# Corrado Romano

## List of Publications by Citations



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|---|---|---|---|
| 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 | Recurrent rearrangements of chromosome 1q21.1 and variable pediatric phenotypes. *New England Journal of Medicine*, **2008**, 359, 1685-99 | 59.2 | 587 |
| 205 | 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 |
| 204 | Disruptive CHD8 mutations define a subtype of autism early in development. *Cell*, **2014**, 158, 263-276 | 56.2 | 467 |
| 203 | A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. *Nature Genetics*, **2008**, 40, 322-8 | 36.3 | 463 |
| 202 | A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. *Nature Genetics*, **2010**, 42, 203-9 | 36.3 | 461 |
| 201 | 15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. *Nature Genetics*, **2009**, 41, 160-2 | 36.3 | 454 |
| 200 | Refining analyses of copy number variation identifies specific genes associated with developmental delay. *Nature Genetics*, **2014**, 46, 1063-71 | 36.3 | 429 |
| 199 | A new chromosome 17q21.31 microdeletion syndrome associated with a common inversion polymorphism. *Nature Genetics*, **2006**, 38, 999-1001 | 36.3 | 355 |
| 198 | Mutations in CEP290, which encodes a centrosomal protein, cause pleiotropic forms of Joubert syndrome. *Nature Genetics*, **2006**, 38, 623-5 | 36.3 | 320 |
| 197 | Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. *Nature Genetics*, **2017**, 49, 515-526 | 36.3 | 283 |
| 196 | Relative burden of large CNVs on a range of neurodevelopmental phenotypes. *PLoS Genetics*, **2011**, 7, e1002334 | 6 | 232 |
| 195 | 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 |
| 194 | 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 |
| 193 | A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. *Nature Genetics*, **2014**, 46, 380-4 | 36.3 | 197 |
| 192 | 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 |
| 191 | Cardiovascular malformations and other cardiovascular abnormalities in neurofibromatosis 1. *American Journal of Medical Genetics Part A*, **2000**, 95, 108-17 | | 176 |
| 190 | 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 |

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|---|---|---|---|
| 189 | 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 |
| 188 | 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 |
| 187 | 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 |
| 186 | Molecular mechanisms generating and stabilizing terminal 22q13 deletions in 44 subjects with Phelan/McDermid syndrome. *PLoS Genetics*, **2011**, 7, e1002173 | 6 | 132 |
| 185 | Targeted Next-Generation Sequencing Analysis of 1,000 Individuals with Intellectual Disability. *Human Mutation*, **2015**, 36, 1197-204 | 4.7 | 122 |
| 184 | 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 |
| 183 | Sulphation deficit in "low-functioning" autistic children: a pilot study. *Biological Psychiatry*, **1999**, 46, 420-4 | 7.9 | 104 |
| 182 | Molecular analysis of aldolase B genes in hereditary fructose intolerance. *Lancet, The*, **1990**, 335, 306-9 | 40 | 103 |
| 181 | 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 |
| 180 | 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 |
| 179 | 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 |
| 178 | Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. *Nature Neuroscience*, **2017**, 20, 1043-1051 | 25.5 | 94 |
| 177 | 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 |
| 176 | Disruptive de novo mutations of DYRK1A lead to a syndromic form of autism and ID. *Molecular Psychiatry*, **2016**, 21, 126-32 | 15.1 | 90 |
| 175 | Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. *American Journal of Human Genetics*, **2016**, 98, 541-552 | 11 | 89 |
| 174 | The molecular landscape of ASPM mutations in primary microcephaly. *Journal of Medical Genetics*, **2009**, 46, 249-53 | 5.8 | 76 |
| 173 | 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 |
| 172 | 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 |

