 AIDS-Resistance Allele by the Coalescence of Haplotypes. American Journal of Human Genetics, 1998, 62, 1507-1515.	2.6	507
391	Incidence of CDKN2A mutations in melanoma families: Inherited susceptibility to several cancers but not always association with dysplastic nevus syndrome. Journal of Dermatological Science, 1998, 16, S145.	1.0	0
392	Uncloned expanded CAG/CTG repeat sequences in autosomal dominant cerebellar ataxia (ADCA) detected by the repeat expansion detection (RED) method.. Journal of Medical Genetics, 1998, 35, 99-102.	1.5	6
393	Large CAG/CTG repeat templates produced by PCR, usefulness for the DIRECT method of cloning genes with CAG/CTG repeat expansions. Nucleic Acids Research, 1998, 26, 1352-1353.	6.5	13
394	Testicular CFTR Splice Variants in Patients with Congenital Absence of the Vas Deferens. Human Molecular Genetics, 1998, 7, 1739-1744.	1.4	74
395	Molecular cloning, expression, and chromosomal localization of a ubiquitously expressed human 6-phosphofructo-2-kinase/ fructose-2,6-bisphosphatase gene (PFKFB3). Cytogenetic and Genome Research, 1998, 83, 214-217.	0.6	71
396	Role of UEV-1, an Inactive Variant of the E2 UbiquitinConjugating Enzymes, in In Vitro Differentiation and Cell Cycle Behavior of HT-29-M6 Intestinal Mucosecretory Cells. Molecular and Cellular Biology, 1998, 18, 576-589.	1.1	142

#	Article	IF	Citations
397	A patient with autistic disorder and a 20/22 chromosomal translocation. Developmental Medicine and Child Neurology, 1998, 40, 492-495.	1.1	16
398	Minibrain (MNBH) is a single copy gene mapping to human chromosome 21q22.2. Cytogenetic and Genome Research, 1997, 77, 182-184.	0.6	3
399	Maternal transmission in sporadic Huntington's disease.. Journal of Neurology, Neurosurgery and Psychiatry, 1997, 62, 535-537.	0.9	14
400	HLA Class II Genes in Soybean Epidemic Asthma Patients. American Journal of Respiratory and Critical Care Medicine, 1997, 156, 1394-1398.	2.5	34
401	Down syndrome: characterisation of a case with partial trisomy of chromosome 21 owing to a paternal balanced translocation (15;21) (q26;q22.1) by FISH.. Journal of Medical Genetics, 1997, 34, 50-54.	1.5	24
402	Connexin26 mutations associated with the most common form of non- syndromic neurosensory autosomal recessive deafness (DFNB1) in Mediterraneans. Human Molecular Genetics, 1997, 6, 1605-1609.	1.4	540
403	Cosmid Contig and Transcriptional Map of Three Regions of Human Chromosome 21q22: Identification of 37 Novel Transcripts by Direct Selection. Genomics, 1997, 45, 59-67.	1.3	11
404	Genomic Organization, Alternative Splicing, and Expression Patterns of theDSCR1(Down Syndrome) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5	4.3	181
405	Confirmation of a Double-Hit Model for the NF1Gene in Benign Neurofibromas. American Journal of Human Genetics, 1997, 61, 512-519.	2.6	217
406	Reduced steady-state levels of mitochondrial RNA and increased mitochondrial DNA amount in human brain with aging. Molecular Brain Research, 1997, 52, 284-289.	2.5	140
407	Absence of Linkage Between Type III Protein S Deficiency and the PROS1 and C4BP Genes in Families Carrying the Protein S Heerlen Allele. Blood, 1997, 89, 2799-2806.	0.6	24
408	Rapid sizing of polymorphic microsatellite markers by capillary array electrophoresis. Journal of Chromatography A, 1997, 781, 295-305.	1.8	27
