Roberto Giorda

List of Publications by Year
in descending order

161 papers	7,009 citations	57758 44 h-index	71685 76 g-index
162 all docs	162 docs citations	162 times ranked	9197 citing authors

#	Article	IF	Citations
1	Expanding Phenotype of Poirierâ€“Bienvenu Syndrome: New Evidence from an Italian Multicentrical Cohort of Patients. Genes, 2022, 13, 276.	2.4	10
2	Comprehensive variant spectrum of the CNGA3 gene in patients affected by achromatopsia. Human Mutation, 2022, 43, 832-858.	2.5	8
3	Respiratory Sinus Arrhythmia (RSA) stress response in preschool age varies by serotonin transporter polymorphism (5-HTTLPR): A preliminary report. Journal of Experimental Child Psychology, 2022, 219, 105413.	1.4	1
4	SCN2A Pathogenic Variants and Epilepsy: Heterogeneous Clinical, Genetic and Diagnostic Features. Brain Sciences, 2022, 12, 18.	2.3	5
5	Serotonin transporter gene methylation and emotional regulation in preschool children born preterm: A longitudinal evaluation of the role of negative emotionality in infancy. Infant Mental Health Journal, 2022, 43, 589-596.	1.8	1
6	Neuroimaging and DNA Methylation: An Innovative Approach to Study the Effects of Early Life Stress on Developmental Plasticity. Frontiers in Psychology, 2021, 12, 672786.	2.1	5
7	Brain Anatomical Mediators of GRIN2B Gene Association with Attention/Hyperactivity Problems: An Integrated Genetic-Neuroimaging Study. Genes, 2021, 12, 1193.	2.4	3
8	ï»¿Hidden pandemic: COVID-19-related stress, SLC6A4 methylation, and infantsâ€™ temperament at 3Âmonths. Scientific Reports, 2021, 11, 15658.	3.3	32
9	Developmental and epilepsy spectrum of Poirierâ€“Bienvenu neurodevelopmental syndrome: Description of a new case study and review of the available literature. Seizure: the Journal of the British Epilepsy Association, 2021, 93, 133-139.	2.0	6
10	RB1CC1 duplication and aberrant overexpression in a patient with schizophrenia: further phenotype delineation and proposal of a pathogenetic mechanism. Molecular Genetics & Genomic Medicine, 2021, 9, e1561.	1.2	2
11	The role of maternal touch in the association between SLC6A4 methylation and stress response in very preterm infants. Developmental Psychobiology, 2021, 63, e22218.	1.6	9
12	Exploring the Contribution of Proximal Family Risk Factors on SLC6A4 DNA Methylation in Children with a History of Maltreatment: A Preliminary Study. International Journal of Environmental Research and Public Health, 2021, 18, 12736.	2.6	3
13	Mild epileptic phenotype associates with de novo eef1a2 mutation: Case report and review. Brain and Development, 2020, 42, 77-82.	1.1	11
14	Deepâ€intronic variants in CNGB3 cause achromatopsia by pseudoexon activation. Human Mutation, 2020, 41, 255-264.	2.5	26
15	Painâ€related increase in serotonin transporter gene methylation associates with emotional regulation in 4.5â€yearâ€old pretermâ€born children. Acta Paediatrica, International Journal of Paediatrics, 2020, 109, 1166-1174.	1.5	23
16	Early Parenting Intervention â€“ Biobehavioral Outcomes in infants with Neurodevelopmental Disabilities (EPI-BOND): study protocol for an Italian multicentre randomised controlled trial. BMJ Open, 2020, 10, e035249.	1.9	19
17	The Mediation Role of Dynamic Multisensory Processing Using Molecular Genetic Data in Dyslexia. Brain Sciences, 2020, 10, 993.	2.3	8
18	Measuring the Outcomes of Maternal COVID-19-related Prenatal Exposure (MOM-COPE): study protocol for a multicentric longitudinal project. BMJ Open, 2020, 10, e044585.	1.9	22

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19	The influence of DCDC2 risk genetic variants on reading: Testing main and haplotypic effects. Neuropsychologia, 2019, 130, 52-58.	1.6	9
20	Telomere length and salivary cortisol stress reactivity in very preterm infants. Early Human Development, 2019, 129, 1-4.	1.8	13
21	Therapeutic effect of Anakinra in the relapsing chronic phase of febrile infectionâ€"related epilepsy syndrome. Epilepsia Open, 2019, 4, 344-350.	2.4	85
