# Corrado Romano

List of Publications by Year
in descending order

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|---|---|---|---|
| 214 papers | 12,701 citations | 41323 49 h-index | 28275 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. | 13.9 | 663 |
| 2 | Disruptive CHD8 Mutations Define a Subtype of Autism Early in Development. Cell, 2014, 158, 263-276. | 13.5 | 637 |
| 3 | Refining analyses of copy number variation identifies specific genes associated with developmental delay. Nature Genetics, 2014, 46, 1063-1071. | 9.4 | 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. | 1.4 | 562 |
| 5 | A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. Nature Genetics, 2010, 42, 203-209. | 9.4 | 539 |
| 6 | 15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. Nature Genetics, 2009, 41, 160-162. | 9.4 | 511 |
| 7 | A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. Nature Genetics, 2008, 40, 322-328. | 9.4 | 509 |
| 8 | Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. Nature Genetics, 2017, 49, 515-526. | 9.4 | 443 |
| 9 | A new chromosome 17q21.31 microdeletion syndrome associated with a common inversion polymorphism. Nature Genetics, 2006, 38, 999-1001. | 9.4 | 418 |
| 10 | Mutations in CEP290, which encodes a centrosomal protein, cause pleiotropic forms of Joubert syndrome. Nature Genetics, 2006, 38, 623-625. | 9.4 | 368 |
| 11 | Relative Burden of Large CNVs on a Range of Neurodevelopmental Phenotypes. PLoS Genetics, 2011, 7, e1002334. | 1.5 | 293 |
| 12 | A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. Nature Genetics, 2014, 46, 380-384. | 9.4 | 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. | 1.5 | 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. | 1.5 | 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. | 1.0 | 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. | 2.6 | 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. | 2.6 | 195 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Molecular Mechanisms Generating and Stabilizing Terminal 22q13 Deletions in 44 Subjects with Phelan/McDermid Syndrome. PLoS Genetics, 2011, 7, e1002173. | 1.5 | 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. | 2.2 | 165 |
| 21 | Targeted Nextâ€Generation Sequencing Analysis of 1,000 Individuals with Intellectual Disability. Human Mutation, 2015, 36, 1197-1204. | 1.1 | 161 |
| 22 | Fourteen new cases contribute to the characterization of the 7q11.23 microduplication syndrome. European Journal of Medical Genetics, 2009, 52, 94-100. | 0.7 | 157 |
| 23 | Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. Nature Neuroscience, 2017, 20, 1043-1051. | 7.1 | 152 |
| 24 | Disruptive de novo mutations of DYRK1A lead to a syndromic form of autism and ID. Molecular Psychiatry, 2016, 21, 126-132. | 4.1 | 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. | 1.4 | 140 |
| 26 | Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. American Journal of Human Genetics, 2016, 98, 541-552. | 2.6 | 132 |
| 27 | Molecular analysis of aldolase B genes in hereditary fructose intolerance. Lancet, The, 1990, 335, 306-309. | 6.3 | 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. | 1.1 | 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. , 2003, 121A, 219-224. | | 124 |
| 30 | Sulphation deficit in â€œlow-functioningâ€•autistic children: a pilot study. Biological Psychiatry, 1999, 46, 420-424. | 0.7 | 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. | 0.9 | 114 |
| 32 | Cryptic telomeric rearrangements in subjects with mental retardation associated with dysmorphism and congenital malformations. Journal of Medical Genetics, 2001, 38, 417-420. | 1.5 | 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. | 0.9 | 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. | 1.4 | 108 |
| 35 | Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297. | 0.7 | 108 |
| 36 | Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932. | 5.8 | 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. | 1.1 | 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. | 1.5 | 93 |