409	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
410	Qualitative and Quantitative Changes in Skeletal Muscle mtDNA and Expression of Mitochondrial-Encoded Genes in the Human Aging Process. Biochemical and Molecular Medicine, 1997, 62, 165-171.	1.5	77
411	Analysis of amino-acid and nucleotide variants in the spinocerebellar ataxia type 1 (SCA1) gene in schizophrenic patients. Human Genetics, 1997, 99, 772-775.	1.8	6
412	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
413	Polymorphisms at 13 expressed human sequences containing CAG/CTG repeats and analysis in autosomal dominant cerebellar ataxia (ADCA) patients. Human Genetics, 1997, 101, 18-21.	1.8	3
414	Alu -splice PCR: a simple method to isolate exon-containing fragments from cloned human genomic DNA. Human Genetics, 1997, 101, 346-350.	1.8	16

#	Article	IF	Citations
415	Inherited susceptibility to several cancers but absence of linkage between dysplastic nevus syndrome and CDKN2A in a melanoma family with a mutation in the CDKN2A (P16INK4A) gene. Human Genetics, 1997, 101, 359-364.	1.8	58
416	High heterogeneity for cystic fibrosis in Spanish families: 75 mutations account for 90% of chromosomes. Human Genetics, 1997, 101, 365-370.	1.8	94
417	SSCP analysis: A blind sensitivity trial. , 1997, 10, 65-70.		60
418	Geographic distribution and regional origin of 272 cystic fibrosis mutations in European populations. , 1997, 10, 135-154.		251
419	Sporadic heteroplasmic single 5.5 Kb mitochondrial DNA deletion associated with cerebellar ataxia, hypogonadotropic hypogonadism, choroidal dystrophy, and mitochondrial respiratory chain complex I deficiency. , 1997, 10, 212-216.		16
420	Missense mutation R1066C in the second transmembrane domain of CFTR causes a severe cystic fibrosis phenotype: Study of 19 heterozygous and 2 homozygous patients. , 1997, 10, 387-392.		12
421	The repeat expansion detection method in the analysis of diseases with CAG/CTG repeat expansion: Usefulness and limitations. Human Mutation, 1997, 10, 486-488.	1.1	9
422	SSCP analysis: A blind sensitivity trial. Human Mutation, 1997, 10, 65-70.	1.1	9
423	Geographic distribution and regional origin of 272 cystic fibrosis mutations in European populations. , 1997, 10, 135.		1
424	Geographic distribution and regional origin of 272 cystic fibrosis mutations in European populations. Human Mutation, 1997, 10, 135-154.	1.1	15
425	Immunohistochemical study of alpha, mu and pi class glutathione S transferase expression in malignant melanoma. British Journal of Dermatology, 1997, 136, 345-350.	1.4	23
426	Genomic Structure and Organization of the HumanrBATGene (SLC3A1). Genomics, 1996, 37, 249-252.	1.3	22
427	The effect of zidovudine on skeletal muscle mtDNA in HIV-1 infected patients with mild or no muscle dysfunction. Brain, 1996, 119, 1357-1364.	3.7	69
428	Homozygosity for R87H missense mutation and for a rare intron 7 DNA variant (7054G???A) in the PROC genes of three siblings initially classified as heterozygotes for protein C deficiency. Blood Coagulation and Fibrinolysis, 1996, 7, 15-23.	0.5	21
429	Aberrant RNA splicing of the protein C and protein S genes in healthy individuals. Blood Coagulation and Fibrinolysis, 1996, 7, 625-631.	0.5	4
430	CFTR haplotypic variability for normal and mutant genes in cystic fibrosis families from southern France. Human Genetics, 1996, 98, 336-344.	1.8	29
