Corrado Romano

https://publons.com/researcher/B-9695-2008/
Web of Science ResearcherID: B-9695-2008

Current affiliation:
- IRCCS Oasi Maria SS from 1988 until present

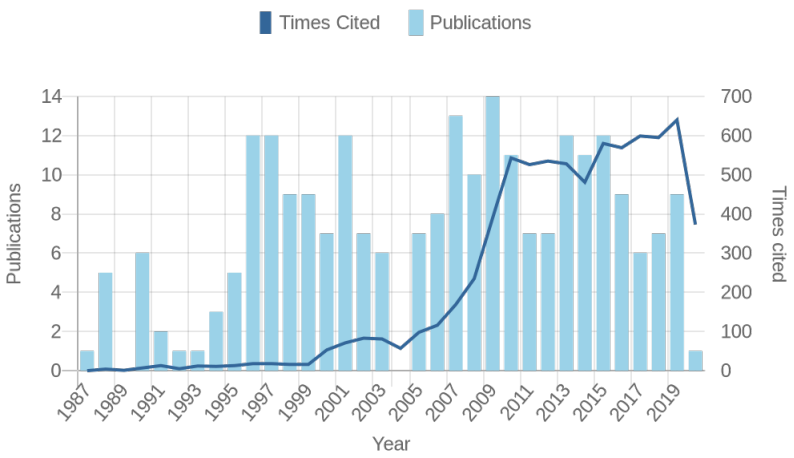
Publications

PUBLICATION METRICS

For all time

CITATIONS	H-INDEX	PUBLICATIONS	*WEB OF SCIENCE* PUBLICATIONS
7456	41	242	198

PUBLICATION IMPACT OVER TIME



PUBLISHING SUMMARY

(6) Human Mutation	WOS	(13) American Journal of Medical Geneti...	WOS
(17) European Journal of Human Geneti...	WOS	(2) Human Molecular Genetics	WOS
(1) The Scientific World Journal		(12) Journal of Medical Genetics	WOS
(4) European Journal of Medical Genetics	WOS	(1) Life Span and Disability	WOS
(2) Epilepsia	WOS	(6) Neurological Sciences	WOS

(1) Current Pharmaceutical Design WOS	(1) Journal of Child Neurology WOS
(2) Biological Psychiatry WOS	(1) Brain and Development WOS
(1) Child's Nervous System WOS	(8) Developmental Brain Dysfunction
(2) PLOS Genetics WOS	(8) The American Journal of Human Gen... WOS
(12) Nature Genetics WOS	(2) Chromosome Research WOS
(12) American Journal of Medical Genetics	(2) Molecular Biology Reports WOS
(1) Genetics and Molecular Research WOS	(2) Giornale Italiano di Dermatologia E V... WOS
(2) Molecular Psychiatry WOS	(1) American Journal of Medical Genetic... WOS
(1) Cell WOS	(1) APMIS WOS
(1) The Lancet WOS	(2) New England Journal of Medicine WOS
(5) Clinical Genetics WOS	(5) Journal of Genetics WOS
(1) Pediatric Infectious Disease Journal WOS	(2) Genetics in Medicine WOS
(2) Clinical Chemistry WOS	(5) Dermatology WOS
(10) Pediatric Dermatology WOS	(2) Journal of the European Academy of ... WOS
(1) Gene WOS	(3) International Journal of Dermatology WOS
(1) BMC Medical Genomics WOS	(2) Pediatrics WOS
(1) Genetic Testing	(3) Gynecological Endocrinology WOS
(1) Neuroscience Letters WOS	(1) European Journal of Pharmacology WOS
(1) Alzheimer's & Dementia: The Journal... WOS	(1) Open Life Sciences WOS
(1) Molecular Cytogenetics WOS	(1) Prenatal Diagnosis WOS
(1) European Journal of Dermatology WOS	(1) British Journal of Dermatology WOS
(2) Cutis WOS	(1) Journal of the American Academy of ... WOS
(3) Cellular and Molecular Biology WOS	(2) Acta Paediatrica WOS
(1) Clinical Chemistry and Laboratory M... WOS	(1) Journal of Neurology, Neurosurgery ... WOS
(1) The American Journal of Clinical Nut... WOS	(2) Journal of Endocrinological Investiga... WOS
(1) The Journal of Maternal-Fetal & Neo... WOS	(1) Journal of Clinical Laboratory Analysi... WOS
(3) Human Cell WOS	(1) Menopause WOS
(1) Metabolism WOS	(1) Epileptic Disorders WOS
(1) Nature Neuroscience WOS	(1) Human Reproduction WOS
(1) Clinica Chimica Acta WOS	(1) Nature Methods WOS
(2) Journal of Pediatric Gastroenterolog... WOS	(1) Central European Journal of Medicine

