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	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. New England Journal of Medicine, 2008, 359, 1685-1699.	27.0	663
2	Disruptive CHD8 Mutations Define a Subtype of Autism Early in Development. Cell, 2014, 158, 263-276.	28.9	637
3	Refining analyses of copy number variation identifies specific genes associated with developmental delay. Nature Genetics, 2014, 46, 1063-1071.	21.4	583
4	PTEN Mutation Spectrum and Genotype-Phenotype Correlations in Bannayan-Riley-Ruvalcaba Syndrome Suggest a Single Entity With Cowden Syndrome. Human Molecular Genetics, 1999, 8, 1461-1472.	2.9	562
5	A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. Nature Genetics, 2010, 42, 203-209.	21.4	539
6	15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. Nature Genetics, 2009, 41, 160-162.	21.4	511
7	A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. Nature Genetics, 2008, 40, 322-328.	21.4	509
8	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. Nature Genetics, 2017, 49, 515-526.	21.4	443
9	A new chromosome 17q21.31 microdeletion syndrome associated with a common inversion polymorphism. Nature Genetics, 2006, 38, 999-1001.	21.4	418
10	Mutations in CEP290, which encodes a centrosomal protein, cause pleiotropic forms of Joubert syndrome. Nature Genetics, 2006, 38, 623-625.	21.4	368
11	Relative Burden of Large CNVs on a Range of Neurodevelopmental Phenotypes. PLoS Genetics, 2011, 7, e1002334.	3.5	293
12	A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. Nature Genetics, 2014, 46, 380-384.	21.4	293
13	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
14	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
15	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
16	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
17	Cardiovascular malformations and other cardiovascular abnormalities in neurofibromatosis 1. American Journal of Medical Genetics Part A, 2000, 95, 108-117.	2.4	214
18	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

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19	Molecular Mechanisms Generating and Stabilizing Terminal 22q13 Deletions in 44 Subjects with Phelan/McDermid Syndrome. PLoS Genetics, 2011, 7, e1002173.	3.5	172
20	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
21	Targeted Nextâ€Generation Sequencing Analysis of 1,000 Individuals with Intellectual Disability. Human Mutation, 2015, 36, 1197-1204.	2.5	161
22	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
23	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. Nature Neuroscience, 2017, 20, 1043-1051.	14.8	152
24	Disruptive de novo mutations of DYRK1A lead to a syndromic form of autism and ID. Molecular Psychiatry, 2016, 21, 126-132.	7.9	142
25	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
26	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. American Journal of Human Genetics, 2016, 98, 541-552.	6.2	132
27	Molecular analysis of aldolase B genes in hereditary fructose intolerance. Lancet, The, 1990, 335, 306-309.	13.7	127
28	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
29	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
30	Sulphation deficit in â€œlow-functioningâ€•autistic children: a pilot study. Biological Psychiatry, 1999, 46, 420-424.	1.3	123
31	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
32	Cryptic telomeric rearrangements in subjects with mental retardation associated with dysmorphism and congenital malformations. Journal of Medical Genetics, 2001, 38, 417-420.	3.2	114
33	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
34	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
35	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	1.3	108
36	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	12.8	105

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37	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
38	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
39	The molecular landscape of ASPM mutations in primary microcephaly. Journal of Medical Genetics, 2009, 46, 249-253.	3.2	91
40	NovelTMEM67mutations and genotype-phenotype correlates in meckelin-related ciliopathies. Human Mutation, 2010, 31, n/a-n/a.	2.5	77
41	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
42	An Updated Survey on Skin Conditions in Down Syndrome. Dermatology, 2002, 205, 234-238.	2.1	72
43	The 2q23.1 microdeletion syndrome: clinical and behavioural phenotype. European Journal of Human Genetics, 2010, 18, 163-170.	2.8	71
44	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
45	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
46	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
47	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
48	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
49	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
50	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
51	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
52	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	8.2	50
53	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
54	Hypersensitivity to Aromatic Anticonvulsants: In Vivo and In Vitro Cross-Reactivity Studies. Current Pharmaceutical Design, 2006, 12, 3373-3381.	1.9	46

