Corrado Romano

List of Publications by Year in Descending Order

206 papers	10,025 citations	48 h-index	97 g-index
225 ext. papers	11,755 ext. citations	6.3 avg, IF	4.72 L-index

#	Paper	IF	Citations
206	The effect of laboratory-verified smoking on SARS-CoV-2 infection: results from the Troina sero-epidemiological survey.. *Internal and Emergency Medicine*, **2022**, 1	3.7	2
205	12q21 Interstitial Deletions: Seven New Syndromic Cases Detected by Array-CGH and Review of the Literature. *Genes*, **2022**, 13, 780	4.2	
204	Clinical spectrum and follow-up in six individuals with Lamb-Shaffer syndrome (SOX5). *American Journal of Medical Genetics, Part A*, **2021**, 185, 608-613	2.5	0
203	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	2.8	1
202	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. *Genome Medicine*, **2021**, 13, 63	14.4	9
201	8p23.2-pter Microdeletions: Seven New Cases Narrowing the Candidate Region and Review of the Literature. *Genes*, **2021**, 12,	4.2	2
200	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 parents' associations. *Italian Journal of Pediatrics*, **2021**, 47, 94	3.2	5
199	Prader-Willi Syndrome with Angelman Syndrome in the Offspring. *Medicina (Lithuania)*, **2021**, 57,	3.1	2
198	CCR3 gene overexpression in patients with Down syndrome. *Molecular Biology Reports*, **2021**, 48, 5335-5338	2.8	1
197	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	2	3
196	Milder presentation of TELO2-related syndrome in two sisters homozygous for the p.Arg609His pathogenic variant. *European Journal of Medical Genetics*, **2021**, 64, 104116	2.6	1
195	gene downregulation in peripheral blood mononuclear cells of patients with Klinefelter syndrome. *Asian Journal of Andrology*, **2021**, 23, 157-162	2.8	
194	Role of long non-coding RNAs in Down syndrome patients: a transcriptome analysis study. *Human Cell*, **2021**, 34, 1662-1670	4.5	2
193	Study of the MDM2 -410T-G polymorphism (rs2279744) by pyrosequencing in mothers of Down Syndrome subjects. *Human Cell*, **2020**, 33, 476-478	4.5	
192	Humanin gene expression in fibroblast of Down syndrome subjects. *International Journal of Medical Sciences*, **2020**, 17, 320-324	3.7	7
191	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. *Nature Communications*, **2020**, 11, 4932	17.4	25
190	Long non-coding RNA GAS5 expression in patients with Down syndrome. *International Journal of Medical Sciences*, **2020**, 17, 1315-1319	3.7	2

189	De novo SMARCA2 variants clustered outside the helicase domain cause a new recognizable syndrome with intellectual disability and blepharophimosis distinct from Nicolaides-Baraitser syndrome. *Genetics in Medicine*, **2020**, 22, 1838-1850	8.1	8
188	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	3.8	2
187	Cerebellar degeneration-related protein 1 expression in fibroblasts of patients affected by down syndrome **2020**, 13, 548-555		
186	TBC1D24 gene mRNA expression in a boy with early infantile epileptic encephalopathy-16. *Acta Neurologica Belgica*, **2020**, 120, 381-383	1.5	2
185	Disruptive variants of associate with autism and interfere with neuronal development and synaptic transmission. *Science Advances*, **2019**, 5, eaax2166	14.3	16
184	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	5.3	11
183	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. *Frontiers in Genetics*, **2019**, 10, 611	4.5	7
182	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	4.3	10
181	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. *Nature Communications*, **2019**, 10, 4679	17.4	21
180	Rare variants in the genetic background modulate cognitive and developmental phenotypes in individuals carrying disease-associated variants. *Genetics in Medicine*, **2019**, 21, 816-825	8.1	71
179	Evidence for long noncoding RNA GAS5 up-regulationin patients with Klinefelter syndrome. *BMC Medical Genetics*, **2019**, 20, 4	2.1	13
178	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	6.3	6
177	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	5.3	8
176	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. *Biological Psychiatry*, **2019**, 85, 287-297	7.9	55
175	The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant. *Annals of Neurology*, **2018**, 83, 926-934	9.4	11
174	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	11	26
173	Mitochondrial mRNA expression in fibroblasts of Down syndrome subjects. *Human Cell*, **2018**, 31, 179-184	4.5	7
172	Expression of miR-132 in Down syndrome subjects. *Human Cell*, **2018**, 31, 268-270	4.5	

