

Corrado Romano

List of Publications by Year in descending order

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214
papers

12,701
citations

41258

49
h-index

28224

105
g-index

225
all docs

225
docs citations

225
times ranked

16165
citing authors

#	ARTICLE	IF	CITATIONS
1	GPR56 gene down-regulation in patients with Klinefelter Syndrome: a candidate for infertility?. <i>Minerva Endocrinology</i> , 2022, 46, .	0.6	0
2	Genetics and Clinical Neuroscience in Intellectual Disability. <i>Brain Sciences</i> , 2022, 12, 338.	1.1	4
3	The effect of laboratory-verified smoking on SARS-CoV-2 infection: results from the Troina sero-epidemiological survey. <i>Internal and Emergency Medicine</i> , 2022, 17, 1617-1630.	1.0	10
4	12q21 Interstitial Deletions: Seven New Syndromic Cases Detected by Array-CGH and Review of the Literature. <i>Genes</i> , 2022, 13, 780.	1.0	0
5	Clinical spectrum and follow-up in six individuals with Lambdâ€“Shaffer syndrome (<scp>SOX5</scp>). <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 608-613.	0.7	6
6	Milder presentation of TELO2-related syndrome in two sisters homozygous for the p.Arg609His pathogenic variant. <i>European Journal of Medical Genetics</i> , 2021, 64, 104116.	0.7	5
7	SOX13 gene downregulation in peripheral blood mononuclear cells of patients with Klinefelter syndrome. <i>Asian Journal of Andrology</i> , 2021, 23, 157.	0.8	0
8	mRNA expression profiling of mitochondrial subunits in subjects with Parkinsonâ€™s disease. <i>Archives of Medical Science</i> , 2021, , .	0.4	4
9	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. <i>Genome Medicine</i> , 2021, 13, 63.	3.6	50
10	8p23.2-pter Microdeletions: Seven New Cases Narrowing the Candidate Region and Review of the Literature. <i>Genes</i> , 2021, 12, 652.	1.0	11
11	The relevance of deep genomic analyses in families with variably expressive CNVs in the era of personalized medicine. <i>Molecular Genetics and Metabolism</i> , 2021, 132, S69.	0.5	0
12	Recommendations for neonatologists and pediatricians working in first level birthing centers on the first communication of genetic disease and malformation syndrome diagnosis: consensus issued by 6 Italian scientific societies and 4 parentsâ€™ associations. <i>Italian Journal of Pediatrics</i> , 2021, 47, 94.	1.0	25
13	Praderâ€“Willi Syndrome with Angelman Syndrome in the Offspring. <i>Medicina (Lithuania)</i> , 2021, 57, 460.	0.8	3
14	CCR3 gene overexpression in patients with Down syndrome. <i>Molecular Biology Reports</i> , 2021, 48, 5335-5338.	1.0	2
15	Seroepidemiological Survey on the Impact of Smoking on SARS-CoV-2 Infection and COVID-19 Outcomes: Protocol for the Troina Study. <i>JMIR Research Protocols</i> , 2021, 10, e32285.	0.5	4
16	Role of long non-coding RNAs in Down syndrome patients: a transcriptome analysis study. <i>Human Cell</i> , 2021, 34, 1662-1670.	1.2	4
17	A study of gene expression by RNA-seq in patients with prostate cancer and in patients with Parkinson disease: an example of inverse comorbidity. <i>Molecular Biology Reports</i> , 2021, 48, 7627-7631.	1.0	5
18	TBC1D24 gene mRNA expression in a boy with early infantile epileptic encephalopathy-16. <i>Acta Neurologica Belgica</i> , 2020, 120, 381-383.	0.5	3