| 39 | The molecular landscape of ASPM mutations in primary microcephaly. Journal of Medical Genetics, 2009, 46, 249-253. | 1.5 | 91 |
| 40 | NovelTMEM67mutations and genotype-phenotype correlates in meckelin-related ciliopathies. Human Mutation, 2010, 31, n/a-n/a. | 1.1 | 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. | 0.4 | 75 |
| 42 | An Updated Survey on Skin Conditions in Down Syndrome. Dermatology, 2002, 205, 234-238. | 0.9 | 72 |
| 43 | The 2q23.1 microdeletion syndrome: clinical and behavioural phenotype. European Journal of Human Genetics, 2010, 18, 163-170. | 1.4 | 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. | 0.7 | 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. | 1.4 | 66 |
| 46 | Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. European Journal of Human Genetics, 2013, 21, 1074-1078. | 1.4 | 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. | 2.6 | 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. | 2.6 | 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. | 0.7 | 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. | 0.7 | 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. | 1.0 | 52 |
| 52 | Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63. | 3.6 | 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. | 0.7 | 49 |
| 54 | Hypersensitivity to Aromatic Anticonvulsants: In Vivo and In Vitro Cross-Reactivity Studies. Current Pharmaceutical Design, 2006, 12, 3373-3381. | 0.9 | 46 |

| # | Article | IF | Citations |
|---|---|---|---|
| 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. | 2.6 | 44 |
| 57 | Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679. | 5.8 | 43 |
| 58 | Perforating Miliaâ€like Idiopathic Calcinosis Cutis and Periorbital Syringomas in a Girl With Down Syndrome. Pediatric Dermatology, 1994, 11, 258-260. | 0.5 | 42 |
| 59 | Homocysteine and related genetic polymorphisms in Down's syndrome IQ. Journal of Neurology, Neurosurgery and Psychiatry, 2005, 76, 706-709. | 0.9 | 42 |
| 60 | Schizophrenia in a patient with subtelomeric duplication of chromosome 22q. Clinical Genetics, 2007, 71, 599-601. | 1.0 | 38 |
| 61 | Expanding CEP290 mutational spectrum in ciliopathies. American Journal of Medical Genetics, Part A, 2009, 149A, 2173-2180. | 0.7 | 38 |
| 62 | LOCALIZED ELASTOSIS PERFORANS SERPIGINOSA IN A BOY WITH DOWN SYNDROME. Pediatric Dermatology, 1997, 14, 244-246. | 0.5 | 36 |
| 63 | Disruptive variants of CSDE1 associate with autism and interfere with neuronal development and synaptic transmission. Science Advances, 2019, 5, eaax2166. | 4.7 | 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. | 1.5 | 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. | 1.5 | 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. | 1.4 | 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. | 1.4 | 32 |
| 68 | Rapid and accurate large-scale genotyping of duplicated genes and discovery of interlocus gene conversions. Nature Methods, 2013, 10, 903-909. | 9.0 | 31 |
| 69 | In utero gene therapy rescues microcephaly caused by Pqbp1-hypofunction in neural stem progenitor cells. Molecular Psychiatry, 2015, 20, 459-471. | 4.1 | 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. | 1.1 | 31 |
| 71 | Celiac Disease in Down's Syndrome with HLA Serological and Molecular Studies. Journal of Pediatric Gastroenterology and Nutrition, 1996, 23, 303-306. | 0.9 | 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. | 0.7 | 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. | 1.1 | 29 |
| 74 | DNMT3B promoter polymorphisms and maternal risk of birth of a child with Down syndrome. Human Reproduction, 2013, 28, 545-550. | 0.4 | 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. | 1.5 | 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. | 1.0 | 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. | 0.4 | 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. | 1.0 | 23 |