(1) Rivista Italiana di Pediatria-italian Journal of ...	(1) Japanese Journal of Human Genetics
(1) International Journal of Medical Scie... WOS	(1) Journal of Alzheimer's Disease WOS
(1) Annals of Neurology WOS	(1) Mutation Research: Fundamental an... WOS
(1) Pharmacological Research WOS	(1) Down's Syndrome, Research and Practice: t...
(2) European Journal of Pediatric Dermatology	(1) Minerva Endocrinologica WOS
(4) Australasian Journal of Dermatology WOS	(2) Brain Dysfunction
(1) Acta Neurologica Belgica WOS	(1) Genetics of Mental Retardation
(1) European Journal of Pediatrics WOS	(1) American Journal of Medical Genetics. Supp...
(2) Human Genetics WOS	(1) Genetic Counseling
(2) Minerva Pediatrica WOS	(1) Frontiers in Genetics WOS
(1) Consensus in Child Neurology	(1) Nature Communications WOS
(1) Science Advances WOS	(2) Rivista Italiana di Pediatria
(1) Clinical Dysmorphology WOS	(1) BMC Medical Genetics WOS
(1) Giornale Italiano di Chimica Clinica	(1) Italian Journal of Mineral and Electrolyte Me...

MANUSCRIPTS PUBLISHED (242)	TIMES CITED (ALL TIME)
TBC1D24 gene mRNA expression in a boy with early infantile epileptic encephalopathy-16 Published: Apr 2020 in Acta Neurologica Belgica DOI: 10.1007/S13760-017-0818-3	0
Evidence for long noncoding RNA GAS5 up-regulationin patients with Klinefelter syndrome Published: Dec 2019 in BMC Medical Genetics DOI: 10.1186/S12881-018-0744-0	4
Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders Published: Dec 2019 in Nature Communications DOI: 10.1038/S41467-019-12435-8	2
Disruptive variants of CSDE1 associate with autism and interfere with neuronal development and synaptic transmission Published: Sep 2019 in Science Advances DOI: 10.1126/SCIADV.AAX2166	0
Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative Published: Jul 2019 in Frontiers in Genetics DOI: 10.3389/FGENE.2019.00611	0

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 Published: 2019 in Human Genetics DOI: 10.1007/S00439-019-01972-3	3
Biallelic intragenic duplication in ADGRB3 (BAI3) gene associated with intellectual disability, cerebellar atrophy, and behavioral disorder Published: 2019 in European Journal of Human Genetics DOI: 10.1038/S41431-018-0321-1	0
De novo variants in FBXO11 cause a syndromic form of intellectual disability with behavioral problems and dysmorphisms Published: 2019 in European Journal of Human Genetics DOI: 10.1038/S41431-018-0292-2	2
Rare variants in the genetic background modulate cognitive and developmental phenotypes in individuals carrying disease-associated variants Published: 2019 in Genetics in Medicine DOI: 10.1038/S41436-018-0266-3	24
Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP Published: 2019 in Biological Psychiatry DOI: 10.1016/J.BIOPSYCH.2018.02.1173	22
Expression of miR-132 in Down syndrome subjects Published: Jul 2018 in Human Cell DOI: 10.1007/S13577-018-0209-Y	0
Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies Published: May 2018 in The American Journal of Human Genetics DOI: 10.1016/J.AJHG.2018.03.004	11
The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant Published: 2018 in Annals of Neurology DOI: 10.1002/ANA.25222	3
A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency Published: 2018 in European Journal of Human Genetics DOI: 10.1038/S41431-017-0039-5	8