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19	Long non-coding RNA GAS5 expression in patients with Down syndrome. <i>International Journal of Medical Sciences</i> , 2020, 17, 1315-1319.	1.1	4
20	De novo SMARCA2 variants clustered outside the helicase domain cause a new recognizable syndrome with intellectual disability and blepharophimosis distinct from Nicolaides-Baraitser syndrome. <i>Genetics in Medicine</i> , 2020, 22, 1838-1850.	1.1	31
21	Structural brain anomalies in Cri-du-Chat syndrome: MRI findings in 14 patients and possible genotype-phenotype correlations. <i>European Journal of Paediatric Neurology</i> , 2020, 28, 110-119.	0.7	3
22	Cerebellar degeneration-related protein 1 expression in fibroblasts of patients affected by down syndrome. <i>International Journal of Transgender Health</i> , 2020, 13, 548-555.	1.1	0
23	Study of the MDM2 -410T-G polymorphism (rs2279744) by pyrosequencing in mothers of Down Syndrome subjects. <i>Human Cell</i> , 2020, 33, 476-478.	1.2	2
24	Humanin gene expression in fibroblast of Down syndrome subjects. <i>International Journal of Medical Sciences</i> , 2020, 17, 320-324.	1.1	12
25	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. <i>Nature Communications</i> , 2020, 11, 4932.	5.8	105
26	GPR56 gene down-regulation in patients with Klinefelter syndrome: a candidate for infertility?. <i>Minerva Endocrinology</i> , 2020, , .	0.6	0
27	Consolidating the Role of TDP2 Mutations in Recessive Spinocerebellar Ataxia Associated with Pediatric Onset Drug Resistant Epilepsy and Intellectual Disability (SCAR23). <i>Cerebellum</i> , 2019, 18, 972-975.	1.4	12
28	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. <i>Nature Communications</i> , 2019, 10, 4679.	5.8	43
29	Disruptive variants of <i>CSDE1</i> associate with autism and interfere with neuronal development and synaptic transmission. <i>Science Advances</i> , 2019, 5, eaax2166.	4.7	35
30	De novo variants in FBXO11 cause a syndromic form of intellectual disability with behavioral problems and dysmorphisms. <i>European Journal of Human Genetics</i> , 2019, 27, 738-746.	1.4	32
31	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. <i>Frontiers in Genetics</i> , 2019, 10, 611.	1.1	14
32	Rare variants in the genetic background modulate cognitive and developmental phenotypes in individuals carrying disease-associated variants. <i>Genetics in Medicine</i> , 2019, 21, 816-825.	1.1	127
33	Evidence for long noncoding RNA GAS5 up-regulation in patients with Klinefelter syndrome. <i>BMC Medical Genetics</i> , 2019, 20, 4.	2.1	20
34	Mutations in ACTL6B, coding for a subunit of the neuron-specific chromatin remodeling complex nBAF, cause early onset severe developmental and epileptic encephalopathy with brain hypomyelination and cerebellar atrophy. <i>Human Genetics</i> , 2019, 138, 187-198.	1.8	12
35	Biallelic intragenic duplication in ADGRB3 (BAI3) gene associated with intellectual disability, cerebellar atrophy, and behavioral disorder. <i>European Journal of Human Genetics</i> , 2019, 27, 594-602.	1.4	15
36	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. <i>Biological Psychiatry</i> , 2019, 85, 287-297.	0.7	108

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37	The epilepsy phenotypic spectrum associated with a recurrent <i>CUX2</i> variant. <i>Annals of Neurology</i> , 2018, 83, 926-934.	2.8	20
38	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. <i>American Journal of Human Genetics</i> , 2018, 102, 985-994.	2.6	59
39	Mitochondrial mRNA expression in fibroblasts of Down syndrome subjects. <i>Human Cell</i> , 2018, 31, 179-181.	1.2	8
40	A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency. <i>European Journal of Human Genetics</i> , 2018, 26, 54-63.	1.4	32
41	Next Generation Sequencing expression profiling of mitochondrial subunits in men with Klinefelter syndrome. <i>International Journal of Medical Sciences</i> , 2018, 15, 31-35.	1.1	11
42	Expression of miR-132 in Down syndrome subjects. <i>Human Cell</i> , 2018, 31, 268-270.	1.2	0
43	Facies: the value of an old diagnostic tip in pediatric dermatology. <i>Giornale Italiano Di Dermatologia E Venereologia</i> , 2018, 153, 716-721.	0.8	0
44	The Methylenetetrahydrofolate Reductase C677T Polymorphism and Risk for Late-Onset Alzheimer's disease: Further Evidence in an Italian Multicenter Study. <i>Journal of Alzheimer's Disease</i> , 2017, 56, 1451-1457.	1.2	20
45	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. <i>Nature Genetics</i> , 2017, 49, 515-526.	9.4	443
46	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. <i>Nature Neuroscience</i> , 2017, 20, 1043-1051.	7.1	152
47	Searching for new pharmacological targets for the treatment of Alzheimer's disease in Down syndrome. <i>European Journal of Pharmacology</i> , 2017, 817, 7-19.	1.7	15
48	A polymorphism (rs1042522) in TP53 gene is a risk factor for Down Syndrome in Sicilian mothers. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2017, 30, 2752-2754.	0.7	2
49	Mutation spectrum of NF1 gene in Italian patients with neurofibromatosis type 1 using Ion Torrent PGM platform. <i>European Journal of Medical Genetics</i> , 2017, 60, 93-99.	0.7	30
50	Killer-specific secretory (Ksp37) gene expression in subjects with Down syndrome. <i>Neurological Sciences</i> , 2016, 37, 793-795.	0.9	5
51	An inflammatory and trophic disconnect biomarker profile revealed in Down syndrome plasma: Relation to cognitive decline and longitudinal evaluation. <i>Alzheimer's and Dementia</i> , 2016, 12, 1132-1148.	0.4	75
52	Low AMH levels as a marker of reduced ovarian reserve in young women affected by Down's syndrome. <i>Menopause</i> , 2016, 23, 1247-1251.	0.8	2
53	Disruptive de novo mutations of DYRK1A lead to a syndromic form of autism and ID. <i>Molecular Psychiatry</i> , 2016, 21, 126-132.	4.1	142
54	Expression of Phosphodiesterase 4B cAMP-specific Gene in Subjects With Cryptorchidism and Down's Syndrome. <i>Journal of Clinical Laboratory Analysis</i> , 2016, 30, 196-199.	0.9	3

