Corrado Romano

List of Publications by Year
in descending order

214 papers	12,701 citations	38742 50 h-index	28297 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?. Minerva Endocrinology, 2022, 46, .	1.1	0
2	Genetics and Clinical Neuroscience in Intellectual Disability. Brain Sciences, 2022, 12, 338.	2.3	4
3	The effect of laboratory-verified smoking on SARS-CoV-2 infection: results from the Troina sero-epidemiological survey. Internal and Emergency Medicine, 2022, 17, 1617-1630.	2.0	10
4	12q21 Interstitial Deletions: Seven New Syndromic Cases Detected by Array-CGH and Review of the Literature. Genes, 2022, 13, 780.	2.4	0
5	Clinical spectrum and followâ€up in six individuals with Lambâ€“Shaffer syndrome (<scp>SOX5</scp>). American Journal of Medical Genetics, Part A, 2021, 185, 608-613.	1.2	6
6	Milder presentation of TELO2-related syndrome in two sisters homozygous for the p.Arg609His pathogenic variant. European Journal of Medical Genetics, 2021, 64, 104116.	1.3	5
7	SOX13 gene downregulation in peripheral blood mononuclear cells of patients with Klinefelter syndrome. Asian Journal of Andrology, 2021, 23, 157.	1.6	0
8	mRNA expression profiling of mitochondrial subunits in subjects with Parkinsonâ€™s disease. Archives of Medical Science, 2021, , .	0.9	4
9	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	8.2	50
10	8p23.2-pter Microdeletions: Seven New Cases Narrowing the Candidate Region and Review of the Literature. Genes, 2021, 12, 652.	2.4	11
11	The relevance of deep genomic analyses in families with variably expressive CNVs in the era of personalized medicine. Molecular Genetics and Metabolism, 2021, 132, S69.	1.1	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. Italian Journal of Pediatrics, 2021, 47, 94.	2.6	25
13	Praderâ€“Willi Syndrome with Angelman Syndrome in the Offspring. Medicina (Lithuania), 2021, 57, 460.	2.0	3
14	CCR3 gene overexpression in patients with Down syndrome. Molecular Biology Reports, 2021, 48, 5335-5338.	2.3	2
15	Seroepidemiological Survey on the Impact of Smoking on SARS-CoV-2 Infection and COVID-19 Outcomes: Protocol for the Troina Study. JMIR Research Protocols, 2021, 10, e32285.	1.0	4
16	Role of long non-coding RNAs in Down syndrome patients: a transcriptome analysis study. Human Cell, 2021, 34, 1662-1670.	2.7	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. Molecular Biology Reports, 2021, 48, 7627-7631.	2.3	5
18	TBC1D24 gene mRNA expression in a boy with early infantile epileptic encephalopathy-16. Acta Neurologica Belgica, 2020, 120, 381-383.	1.1	3

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19	Long non-coding RNA GAS5 expression in patients with Down syndrome. International Journal of Medical Sciences, 2020, 17, 1315-1319.	2.5	4
20	De novo SMARCA2 variants clustered outside the helicase domain cause a new recognizable syndrome with intellectual disability and blepharophimosis distinct from Nicolaidesâ€“Baraitser syndrome. Genetics in Medicine, 2020, 22, 1838-1850.	2.4	31
21	Structural brain anomalies in Cri-du-Chat syndrome: MRI findings in 14 patients and possible genotype-phenotype correlations. European Journal of Paediatric Neurology, 2020, 28, 110-119.	1.6	3
22	Cerebellar degeneration-related protein 1 expression in fibroblasts of patients affected by down syndrome. International Journal of Transgender Health, 2020, 13, 548-555.	2.3	0
23	Study of the MDM2 -410T-G polymorphism (rs2279744) by pyrosequencing in mothers of Down Syndrome subjects. Human Cell, 2020, 33, 476-478.	2.7	2
24	Humanin gene expression in fibroblast of Down syndrome subjects. International Journal of Medical Sciences, 2020, 17, 320-324.	2.5	12
25	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	12.8	105
26	GPR56 gene down-regulation in patients with Klinefelter syndrome: a candidate for infertility?. Minerva Endocrinology, 2020, , .	1.1	0
27	Consolidating the Role of TDP2 Mutations in Recessive Spinocerebellar Ataxia Associated with Pediatric Onset Drug Resistant Epilepsy and Intellectual Disability (SCAR23). Cerebellum, 2019, 18, 972-975.	2.5	12
28	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	12.8	43