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|---|---|---|---|
| 171 | 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 |
| 170 | An updated survey on skin conditions in Down syndrome. *Dermatology*, **2002**, 205, 234-8 | 4.4 | 66 |
| 169 | The 2q23.1 microdeletion syndrome: clinical and behavioural phenotype. *European Journal of Human Genetics*, **2010**, 18, 163-70 | 5.3 | 65 |
| 168 | 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 |
| 167 | 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 |
| 166 | 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 |
| 165 | Novel TMEM67 mutations and genotype-phenotype correlates in meckelin-related ciliopathies. *Human Mutation*, **2010**, 31, E1319-31 | 4.7 | 57 |
| 164 | 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 |
| 163 | Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. *Biological Psychiatry*, **2019**, 85, 287-297 | 7.9 | 55 |
| 162 | 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 |
| 161 | 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 |
| 160 | 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 |
| 159 | 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 |
| 158 | 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 |
| 157 | A gene for FG syndrome maps in the Xq12-q21.31 region **1997**, 73, 87-90 | | 40 |
| 156 | 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 |
| 155 | Hypersensitivity to aromatic anticonvulsants: in vivo and in vitro cross-reactivity studies. *Current Pharmaceutical Design*, **2006**, 12, 3373-81 | 3.3 | 37 |
| 154 | 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 |

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|---|---|---|---|
| 153 | Perforating milia-like idiopathic calcinosis cutis and periorbital syringomas in a girl with Down syndrome. *Pediatric Dermatology*, **1994**, 11, 258-60 | 1.9 | 36 |
| 152 | Localized elastosis perforans serpiginosa in a boy with Down syndrome. *Pediatric Dermatology*, **1997**, 14, 244-6 | 1.9 | 32 |
| 151 | Schizophrenia in a patient with subtelomeric duplication of chromosome 22q. *Clinical Genetics*, **2007**, 71, 599-601 | 4 | 32 |
| 150 | Homocysteine and related genetic polymorphisms in Down's syndrome IQ. *Journal of Neurology, Neurosurgery and Psychiatry*, **2005**, 76, 706-9 | 5.5 | 32 |
| 149 | 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 |
| 148 | Rapid and accurate large-scale genotyping of duplicated genes and discovery of interlocus gene conversions. *Nature Methods*, **2013**, 10, 903-9 | 21.6 | 30 |
| 147 | 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 | 12.7 | 27 |
| 146 | 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 |
| 145 | Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. *Nature Communications*, **2020**, 11, 4932 | 17.4 | 25 |
| 144 | Expanding CEP290 mutational spectrum in ciliopathies. *American Journal of Medical Genetics, Part A* , **2009**, 149A, 2173-80 | 2.5 | 24 |
| 143 | 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 |
| 142 | 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 |
| 141 | In utero gene therapy rescues microcephaly caused by Pqbp1-hypofunction in neural stem progenitor cells. *Molecular Psychiatry*, **2015**, 20, 459-71 | 15.1 | 22 |
| 140 | Ichthyosis and neutral lipid storage disease. *American Journal of Medical Genetics Part A*, **1988**, 29, 377-82 | | 22 |
| 139 | Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. *Nature Communications*, **2019**, 10, 4679 | 17.4 | 21 |
| 138 | DNMT3B promoter polymorphisms and maternal risk of birth of a child with Down syndrome. *Human Reproduction*, **2013**, 28, 545-50 | 5.7 | 21 |
| 137 | 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 |
| 136 | Celiac disease in Down's syndrome with HLA serological and molecular studies. *Journal of Pediatric Gastroenterology and Nutrition*, **1996**, 23, 303-6 | 2.8 | 19 |