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55	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. <i>American Journal of Human Genetics</i> , 2016, 98, 541-552.	2.6	132
56	MECP2 missense mutations outside the canonical MBD and TRD domains in males with intellectual disability. <i>Journal of Human Genetics</i> , 2016, 61, 95-101.	1.1	29
57	The Koolen-de Vries syndrome: a phenotypic comparison of patients with a 17q21.31 microdeletion versus a KANSL1 sequence variant. <i>European Journal of Human Genetics</i> , 2016, 24, 652-659.	1.4	108
58	A Multiplex PCR-Based Next-Generation Sequencing Approach Has Detected a Common Large Deletion in STS Gene in a Patient with X-Linked Ichthyosis. <i>Journal of Biomedical Science and Engineering</i> , 2016, 09, 337-341.	0.2	0
59	A novel splice acceptor site mutation in the ATP2A2 gene in a family with Darier disease. <i>Giornale Italiano Di Dermatologia E Venereologia</i> , 2016, 151, 582-5.	0.8	0
60	LDOC1 expression in fibroblasts of patients with Down syndrome. <i>Open Life Sciences</i> , 2015, 10, .	0.6	0
61	Recurrent duplications of 17q12 associated with variable phenotypes. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 3038-3045.	0.7	22
62	Response to Phelan K. et al.: Letter to the Editor Regarding Disciglio et al: Interstitial 22q13 deletions not involving <i>SHANK3</i> gene: A new contiguous gene syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1681-1681.	0.7	2
63	Targeted Next-Generation Sequencing Analysis of 1,000 Individuals with Intellectual Disability. <i>Human Mutation</i> , 2015, 36, 1197-1204.	1.1	161
64	Excess of runs of homozygosity is associated with severe cognitive impairment in intellectual disability. <i>Genetics in Medicine</i> , 2015, 17, 396-399.	1.1	19
65	NF-kB1 gene expression in Down syndrome patients. <i>Neurological Sciences</i> , 2015, 36, 1065-1066.	0.9	4
66	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. <i>American Journal of Human Genetics</i> , 2015, 97, 343-352.	2.6	230
67	Target sequencing approach intended to discover new mutations in non-syndromic intellectual disability. <i>Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis</i> , 2015, 781, 32-36.	0.4	10
68	In utero gene therapy rescues microcephaly caused by Pqbp1-hypofunction in neural stem progenitor cells. <i>Molecular Psychiatry</i> , 2015, 20, 459-471.	4.1	31
69	The transcriptional regulator <i>ADNP</i> links the BAF (SWI/SNF) complexes with autism. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2014, 166, 315-326.	0.7	68
70	Definition of 5q11.2 microdeletion syndrome reveals overlap with CHARGE syndrome and 22q11 deletion syndrome phenotypes. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2843-2848.	0.7	8
71	Gene expression profiling and qRT-PCR expression of RRP1B, PCNT, KIF21A and ADRB2 in leucocytes of Down TM s syndrome subjects. <i>Journal of Genetics</i> , 2014, 93, 18-23.	0.4	4
72	CASP3 protein expression by flow cytometry in Down TM s syndrome subjects. <i>Human Cell</i> , 2014, 27, 43-45.	1.2	2