29	Disruptive variants of CSDE1 associate with autism and interfere with neuronal development and synaptic transmission. Science Advances, 2019, 5, eaax2166.	10.3	35
30	De novo variants in FBXO11 cause a syndromic form of intellectual disability with behavioral problems and dysmorphisms. European Journal of Human Genetics, 2019, 27, 738-746.	2.8	32
31	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	2.3	14
32	Rare variants in the genetic background modulate cognitive and developmental phenotypes in individuals carrying disease-associated variants. Genetics in Medicine, 2019, 21, 816-825.	2.4	127
33	Evidence for long noncoding RNA GAS5 up-regulationin patients with Klinefelter syndrome. BMC Medical Genetics, 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. Human Genetics, 2019, 138, 187-198.	3.8	12
35	Biallelic intragenic duplication in ADGRB3 (BAI3) gene associated with intellectual disability, cerebellar atrophy, and behavioral disorder. European Journal of Human Genetics, 2019, 27, 594-602.	2.8	15
36	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	1.3	108

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37	The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant. Annals of Neurology, 2018, 83, 926-934.	5.3	20
38	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. American Journal of Human Genetics, 2018, 102, 985-994.	6.2	59
39	Mitochondrial mRNA expression in fibroblasts of Down syndrome subjects. Human Cell, 2018, 31, 179-181.	2.7	8
40	A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency. European Journal of Human Genetics, 2018, 26, 54-63.	2.8	32
41	Next Generation Sequencing expression profiling of mitochondrial subunits in men with Klinefelter syndrome. International Journal of Medical Sciences, 2018, 15, 31-35.	2.5	11
42	Expression of miR-132 in Down syndrome subjects. Human Cell, 2018, 31, 268-270.	2.7	0
43	Facies: the value of an old diagnostic tip in pediatric dermatology. Giornale Italiano Di Dermatologia E Venereologia, 2018, 153, 716-721.	0.8	0
44	The Methylenetetrahydrofolate Reductase C677T Polymorphism and Risk for Late-Onset Alzheimerâ€™s disease: Further Evidence in an Italian Multicenter Study. Journal of Alzheimer's Disease, 2017, 56, 1451-1457.	2.6	20
45	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. Nature Genetics, 2017, 49, 515-526.	21.4	443
46	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. Nature Neuroscience, 2017, 20, 1043-1051.	14.8	152
47	Searching for new pharmacological targets for the treatment of Alzheimer's disease in Down syndrome. European Journal of Pharmacology, 2017, 817, 7-19.	3.5	15
48	A polymorphism (rs1042522) in TP53 gene is a risk factor for Down Syndrome in Sicilian mothers. Journal of Maternal-Fetal and Neonatal Medicine, 2017, 30, 2752-2754.	1.5	2
49	Mutation spectrum of NF1 gene in Italian patients with neurofibromatosis type 1 using Ion Torrent PGMâ„¢ platform. European Journal of Medical Genetics, 2017, 60, 93-99.	1.3	30
50	Killer-specific secretory (Ksp37) gene expression in subjects with Downâ€™s syndrome. Neurological Sciences, 2016, 37, 793-795.	1.9	5
51	An inflammatory and trophic disconnect biomarker profile revealed in Down syndrome plasma: Relation to cognitive decline and longitudinal evaluation. Alzheimer's and Dementia, 2016, 12, 1132-1148.	0.8	75
52	Low AMH levels as a marker of reduced ovarian reserve in young women affected by Down's syndrome. Menopause, 2016, 23, 1247-1251.	2.0	2
53	Disruptive de novo mutations of DYRK1A lead to a syndromic form of autism and ID. Molecular Psychiatry, 2016, 21, 126-132.	7.9	142
54	Expression of Phosphodiesterase 4B cAMPâ€Specific Gene in Subjects With Cryptorchidism and Down's Syndrome. Journal of Clinical Laboratory Analysis, 2016, 30, 196-199.	2.1	3

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55	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. American Journal of Human Genetics, 2016, 98, 541-552.	6.2	132
56	MECP2 missense mutations outside the canonical MBD and TRD domains in males with intellectual disability. Journal of Human Genetics, 2016, 61, 95-101.	2.3	29