431	A female compound heterozygote (pre- and full mutation) for the CGG FMR1 expansion. Human Genetics, 1996, 98, 419-421.	1.8	22
432	Molecular analysis of the cystinuria disease gene: identification of four new mutations, one large deletion, and one polymorphism. Human Genetics, 1996, 98, 447-451.	1.8	44

#	Article	IF	Citations
433	YAC and cosmid FISH mapping of an unbalanced chromosomal translocation causing partial trisomy 21 and Down syndrome. Human Genetics, 1996, 98, 460-466.	1.8	18
434	Sex differences in mutational rate and mutational mechanism in the NF1 gene in neurofibromatosis type 1 patients. Human Genetics, 1996, 98, 696-699.	1.8	97
435	Genomic organization of TUPLE1/HIRA: a gene implicated in DiGeorge syndrome. Mammalian Genome, 1996, 7, 911-914.	1.0	11
436	PRENATAL DIAGNOSIS OF CYSTIC FIBROSIS IN A HIGHLY HETEROGENEOUS POPULATION. , 1996, 16, 215-222.		12
437	Recurrence of thePROC gene mutation R178Q: Independent origins in Spanish protein C deficiency patients. , 1996, 8, 71-73.		3
438	Haplotype analysis of 94 cystic fibrosis mutations with seven polymorphicCFTR DNA markers. , 1996, 8, 149-159.		42
439	Identification of two new nonsense mutations Q311X and W326X in Exon 2 of the adrenoleukodystrophy (ALD) gene. , 1996, 8, 286-287.		4
440	Complexity in a monogenic disease. Nature Genetics, 1996, 12, 348-350.	9.4	107
441	Mosaicism for the fragile X syndrome full mutation and deletions within the CGG repeat of the FMR1 gene.. Journal of Medical Genetics, 1996, 33, 338-340.	1.5	37
442	Huntington's disease: confirmation of diagnosis and presymptomatic testing in Spanish families by genetic analysis.. Journal of Neurology, Neurosurgery and Psychiatry, 1996, 61, 625-627.	0.9	8
443	Development in a 46 XX Boy with Positive SRY Gene. Journal of Pediatric Endocrinology and Metabolism, 1996, 9, 623-6.	0.4	16
444	A human homologue of Drosophila minibrain (MNB) is expressed in the neuronal regions affected in Down syndrome and maps to the critical region. Human Molecular Genetics, 1996, 5, 1305-1310.	1.4	197
445	Thirteen cystic fibrosis patients, 12 compound heterozygous and one homozygous for the missense mutation G85E: a pancreatic sufficiency/insufficiency mutation with variable clinical presentation.. Journal of Medical Genetics, 1996, 33, 820-822.	1.5	12
446	Cloning (CAG/GTC)n STSs by an Alu-(CAG/GTC)n PCR method: an approach to human chromosome 12 and spinocerebellar ataxia 2 (SCA2). Nucleic Acids Research, 1996, 24, 3651-3652.	6.5	2
447	Renal-hepatic-pancreatic dysplasia: an autosomal recessive malformation.. Journal of Medical Genetics, 1996, 33, 409-412.	1.5	35
448	Haplotype analysis of 94 cystic fibrosis mutations with seven polymorphic CFTR DNA markers. Human Mutation, 1996, 8, 149-159.	1.1	5
449	Geographic distribution and origin of CFTR mutations in Germany. Human Genetics, 1996, 97, 727-731.	1.8	3
450	Ectopic Transcript Analysis Indicates that Allelic Exclusion is an Important Cause of Type I Protein C Deficiency in Patients with Nonsense and Frameshift Mutations in the PROC Gene. Thrombosis and Haemostasis, 1996, 75, 870-876.	1.8	7

#	Article	IF	Citations
451	Integration of 30 CA-Repeat Markers into the Cytogenetic, Genetic and YAC Maps of Human Chromosome 21. European Journal of Human Genetics, 1996, 4, 135-142.	1.4	6