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73	A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. <i>Nature Genetics</i> , 2014, 46, 380-384.	9.4	293
74	Definition of minimal duplicated region encompassing the <i>XIAP</i> and <i>STAG2</i> genes in the Xq25 microduplication syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1923-1930.	0.7	15
75	Disruption of the ASTN2/TRIM32 locus at 9q33.1 is a risk factor in males for autism spectrum disorders, ADHD and other neurodevelopmental phenotypes. <i>Human Molecular Genetics</i> , 2014, 23, 2752-2768.	1.4	140
76	Refining analyses of copy number variation identifies specific genes associated with developmental delay. <i>Nature Genetics</i> , 2014, 46, 1063-1071.	9.4	583
77	The MTRR 66A>G polymorphism and maternal risk of birth of a child with Down syndrome in Caucasian women: a case-control study and a meta-analysis. <i>Molecular Biology Reports</i> , 2014, 41, 5571-5583.	1.0	23
78	Disruptive CHD8 Mutations Define a Subtype of Autism Early in Development. <i>Cell</i> , 2014, 158, 263-276.	13.5	637
79	Interstitial 22q13 deletions not involving SHANK3 gene: A new contiguous gene syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1666-1676.	0.7	49
80	Increased FGF3 and FGF4 gene dosage is a risk factor for craniosynostosis. <i>Gene</i> , 2014, 534, 435-439.	1.0	19
81	6p22.3 deletion: report of a patient with autism, severe intellectual disability and electroencephalographic anomalies. <i>Molecular Cytogenetics</i> , 2013, 6, 4.	0.4	23
82	Identification of pathogenic gene variants in small families with intellectually disabled siblings by exome sequencing. <i>Journal of Medical Genetics</i> , 2013, 50, 802-811.	1.5	93
83	Poly (ADP-ribose) polymerase 1 expression in fibroblasts of Down syndrome subjects. <i>Open Medicine (Poland)</i> , 2013, 8, 762-765.	0.6	0
84	3q29 microdeletion syndrome: Cognitive and behavioral phenotype in four patients. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 3018-3022.	0.7	19
85	The duplication 17p13.3 phenotype: Analysis of 21 families delineates developmental, behavioral and brain abnormalities, and rare variant phenotypes. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 1833-1852.	0.7	53
86	The MTR 2756A>G polymorphism and maternal risk of birth of a child with Down syndrome: a case-control study and a meta-analysis. <i>Molecular Biology Reports</i> , 2013, 40, 6913-6925.	1.0	17
87	Pericentrin expression in Down's syndrome. <i>Neurological Sciences</i> , 2013, 34, 2023-2025.	0.9	5
88	SPAG5 mRNA is over-expressed in peripheral blood leukocytes of patients with Down's syndrome and cryptorchidism. <i>Neurological Sciences</i> , 2013, 34, 549-551.	0.9	8
89	KIF21A mRNA expression in patients with Down syndrome. <i>Neurological Sciences</i> , 2013, 34, 569-571.	0.9	7
90	Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. <i>European Journal of Human Genetics</i> , 2013, 21, 1074-1078.	1.4	64