22	Novel epilepsy phenotype associated to a known SCN8A mutation. Seizure: the Journal of the British Epilepsy Association, 2019, 67, 15-17.	2.0	6
23	Pain exposure associates with telomere length erosion in very preterm infants. Psychoneuroendocrinology, 2018, 89, 113-119.	2.7	15
24	Chromothripsis and ring chromosome 22: a paradigm of genomic complexity in the Phelan-McDermid syndrome (22q13 deletion syndrome). Journal of Medical Genetics, 2018, 55, 269-277.	3.2	22
25	From CNTNAP2 to Early Expressive Language in Infancy: The Mediation Role of Rapid Auditory Processing. Cerebral Cortex, 2018, 28, 2100-2108.	2.9	15
26	De novo unbalanced translocations have a complex history/aetiology. Human Genetics, 2018, 137, 817-829.	3.8	23
27	Very preterm birth is associated with PLAGL1 gene hypomethylation at birth and discharge. Epigenomics, 2018, 10, 1121-1130.	2.1	7
28	From early stress to 12-month development in very preterm infants: Preliminary findings on epigenetic mechanisms and brain growth. PLoS ONE, 2018, 13, e0190602.	2.5	60
29	The role of READ1 and KIAA0319 genetic variations in developmental dyslexia: testing main and interactive effects. Journal of Human Genetics, 2017, 62, 949-955.	2.3	8
30	Complex effects of dyslexia risk factors account for <scp>ADHD</scp> traits: evidence from two independent samples. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2017, 58, 75-82.	5.2	28
31	Clinical Characterization, Genetics, and Long-Term Follow-up of a Large Cohort of Patients With Agenesis of the Corpus Callosum. Journal of Child Neurology, 2017, 32, 60-71.	1.4	34
32	Maternal Sensitivity Buffers the Association between SLC6A4 Methylation and Socio-Emotional Stress Response in 3-Month-Old Full Term, but not very Preterm Infants. Frontiers in Psychiatry, 2017, 8, 171.	2.6	28
33	Telomere Length in Preterm Infants: A Promising Biomarker of Early Adversity and Care in the Neonatal Intensive Care Unit?. Frontiers in Endocrinology, 2017, 8, 295.	3.5	7
34	Serotonin Transporter Gene (SLC6A4) Methylation Associates With NeonatalÂIntensive Care Unit Stay and 3â€MonthâEOld Temperament in Preterm Infants. Child Development, 2016, 87, 38-48.	3.0	73
35	SLC6A4 methylation as an epigenetic marker of life adversity exposures in humans: A systematic review of literature. Neuroscience and Biobehavioral Reviews, 2016, 71, 7-20.	6.1	105
36	SLC6A4 promoter region methylation and socio-emotional stress response in very preterm and full-term infants. Epigenomics, 2016, 8, 895-907.	2.1	37

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37	Partial deletion of <scp>DEPDC</scp>5 in a child with focal epilepsy. Epilepsia Open, 2016, 1, 140-144.	2.4	1
38	Effect of family structure and TPH2 G-703T on the stability of dysregulation profile throughout adolescence. Journal of Affective Disorders, 2016, 190, 576-584.	4.1	7
39	Long-term follow-up of a patient with 5q31.3 microdeletion syndrome and the smallest de novo 5q31.2q31.3 deletion involving PURA. Molecular Cytogenetics, 2015, 8, 89.	0.9	14
40	Pain-related stress during the Neonatal Intensive Care Unit stay and SLC6A4 methylation in very preterm infants. Frontiers in Behavioral Neuroscience, 2015, 9, 99.	2.0	78
41	A novel mutation in COL4A1 gene: A possible cause of early postnatal cerebrovascular events. American Journal of Medical Genetics, Part A, 2015, 167, 810-815.	1.2	7
42	GRIN2B predicts attention problems among disadvantaged children. European Child and Adolescent Psychiatry, 2015, 24, 827-836.	4.7	18
43	An assessment of gene-by-gene interactions as a tool to unfold missing heritability in dyslexia. Human Genetics, 2015, 134, 749-760.	3.8	20
44	The role of DCDC2 genetic variants and low socioeconomic status in vulnerability to attention problems. European Child and Adolescent Psychiatry, 2015, 24, 309-318.	4.7	13