452	A nuclear defect in the 4p16 region predisposes to multiple mitochondrial DNA deletions in families with Wolfram syndrome.. Journal of Clinical Investigation, 1996, 97, 1570-1576.	3.9	95
453	Abdominal aortic aneurysms and autosomal dominant polycystic kidney disease.. Journal of the American Society of Nephrology: JASN, 1996, 7, 2483-2486.	3.0	56
454	The molecular basis of cystinuria: the role of the rBAT gene. Amino Acids, 1996, 11, 225-246.	1.2	13
455	Genetic heterogeneity in cystinuria: the SLC3A1 gene is linked to type I but not to type III cystinuria.. Proceedings of the National Academy of Sciences of the United States of America, 1995, 92, 9667-9671.	3.3	98
456	Clinical, neuropathologic, and genetic studies of a large spinocerebellar ataxia type 1 (SCA1) kindred. Neurology, 1995, 45, 24-30.	1.5	141
457	Simultaneous On-line DNA Sequencing on Both Strands with Two Fluorescent Dyes. Analytical Biochemistry, 1995, 224, 117-121.	1.1	40
458	Predominant occurrence of somatic mutations of theNF2 gene in meningiomas and schwannomas. Genes Chromosomes and Cancer, 1995, 13, 211-216.	1.5	132
459	Amplifying dinucleotide microsatellite loci from bone and tooth samples of up to 5000 years of age: more inconsistency than usefulness. Human Genetics, 1995, 96, 205-212.	1.8	44
460	The 9-bp deletion in region V of mitochondrial DNA: evidence of mutation recurrence. Human Genetics, 1995, 96, 225-228.	1.8	12
461	A novel polymorphism (6376 G/T) in intron 7 of the human protein C gene. Human Genetics, 1995, 96, 243-244.	1.8	1
462	Extensive analysis of 40 infertile patients with congenital absence of the vas deferens: in 50% of cases only one CFTR allele could be detected. Human Genetics, 1995, 95, 205-11.	1.8	96
463	De novo missense mutation Y174S in exon 1 of the adrenoleukodystrophy (ALD) gene. Human Genetics, 1995, 95, 235-7.	1.8	8
464	Assignment of the gene responsible for cystinuria (rBAT) and of markers D2S119 and D2S177 to 2p16 by fluorescence in situ hybridization. Human Genetics, 1995, 95, 633-6.	1.8	30
465	Two further cases of mutation R1947X in the NF1 gene: screening for a relatively common recurrent mutation. Human Genetics, 1995, 96, 361-3.	1.8	16
466	Characterisation of three microsatellite polymorphisms (D21S1262, D21S1419 and D21S1421) from band 21q22.1. Human Genetics, 1995, 95, 596-8.	1.8	2
467	Five new microsatellite polymorphisms at the q21 region of human chromosome 21. Human Genetics, 1995, 95, 119-22.	1.8	1
468	Clinical characteristics of 16 cystic fibrosis patients with the missense mutation R334W, a pancreatic insufficiency mutation with variable age of onset and interfamilial clinical differences. Human Genetics, 1995, 95, 331-6.	1.8	36

#	Article	IF	Citations
469	Identification of two highly polymorphic CA-repeats (D21S1224 and D21S1261) on human chromosome 21q22.3. Human Genetics, 1995, 95, 367-9.	1.8	3
470	Prenatal diagnosis of neurofibromatosis type 1: From flanking rflps to intragenic microsatellite markers. Prenatal Diagnosis, 1995, 15, 129-134.	1.1	8
471	Rapid fetal karyotype from cystic hygroma and pleural effusions. Prenatal Diagnosis, 1995, 15, 141-148.	1.1	10
472	Prenatal diagnosis of fragile x syndrome: (cgg)n expansion and methylation of chorionic villus samples. Prenatal Diagnosis, 1995, 15, 801-807.	1.1	20
473	Phenotype of glutathione S-transferase Mu (GSTM1) and susceptibility to malignant melanoma. British Journal of Cancer, 1995, 72, 324-326.	2.9	44