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91	DNMT3B promoter polymorphisms and maternal risk of birth of a child with Down syndrome. <i>Human Reproduction</i> , 2013, 28, 545-550.	0.4	27
92	Rapid and accurate large-scale genotyping of duplicated genes and discovery of interlocus gene conversions. <i>Nature Methods</i> , 2013, 10, 903-909.	9.0	31
93	Multiplex ligation-dependent probe amplification detection of an unknown large deletion of the CREB-binding protein gene in a patient with Rubinstein-Taybi Syndrome. <i>Genetics and Molecular Research</i> , 2013, 12, 2809-15.	0.3	5
94	Expression of STRBP mRNA in patients with cryptorchidism and Down's syndrome. <i>Journal of Endocrinological Investigation</i> , 2012, 35, 5-7.	1.8	14
95	A de novo 8q22.2-24.3 duplication in a patient with mild phenotype. <i>European Journal of Medical Genetics</i> , 2012, 55, 67-70.	0.7	19
96	PTEN Gene: A Model for Genetic Diseases in Dermatology. <i>Scientific World Journal</i> , The, 2012, 2012, 1-8.	0.8	18
97	Expression of LDOC1 mRNA in leucocytes of patients with Down's syndrome. <i>Journal of Genetics</i> , 2012, 91, 95-98.	0.4	3
98	Expression of LDOC1 mRNA in leucocytes of patients with Down's syndrome. <i>Journal of Genetics</i> , 2012, 91, 95-8.	0.4	1
99	Gene expression profiling and qRT-PCR expression of RRP1B, PCNT, KIF21A and ADRB2 in leucocytes of Down's syndrome subjects. <i>Journal of Genetics</i> , 2012, 91, e18-23.	0.4	5
100	Definition of the neurological phenotype associated with dup (X)(p11.22-p11.23). <i>Epileptic Disorders</i> , 2011, 13, 240-251.	0.7	8
101	Differential expression of PARP1 mRNA in leucocytes of patients with Down's syndrome. <i>Journal of Genetics</i> , 2011, 90, 469-472.	0.4	6
102	Assessment of 2q23.1 Microdeletion Syndrome Implicates MBD5 as a Single Causal Locus of Intellectual Disability, Epilepsy, and Autism Spectrum Disorder. <i>American Journal of Human Genetics</i> , 2011, 89, 551-563.	2.6	195
103	The Pitt-Hopkins syndrome: Report of 16 new patients and clinical diagnostic criteria. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1536-1545.	0.7	55
104	Molecular Mechanisms Generating and Stabilizing Terminal 22q13 Deletions in 44 Subjects with Phelan/McDermid Syndrome. <i>PLoS Genetics</i> , 2011, 7, e1002173.	1.5	172
105	Relative Burden of Large CNVs on a Range of Neurodevelopmental Phenotypes. <i>PLoS Genetics</i> , 2011, 7, e1002334.	1.5	293
106	An unusual presentation of Becker Nevus. <i>European Journal of Dermatology</i> , 2010, 20, 522-523.	0.3	6
107	The Clinical Evaluation of Patients with Mental Retardation/Intellectual Disability. <i>Monographs in Human Genetics</i> , 2010, , 57-66.	0.5	3
108	Common pathological mutations in <i>PQBP1</i> induce nonsense-mediated mRNA decay and enhance exclusion of the mutant exon. <i>Human Mutation</i> , 2010, 31, 90-98.	1.1	18

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109	Novel <i>TMEM67</i> mutations and genotype-phenotype correlates in meckelin-related ciliopathies. <i>Human Mutation</i> , 2010, 31, n/a-n/a.	1.1	77
110	Decreased expression of <i>GRAF1/OPHN-1-L</i> in the X-linked alpha thalassemia mental retardation syndrome. <i>BMC Medical Genomics</i> , 2010, 3, 28.	0.7	12
111	The 2q23.1 microdeletion syndrome: clinical and behavioural phenotype. <i>European Journal of Human Genetics</i> , 2010, 18, 163-170.	1.4	71
112	A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. <i>Nature Genetics</i> , 2010, 42, 203-209.	9.4	539
113	Familial 1.1Mb deletion in chromosome Xq22.1 associated with mental retardation and behavioural disorders in female patients. <i>European Journal of Medical Genetics</i> , 2010, 53, 113-116.	0.7	20
114	The molecular landscape of <i>ASPM</i> mutations in primary microcephaly. <i>Journal of Medical Genetics</i> , 2009, 46, 249-253.	1.5	91
115	Expanding <i>CEP290</i> mutational spectrum in ciliopathies. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 2173-2180.	0.7	38
116	<i>MKS3/TMEM67</i> mutations are a major cause of COACH Syndrome, a Joubert Syndrome related disorder with liver involvement. <i>Human Mutation</i> , 2009, 30, E432-E442.	1.1	96
117	<i>SPANX-B</i> and <i>SPANX-C</i> (Xq27 region) gene dosage analysis in Down's syndrome subjects with undescended testes. <i>Journal of Genetics</i> , 2009, 88, 93-97.	0.4	3
118	15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. <i>Nature Genetics</i> , 2009, 41, 160-162.	9.4	511
119	Genome rearrangements in patients with blepharophimosis, mental retardation and hypothyroidism, so-called Young's Simpson syndrome. <i>Clinical Genetics</i> , 2009, 76, 210-213.	1.0	3
120	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. <i>American Journal of Human Genetics</i> , 2009, 85, 394-400.	2.6	60
121	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. <i>American Journal of Human Genetics</i> , 2009, 85, 419.	2.6	2
122	Fourteen new cases contribute to the characterization of the 7q11.23 microduplication syndrome. <i>European Journal of Medical Genetics</i> , 2009, 52, 94-100.	0.7	157
123	Further delineation of the 15q13 microdeletion and duplication syndromes: a clinical spectrum varying from non-pathogenic to a severe outcome. <i>Journal of Medical Genetics</i> , 2009, 46, 511-523.	1.5	250
124	Inflammatory bowel disease in children and adolescents in Italy: Data from the pediatric national IBD register (1996-2003). <i>Inflammatory Bowel Diseases</i> , 2008, 14, 1246-1252.	0.9	112
125	12q12 deletion: A new patient contributing to genotype-phenotype correlation. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 1354-1357.	0.7	10
126	A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. <i>Nature Genetics</i> , 2008, 40, 322-328.	9.4	509