57	The Koolen-de Vries syndrome: a phenotypic comparison of patients with a 17q21.31 microdeletion versus a KANSL1 sequence variant. European Journal of Human Genetics, 2016, 24, 652-659.	2.8	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. Journal of Biomedical Science and Engineering, 2016, 09, 337-341.	0.4	0
59	A novel splice acceptor site mutation in the ATP2A2 gene in a family with Darier disease. Giornale Italiano Di Dermatologia E Venereologia, 2016, 151, 582-5.	0.8	0
60	LDOC1 expression in fibroblasts of patients with Down syndrome. Open Life Sciences, 2015, 10, .	1.4	0
61	Recurrent duplications of 17q12 associated with variable phenotypes. American Journal of Medical Genetics, Part A, 2015, 167, 3038-3045.	1.2	22
62	Response to Phelan K. et al.: Letter to the Editor Regarding Disciglio et al: Interstitial 22q13 deletions not involving SHANK3 gene: A new contiguous gene syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1681-1681.	1.2	2
63	Targeted Nextâ€Generation Sequencing Analysis of 1,000 Individuals with Intellectual Disability. Human Mutation, 2015, 36, 1197-1204.	2.5	161
64	Excess of runs of homozygosity is associated with severe cognitive impairment in intellectual disability. Genetics in Medicine, 2015, 17, 396-399.	2.4	19
65	NF-kB1 gene expression in Down syndrome patients. Neurological Sciences, 2015, 36, 1065-1066.	1.9	4
66	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. American Journal of Human Genetics, 2015, 97, 343-352.	6.2	230
67	Target sequencing approach intended to discover new mutations in non-syndromic intellectual disability. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2015, 781, 32-36.	1.0	10
68	In utero gene therapy rescues microcephaly caused by Pqbp1-hypofunction in neural stem progenitor cells. Molecular Psychiatry, 2015, 20, 459-471.	7.9	31
69	The transcriptional regulator ADNP links the BAF (SWI/SNF) complexes with autism. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 315-326.	1.6	68
70	Definition of 5q11.2 microdeletion syndrome reveals overlap with CHARGE syndrome and 22q11 deletion syndrome phenotypes. American Journal of Medical Genetics, Part A, 2014, 164, 2843-2848.	1.2	8
71	Gene expression profiling and qRT-PCR expression of RRP1B, PCNT, KIF21A and ADRB2 in leucocytes of Downâ€™s syndrome subjects. Journal of Genetics, 2014, 93, 18-23.	0.7	4
72	CASP3 protein expression by flow cytometry in Downâ€™s syndrome subjects. Human Cell, 2014, 27, 43-45.	2.7	2

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73	A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. Nature Genetics, 2014, 46, 380-384.	21.4	293
74	Definition of minimal duplicated region encompassing the XIAP and STAG2 genes in the Xq25 microduplication syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 1923-1930.	1.2	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. Human Molecular Genetics, 2014, 23, 2752-2768.	2.9	140
76	Refining analyses of copy number variation identifies specific genes associated with developmental delay. Nature Genetics, 2014, 46, 1063-1071.	21.4	583
77	The MTRR 66A>G polymorphism and maternal risk of birth of a child with Down syndrome in Caucasian women: a caseâ€“control study and a meta-analysis. Molecular Biology Reports, 2014, 41, 5571-5583.	2.3	23
78	Disruptive CHD8 Mutations Define a Subtype of Autism Early in Development. Cell, 2014, 158, 263-276.	28.9	637
79	Interstitial 22q13 deletions not involving SHANK3 gene: A new contiguous gene syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 1666-1676.	1.2	49
80	Increased FGF3 and FGF4 gene dosage is a risk factor for craniosynostosis. Gene, 2014, 534, 435-439.	2.2	19
81	6p22.3 deletion: report of a patient with autism, severe intellectual disability and electroencephalographic anomalies. Molecular Cytogenetics, 2013, 6, 4.	0.9	23
82	Identification of pathogenic gene variants in small families with intellectually disabled siblings by exome sequencing. Journal of Medical Genetics, 2013, 50, 802-811.	3.2	93
83	Poly (ADP-ribose) polymerase 1 expression in fibroblasts of Down syndrome subjects. Open Medicine (Poland), 2013, 8, 762-765.	1.3	0