171	Facies: the value of an old diagnostic tip in pediatric dermatology. *Giornale Italiano Di Dermatologia E Venereologia*, **2018**, 153, 716-721	0.8	
170	A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency. *European Journal of Human Genetics*, **2018**, 26, 54-63	5.3	23
169	Next Generation Sequencing expression profiling of mitochondrial subunits in men with Klinefelter syndrome. *International Journal of Medical Sciences*, **2018**, 15, 31-35	3.7	7
168	The Methylenetetrahydrofolate Reductase C677T Polymorphism and Risk for Late-Onset Alzheimer's disease: Further Evidence in an Italian Multicenter Study. *Journal of Alzheimerls Disease*, **2017**, 56, 1451-1457	4.3	17
167	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. *Nature Genetics*, **2017**, 49, 515-526	36.3	283
166	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. *Nature Neuroscience*, **2017**, 20, 1043-1051	25.5	94
165	Searching for new pharmacological targets for the treatment of Alzheimer's disease in Down syndrome. *European Journal of Pharmacology*, **2017**, 817, 7-19	5.3	10
164	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	2	1
163	Mutation spectrum of NF1 gene in Italian patients with neurofibromatosis type 1 using Ion Torrent PGM platform. *European Journal of Medical Genetics*, **2017**, 60, 93-99	2.6	23
162	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, 652-9	5.3	57
161	Low AMH levels as a marker of reduced ovarian reserve in young women affected by Down's syndrome. *Menopause*, **2016**, 23, 1247-1251	2.5	2
160	Disruptive de novo mutations of DYRK1A lead to a syndromic form of autism and ID. *Molecular Psychiatry*, **2016**, 21, 126-32	15.1	90
159	Expression of Phosphodiesterase 4B cAMP-Specific Gene in Subjects With Cryptorchidism and Down's Syndrome. *Journal of Clinical Laboratory Analysis*, **2016**, 30, 196-9	3	3
158	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. *American Journal of Human Genetics*, **2016**, 98, 541-552	11	89
157	MECP2 missense mutations outside the canonical MBD and TRD domains in males with intellectual disability. *Journal of Human Genetics*, **2016**, 61, 95-101	4.3	18
156	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	0.7	
155	Killer-specific secretory (Ksp37) gene expression in subjects with Down's syndrome. *Neurological Sciences*, **2016**, 37, 793-5	3.5	4
154	An inflammatory and trophic disconnect biomarker profile revealed in Down syndrome plasma: Relation to cognitive decline and longitudinal evaluation. *Alzheimerls and Dementia*, **2016**, 12, 1132-1148	1.2	62