45	GRIN2B mediates susceptibility to intelligence quotient and cognitive impairments in developmental dyslexia. Psychiatric Genetics, 2015, 25, 9-20.	1.1	32
46	Testis development in the absence of SRY: chromosomal rearrangements at SOX9 and SOX3. European Journal of Human Genetics, 2015, 23, 1025-1032.	2.8	59
47	Effect of the serotonin transporter gene and of environment on the continuity of anxiety and depression traits throughout adolescence. Epidemiology and Psychiatric Sciences, 2014, 23, 399-409.	3.9	8
48	A new patient with a terminal de novo 2p25.3 deletion of 1.9ÂMb associated with early-onset of obesity, intellectual disabilities and hyperkinetic disorder. Molecular Cytogenetics, 2014, 7, 53.	0.9	19
49	Genotypeâ€“phenotype relationship in a child with 2.3ÂMb de novo interstitial 12p13.33-p13.32 deletion. European Journal of Medical Genetics, 2014, 57, 334-338.	1.3	13
50	The DCDC2/intron 2 deletion and white matter disorganization: Focus on developmental dyslexia. Cortex, 2014, 57, 227-243.	2.4	40
51	KIAA0319 and ROBO1: evidence on association with reading and pleiotropic effects on language and mathematics abilities in developmental dyslexia. Journal of Human Genetics, 2014, 59, 189-197.	2.3	52
52	8q12 microduplication including CHD7: clinical report on a new patient with Duane retraction syndrome type 3. Molecular Cytogenetics, 2013, 6, 49.	0.9	3
53	5p13 microduplication syndrome: A new case and better clinical definition of the syndrome. European Journal of Medical Genetics, 2013, 56, 54-58.	1.3	14
54	An assessment of geneâ€byâ€environment interactions inÂdevelopmental dyslexiaâ€related phenotypes. Genes, Brain and Behavior, 2013, 12, 47-55.	2.2	55

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55	Low-copy repeats at the human VIPR2 gene predispose to recurrent and nonrecurrent rearrangements. European Journal of Human Genetics, 2013, 21, 757-761.	2.8	21
56	DCDC2 genetic variants and susceptibility to developmental dyslexia. Psychiatric Genetics, 2012, 22, 25-30.	1.1	71
57	Pediatric Biobanking: A Pilot Qualitative Survey of Practices, Rules, and Researcher Opinions in Ten European Countries. Biopreservation and Biobanking, 2012, 10, 29-36.	1.0	22
58	Influence of the OPRM1 gene polymorphism upon children's degree of withdrawal and brain activation in response to facial expressions. Developmental Cognitive Neuroscience, 2012, 2, 103-109.	4.0	27
59	CEREBRAL RESPONSES TO EMOTIONAL EXPRESSIONS AND THE DEVELOPMENT OF SOCIAL ANXIETY DISORDER: A PRELIMINARY LONGITUDINAL STUDY. Depression and Anxiety, 2012, 29, 54-61.	4.1	39
60	De Novo Unbalanced Translocations in Prader-Willi and Angelman Syndrome Might Be the Reciprocal Product of inv dup(15)s. PLoS ONE, 2012, 7, e39180.	2.5	5
61	An Assessment of Gene-by-Environment Interactions in Developmental Dyslexia-Related Phenotypes. Genes, Brain and Behavior, 2012, , n/a-n/a.	2.2	0
62	A de novo balanced translocation t(7;12)(p21.2;p12.3) in a patient with Saethreâ€“Chotzen-like phenotype downregulates TWIST and an osteoclastic protein-tyrosine phosphatase, PTP-oc. European Journal of Medical Genetics, 2011, 54, e478-e483.	1.3	5
63	Definition of the neurological phenotype associated with dup (X)(p11.22-p11.23). Epileptic Disorders, 2011, 13, 240-251.	1.3	8
64	Pleiotropic Effects of DCDC2 and DYX1C1 Genes on Language and Mathematics Traits in Nuclear Families of Developmental Dyslexia. Behavior Genetics, 2011, 41, 67-76.	2.1	43
65	Common structural features characterize interstitial intrachromosomal Xp and 18q triplications. American Journal of Medical Genetics, Part A, 2011, 155, 2681-2687.	1.2	6
66	XX males SRY negative: a confirmed cause of infertility. Journal of Medical Genetics, 2011, 48, 710-712.	3.2	86
67	Rare familial 16q21 microdeletions under a linkage peak implicate cadherin 8 (CDH8) in susceptibility to autism and learning disability. Journal of Medical Genetics, 2011, 48, 48-54.	3.2	94