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|---|---|---|---|
| 135 | 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 |
| 134 | 6p22.3 deletion: report of a patient with autism, severe intellectual disability and electroencephalographic anomalies. *Molecular Cytogenetics*, **2013**, 6, 4 | 2 | 18 |
| 133 | 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 |
| 132 | How microsatellite analysis can be exploited for subtelomeric chromosomal rearrangement analysis in mental retardation. *Journal of Medical Genetics*, **2001**, 38, E1 | 5.8 | 18 |
| 131 | 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 |
| 130 | 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 |
| 129 | Recurrent duplications of 17q12 associated with variable phenotypes. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 3038-45 | 2.5 | 17 |
| 128 | Prevalence of atopic dermatitis in patients with Down syndrome: a clinical survey. *Journal of the American Academy of Dermatology*, **1997**, 36, 1019-21 | 4.5 | 17 |
| 127 | Skin-picking: the best cutaneous feature in the recognization of Prader-Willi syndrome. *International Journal of Dermatology*, **1994**, 33, 866-7 | 1.7 | 17 |
| 126 | Disruptive variants of associate with autism and interfere with neuronal development and synaptic transmission. *Science Advances*, **2019**, 5, eaax2166 | 14.3 | 16 |
| 125 | Excess of runs of homozygosity is associated with severe cognitive impairment in intellectual disability. *Genetics in Medicine*, **2015**, 17, 396-9 | 8.1 | 16 |
| 124 | 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 |
| 123 | 3q29 microdeletion syndrome: Cognitive and behavioral phenotype in four patients. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 3018-22 | 2.5 | 15 |
| 122 | Seizures in patients with trisomy 21. *American Journal of Medical Genetics Part A*, **1990**, 7, 298-300 | | 15 |
| 121 | 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 |
| 120 | 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 |
| 119 | PTEN gene: a model for genetic diseases in dermatology. *Scientific World Journal, The*, **2012**, 2012, 252457 | 2.2 | 14 |
| 118 | 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 |

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| 117 | Piezogenic pedal papules during Prader-Willi syndrome. *Journal of the European Academy of Dermatology and Venereology*, **2005**, 19, 136-7 | 4.6 | 14 |
| 116 | Alopecia areata in Down syndrome: a clinical evaluation. *Journal of the European Academy of Dermatology and Venereology*, **2005**, 19, 769-70 | 4.6 | 14 |
| 115 | Increased FGF3 and FGF4 gene dosage is a risk factor for craniosynostosis. *Gene*, **2014**, 534, 435-9 | 3.8 | 13 |
| 114 | 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 |
| 113 | A further family with epilepsy, dementia and yellow teeth: the Kohlschütter syndrome. *Brain and Development*, **1995**, 17, 133-8; discussion 142-3 | 2.2 | 13 |
| 112 | Evidence for long noncoding RNA GAS5 up-regulationin patients with Klinefelter syndrome. *BMC Medical Genetics*, **2019**, 20, 4 | 2.1 | 13 |
| 111 | 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 |
| 110 | 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 |
| 109 | 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 |
| 108 | Biochemical diagnosis and outcome of 2 years treatment in a patient with combined methylmalonic aciduria and homocystinuria. *European Journal of Pediatrics*, **1992**, 151, 818-20 | 4.1 | 12 |
| 107 | The fragile X in Sicily: an epidemiological survey. *American Journal of Medical Genetics Part A*, **1988**, 30, 665-72 | | 12 |
| 106 | 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, 953-6 | 3.1 | 12 |
| 105 | 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 |
| 104 | The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant. *Annals of Neurology*, **2018**, 83, 926-934 | 9.4 | 11 |
| 103 | Skewed X-inactivation in a family with mental retardation and PQBP1 gene mutation. *Clinical Genetics*, **2005**, 67, 446-7 | 4 | 11 |
| 102 | An additional case of macular phylloid mosaicism. *Dermatology*, **2001**, 202, 73 | 4.4 | 11 |
| 101 | Cutaneous findings in the mentally retarded. *International Journal of Dermatology*, **1996**, 35, 317-22 | 1.7 | 11 |
| 100 | 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 |