| 81 | Recurrent duplications of 17q12 associated with variable phenotypes. American Journal of Medical Genetics, Part A, 2015, 167, 3038-3045. | 0.7 | 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. | 0.7 | 22 |
| 83 | Alopecia areata in Down syndrome: a clinical evaluation. Journal of the European Academy of Dermatology and Venereology, 2005, 19, 769-770. | 1.3 | 21 |
| 84 | Narrowing the Candidate Region for Congenital Diaphragmatic Hernia in Chromosome 15q26: Contradictory Results. American Journal of Human Genetics, 2005, 77, 892-894. | 2.6 | 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. | 0.7 | 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. | 1.2 | 20 |
| 87 | The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant. Annals of Neurology, 2018, 83, 926-934. | 2.8 | 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. | 0.6 | 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. | 0.7 | 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. | 0.7 | 19 |
| 92 | Increased FGF3 and FGF4 gene dosage is a risk factor for craniosynostosis. Gene, 2014, 534, 435-439. | 1.0 | 19 |
| 93 | Excess of runs of homozygosity is associated with severe cognitive impairment in intellectual disability. Genetics in Medicine, 2015, 17, 396-399. | 1.1 | 19 |
| 94 | SKIN-PICKING: THE BEST CUTANEOUS FEATURE IN THE RECOGNIZATION OF PRADER-WILLI SYNDROME. International Journal of Dermatology, 1994, 33, 866-867. | 0.5 | 18 |
| 95 | A further family with epilepsy, dementia and yellow teeth: the KohlschÃ¼tter syndrome. Brain and Development, 1995, 17, 133-138. | 0.6 | 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. | 1.0 | 18 |
| 97 | Common pathological mutations inPQBP1induce nonsense-mediated mRNA decay and enhance exclusion of the mutant exon. Human Mutation, 2010, 31, 90-98. | 1.1 | 18 |
| 98 | PTEN Gene: A Model for Genetic Diseases in Dermatology. Scientific World Journal, The, 2012, 2012, 1-8. | 0.8 | 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. | 1.3 | 17 |
| 100 | Milia-like idiopathic calcinosis cutis: an unusual dermatosis associated with Down syndrome. British Journal of Dermatology, 1996, 134, 143-146. | 1.4 | 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. | 1.1 | 17 |
| 102 | Piezogenic pedal papules during Prader-Willi syndrome. Journal of the European Academy of Dermatology and Venereology, 2005, 19, 136-137. | 1.3 | 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. | 1.0 | 17 |
| 104 | An Additional Case of Macular Phylloid Mosaicism. Dermatology, 2001, 202, 73-73. | 0.9 | 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. | 0.7 | 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. | 1.7 | 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. | 1.4 | 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. | 0.9 | 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. | 1.4 | 14 |
| 111 | Expression of STRBP mRNA in patients with cryptorchidism and Downâ€™s syndrome. Journal of Endocrinological Investigation, 2012, 35, 5-7. | 1.8 | 14 |
| 112 | Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611. | 1.1 | 14 |
| 113 | An intriguing case of LEOPARD syndrome.. Pediatric Dermatology, 1998, 15, 125-128. | 0.5 | 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. | 0.7 | 12 |
| 116 | CUTANEOUS FINDINGS IN THE MENTALLY RETARDED. International Journal of Dermatology, 1996, 35, 317-322. | 0.5 | 12 |
| 117 | Decreased expression of GRAF1/OPHN-1-L in the X-linked alpha thalassemia mental retardation syndrome. BMC Medical Genomics, 2010, 3, 28. | 0.7 | 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. | 1.4 | 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. | 1.8 | 12 |
| 120 | Humanin gene expression in fibroblast of Down syndrome subjects. International Journal of Medical Sciences, 2020, 17, 320-324. | 1.1 | 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. | 1.1 | 11 |
| 122 | Skewed X-inactivation in a family with mental retardation and PQBP1 gene mutation. Clinical Genetics, 2005, 67, 446-447. | 1.0 | 11 |