68	Molecular Mechanisms Generating and Stabilizing Terminal 22q13 Deletions in 44 Subjects with Phelan/McDermid Syndrome. PLoS Genetics, 2011, 7, e1002173.	3.5	172
69	Chromosome 22q13 Rearrangements Causing Global Developmental Delay and Autistic Spectrum Disorder. Monographs in Human Genetics, 2010, , 137-150.	0.5	2
70	COMT Val158Met polymorphism and socioeconomic status interact to predict attention deficit/hyperactivity problems in children aged 10â€“14. European Child and Adolescent Psychiatry, 2010, 19, 549-557.	4.7	43
71	Breakpoint determination of 15 large deletions in Peutzâ€“Jeghers subjects. Human Genetics, 2010, 128, 373-382.	3.8	26
72	Mutations in SOX17 are associated with congenital anomalies of the kidney and the urinary tract. Human Mutation, 2010, 31, 1352-1359.	2.5	54

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73	Genotypeâ€“phenotype relationship in three cases with overlapping 19p13.12 microdeletions. European Journal of Human Genetics, 2010, 18, 1302-1309.	2.8	46
74	Refining the phenotype associated with MEF2C haploinsufficiency. Clinical Genetics, 2010, 78, 471-477.	2.0	85
75	Olfactory Receptor-Related Duplicons Mediate a Microdeletion at 11q13.2q13.4 Associated with a Syndromic Phenotype. Molecular Syndromology, 2010, 1, 176-184.	0.8	30
76	A novelCLN8mutation in late-infantile-onset neuronal ceroid lipofuscinosis (LINCL) reveals aspects of CLN8 neurobiological function. Human Mutation, 2009, 30, 1104-1116.	2.5	53
77	Different molecular mechanisms causing 9p21 deletions in acute lymphoblastic leukemia of childhood. Human Genetics, 2009, 126, 511-520.	3.8	39
78	Complex pathogenesis of Hirschsprung's disease in a patient with hydrocephalus, vesico-ureteral reflux and a balanced translocation t(3;17)(p12;q11). European Journal of Human Genetics, 2009, 17, 483-490.	2.8	26
79	Mosaic 22q13 deletions: evidence for concurrent mosaic segmental isodisomy and gene conversion. European Journal of Human Genetics, 2009, 17, 426-433.	2.8	16
80	Inverted duplications deletions: underdiagnosed rearrangements??. Clinical Genetics, 2009, 75, 505-513.	2.0	64
81	The influence of family structure, the TPH2â€ƒGâ€703T and the 5â€HTTLPR serotonergic genes upon affective problems in children aged 10â€“14â€ƒyears. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2009, 50, 317-325.	5.2	44
82	The role played by the interaction between genetic factors and attachment in the stress response in infancy. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2009, 50, 1513-1522.	5.2	46
83	Complex Segmental Duplications Mediate a Recurrent dup(X)(p11.22-p11.23) Associated with Mental Retardation, Speech Delay, and EEG Anomalies in Males and Females. American Journal of Human Genetics, 2009, 85, 394-400.	6.2	60
84	Complex Segmental Duplications Mediate a Recurrent dup(X)(p11.22-p11.23) Associated with Mental Retardation, Speech Delay, and EEG Anomalies in Males and Females. American Journal of Human Genetics, 2009, 85, 419.	6.2	2
85	The tumor suppressor gene TRC8/RNF139 is disrupted by a constitutional balanced translocation t(8;22)(q24.13;q11.21) in a young girl with dysgerminoma. Molecular Cancer, 2009, 8, 52.	19.2	24
86	A familial inverted duplication/deletion of 2p25.1â€“25.3 provides new clues on the genesis of inverted duplications. European Journal of Human Genetics, 2009, 17, 179-186.	2.8	37
87	Mutations in CNGA3 impair trafficking or function of cone cyclic nucleotide-gated channels, resulting in achromatopsia. Human Mutation, 2008, 29, 1228-1236.	2.5	54
88	Detailed phenotypeâ€“genotype study in five patients with chromosome 6q16 deletion: narrowing the critical region for Praderâ€“Willi-like phenotype. European Journal of Human Genetics, 2008, 16, 1443-1449.	2.8	74
89	Molecular and cytogenetic analysis of the spreading of X inactivation in a girl with microcephaly, mild dysmorphic features and t(X;5)(q22.1;q31.1). European Journal of Human Genetics, 2008, 16, 897-905.	2.8	15