153	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	
152	NF-kB1 gene expression in Down syndrome patients. *Neurological Sciences*, **2015**, 36, 1065-6	3.5	4
151	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, 343-52	11	136
150	Target sequencing approach intended to discover new mutations in non-syndromic intellectual disability. *Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis*, **2015**, 781, 32-6	3.3	6
149	In utero gene therapy rescues microcephaly caused by Pqbp1-hypofunction in neural stem progenitor cells. *Molecular Psychiatry*, **2015**, 20, 459-71	15.1	22
148	Recurrent duplications of 17q12 associated with variable phenotypes. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 3038-45	2.5	17
147	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	2.5	1
146	Targeted Next-Generation Sequencing Analysis of 1,000 Individuals with Intellectual Disability. *Human Mutation*, **2015**, 36, 1197-204	4.7	122
145	Excess of runs of homozygosity is associated with severe cognitive impairment in intellectual disability. *Genetics in Medicine*, **2015**, 17, 396-9	8.1	16
144	A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. *Nature Genetics*, **2014**, 46, 380-4	36.3	197
143	Definition of minimal duplicated region encompassing the XIAP and STAG2 genes in the Xq25 microduplication syndrome. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 1923-30	2.5	12
142	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, 2752-68	5.6	104
141	Refining analyses of copy number variation identifies specific genes associated with developmental delay. *Nature Genetics*, **2014**, 46, 1063-71	36.3	429
140	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. *Molecular Biology Reports*, **2014**, 41, 5571-83	2.8	17
139	Disruptive CHD8 mutations define a subtype of autism early in development. *Cell*, **2014**, 158, 263-276	56.2	467
138	Interstitial 22q13 deletions not involving SHANK3 gene: a new contiguous gene syndrome. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 1666-76	2.5	36
137	Increased FGF3 and FGF4 gene dosage is a risk factor for craniosynostosis. *Gene*, **2014**, 534, 435-9	3.8	13
136	The transcriptional regulator ADNP links the BAF (SWI/SNF) complexes with autism. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2014**, 166C, 315-26	3.1	53

135	Definition of 5q11.2 microdeletion syndrome reveals overlap with CHARGE syndrome and 22q11 deletion syndrome phenotypes. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 2843-8	2.5	7
134	Gene expression profiling and qRT-PCR expression of RRP1B, PCNT, KIF21A and ADRB2 in leucocytes of Down syndrome subjects. *Journal of Genetics*, **2014**, 93, 18-23	1.2	1
133	CASP3 protein expression by flow cytometry in Down's syndrome subjects. *Human Cell*, **2014**, 27, 43-5	4.5	1
132	6p22.3 deletion: report of a patient with autism, severe intellectual disability and electroencephalographic anomalies. *Molecular Cytogenetics*, **2013**, 6, 4	2	18
131	Identification of pathogenic gene variants in small families with intellectually disabled siblings by exome sequencing. *Journal of Medical Genetics*, **2013**, 50, 802-11	5.8	70
130	Poly (ADP-ribose) polymerase 1 expression in fibroblasts of Down syndrome subjects. *Open Medicine (Poland)*, **2013**, 8, 762-765	2.2	
129	3q29 microdeletion syndrome: Cognitive and behavioral phenotype in four patients. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 3018-22	2.5	15
128	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**, 161A, 1833-52	2.5	42
127	The MTR 2756A>G polymorphism and maternal risk of birth of a child with Down syndrome: a case-control study and a meta-analysis. *Molecular Biology Reports*, **2013**, 40, 6913-25	2.8	12
126	Pericentrin expression in Down's syndrome. *Neurological Sciences*, **2013**, 34, 2023-5	3.5	2
125	SPAG5 mRNA is over-expressed in peripheral blood leukocytes of patients with Down's syndrome and cryptorchidism. *Neurological Sciences*, **2013**, 34, 549-51	3.5	8
124	KIF21A mRNA expression in patients with Down syndrome. *Neurological Sciences*, **2013**, 34, 569-71	3.5	6
123	Phenotypic spectrum and prevalence of INPP5E mutations in Joubert syndrome and related disorders. *European Journal of Human Genetics*, **2013**, 21, 1074-8	5.3	45
122	DNMT3B promoter polymorphisms and maternal risk of birth of a child with Down syndrome. *Human Reproduction*, **2013**, 28, 545-50	5.7	21
121	Rapid and accurate large-scale genotyping of duplicated genes and discovery of interlocus gene conversions. *Nature Methods*, **2013**, 10, 903-9	21.6	30
120	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	1.2	5
119	Expression of STRBP mRNA in patients with cryptorchidism and Down's syndrome. *Journal of Endocrinological Investigation*, **2012**, 35, 5-7	5.2	10
118	A de novo 8q22.2-24.3 duplication in a patient with mild phenotype. *European Journal of Medical Genetics*, **2012**, 55, 67-70	2.6	16