84	3q29 microdeletion syndrome: Cognitive and behavioral phenotype in four patients. American Journal of Medical Genetics, Part A, 2013, 161, 3018-3022.	1.2	19
85	The duplication 17p13.3 phenotype: Analysis of 21 families delineates developmental, behavioral and brain abnormalities, and rare variant phenotypes. American Journal of Medical Genetics, Part A, 2013, 161, 1833-1852.	1.2	53
86	The MTR 2756A>G polymorphism and maternal risk of birth of a child with Down syndrome: a caseâ€“control study and a meta-analysis. Molecular Biology Reports, 2013, 40, 6913-6925.	2.3	17
87	Pericentrin expression in Downâ€™s syndrome. Neurological Sciences, 2013, 34, 2023-2025.	1.9	5
88	SPAG5 mRNA is over-expressed in peripheral blood leukocytes of patients with Downâ€™s syndrome and cryptorchidism. Neurological Sciences, 2013, 34, 549-551.	1.9	8
89	KIF21A mRNA expression in patients with Down syndrome. Neurological Sciences, 2013, 34, 569-571.	1.9	7
90	Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. European Journal of Human Genetics, 2013, 21, 1074-1078.	2.8	64

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91	DNMT3B promoter polymorphisms and maternal risk of birth of a child with Down syndrome. Human Reproduction, 2013, 28, 545-550.	0.9	27
92	Rapid and accurate large-scale genotyping of duplicated genes and discovery of interlocus gene conversions. Nature Methods, 2013, 10, 903-909.	19.0	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. Genetics and Molecular Research, 2013, 12, 2809-15.	0.2	5
94	Expression of STRBP mRNA in patients with cryptorchidism and Downâ€™s syndrome. Journal of Endocrinological Investigation, 2012, 35, 5-7.	3.3	14
95	A de novo 8q22.2-24.3 duplication in a patient with mild phenotype. European Journal of Medical Genetics, 2012, 55, 67-70.	1.3	19
96	PTEN Gene: A Model for Genetic Diseases in Dermatology. Scientific World Journal, The, 2012, 2012, 1-8.	2.1	18
97	Expression of LDOC1 mRNA in leucocytes of patients with Downâ€™s syndrome. Journal of Genetics, 2012, 91, 95-98.	0.7	3
98	Expression of LDOC1 mRNA in leucocytes of patients with Down's syndrome. Journal of Genetics, 2012, 91, 95-8.	0.7	1
99	Gene expression profiling and qRT-PCR expression of RRP1B, PCNT, KIF21A and ADRB2 in leucocytes of Down's syndrome subjects. Journal of Genetics, 2012, 91, e18-23.	0.7	5
100	Definition of the neurological phenotype associated with dup (X)(p11.22-p11.23). Epileptic Disorders, 2011, 13, 240-251.	1.3	8
101	Differential expression of PARP1 mRNA in leucocytes of patients with Downâ€™s syndrome. Journal of Genetics, 2011, 90, 469-472.	0.7	6
102	Assessment of 2q23.1 Microdeletion Syndrome Implicates MBD5 as a Single Causal Locus of Intellectual Disability, Epilepsy, and Autism Spectrum Disorder. American Journal of Human Genetics, 2011, 89, 551-563.	6.2	195
103	The Pittâ€Hopkins syndrome: Report of 16 new patients and clinical diagnostic criteria. American Journal of Medical Genetics, Part A, 2011, 155, 1536-1545.	1.2	55
104	Molecular Mechanisms Generating and Stabilizing Terminal 22q13 Deletions in 44 Subjects with Phelan/McDermid Syndrome. PLoS Genetics, 2011, 7, e1002173.	3.5	172
105	Relative Burden of Large CNVs on a Range of Neurodevelopmental Phenotypes. PLoS Genetics, 2011, 7, e1002334.	3.5	293
106	An unusual presentation ofÂBecker Nevus. European Journal of Dermatology, 2010, 20, 522-523.	0.6	6
107	The Clinical Evaluation of Patients with Mental Retardation/Intellectual Disability. Monographs in Human Genetics, 2010, , 57-66.	0.5	3
108	Common pathological mutations inPQBP1induce nonsense-mediated mRNA decay and enhance exclusion of the mutant exon. Human Mutation, 2010, 31, 90-98.	2.5	18

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109	NovelTMEM67mutations and genotype-phenotype correlates in meckelin-related ciliopathies. Human Mutation, 2010, 31, n/a-n/a.	2.5	77
110	Decreased expression of GRAF1/OPHN-1-L in the X-linked alpha thalassemia mental retardation syndrome. BMC Medical Genomics, 2010, 3, 28.	1.5	12
111	The 2q23.1 microdeletion syndrome: clinical and behavioural phenotype. European Journal of Human Genetics, 2010, 18, 163-170.	2.8	71
112	A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. Nature Genetics, 2010, 42, 203-209.	21.4	539