474	WASP gene mutations in Wiskott-Aldrich syndrome and X-linked thrombocytopenia. Human Molecular Genetics, 1995, 4, 1127-1135.	1.4	142
475	A new human gene from the Down syndrome critical region encodes a proline-rich protein highly expressed in fetal brain and heart. Human Molecular Genetics, 1995, 4, 1935-1944.	1.4	250
476	The human collagenase-3 (CLG3) gene is located on chromosome 11q22.3 clustered to other members of the matrix metalloproteinase gene family. Genomics, 1995, 26, 615-618.	1.3	48
477	Mutations in the Cystic Fibrosis Gene in Patients with Congenital Absence of the Vas Deferens. New England Journal of Medicine, 1995, 332, 1475-1480.	13.9	959
478	Severe Type I Protein C Deficiency in a Compound Heterozygote for Y124C and Q132X Mutations in Exon 6 of the PROC Gene. Thrombosis and Haemostasis, 1995, 74, 1215-1220.	1.8	10
479	A cystic fibrosis patient homozygous for the new frameshift mutation 936delTA: description and clinical data.. Journal of Medical Genetics, 1994, 31, 369-370.	1.5	3
480	Chemiluminescent detection of blotted PCR products (CB-PCR) of two CAG dynamic mutations (Huntington's disease and spinocerebellar ataxia type 1).. Journal of Medical Genetics, 1994, 31, 654-655.	1.5	11
481	Multiple deletions of mtDNA in two brothers with sideroblastic anemia and motochondrial myopathy and in their asymptomatic mother. Human Molecular Genetics, 1994, 3, 1945-1949.	1.4	72
482	A cluster of cystic fibrosis mutations in exon 17b of the CFTR gene: a site for rare mutations.. Journal of Medical Genetics, 1994, 31, 731-734.	1.5	7
483	Identification of a new frameshift mutation (1801delAG) in the ALD gene. Human Molecular Genetics, 1994, 3, 1889-1890.	1.4	26
484	Two novel mutations in exon 5 of the protein C gene in two Spanish families with thrombophilia due to protein C deficiency. Human Molecular Genetics, 1994, 3, 1205-1206.	1.4	1
485	Neurofibromatosis Type 1 Due to Germ-Line Mosaicism in a Clinically Normal Father. New England Journal of Medicine, 1994, 331, 1403-1407.	13.9	146
486	Molecular analysis of the (CGG)n expansion in the FMR-1 gene in 59 Spanish fragile X syndrome families. Human Genetics, 1994, 94, 395-400.	1.8	22

#	Article	IF	Citations
487	Analysis of the CFTR gene confirms the high genetic heterogeneity of the Spanish population: 43 mutations account for only 78% of CF chromosomes. Human Genetics, 1994, 93, 447-51.	1.8	65
488	Complete detection of mutations in cystic fibrosis patients of Native American origin. Human Genetics, 1994, 94, 629-32.	1.8	35
489	Cystic fibrosis in a low-incidence population: two major mutations in Finland. Human Genetics, 1994, 93, 162-166.	1.8	35
490	Two CA/GT repeat polymorphisms in intron 27 of the human neurofibromatosis type 1 (NF1) gene. Human Genetics, 1994, 93, 351-352.	1.8	81
491	Three CA/GT repeat polymorphisms from loci D21S414 and D21S1234 on human chromosome 21. Human Genetics, 1994, 93, 359-360.	1.8	3
492	Carrier detection and microsatellite analysis of duchenne and becker muscular dystrophy in spanish families. Prenatal Diagnosis, 1994, 14, 123-130.	1.1	4
493	Prenatal diagnosis of werdnig-hoffmann disease: DNA analysis of a mummified umbilical cord using closely linked microsatellite markers. Prenatal Diagnosis, 1994, 14, 219-222.	1.1	5