117	PTEN gene: a model for genetic diseases in dermatology. *Scientific World Journal, The*, **2012**, 2012, 252457	2.2	14
116	Expression of LDOC1 mRNA in leucocytes of patients with Down syndrome. *Journal of Genetics*, **2012**, 91, 95-98	1.2	3
115	Expression of LDOC1 mRNA in leucocytes of patients with Down's syndrome. *Journal of Genetics*, **2012**, 91, 95-8	1.2	1
114	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	1.2	5
113	Definition of the neurological phenotype associated with dup (X)(p11.22-p11.23). *Epileptic Disorders*, **2011**, 13, 240-51	1.9	6
112	Differential expression of PARP1 mRNA in leucocytes of patients with Down's syndrome. *Journal of Genetics*, **2011**, 90, 469-72	1.2	5
111	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, 551-63	11	166
110	The Pitt-Hopkins syndrome: report of 16 new patients and clinical diagnostic criteria. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1536-45	2.5	46
109	Molecular mechanisms generating and stabilizing terminal 22q13 deletions in 44 subjects with Phelan/McDermid syndrome. *PLoS Genetics*, **2011**, 7, e1002173	6	132
108	Relative burden of large CNVs on a range of neurodevelopmental phenotypes. *PLoS Genetics*, **2011**, 7, e1002334	6	232
107	The 2q23.1 microdeletion syndrome: clinical and behavioural phenotype. *European Journal of Human Genetics*, **2010**, 18, 163-70	5.3	65
106	A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. *Nature Genetics*, **2010**, 42, 203-9	36.3	461
105	Familial 1.1 Mb deletion in chromosome Xq22.1 associated with mental retardation and behavioural disorders in female patients. *European Journal of Medical Genetics*, **2010**, 53, 113-6	2.6	18
104	An unusual presentation of Becker Nevus. *European Journal of Dermatology*, **2010**, 20, 522-3	0.8	4
103	The Clinical Evaluation of Patients with Mental Retardation/Intellectual Disability. *Monographs in Human Genetics*, **2010**, 57-66		2
102	Common pathological mutations in PQBP1 induce nonsense-mediated mRNA decay and enhance exclusion of the mutant exon. *Human Mutation*, **2010**, 31, 90-8	4.7	14
101	Novel TMEM67 mutations and genotype-phenotype correlates in meckelin-related ciliopathies. *Human Mutation*, **2010**, 31, E1319-31	4.7	57
100	Decreased expression of GRAF1/OPHN-1-L in the X-linked alpha thalassemia mental retardation syndrome. *BMC Medical Genomics*, **2010**, 3, 28	3.7	10

99	The molecular landscape of ASPM mutations in primary microcephaly. *Journal of Medical Genetics*, **2009**, 46, 249-53	5.8	76
98	Expanding CEP290 mutational spectrum in ciliopathies. *American Journal of Medical Genetics, Part A* , **2009**, 149A, 2173-80	2.5	24
97	MKS3/TMEM67 mutations are a major cause of COACH Syndrome, a Joubert Syndrome related disorder with liver involvement. *Human Mutation*, **2009**, 30, E432-42	4.7	72
96	SPANX-B and SPANX-C (Xq27 region) gene dosage analysis in Down's syndrome subjects with undescended testes. *Journal of Genetics*, **2009**, 88, 93-7	1.2	1
95	15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. *Nature Genetics*, **2009**, 41, 160-2	36.3	454
94	Genome rearrangements in patients with blepharophimosis, mental retardation and hypothyroidism, so-called Young-Simpson syndrome. *Clinical Genetics*, **2009**, 76, 210-3	4	3
93	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	11	56
92	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	11	2
91	Fourteen new cases contribute to the characterization of the 7q11.23 microduplication syndrome. *European Journal of Medical Genetics*, **2009**, 52, 94-100	2.6	132
90	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, 511-23	5.8	226
89	A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. *Nature Genetics*, **2008**, 40, 322-8	36.3	463
88	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	5.3	12
87	Three new patients with dup(17)(p11.2p11.2) without autism. *Clinical Genetics*, **2008**, 73, 294-6	4	7
86	Cerebriform plantar hyperplasia: the major cutaneous feature of Proteus syndrome. *International Journal of Dermatology*, **2008**, 47, 374-6	1.7	8
85	Recurrent rearrangements of chromosome 1q21.1 and variable pediatric phenotypes. *New England Journal of Medicine*, **2008**, 359, 1685-99	59.2	587
84	Further delineation of deletion 1p36 syndrome in 60 patients: a recognizable phenotype and common cause of developmental delay and mental retardation. *Pediatrics*, **2008**, 121, 404-10	7.4	197
83	Inflammatory bowel disease in children and adolescents in Italy: data from the pediatric national IBD register (1996-2003). *Inflammatory Bowel Diseases*, **2008**, 14, 1246-52	4.5	97
82	12q12 deletion: a new patient contributing to genotype-phenotype correlation. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1354-7	2.5	10