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|---|---|---|---|
| 99 | 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 | 4.2 | 10 |
| 98 | Expression of STRBP mRNA in patients with cryptorchidism and Down's syndrome. *Journal of Endocrinological Investigation*, **2012**, 35, 5-7 | 5.2 | 10 |
| 97 | 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 |
| 96 | Severe complex I deficiency in a case of neonatal-onset lactic acidosis and fatal liver failure. *Acta Paediatrica, International Journal of Paediatrics*, **1997**, 86, 326-9 | 3.1 | 10 |
| 95 | A balanced complex chromosomal rearrangement (BCCR) with phenotypic effect. *Clinical Genetics*, **1991**, 40, 57-61 | 4 | 10 |
| 94 | 12q12 deletion: a new patient contributing to genotype-phenotype correlation. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1354-7 | 2.5 | 10 |
| 93 | An intronic deletion leading to skipping of exon 21 of COL1A2 in a boy with mild osteogenesis imperfecta. *Connective Tissue Research*, **1993**, 29, 31-40 | 3.3 | 10 |
| 92 | Phenotypic and phoniatric findings in mosaic cri du chat syndrome. *American Journal of Medical Genetics Part A*, **1991**, 39, 391-5 | | 10 |
| 91 | Basal body temperature curves and endocrine pattern of menstrual cycles in Down syndrome. *Gynecological Endocrinology*, **1996**, 10, 133-7 | 2.4 | 9 |
| 90 | Facial midline defect in the fetal alcohol syndrome: embryogenetic considerations in two clinical cases. *American Journal of Medical Genetics Part A*, **1988**, 29, 477-82 | | 9 |
| 89 | Evaluation of a mutation screening strategy for sporadic cases of ATR-X syndrome. *Journal of Medical Genetics*, **1999**, 36, 183-6 | 5.8 | 9 |
| 88 | Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. *Genome Medicine*, **2021**, 13, 63 | 14.4 | 9 |
| 87 | 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 |
| 86 | Cerebriform plantar hyperplasia: the major cutaneous feature of Proteus syndrome. *International Journal of Dermatology*, **2008**, 47, 374-6 | 1.7 | 8 |
| 85 | An intriguing case of LEOPARD syndrome. *Pediatric Dermatology*, **1998**, 15, 125-8 | 1.9 | 8 |
| 84 | An additional case of linear and whorled nevoid hypermelanosis associated with birth defects and mental retardation. *Pediatric Dermatology*, **1999**, 16, 71-3 | 1.9 | 8 |
| 83 | 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 |
| 82 | 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 |

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|---|---|---|---|
| 81 | Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. *Frontiers in Genetics*, **2019**, 10, 611 | 4.5 | 7 |
| 80 | Humanin gene expression in fibroblast of Down syndrome subjects. *International Journal of Medical Sciences*, **2020**, 17, 320-324 | 3.7 | 7 |
| 79 | Mitochondrial mRNA expression in fibroblasts of Down syndrome subjects. *Human Cell*, **2018**, 31, 179-184 | 4.5 | 7 |
| 78 | 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 |
| 77 | Hypomelanosis of Ito: a syndrome requiring a multisystem approach. *Australasian Journal of Dermatology*, **1997**, 38, 65-70 | 1.3 | 7 |
| 76 | Failure of fluoxetine to modify the skin-picking behaviour of Prader-Willi syndrome. *Australasian Journal of Dermatology*, **1998**, 39, 57-8 | 1.3 | 7 |
| 75 | Three new patients with dup(17)(p11.2p11.2) without autism. *Clinical Genetics*, **2008**, 73, 294-6 | 4 | 7 |
| 74 | Rubinstein-Taybi syndrome with epidermal nevus: a case report. *Pediatric Dermatology*, **2001**, 18, 34-7 | 1.9 | 7 |
| 73 | Saethre-Chotzen syndrome: a clinical, EEG and neuroradiological study. *Childls Nervous System*, **1996**, 12, 699-704 | 1.7 | 7 |
| 72 | Is there a relationship between zinc and the peculiar comorbidities of Down syndrome?. *Down Syndrome Research and Practice*, **2002**, 8, 25-8 | | 7 |
| 71 | 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 |
| 70 | 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 |
| 69 | KIF21A mRNA expression in patients with Down syndrome. *Neurological Sciences*, **2013**, 34, 569-71 | 3.5 | 6 |
| 68 | Definition of the neurological phenotype associated with dup (X)(p11.22-p11.23). *Epileptic Disorders*, **2011**, 13, 240-51 | 1.9 | 6 |
| 67 | 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 |
| 66 | Neuroendocrine features of pubertal development in females with mental retardation. *Gynecological Endocrinology*, **2001**, 15, 178-183 | 2.4 | 6 |
| 65 | Medial telangiectatic sacral nevi (Types A and C) associated with Williams syndrome. *Dermatology*, **2000**, 201, 285-6 | 4.4 | 6 |
| 64 | Cardiofaciocutaneous (CFC) syndrome. *Australasian Journal of Dermatology*, **1999**, 40, 111-3 | 1.3 | 6 |