494	Analysis of the CFTR gene in the Spanish population: SSCP-screening for 60 known mutations and identification of four new mutations (Q30X, A120T, 1812-1 Gâ†'A, and 3667de14). Human Mutation, 1994, 3, 223-230.	1.1	31
495	New alleles at microsatellite loci in CEPH families mainly arise from somatic mutations in the lymphoblastoid cell lines. Human Mutation, 1994, 3, 365-372.	1.1	44
496	Cystinuria caused by mutations in rBAT, a gene involved in the transport of cystine. Nature Genetics, 1994, 6, 420-425.	9.4	366
497	The origin of the major cystic fibrosis mutation (Î"F508) in European populations. Nature Genetics, 1994, 7, 169-175.	9.4	323
498	Reply to â€" Age of the Î"F508 cystic fibrosis mutation. Nature Genetics, 1994, 8, 216-218.	9.4	25
499	Mapping of the human Zn-α₂-glycoprotein gene (AZGP1) to chromosome 7q22 by in situ hybridization. Cytogenetic and Genome Research, 1994, 66, 263-266.	0.6	23
500	Acceptor splice site mutation in the invariant AG of intron 5 of the protein C gene, causing type I protein C deficiency. Human Genetics, 1993, 92, 506-508.	1.8	4
501	Cystic fibrosis in Spain: high frequency of mutation G542X in the Mediterranean coastal area. Human Genetics, 1993, 91, 66-70.	1.8	35
502	A highly informative CA/GT repeat polymorphism in intron 38 of the human neurofibromatosis type 1 (NF1) gene. Human Genetics, 1993, 92, 429-430.	1.8	85
503	Genetic variation of microsatellite markers D1S117, D6S89, D11S35, APOC2, and D21S168 in the Spanish population. International Journal of Legal Medicine, 1993, 105, 271-277.	1.2	10
504	Novel alleles, hemizygosity and deletions at an Alu-repeat within the neurofibromatosis type 1 (NF1) gene. Human Molecular Genetics, 1993, 2, 725-730.	1.4	47

#	Article	IF	Citations
505	Isolation and Characterization of 14 CA-Repeat Microsatellites from Human Chromosome 21. Genomics, 1993, 18, 151-155.	1.3	12
506	A new missense mutation (E92K) in the first transmembrane domain of the CFTR gene causes a benign cystic fibrosis phenotype. Human Molecular Genetics, 1993, 2, 79-80.	1.4	27
507	Microsatellite haplotypes for cystic fibrosis: mutation frameworks and evolutionary tracers. Human Molecular Genetics, 1993, 2, 1015-1022.	1.4	97
508	Identification of two new missense mutations (K58N and R121Q) in the Norrie disease (ND) gene in two Spanish families. Human Molecular Genetics, 1993, 2, 1953-1955.	1.4	31
509	Haplotype analysis to determine the position of a mutation among closely linked DNA markers. Human Molecular Genetics, 1993, 2, 1007-1014.	1.4	19
510	Identification of a new missense mutation (P205S) in the first transmembrane domain of the CFTR gene associated with a mild cystic fibrosis phenotype. Human Molecular Genetics, 1993, 2, 1741-1742.	1.4	18
511	Two dinucleotide repeat polymorphisms at 21q22.3 (D21S416 and D21S1235). Human Molecular Genetics, 1993, 2, 1744-1744.	1.4	3
512	Identification of a 31-bp insertion (3860ins31) in exon 20 of the cysticfibrosis (CFTR) gene. Human Molecular Genetics, 1993, 2, 1317-1318.	1.4	7
513	Uniparental inheritance of microsatellite alleles of the cystic fibrosis gene (CFTR): identification of a 50 kilobase deletion. Human Molecular Genetics, 1993, 2, 677-681.	1.4	32
514	Presymptomatic analysis of spinocerebellar ataxia type 1 (SCA1) via the expansion of the SCA1 CAG-repeat in a large pedigree displaying anticipation and parental male bias. Human Molecular Genetics, 1993, 2, 2123-2128.	1.4	92