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127	Identification of non-recurrent submicroscopic genome imbalances: the advantage of genome-wide microarrays over targeted approaches. <i>European Journal of Human Genetics</i> , 2008, 16, 395-400.	1.4	14
128	Three new patients with dup(17)(p11.2p11.2) without autism. <i>Clinical Genetics</i> , 2008, 73, 294-296.	1.0	7
129	Cerebriform plantar hyperplasia: the major cutaneous feature of Proteus syndrome. <i>International Journal of Dermatology</i> , 2008, 47, 374-376.	0.5	10
130	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. <i>New England Journal of Medicine</i> , 2008, 359, 1685-1699.	13.9	663
131	Further Delineation of Deletion 1p36 Syndrome in 60 Patients: A Recognizable Phenotype and Common Cause of Developmental Delay and Mental Retardation. <i>Pediatrics</i> , 2008, 121, 404-410.	1.0	233
132	Bannayan-Riley-Ruvalcaba Syndrome. , 2008, , 511-515.		0
133	Genetics of Pten Hamartoma Tumor Syndrome (PHTS). , 2008, , 483-489.		0
134	A New 6-bp SOX-3 Polyalanine Tract Deletion Does Not Segregate with Mental Retardation. <i>Genetic Testing and Molecular Biomarkers</i> , 2007, 11, 124-127.	1.7	7
135	Environmental influence on the worldwide prevalence of a 776C->G variant in the transcobalamin gene (TCN2). <i>Journal of Medical Genetics</i> , 2007, 44, 363-367.	1.5	33
136	Cryptic deletions are a common finding in "balanced" reciprocal and complex chromosome rearrangements: a study of 59 patients. <i>Journal of Medical Genetics</i> , 2007, 44, 750-762.	1.5	244
137	RSK2 enzymatic assay as a second level diagnostic tool in Coffin-Lowry syndrome. <i>Clinica Chimica Acta</i> , 2007, 384, 35-40.	0.5	8
138	Free and total leptin serum levels and soluble leptin receptors levels in two models of genetic obesity: the Prader-Willi and the Down syndromes. <i>Metabolism: Clinical and Experimental</i> , 2007, 56, 1076-1080.	1.5	32
139	The Italian XLMR bank: a clinical and molecular database. <i>Human Mutation</i> , 2007, 28, 13-18.	1.1	2
140	1.5 Mb de novo 22q11.21 microduplication in a patient with cognitive deficits and dysmorphic facial features. <i>Clinical Genetics</i> , 2007, 71, 177-182.	1.0	52
141	Schizophrenia in a patient with subtelomeric duplication of chromosome 22q. <i>Clinical Genetics</i> , 2007, 71, 599-601.	1.0	38
142	Prevalence of methylenetetrahydrofolate reductase 677T and 1298C alleles and folate status: a comparative study in Mexican, West African, and European populations. <i>American Journal of Clinical Nutrition</i> , 2006, 83, 701-707.	2.2	165
143	6q Terminal Deletion Syndrome Associated with a Distinctive EEG and Clinical Pattern: A Report of Five Cases. <i>Epilepsia</i> , 2006, 47, 830-838.	2.6	44
144	Mutations in CEP290, which encodes a centrosomal protein, cause pleiotropic forms of Joubert syndrome. <i>Nature Genetics</i> , 2006, 38, 623-625.	9.4	368

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145	A new chromosome 17q21.31 microdeletion syndrome associated with a common inversion polymorphism. <i>Nature Genetics</i> , 2006, 38, 999-1001.	9.4	418
146	Nail aplasia, microcephaly, severe mental retardation and MRI abnormalities: report of two unrelated cases. <i>Neurological Sciences</i> , 2006, 27, 425-431.	0.9	6
147	Hypersensitivity to Aromatic Anticonvulsants: In Vivo and In Vitro Cross-Reactivity Studies. <i>Current Pharmaceutical Design</i> , 2006, 12, 3373-3381.	0.9	46
148	Piezogenic pedal papules during Prader-Willi syndrome. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2005, 19, 136-137.	1.3	17
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