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| 63 | Milia-like idiopathic calcinosis cutis: an unusual dermatosis associated with Down syndrome. *British Journal of Dermatology*, **1996**, 134, 143-146 | 4 | 6 |
| 62 | 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 |
| 61 | Differential expression of PARP1 mRNA in leucocytes of patients with Down's syndrome. *Journal of Genetics*, **2011**, 90, 469-72 | 1.2 | 5 |
| 60 | RSK2 enzymatic assay as a second level diagnostic tool in Coffin-Lowry syndrome. *Clinica Chimica Acta*, **2007**, 384, 35-40 | 6.2 | 5 |
| 59 | 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 |
| 58 | 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 |
| 57 | 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 |
| 56 | 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 |
| 55 | NF-kB1 gene expression in Down syndrome patients. *Neurological Sciences*, **2015**, 36, 1065-6 | 3.5 | 4 |
| 54 | An unusual presentation of Becker Nevus. *European Journal of Dermatology*, **2010**, 20, 522-3 | 0.8 | 4 |
| 53 | Medial telangiectatic sacral nevi and MCA/MR syndromes. *Pediatric Dermatology*, **2003**, 20, 370-1 | 1.9 | 4 |
| 52 | Killer-specific secretory (Ksp37) gene expression in subjects with Down's syndrome. *Neurological Sciences*, **2016**, 37, 793-5 | 3.5 | 4 |
| 51 | 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 |
| 50 | Expression of LDOC1 mRNA in leucocytes of patients with Down syndrome. *Journal of Genetics*, **2012**, 91, 95-98 | 1.2 | 3 |
| 49 | What syndrome is this? Lesch-Nyhan syndrome. *Pediatric Dermatology*, **1996**, 13, 169-70 | 1.9 | 3 |
| 48 | Genome rearrangements in patients with blepharophimosis, mental retardation and hypothyroidism, so-called Young-Simpson syndrome. *Clinical Genetics*, **2009**, 76, 210-3 | 4 | 3 |
| 47 | 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 |
| 46 | 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 |

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| 45 | 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 |
| 44 | 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 |
| 43 | Pericentrin expression in Down's syndrome. *Neurological Sciences*, **2013**, 34, 2023-5 | 3.5 | 2 |
| 42 | 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 |
| 41 | The Clinical Evaluation of Patients with Mental Retardation/Intellectual Disability. *Monographs in Human Genetics*, **2010**, 57-66 | | 2 |
| 40 | The Italian XLMR bank: a clinical and molecular database. *Human Mutation*, **2007**, 28, 13-8 | 4.7 | 2 |
| 39 | Nail aplasia, microcephaly, severe mental retardation and MRI abnormalities: report of two unrelated cases. *Neurological Sciences*, **2006**, 27, 425-31 | 3.5 | 2 |
| 38 | Denaturing HPLC-based assay for detection of ATRX gene mutations. *Clinical Chemistry*, **2005**, 51, 1314-5 | 5.5 | 2 |
| 37 | IGG Antibodies to Beta-Lactoglobulin and Cow's Milk Protein Intolerance in Down Syndrome. *Down Syndrome Research and Practice*, **1998**, 5, 120-122 | | 2 |
| 36 | Long non-coding RNA GAS5 expression in patients with Down syndrome. *International Journal of Medical Sciences*, **2020**, 17, 1315-1319 | 3.7 | 2 |
| 35 | 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 |
| 34 | 8p23.2-pter Microdeletions: Seven New Cases Narrowing the Candidate Region and Review of the Literature. *Genes*, **2021**, 12, | 4.2 | 2 |
| 33 | Prader-Willi Syndrome with Angelman Syndrome in the Offspring. *Medicina (Lithuania)*, **2021**, 57, | 3.1 | 2 |
| 32 | TBC1D24 gene mRNA expression in a boy with early infantile epileptic encephalopathy-16. *Acta Neurologica Belgica*, **2020**, 120, 381-383 | 1.5 | 2 |
| 31 | Role of long non-coding RNAs in Down syndrome patients: a transcriptome analysis study. *Human Cell*, **2021**, 34, 1662-1670 | 4.5 | 2 |
| 30 | 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 |
| 29 | 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 |
| 28 | 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 |