515	Prenatal diagnosis of sporadic neurofibromatosis 1. Lancet, The, 1992, 339, 119-120.	6.3	9
516	Multiplex PCR amplification of three microsatellites within the CFTR gene. Genomics, 1992, 13, 1362-1364.	1.3	73
517	Mutation analysis of genetic diseases by asymmetric-PCR SSCP and ethidium bromide staining: application to neurofibromatosis and cystic fibrosis. Molecular and Cellular Probes, 1992, 6, 357-359.	0.9	13
518	Dinucleotide (CA/GT) repeat polymorphism in intron 17B of the cystic fibrosis transmembrane conductance regulator (CFTR) gene. Human Genetics, 1992, 88, 356.	1.8	21
519	A rare DNA variant in exon 15 of the cystic fibrosis transmembrane conductance regulator (CFTR) gene. Human Genetics, 1992, 90, 474.	1.8	6
520	Incidence and expression of the N1303K mutation of the cystic fibrosis (CFTR) gene. Human Genetics, 1992, 89, 653-658.	1.8	69
521	Continuum of overlapping clones spanning the entire human chromosome 21q. Nature, 1992, 359, 380-387.	13.7	436
522	Identification of a frameshift mutation (1609delCA) in exon 10 of the CFTR gene in seven Spanish cystic fibrosis patients. Human Mutation, 1992, 1, 75-76.	1.1	12

#	Article	IF	Citations
523	Protein C deficiency: Identification of a novel two-base pair insertion and two point mutations in exon 7 of the protein C gene in Spanish families. Human Mutation, 1992, 1, 428-431.	1.1	5
524	CAGT Microsatellite alleles within the cystic fibrosis transmembrane conductance regulator (CFTR) gene are not generated by unequal crossingover. Genomics, 1991, 10, 692-698.	1.3	129
525	High conservation of sequences involved in cystic fibrosis mutations in five mammalian species. Genomics, 1991, 10, 1070-1072.	1.3	10
526	The search for South European cystic fibrosis mutations: Identification of two new mutations, four variants, and intronic sequences. Genomics, 1991, 10, 193-200.	1.3	117
527	Prenatal diagnosis of cystic fibrosis by multiplex PCR of mutation and microsatellite alleles. Lancet, The, 1991, 338, 458.	6.3	12
528	Prenatal diagnosis of cystic fibrosis by simultaneous analysis of two different mutations. Prenatal Diagnosis, 1991, 11, 671-672.	1.1	1
529	A tetranucleotide repeat polymorphism in the cystic fibrosis gene. Human Genetics, 1991, 86, 625.	1.8	35
530	Analysis of 14 cystic fibrosis mutations in five South European populations. Human Genetics, 1991, 87, 737-8.	1.8	52
531	Recurrence of a nonsense mutation in the NF1 gene causing classical neurofibromatosis type 1. Human Genetics, 1991, 88, 185-8.	1.8	39
532	MspI restriction fragment length polymorphism near exon 10 of cystic fibrosis (CFTR) gene. Nucleic Acids Research, 1991, 19, 1719-1719.	6.5	0
533	Mutation and linkage disequilibrium analysis in genetic counselling of Spanish cystic fibrosis families.. Journal of Medical Genetics, 1991, 28, 771-776.	1.5	11
534	SSCP-polymorphism in intron 12 of the CFTR gene recognized by BclI. Nucleic Acids Research, 1991, 19, 6343-6343.	6.5	14
535	Genetic Analysis of Cystic Fibrosis. Advances in Experimental Medicine and Biology, 1991, 290, 31-38.	0.8	0
536	PCR detection of the pKM.19/ScrfI RFLP (D7S23), a marker closely linked to the cystic fibrosis mutation. Nucleic Acids Research, 1990, 18, 4957-4957.	6.5	3