113	Familial 1.1ÂMb deletion in chromosome Xq22.1 associated with mental retardation and behavioural disorders in female patients. European Journal of Medical Genetics, 2010, 53, 113-116.	1.3	20
114	The molecular landscape of ASPM mutations in primary microcephaly. Journal of Medical Genetics, 2009, 46, 249-253.	3.2	91
115	Expanding CEP290 mutational spectrum in ciliopathies. American Journal of Medical Genetics, Part A, 2009, 149A, 2173-2180.	1.2	38
116	MKS3/TMEM67mutations are a major cause of COACH Syndrome, a Joubert Syndrome related disorder with liver involvement. Human Mutation, 2009, 30, E432-E442.	2.5	96
117	SPANX-B and SPANX-C (Xq27 region) gene dosage analysis in Downâ€™s syndrome subjects with undescended testes. Journal of Genetics, 2009, 88, 93-97.	0.7	3
118	15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. Nature Genetics, 2009, 41, 160-162.	21.4	511
119	Genome rearrangements in patients with blepharophimosis, mental retardation and hypothyroidism, soâ€called Youngâ€Simpson syndrome. Clinical Genetics, 2009, 76, 210-213.	2.0	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. American Journal of Human Genetics, 2009, 85, 394-400.	6.2	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. American Journal of Human Genetics, 2009, 85, 419.	6.2	2
122	Fourteen new cases contribute to the characterization of the 7q11.23 microduplication syndrome. European Journal of Medical Genetics, 2009, 52, 94-100.	1.3	157
123	Further delineation of the 15q13 microdeletion and duplication syndromes: a clinical spectrum varying from non-pathogenic to a severe outcome. Journal of Medical Genetics, 2009, 46, 511-523.	3.2	250
124	Inflammatory bowel disease in children and adolescents in Italy: Data from the pediatric national IBD register (1996â€“2003). Inflammatory Bowel Diseases, 2008, 14, 1246-1252.	1.9	112
125	12q12 deletion: A new patient contributing to genotypeâ€“phenotype correlation. American Journal of Medical Genetics, Part A, 2008, 146A, 1354-1357.	1.2	10
126	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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127	Identification of non-recurrent submicroscopic genome imbalances: the advantage of genome-wide microarrays over targeted approaches. European Journal of Human Genetics, 2008, 16, 395-400.	2.8	14
128	Three new patients with dup(17)(p11.2p11.2) without autism. Clinical Genetics, 2008, 73, 294-296.	2.0	7
129	Cerebriform plantar hyperplasia: the major cutaneous feature of Proteus syndrome. International Journal of Dermatology, 2008, 47, 374-376.	1.0	10
130	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. New England Journal of Medicine, 2008, 359, 1685-1699.	27.0	663
131	Further Delineation of Deletion 1p36 Syndrome in 60 Patients: A Recognizable Phenotype and Common Cause of Developmental Delay and Mental Retardation. Pediatrics, 2008, 121, 404-410.	2.1	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. Genetic Testing and Molecular Biomarkers, 2007, 11, 124-127.	1.7	7
135	Environmental influence on the worldwide prevalence of a 776C->G variant in the transcobalamin gene (TCN2). Journal of Medical Genetics, 2007, 44, 363-367.	3.2	33
136	Cryptic deletions are a common finding in "balanced" reciprocal and complex chromosome rearrangements: a study of 59 patients. Journal of Medical Genetics, 2007, 44, 750-762.	3.2	244
137	RSK2 enzymatic assay as a second level diagnostic tool in Coffin-Lowry syndrome. Clinica Chimica Acta, 2007, 384, 35-40.	1.1	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. Metabolism: Clinical and Experimental, 2007, 56, 1076-1080.	3.4	32
139	The Italian XLMR bank: a clinical and molecular database. Human Mutation, 2007, 28, 13-18.	2.5	2
140	1.5 Mb de novo 22q11.21 microduplication in a patient with cognitive deficits and dysmorphic facial features. Clinical Genetics, 2007, 71, 177-182.	2.0	52
141	Schizophrenia in a patient with subtelomeric duplication of chromosome 22q. Clinical Genetics, 2007, 71, 599-601.	2.0	38