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55	A gene for FG syndrome maps in the Xq12-q21.31 region. , 1997, 73, 87-90.		45
56	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
57	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	12.8	43
58	Perforating Miliaâ€like Idiopathic Calcinosis Cutis and Periorbital Syringomas in a Girl With Down Syndrome. Pediatric Dermatology, 1994, 11, 258-260.	0.9	42
59	Homocysteine and related genetic polymorphisms in Down's syndrome IQ. Journal of Neurology, Neurosurgery and Psychiatry, 2005, 76, 706-709.	1.9	42
60	Schizophrenia in a patient with subtelomeric duplication of chromosome 22q. Clinical Genetics, 2007, 71, 599-601.	2.0	38
61	Expanding CEP290 mutational spectrum in ciliopathies. American Journal of Medical Genetics, Part A, 2009, 149A, 2173-2180.	1.2	38
62	LOCALIZED ELASTOSIS PERFORANS SERPIGINOSA IN A BOY WITH DOWN SYNDROME. Pediatric Dermatology, 1997, 14, 244-246.	0.9	36
63	Disruptive variants of CSDE1 associate with autism and interfere with neuronal development and synaptic transmission. Science Advances, 2019, 5, eaax2166.	10.3	35
64	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
65	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
66	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
67	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
68	Rapid and accurate large-scale genotyping of duplicated genes and discovery of interlocus gene conversions. Nature Methods, 2013, 10, 903-909.	19.0	31
69	In utero gene therapy rescues microcephaly caused by Pqbp1-hypofunction in neural stem progenitor cells. Molecular Psychiatry, 2015, 20, 459-471.	7.9	31
70	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
71	Celiac Disease in Down's Syndrome with HLA Serological and Molecular Studies. Journal of Pediatric Gastroenterology and Nutrition, 1996, 23, 303-306.	1.8	31
72	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

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73	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
74	DNMT3B promoter polymorphisms and maternal risk of birth of a child with Down syndrome. Human Reproduction, 2013, 28, 545-550.	0.9	27
75	Seizures in patients with trisomy 21. American Journal of Medical Genetics Part A, 2005, 37, 298-300.	2.4	26
76	How microsatellite analysis can be exploited for subtelomeric chromosomal rearrangement analysis in mental retardation. Journal of Medical Genetics, 2001, 38, e1-e1.	3.2	25
77	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
78	Ichthyosis and neutral lipid storage disease. American Journal of Medical Genetics Part A, 1988, 29, 377-382.	2.4	24
79	6p22.3 deletion: report of a patient with autism, severe intellectual disability and electroencephalographic anomalies. Molecular Cytogenetics, 2013, 6, 4.	0.9	23
80	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
81	Recurrent duplications of 17q12 associated with variable phenotypes. American Journal of Medical Genetics, Part A, 2015, 167, 3038-3045.	1.2	22
82	A prevalence study of celiac disease in persons with Down syndrome residing in the United States of America. Acta Paediatrica, International Journal of Paediatrics, 1999, 88, 953-956.	1.5	22
83	Alopecia areata in Down syndrome: a clinical evaluation. Journal of the European Academy of Dermatology and Venereology, 2005, 19, 769-770.	2.4	21
84	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
85	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
86	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
87	The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant. Annals of Neurology, 2018, 83, 926-934.	5.3	20
88	Evidence for long noncoding RNA GAS5 up-regulationin patients with Klinefelter syndrome. BMC Medical Genetics, 2019, 20, 4.	2.1	20
89	Prevalence of atopic dermatitis in patients with Down syndrome: A clinical survey. Journal of the American Academy of Dermatology, 1997, 36, 1019-1021.	1.2	19
90	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