81	Bannayan-Riley-Ruvalcaba Syndrome **2008**, 511-515		
80	Genetics of Pten Hamartoma Tumor Syndrome (PHTS) **2008**, 483-489		
79	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, 1076-80	13.7	27
78	The Italian XLMR bank: a clinical and molecular database. *Human Mutation*, **2007**, 28, 13-8	4.7	2
77	1.5 Mb de novo 22q11.21 microduplication in a patient with cognitive deficits and dysmorphic facial features. *Clinical Genetics*, **2007**, 71, 177-82	4	48
76	Schizophrenia in a patient with subtelomeric duplication of chromosome 22q. *Clinical Genetics*, **2007**, 71, 599-601	4	32
75	A new 6-bp SOX-3 polyalanine tract deletion does not segregate with mental retardation. *Genetic Testing and Molecular Biomarkers*, **2007**, 11, 124-7		6
74	Environmental influence on the worldwide prevalence of a 776C->G variant in the transcobalamin gene (TCN2). *Journal of Medical Genetics*, **2007**, 44, 363-7	5.8	31
73	Cryptic deletions are a common finding in "balanced" reciprocal and complex chromosome rearrangements: a study of 59 patients. *Journal of Medical Genetics*, **2007**, 44, 750-62	5.8	206
72	RSK2 enzymatic assay as a second level diagnostic tool in Coffin-Lowry syndrome. *Clinica Chimica Acta*, **2007**, 384, 35-40	6.2	5
71	Hypersensitivity to aromatic anticonvulsants: in vivo and in vitro cross-reactivity studies. *Current Pharmaceutical Design*, **2006**, 12, 3373-81	3.3	37
70	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, 701-7	7	142
69	6q terminal deletion syndrome associated with a distinctive EEG and clinical pattern: a report of five cases. *Epilepsia*, **2006**, 47, 830-8	6.4	39
68	Mutations in CEP290, which encodes a centrosomal protein, cause pleiotropic forms of Joubert syndrome. *Nature Genetics*, **2006**, 38, 623-5	36.3	320
67	A new chromosome 17q21.31 microdeletion syndrome associated with a common inversion polymorphism. *Nature Genetics*, **2006**, 38, 999-1001	36.3	355
66	Nail aplasia, microcephaly, severe mental retardation and MRI abnormalities: report of two unrelated cases. *Neurological Sciences*, **2006**, 27, 425-31	3.5	2
65	Narrowing the candidate region for congenital diaphragmatic hernia in chromosome 15q26: contradictory results. *American Journal of Human Genetics*, **2005**, 77, 892-4; author reply 894-5	11	20
64	Piezogenic pedal papules during Prader-Willi syndrome. *Journal of the European Academy of Dermatology and Venereology*, **2005**, 19, 136-7	4.6	14