537	Distribution of the Î”F508 mutation in 194 Spanish cystic fibrosis families. Human Genetics, 1990, 85, 396-397.	1.8	17
538	Cystic fibrosis mutation Î”F508 in Finland: other mutations predominate. Human Genetics, 1990, 85, 413-415.	1.8	12
539	Î” F508 gene deletion and prenatal diagnosis of cystic fibrosis in Italian and Spanish families. Prenatal Diagnosis, 1990, 10, 413-414.	1.1	8
540	Genetic differences in cystic fibrosis patients with and without pancreatic insufficiency. Human Genetics, 1990, 84, 435-8.	1.8	10

#	Article	IF	Citations
541	Scrfl restriction fragment length polymorphism at the D7S23 locus (probe pKM.19), closely linked to cystic fibrosis. Nucleic Acids Research, 1990, 18, 1318-1318.	6.5	3
542	The genotype of a new linked DNA marker, MP6d-9, is related to the clinical course of cystic fibrosis.. Journal of Medical Genetics, 1990, 27, 17-20.	1.5	9
543	A new polymorphic locus, D7S411, isolated by cloning from preparative pulse-field gels is close to the mutation causing cystic fibrosis. Genomics, 1990, 6, 39-47.	1.3	15
544	Polymerase chain reaction for detection of the pMP6d-9/MspI RFLP, a marker closely linked to the cystic fibrosis mutation. Nucleic Acids Research, 1989, 17, 7118-7118.	6.5	27
545	Cystic fibrosis in Finland: a molecular and genealogical study. Human Genetics, 1989, 83, 20-25.	1.8	25
546	Detection of a rare allele with the pMP6d-9/MspI RFLP near the cystic fibrosis locus. Human Genetics, 1989, 83, 305-306.	1.8	2
547	Linkage disequilibrium for DNA haplotypes near the cystic fibrosis locus in two South European populations. Human Genetics, 1989, 83, 175-178.	1.8	17
548	Î”F508 GENE DELETION IN CYSTIC FIBROSIS IN SOUTHERN EUROPE. Lancet, The, 1989, 334, 1404.	6.3	56
549	Detection of a rare-cutter RFLP in a CpG-rich island near the cystic fibrosis locus. Human Genetics, 1988, 80, 309-310.	1.8	3
550	GENETIC DIFFERENCES BETWEEN CYSTIC FIBROSIS WITH AND WITHOUT MECONIUM ILEUS. Lancet, The, 1988, 331, 376-378.	6.3	54
551	Experience with New DNA Markers for the Diagnosis of Cystic Fibrosis. New England Journal of Medicine, 1988, 318, 50-51.	13.9	55
552	Isolation and mapping of a polymorphic DNA sequence pXH3 on chromosome X [DXS235]. Nucleic Acids Research, 1988, 16, 2740-2740.	6.5	0
553	A rapid method to identify cosmids containing rare restriction site. Nucleic Acids Research, 1987, 15, 1415-1425.	6.5	23
554	Physical and genetic analysis of cosmids from the vicinity of the cystic fibrosis locus. Nucleic Acids Research, 1987, 15, 3639-3652.	6.5	20
555	The Cystic Fibrosis Locus. Enzyme, 1987, 38, 8-13.	0.7	2
556	A candidate for the cystic fibrosis locus isolated by selection for methylation-free islands. Nature, 1987, 326, 840-845.	13.7	364
557	Translocation of the c-myc oncogene 3â€² to the immunoglobin heavy chain enhancer in the tumor DNA from a Burkitt lymphoma. Cell Biology International Reports, 1986, 10, 148-148.	0.7	0
558	Chromosome assignment and restriction fragment length polymorphism analysis of the anonymous DNA probe B79a at 7q22 (HMG8 assignment D7S13). Human Genetics, 1986, 74, 320-2.	1.8	25

#	Article	IF	Citations
559	CHRONIC T-CELL LYMPHOCYTOSIS ASSOCIATED WITH PURE RED CELL APLASIA, THYMOMA AND HYPOGAMMAGLOBULINAEMIA. British Journal of Haematology, 1985, 61, 582-584.	1.2	16
560	Plasmatic and Urinary Protein C Levels in Nephrotic Syndrome. Thrombosis and Haemostasis, 1985, 54, 900-900.	1.8	5