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91	3q29 microdeletion syndrome: Cognitive and behavioral phenotype in four patients. American Journal of Medical Genetics, Part A, 2013, 161, 3018-3022.	1.2	19
92	Increased FGF3 and FGF4 gene dosage is a risk factor for craniosynostosis. Gene, 2014, 534, 435-439.	2.2	19
93	Excess of runs of homozygosity is associated with severe cognitive impairment in intellectual disability. Genetics in Medicine, 2015, 17, 396-399.	2.4	19
94	SKIN-PICKING: THE BEST CUTANEOUS FEATURE IN THE RECOGNIZATION OF PRADER-WILLI SYNDROME. International Journal of Dermatology, 1994, 33, 866-867.	1.0	18
95	A further family with epilepsy, dementia and yellow teeth: the KohlschÃ¼tter syndrome. Brain and Development, 1995, 17, 133-138.	1.1	18
96	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
97	Common pathological mutations inPQBP1induce nonsense-mediated mRNA decay and enhance exclusion of the mutant exon. Human Mutation, 2010, 31, 90-98.	2.5	18
98	PTEN Gene: A Model for Genetic Diseases in Dermatology. Scientific World Journal, The, 2012, 2012, 1-8.	2.1	18
99	Biochemical diagnosis and outcome of 2 years treatment in a patient with combined methylmalonic aciduria and homocystinuria. European Journal of Pediatrics, 1992, 151, 818-820.	2.7	17
100	Milia-like idiopathic calcinosis cutis: an unusual dermatosis associated with Down syndrome. British Journal of Dermatology, 1996, 134, 143-146.	1.5	17
101	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, 747-751.	2.3	17
102	Piezogenic pedal papules during Praderâ€“Willi syndrome. Journal of the European Academy of Dermatology and Venereology, 2005, 19, 136-137.	2.4	17
103	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
104	An Additional Case of Macular Phylloid Mosaicism. Dermatology, 2001, 202, 73-73.	2.1	15
105	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
106	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
107	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
108	The fragile X in sicily: An epidemiological survey. American Journal of Medical Genetics Part A, 1988, 30, 665-672.	2.4	14

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109	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, 72-73.	2.1	14
110	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
111	Expression of STRBP mRNA in patients with cryptorchidism and Downâ€™s syndrome. Journal of Endocrinological Investigation, 2012, 35, 5-7.	3.3	14
112	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	2.3	14
113	An intriguing case of LEOPARD syndrome.. Pediatric Dermatology, 1998, 15, 125-128.	0.9	13
114	Phenotypic and phoniatric findings in mosaic cri du chat syndrome. American Journal of Medical Genetics Part A, 1991, 39, 391-395.	2.4	12
115	Basal body temperature curves and endocrine pattern of menstrual cycles in Down syndrome. Gynecological Endocrinology, 1996, 10, 133-137.	1.7	12
116	CUTANEOUS FINDINGS IN THE MENTALLY RETARDED. International Journal of Dermatology, 1996, 35, 317-322.	1.0	12
117	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
118	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
119	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
120	Humanin gene expression in fibroblast of Down syndrome subjects. International Journal of Medical Sciences, 2020, 17, 320-324.	2.5	12
121	An Intronic Deletion Leading to Skipping of Exon 21 ofCol1a2in a Boy with Mild Osteogenesis Imperfecta. Connective Tissue Research, 1993, 29, 31-40.	2.3	11
122	Skewed X-inactivation in a family with mental retardation and PQBP1 gene mutation. Clinical Genetics, 2005, 67, 446-447.	2.0	11
123	A balanced complex chromosomal rearrangement (BCCR) with phenotypic effect. Clinical Genetics, 1991, 40, 57-61.	2.0	11
124	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
125	8p23.2-pter Microdeletions: Seven New Cases Narrowing the Candidate Region and Review of the Literature. Genes, 2021, 12, 652.	2.4	11
126	Severe complex I deficiency in a case of neonatal-onset lactic acidosis and fatal liver failure. Acta Paediatrica, International Journal of Paediatrics, 1997, 86, 326-329.	1.5	10