63	Alopecia areata in Down syndrome: a clinical evaluation. *Journal of the European Academy of Dermatology and Venereology*, **2005**, 19, 769-70	4.6	14
62	Skewed X-inactivation in a family with mental retardation and PQBP1 gene mutation. *Clinical Genetics*, **2005**, 67, 446-7	4	11
61	Identification of novel mutations in patients with Coffin-Lowry syndrome by a denaturing HPLC-based assay. *Clinical Chemistry*, **2005**, 51, 2356-8	5.5	5
60	Homocysteine and related genetic polymorphisms in Down's syndrome IQ. *Journal of Neurology, Neurosurgery and Psychiatry*, **2005**, 76, 706-9	5.5	32
59	Denaturing HPLC-based assay for detection of ATRX gene mutations. *Clinical Chemistry*, **2005**, 51, 1314-5	5.5	2
58	Methionine synthase (MTR) 2756 (A --> 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 **2003**, 121A, 219-24		102
57	Medial telangiectatic sacral nevi and MCA/MR syndromes. *Pediatric Dermatology*, **2003**, 20, 370-1	1.9	4
56	Skewed X chromosome inactivation in carriers is not a constant finding in FG syndrome. *European Journal of Human Genetics*, **2003**, 11, 352-6	5.3	3
55	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	5.9	58
54	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, 276-80	5.8	3
53	An updated survey on skin conditions in Down syndrome. *Dermatology*, **2002**, 205, 234-8	4.4	66
52	Is there a relationship between zinc and the peculiar comorbidities of Down syndrome?. *Down Syndrome Research and Practice*, **2002**, 8, 25-8		7
51	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, 296-8	2.5	
50	Rubinstein-Taybi syndrome with epidermal nevus: a case report. *Pediatric Dermatology*, **2001**, 18, 34-7	1.9	7
49	Prenatal diagnosis of ATR-X syndrome in a fetus with a new G>T splicing mutation in the XNP/ATR-X gene. *Prenatal Diagnosis*, **2001**, 21, 747-51	3.2	15
48	Neuroendocrine features of pubertal development in females with mental retardation. *Gynecological Endocrinology*, **2001**, 15, 178-183	2.4	6
47	How microsatellite analysis can be exploited for subtelomeric chromosomal rearrangement analysis in mental retardation. *Journal of Medical Genetics*, **2001**, 38, E1	5.8	18
46	Allele varepsilon4 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, 129-31	3.3	15

45	An additional case of macular phylloid mosaicism. *Dermatology*, **2001**, 202, 73	4.4	11
44	Prevalence and clinical picture of celiac disease in italian down syndrome patients: a multicenter study. *Journal of Pediatric Gastroenterology and Nutrition*, **2001**, 33, 139-43	2.8	95
43	Cryptic telomeric rearrangements in subjects with mental retardation associated with dysmorphism and congenital malformations. *Journal of Medical Genetics*, **2001**, 38, 417-20	5.8	92
42	Neuroendocrine features of pubertal development in females with mental retardation. *Gynecological Endocrinology*, **2001**, 15, 178-183	2.4	
41	Cardiovascular malformations and other cardiovascular abnormalities in neurofibromatosis 1. *American Journal of Medical Genetics Part A*, **2000**, 95, 108-17		176
40	Medial telangiectatic sacral nevi (Types A and C) associated with Williams syndrome. *Dermatology*, **2000**, 201, 285-6	4.4	6
39	PTEN mutation spectrum and genotype-phenotype correlations in Bannayan-Riley-Ruvalcaba syndrome suggest a single entity with Cowden syndrome. *Human Molecular Genetics*, **1999**, 8, 1461-72	5.6	483
38	Cardiofaciocutaneous (CFC) syndrome. *Australasian Journal of Dermatology*, **1999**, 40, 111-3	1.3	6
37	Sulphation deficit in "low-functioning" autistic children: a pilot study. *Biological Psychiatry*, **1999**, 46, 420-4	7.9	104
36	Progressive cribriform and zosteriform hyperpigmentation: the late-onset feature of linear and whorled nevoid hypermelanosis associated with congenital neurological, skeletal and cutaneous anomalies. *Dermatology*, **1999**, 199, 72-3	4.4	13
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