142	Prevalence of methylenetetrahydrofolate reductase 677T and 1298C alleles and folate status: a comparative study in Mexican, West African, and European populations. American Journal of Clinical Nutrition, 2006, 83, 701-707.	4.7	165
143	6q Terminal Deletion Syndrome Associated with a Distinctive EEG and Clinical Pattern: A Report of Five Cases. Epilepsia, 2006, 47, 830-838.	5.1	44
144	Mutations in CEP290, which encodes a centrosomal protein, cause pleiotropic forms of Joubert syndrome. Nature Genetics, 2006, 38, 623-625.	21.4	368

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145	A new chromosome 17q21.31 microdeletion syndrome associated with a common inversion polymorphism. Nature Genetics, 2006, 38, 999-1001.	21.4	418
146	Nail aplasia, microcephaly, severe mental retardation and MRI abnormalities: report of two unrelated cases. Neurological Sciences, 2006, 27, 425-431.	1.9	6
147	Hypersensitivity to Aromatic Anticonvulsants: In Vivo and In Vitro Cross-Reactivity Studies. Current Pharmaceutical Design, 2006, 12, 3373-3381.	1.9	46
148	Piezogenic pedal papules during Praderâ€“Willi syndrome. Journal of the European Academy of Dermatology and Venereology, 2005, 19, 136-137.	2.4	17
149	Alopecia areata in Down syndrome: a clinical evaluation. Journal of the European Academy of Dermatology and Venereology, 2005, 19, 769-770.	2.4	21
150	Skewed X-inactivation in a family with mental retardation and PQBP1 gene mutation. Clinical Genetics, 2005, 67, 446-447.	2.0	11
151	Seizures in patients with trisomy 21. American Journal of Medical Genetics Part A, 2005, 37, 298-300.	2.4	26
152	Identification of Novel Mutations in Patients with Coffinâ€“Lowry Syndrome by a Denaturing HPLC-Based Assay. Clinical Chemistry, 2005, 51, 2356-2358.	3.2	5
153	Homocysteine and related genetic polymorphisms in Down's syndrome IQ. Journal of Neurology, Neurosurgery and Psychiatry, 2005, 76, 706-709.	1.9	42
154	Denaturing HPLC-Based Assay for Detection of ATRX Gene Mutations. Clinical Chemistry, 2005, 51, 1314-1315.	3.2	2
155	Narrowing the Candidate Region for Congenital Diaphragmatic Hernia in Chromosome 15q26: Contradictory Results. American Journal of Human Genetics, 2005, 77, 892-894.	6.2	20
156	Methionine synthase (MTR) 2756 (Aâ€‰â†’â€‰G) polymorphism, double heterozygosity methionine synthase 2756 AG/methionine synthase reductase (MTRR) 66 AG, and elevated homocysteinemia are three risk factors for having a child with Down syndrome. American Journal of Medical Genetics, Part A, 2003, 121A, 219-224.	1.2	124
157	Medial Telangiectatic Sacral Nevi and Mca/Mr Syndromes. Pediatric Dermatology, 2003, 20, 370-371.	0.9	6
158	Skewed X chromosome inactivation in carriers is not a constant finding in FG syndrome. European Journal of Human Genetics, 2003, 11, 352-356.	2.8	3
159	Genetic Determinants of Folate and Vitamin B12 Metabolism: A Common Pathway in Neural Tube Defect and Down Syndrome?. Clinical Chemistry and Laboratory Medicine, 2003, 41, 1473-7.	2.3	66
160	A new MRXS locus maps to the X chromosome pericentromeric region: a new syndrome or narrow definition of Sutherland-Haan genetic locus?. Journal of Medical Genetics, 2002, 39, 276-280.	3.2	3
161	An Updated Survey on Skin Conditions in Down Syndrome. Dermatology, 2002, 205, 234-238.	2.1	72
162	Is there a relationship between zinc and the peculiar comorbidities of Down syndrome?. Down Syndrome Research and Practice, 2002, 8, 25-28.	0.3	10

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163	Allele Îµ4 of apolipoprotein E gene is less frequent in Down syndrome patient of the Sicilian population and has no influence on the grade of mental retardation. Neuroscience Letters, 2001, 306, 129-131.	2.1	18
164	An Additional Case of Macular Phylloid Mosaicism. Dermatology, 2001, 202, 73-73.	2.1	15
165	Prevalence and Clinical Picture of Celiac Disease in Italian Down Syndrome Patients: A Multicenter Study. Journal of Pediatric Gastroenterology and Nutrition, 2001, 33, 139-143.	1.8	114
166	Facial and Skeletal Malformations, Mental Retardation, Aganglionosis, and Neurogenic Muscle Weakness: A Variant of Niikawa-Kuroki Syndrome or a New Syndrome?. Journal of Child Neurology, 2001, 16, 296-296.	1.4	0
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