90	A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. Nature Genetics, 2008, 40, 322-328.	21.4	509

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91	Concurrent transposition of distal 6p and 20q to the 22q telomere: A recurrent benign chromosomal variant. European Journal of Medical Genetics, 2008, 51, 148-155.	1.3	2
92	A 12Mb deletion at 7q33â€“q35 associated with autism spectrum disorders and primary amenorrhea. European Journal of Medical Genetics, 2008, 51, 631-638.	1.3	68
93	Deletion of a 760 kb region at 4p16 determines the prenatal and postnatal growth retardation characteristic of Wolf-Hirschhorn syndrome. Journal of Medical Genetics, 2007, 44, 647-650.	3.2	21
94	Subtelomeric trisomy 21q: A new benign chromosomal variant. European Journal of Medical Genetics, 2007, 50, 54-59.	1.3	7
95	A large analphoid invdup(3)(q22.3qter) marker chromosome characterized by array-CGH in a child with malformations, mental retardation, ambiguous genitalia and Blaschko's lines. European Journal of Medical Genetics, 2007, 50, 264-273.	1.3	10
96	Socioeconomic status mediates the genetic contribution of the dopamine receptor D4 and serotonin transporter linked promoter region repeat polymorphisms to externalization in preadolescence. Development and Psychopathology, 2007, 19, 1147-1160.	2.3	62
97	Two classes of low-copy repeats comediate a new recurrent rearrangement consisting of duplication at 8p23.1 and triplication at 8p23.2. Human Mutation, 2007, 28, 459-468.	2.5	41
98	Overexpression of the C-type natriuretic peptide (CNP) is associated with overgrowth and bone anomalies in an individual with balanced t(2;7) translocation. Human Mutation, 2007, 28, 724-731.	2.5	118
99	DNA methylation regulates tissue-specific expression of Shank3. Journal of Neurochemistry, 2007, 101, 1380-1391.	3.9	67
100	Effect of the catechol-O-methyltransferase val158met genotype on children?s early phases of facial stimuli processing. Genes, Brain and Behavior, 2007, 6, 364-374.	2.2	14
101	Association of short-term memory with a variant within DYX1C1 in developmental dyslexia. Genes, Brain and Behavior, 2007, 6, 640-646.	2.2	79
102	The Italian Preadolescent Mental Health Project (PrISMA): rationale and methods. International Journal of Methods in Psychiatric Research, 2006, 15, 22-35.	2.1	63
103	A 46,X,inv(Y) young woman with gonadal dysgenesis and gonadoblastoma: Cytogenetics, molecular, and methylation studies. American Journal of Medical Genetics, Part A, 2006, 140A, 40-45.	1.2	15
104	A novel familial MECP2 mutation in a young boy: Clinical and molecular findings. Neurology, 2006, 67, 867-868.	1.1	6
105	Identification of a recurrent breakpoint within the SHANK3 gene in the 22q13.3 deletion syndrome. Journal of Medical Genetics, 2006, 43, 822-828.	3.2	155
106	Influence of the Serotonin Transporter Promoter Gene and Shyness on Childrenâ€™s Cerebral Responses to Facial Expressions. Archives of General Psychiatry, 2005, 62, 85.	12.3	169
107	A 2.3â€‰Mb duplication of chromosome 8q24.3 associated with severe mental retardation and epilepsy detected by standard karyotype. European Journal of Human Genetics, 2005, 13, 586-591.	2.8	45
108	A family-based association study does not support DYX1C1 on 15q21.3 as a candidate gene in developmental dyslexia. European Journal of Human Genetics, 2005, 13, 491-499.	2.8	81

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109	Molecular characterization of a t(2;6) balanced translocation that is associated with a complex phenotype and leads to truncation of theTCBA1gene. Human Mutation, 2005, 26, 426-436.	2.5	25
110	Loss-of-function mutation of the AF9/MLLT3 gene in a girl with neuromotor development delay, cerebellar ataxia, and epilepsy. Human Genetics, 2005, 118, 76-81.	3.8	27
111	Direct duplication 12p11.21â€“p13.31 mediated by segmental duplications: a new recurrent rearrangement?. Human Genetics, 2005, 118, 207-213.	3.8	6