| 123 | A balanced complex chromosomal rearrangement (BCCR) with phenotypic effect. Clinical Genetics, 1991, 40, 57-61. | 1.0 | 11 |
| 124 | Next Generation Sequencing expression profiling of mitochondrial subunits in men with Klinefelter syndrome. International Journal of Medical Sciences, 2018, 15, 31-35. | 1.1 | 11 |
| 125 | 8p23.2-pter Microdeletions: Seven New Cases Narrowing the Candidate Region and Review of the Literature. Genes, 2021, 12, 652. | 1.0 | 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. | 0.7 | 10 |

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|---|---|---|---|
| 127 | Rubinsteinâ€“Taybi Syndrome with Epidermal Nevus: A Case Report. Pediatric Dermatology, 2001, 18, 34-37. | 0.5 | 10 |
| 128 | 12q12 deletion: A new patient contributing to genotypeâ€“phenotype correlation. American Journal of Medical Genetics, Part A, 2008, 146A, 1354-1357. | 0.7 | 10 |
| 129 | Cerebriform plantar hyperplasia: the major cutaneous feature of Proteus syndrome. International Journal of Dermatology, 2008, 47, 374-376. | 0.5 | 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. | 0.4 | 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. | 1.5 | 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. | 1.0 | 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. | 0.6 | 9 |
| 136 | AN ADDITIONAL CASE OF LINEAR AND WHORLED NEVOID HYPERMELANOSIS ASSOCIATED WITH BIRTH DEFECTS AND MENTAL RETARDATION. Pediatric Dermatology, 1999, 16, 71-73. | 0.5 | 9 |
| 137 | Failure of fluoxetine to modify the skin-picking behaviour of Prader-Willi syndrome. Australasian Journal of Dermatology, 1998, 39, 57-60. | 0.4 | 8 |
| 138 | RSK2 enzymatic assay as a second level diagnostic tool in Coffin-Lowry syndrome. Clinica Chimica Acta, 2007, 384, 35-40. | 0.5 | 8 |
| 139 | Definition of the neurological phenotype associated with dup (X)(p11.22-p11.23). Epileptic Disorders, 2011, 13, 240-251. | 0.7 | 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. | 0.9 | 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. | 0.7 | 8 |
| 142 | Mitochondrial mRNA expression in fibroblasts of Down syndrome subjects. Human Cell, 2018, 31, 179-181. | 1.2 | 8 |
| 143 | Hypomelanosis of Ito: A syndrome requiring a multisystem approach. Australasian Journal of Dermatology, 1997, 38, 65-70. | 0.4 | 7 |
| 144 | Cardiofaciocutaneous (CFC) syndrome. Australasian Journal of Dermatology, 1999, 40, 111-113. | 0.4 | 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. | 1.0 | 7 |
| 147 | KIF21A mRNA expression in patients with Down syndrome. Neurological Sciences, 2013, 34, 569-571. | 0.9 | 7 |
| 148 | Medial Telangiectatic Sacral Nevi (Types A and C) Associated with Williams Syndrome. Dermatology, 2000, 201, 285-286. | 0.9 | 6 |
| 149 | Neuroendocrine features of pubertal development in females with mental retardation. Gynecological Endocrinology, 2001, 15, 178-183. | 0.7 | 6 |
| 150 | Medial Telangiectatic Sacral Nevi and Mca/Mr Syndromes. Pediatric Dermatology, 2003, 20, 370-371. | 0.5 | 6 |
| 151 | Nail aplasia, microcephaly, severe mental retardation and MRI abnormalities: report of two unrelated cases. Neurological Sciences, 2006, 27, 425-431. | 0.9 | 6 |
| 152 | An unusual presentation ofÂBecker Nevus. European Journal of Dermatology, 2010, 20, 522-523. | 0.3 | 6 |
| 153 | Differential expression of PARP1 mRNA in leucocytes of patients with Downâ€™s syndrome. Journal of Genetics, 2011, 90, 469-472. | 0.4 | 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. | 0.7 | 6 |
| 155 | Identification of Novel Mutations in Patients with Coffinâ€“Lowry Syndrome by a Denaturing HPLC-Based Assay. Clinical Chemistry, 2005, 51, 2356-2358. | 1.5 | 5 |
| 156 | Pericentrin expression in Downâ€™s syndrome. Neurological Sciences, 2013, 34, 2023-2025. | 0.9 | 5 |
| 157 | Killer-specific secretory (Ksp37) gene expression in subjects with Downâ€™s syndrome. Neurological Sciences, 2016, 37, 793-795. | 0.9 | 5 |
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