| | | | |
|---|---|---|---|
| 27 | 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 |
| 26 | CASP3 protein expression by flow cytometry in Down's syndrome subjects. *Human Cell*, **2014**, 27, 43-5 | 4.5 | 1 |
| 25 | 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 |
| 24 | A case of FG syndrome with gingival hyperplasia and keloids. *Pediatric Dermatology*, **1995**, 12, 387-9 | 1.9 | 1 |
| 23 | Growth hormone subnormality in Down syndrome. *American Journal of Medical Genetics Part A*, **1992**, 43, 894-5 | | 1 |
| 22 | A dermatoglyphic study of a group of Sicilian children with fragile-X syndrome. *American Journal of Medical Genetics Part A*, **1988**, 30, 177-83 | | 1 |
| 21 | 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 |
| 20 | CCR3 gene overexpression in patients with Down syndrome. *Molecular Biology Reports*, **2021**, 48, 5335-5338 | 2.8 | 1 |
| 19 | 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 |
| 18 | Expression of LDOC1 mRNA in leucocytes of patients with Down's syndrome. *Journal of Genetics*, **2012**, 91, 95-8 | 1.2 | 1 |
| 17 | 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 |
| 16 | Study of the MDM2 -410T-G polymorphism (rs2279744) by pyrosequencing in mothers of Down Syndrome subjects. *Human Cell*, **2020**, 33, 476-478 | 4.5 | |
| 15 | Expression of miR-132 in Down syndrome subjects. *Human Cell*, **2018**, 31, 268-270 | 4.5 | |
| 14 | Poly (ADP-ribose) polymerase 1 expression in fibroblasts of Down syndrome subjects. *Open Medicine (Poland)*, **2013**, 8, 762-765 | 2.2 | |
| 13 | 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 | |
| 12 | Prevention and screening. *Journal of Perinatal Medicine*, **1994**, 22 Suppl 1, 5-8 | 2.7 | |
| 11 | RFLP analysis in 5 Sicilian families with the fragile X syndrome. *American Journal of Medical Genetics Part A*, **1991**, 38, 347-8 | | |
| 10 | Brain dysfunction and the immune system: lymphocyte's beta-adrenergic receptor in Down syndrome. *Pharmacological Research*, **1990**, 22 Suppl 1, 49-50 | 10.2 | |

| | | |
|---|---|---|
| 9 | Bannayan-Riley-Ruvalcaba Syndrome **2008**, 511-515 | |
| 8 | Genetics of Pten Hamartoma Tumor Syndrome (PHTS) **2008**, 483-489 | |
| 7 | Facies: the value of an old diagnostic tip in pediatric dermatology. *Giornale Italiano Di Dermatologia E Venereologia*, **2018**, 153, 716-721 | 0.8 |
| 6 | 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 |
| 5 | Cerebellar degeneration-related protein 1 expression in fibroblasts of patients affected by down syndrome **2020**, 13, 548-555 | |
| 4 | gene downregulation in peripheral blood mononuclear cells of patients with Klinefelter syndrome. *Asian Journal of Andrology*, **2021**, 23, 157-162 | 2.8 |
| 3 | 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 |
| 2 | Neuroendocrine features of pubertal development in females with mental retardation. *Gynecological Endocrinology*, **2001**, 15, 178-183 | 2.4 |
| 1 | 12q21 Interstitial Deletions: Seven New Syndromic Cases Detected by Array-CGH and Review of the Literature. *Genes*, **2022**, 13, 780 | 4.2 |