112	Reciprocal translocations: a trap for cytogenetists?. Human Genetics, 2005, 117, 571-582.	3.8	54
113	Inversion polymorphisms and non-contiguous terminal deletions: the cause and the (unpredicted) effect of our genome architecture. Journal of Medical Genetics, 2005, 43, e19-e19.	3.2	27
114	O16: Few duplicons make theÂmost noise. European Journal of Medical Genetics, 2005, 48, 479-480.	1.3	0
115	A locus on 15q15-15qter influences dyslexia: further support from a transmission/disequilibrium study in an Italian speaking population. Journal of Medical Genetics, 2004, 41, 42-46.	3.2	37
116	Silencer elements as possible inhibitors of pseudoexon splicing. Nucleic Acids Research, 2004, 32, 1783-1791.	14.5	120
117	Selective disruption of muscle and brain-specific BPAG1 isoforms in a girl with a 6;15 translocation, cognitive and motor delay, and tracheo-oesophageal atresia. Journal of Medical Genetics, 2004, 41, e71-e71.	3.2	41
118	Inverted duplications: how many of them are mosaic?. European Journal of Human Genetics, 2004, 12, 713-717.	2.8	33
119	An Assessment of Transmission Disequilibrium Between Quantitative Measures of Childhood Problem Behaviors and DRD2/TaqI and DRD4/48bp-Repeat Polymorphisms. Behavior Genetics, 2004, 34, 495-502.	2.1	30
120	A case-control and family-based association study of the 5-HTTLPR in pediatric-onset depressive disorders. Biological Psychiatry, 2004, 56, 292-295.	1.3	42
121	Over-representation of exonic splicing enhancers in human intronless genes suggests multiple functions in mRNA processing. Biochemical and Biophysical Research Communications, 2004, 322, 470-476.	2.1	13
122	Relevance of sequence and structure elements for deletion events in the dystrophin gene major hot-spot. Human Genetics, 2003, 112, 272-288.	3.8	24
123	The 129 codon polymorphism of the Prion Protein gene influences earlier cognitive performance in Down syndrome subjects. Journal of Neurology, 2003, 250, 688-692.	3.6	29
124	No evidence for association and linkage disequilibrium between dyslexia and markers of four dopamine-related genes. European Child and Adolescent Psychiatry, 2003, 12, 198-202.	4.7	29
125	Trans -acting factors may cause dystrophin splicing misregulation in BMD skeletal muscles. FEBS Letters, 2003, 537, 30-34.	2.8	16
126	Molecular analysis of LGMD-2B and MM patients: identification of novel DYSF mutations and possible founder effect in the Italian population. Neuromuscular Disorders, 2003, 13, 788-795.	0.6	45

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127	Neocentromeres in 15q24-26 Map to Duplicons Which Flanked an Ancestral Centromere in 15q25. Genome Research, 2003, 13, 2059-2068.	5.5	107
128	Distal trisomy 6p and 20q owing to the concurrent transposition of distal 6p and 20q to the 22q telomere: a genomic polymorphism?. Journal of Medical Genetics, 2003, 40, 94e-94.	3.2	11
129	Unusual cognitive and behavioural profile in a Williams syndrome patient with atypical 7q11.23 deletion. Journal of Medical Genetics, 2003, 40, 526-530.	3.2	65
130	Synthesis of ribosomal proteins in developingDictyostelium discoideumcells is controlled by the methylation of proteins S24 and S31. Biochemistry and Cell Biology, 2002, 80, 261-270.	2.0	5
131	The dystrophin gene is alternatively spliced throughout its coding sequence. FEBS Letters, 2002, 517, 163-166.	2.8	33
132	Heterozygous Submicroscopic Inversions Involving Olfactory Receptorâ€“Gene Clusters Mediate the Recurrent t(4;8)(p16;p23) Translocation. American Journal of Human Genetics, 2002, 71, 276-285.	6.2	185
133	20â€Mb duplication of chromosome 9p in a girl with minimal physical findings and normal IQ: Narrowing of the 9p duplication critical region to 6 Mb. American Journal of Medical Genetics Part A, 2002, 112, 154-159.	2.4	19
134	Olfactory Receptorâ€“Gene Clusters, Genomic-Inversion Polymorphisms, and Common Chromosome Rearrangements. American Journal of Human Genetics, 2001, 68, 874-883.	6.2	338