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127	Rubinsteinâ€“Taybi Syndrome with Epidermal Nevus: A Case Report. Pediatric Dermatology, 2001, 18, 34-37.	0.9	10
128	12q12 deletion: A new patient contributing to genotypeâ€“phenotype correlation. American Journal of Medical Genetics, Part A, 2008, 146A, 1354-1357.	1.2	10
129	Cerebriform plantar hyperplasia: the major cutaneous feature of Proteus syndrome. International Journal of Dermatology, 2008, 47, 374-376.	1.0	10
130	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
131	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
132	Evaluation of a mutation screening strategy for sporadic cases of ATR-X syndrome. Journal of Medical Genetics, 1999, 36, 183-6.	3.2	10
133	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
134	Facial midline defect in the fetal alcohol syndrome. Embryogenetic considerations in two clinical cases. American Journal of Medical Genetics Part A, 1988, 29, 477-482.	2.4	9
135	Saethre-Chotzen syndrome: a clinical, EEG and neuroradiological study. Child's Nervous System, 1996, 12, 699-704.	1.1	9
136	AN ADDITIONAL CASE OF LINEAR AND WHORLED NEVOID HYPERMELANOSIS ASSOCIATED WITH BIRTH DEFECTS AND MENTAL RETARDATION. Pediatric Dermatology, 1999, 16, 71-73.	0.9	9
137	Failure of fluoxetine to modify the skin-picking behaviour of Prader-Willi syndrome. Australasian Journal of Dermatology, 1998, 39, 57-60.	0.7	8
138	RSK2 enzymatic assay as a second level diagnostic tool in Coffin-Lowry syndrome. Clinica Chimica Acta, 2007, 384, 35-40.	1.1	8
139	Definition of the neurological phenotype associated with dup (X)(p11.22-p11.23). Epileptic Disorders, 2011, 13, 240-251.	1.3	8
140	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
141	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
142	Mitochondrial mRNA expression in fibroblasts of Down syndrome subjects. Human Cell, 2018, 31, 179-181.	2.7	8
143	Hypomelanosis of Ito: A syndrome requiring a multisystem approach. Australasian Journal of Dermatology, 1997, 38, 65-70.	0.7	7
144	Cardiofaciocutaneous (CFC) syndrome. Australasian Journal of Dermatology, 1999, 40, 111-113.	0.7	7

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145	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
146	Three new patients with dup(17)(p11.2p11.2) without autism. Clinical Genetics, 2008, 73, 294-296.	2.0	7
147	KIF21A mRNA expression in patients with Down syndrome. Neurological Sciences, 2013, 34, 569-571.	1.9	7
148	Medial Telangiectatic Sacral Nevi (Types A and C) Associated with Williams Syndrome. Dermatology, 2000, 201, 285-286.	2.1	6
149	Neuroendocrine features of pubertal development in females with mental retardation. Gynecological Endocrinology, 2001, 15, 178-183.	1.7	6
150	Medial Telangiectatic Sacral Nevi and Mca/Mr Syndromes. Pediatric Dermatology, 2003, 20, 370-371.	0.9	6
151	Nail aplasia, microcephaly, severe mental retardation and MRI abnormalities: report of two unrelated cases. Neurological Sciences, 2006, 27, 425-431.	1.9	6
152	An unusual presentation ofÂBecker Nevus. European Journal of Dermatology, 2010, 20, 522-523.	0.6	6
153	Differential expression of PARP1 mRNA in leucocytes of patients with Downâ€™s syndrome. Journal of Genetics, 2011, 90, 469-472.	0.7	6
154	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
155	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
156	Pericentrin expression in Downâ€™s syndrome. Neurological Sciences, 2013, 34, 2023-2025.	1.9	5
157	Killer-specific secretory (Ksp37) gene expression in subjects with Downâ€™s syndrome. Neurological Sciences, 2016, 37, 793-795.	1.9	5
158	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
159	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
160	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
161	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
162	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

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163	NF-kB1 gene expression in Down syndrome patients. Neurological Sciences, 2015, 36, 1065-1066.	1.9	4
164	Long non-coding RNA GAS5 expression in patients with Down syndrome. International Journal of Medical Sciences, 2020, 17, 1315-1319.	2.5	4
165	mRNA expression profiling of mitochondrial subunits in subjects with Parkinsonâ€™s disease. Archives of Medical Science, 2021, , .	0.9	4
166	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
167	Role of long non-coding RNAs in Down syndrome patients: a transcriptome analysis study. Human Cell, 2021, 34, 1662-1670.	2.7	4
168	Genetics and Clinical Neuroscience in Intellectual Disability. Brain Sciences, 2022, 12, 338.	2.3	4
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