135	Disruption of the ProSAP2 Gene in a t(12;22)(q24.1;q13.3) Is Associated with the 22q13.3 Deletion Syndrome. American Journal of Human Genetics, 2001, 69, 261-268.	6.2	273
136	CNGA3 Mutations in Hereditary Cone Photoreceptor Disorders. American Journal of Human Genetics, 2001, 69, 722-737.	6.2	294
137	Primary beta-sarcoglycanopathy manifesting as recurrent exercise-induced myoglobinuria. Neuromuscular Disorders, 2001, 11, 389-394.	0.6	33
138	Clinical and genetic analysis of a family with X-linked congenital nystagmus (NYS1). Ophthalmic Genetics, 2001, 22, 241-248.	1.2	32
139	Inverted duplications are recurrent rearrangements always associated with a distal deletion: description of a new case involving 2q. European Journal of Human Genetics, 2000, 8, 597-603.	2.8	66
140	Cell type specificity and mechanism of control of a gene may be reverted in different strains of Dictyostelium discoideum. Biochimica Et Biophysica Acta Gene Regulatory Mechanisms, 2000, 1492, 23-30.	2.4	1
141	Effects of Serotonin Transporter Promoter Genotype on Platelet Serotonin Transporter Functionality in Depressed Children and Adolescents. Journal of the American Academy of Child and Adolescent Psychiatry, 1999, 38, 1396-1402.	0.5	70
142	The cytoplasmic domain of rat NKR-P1 receptor interacts with the N-terminal domain of p56lck via cysteine residues. European Journal of Immunology, 1997, 27, 72-77.	2.9	33
143	Analysis of the Structure and Expression of the Augmenter of Liver Regeneration (ALR) Gene. Molecular Medicine, 1996, 2, 97-108.	4.4	86
144	A NEW XENOGENEIC RADIATION CHIMERA (HAMSTER-TO-RAT). Transplantation, 1994, 57, 1528-1531.	1.0	9

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145	Dinucleotide repeat polymorphism at the DXS1146 locus. Human Molecular Genetics, 1993, 2, 1078-1078.	2.9	2
146	Microsatellite repeat polymorphism at the D13S197 locus. Human Molecular Genetics, 1993, 2, 337-337.	2.9	4
147	Dinucleotide repeat polymorphisms at the D13S192 and D13S193 loci. Human Molecular Genetics, 1993, 2, 86-86.	2.9	4
148	A novel HLA-DR4 haplotype generated by a rare recombinational event between DRB1 and DQA1 loci. Immunogenetics, 1992, 36, 338-40.	2.4	12
149	Glutamic acid decarboxylase expression in islets and brain. Lancet, The, 1991, 338, 1469-1470.	13.7	25
150	Dictyostelium discoideum gene family contains a long internal amino acid repeat. Genesis, 1991, 12, 133-138.	2.1	6
151	Full length ? chain cDNAs of DQw9 and DQw8 molecules encode proteins that differ only at amino acid 57. Immunogenetics, 1991, 33, 404-408.	2.4	11
152	NKR-P1, a signal transduction molecule on natural killer cells. Science, 1990, 249, 1298-1300.	12.6	235
153	A shared internal threonine-glutamic acid-threonine-proline repeat defines a family of Dictyostelium discoideum spore germination specific proteins. Biochemistry, 1990, 29, 7264-7269.	2.5	46
154	Nucleotide sequences of Dictyostelium discoideum developmentally regulated cDNAs rich in (AAC) imply proteins that contain clusters of asparagine, glutamine, or threonine. Molecular Genetics and Genomics, 1989, 218, 453-459.	2.4	42
155	Molecular organization of developmentally regulated Dictyostelium discoideum ubiquitin cDNAs. Biochemistry, 1989, 28, 5226-5231.	2.5	44
156	Organization of a gene family developmentally regulated during Dictyostelium discoideum spore germination. Journal of Molecular Biology, 1989, 205, 63-69.	4.2	23
157	Rapid detection of IDDM susceptibility with HLA-DQ beta-alleles as markers. Diabetes, 1989, 38, 1617-1622.	0.6	11
158	Characterization of genes that are developmentally regulated duringDictyostelium discoideum spore germination. Genesis, 1988, 9, 303-313.	2.1	9
159	Structure of two developmentally regulated Dictyostelium discoideum ubiquitin genes.. Molecular and Cellular Biology, 1987, 7, 2097-2103.	2.3	46
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