Next Generation Sequencing expression profiling of mitochondrial subunits in men with Klinefelter syndrome Published: 2018 in International Journal of Medical Sciences DOI: 10.7150/IJMS.21075	4
Mitochondrial mRNA expression in fibroblasts of Down syndrome subjects Published: 2018 in Human Cell DOI: 10.1007/S13577-018-0205-2	2
Facies: the value of an old diagnostic tip in pediatric dermatology Published: 2018 in Giornale Italiano di Dermatologia E Venereologia DOI: 10.23736/S0392-0488.17.05626-7	0
Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains Published: Aug 2017 in Nature Neuroscience DOI: 10.1038/NN.4589	48
Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases Published: Apr 2017 in Nature Genetics DOI: 10.1038/NG.3792	141
Mutation spectrum of NF1 gene in Italian patients with neurofibromatosis type 1 using Ion Torrent PGM (TM) platform Published: 2017 in European Journal of Medical Genetics DOI: 10.1016/J.EJMG.2016.11.001	9
The Methylenetetrahydrofolate Reductase C677T Polymorphism and Risk for Late-Onset Alzheimer's disease: Further Evidence in an Italian Multicenter Study Published: 2017 in Journal of Alzheimer's Disease DOI: 10.3233/JAD-161081	6
A polymorphism (rs1042522) in TP53 gene is a risk factor for Down Syndrome in Sicilian mothers Published: 2017 in The Journal of Maternal-Fetal & Neonatal Medicine DOI: 10.1080/14767058.2016.1262343	0
Searching for new pharmacological targets for the treatment of Alzheimer's disease in Down syndrome Published: 2017 in European Journal of Pharmacology DOI: 10.1016/J.EJPHAR.2017.10.004	2
Low AMH levels as a marker of reduced ovarian reserve in young women affected by Down's syndrome Published: Nov 2016 in Menopause DOI: 10.1097/GME.0000000000000695	1

Expression of Phosphodiesterase 4B cAMP-Specific Gene in Subjects With Cryptorchidism and Down's Syndrome Published: May 2016 in Journal of Clinical Laboratory Analysis DOI: 10.1002/JCLA.21835	2
Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders Published: Mar 2016 in The American Journal of Human Genetics DOI: 10.1016/J.AJHG.2016.02.004	48
A novel splice acceptor site mutation in the ATP2A2 gene in a family with Darier disease Published: 2016 in Giornale Italiano di Dermatologia E Venereologia	0
Disruptive de novo mutations of DYRK1A lead to a syndromic form of autism and ID Published: 2016 in Molecular Psychiatry DOI: 10.1038/MP.2015.5	59
The Koolen-de Vries syndrome: a phenotypic comparison of patients with a 17q21.31 microdeletion versus a KANSL1 sequence variant Published: 2016 in European Journal of Human Genetics DOI: 10.1038/EJHG.2015.178	27
An inflammatory and trophic disconnect biomarker profile revealed in Down syndrome plasma: Relation to cognitive decline and longitudinal evaluation Published: 2016 in Alzheimer's & Dementia: The Journal of the Alzheimer's Association DOI: 10.1016/J.JALZ.2016.05.001	33
MECP2 missense mutations outside the canonical MBD and TRD domains in males with intellectual disability Published: 2016 in Japanese Journal of Human Genetics DOI: 10.1038/JHG.2015.118	15
Killer-specific secretory (Ksp37) gene expression in subjects with Down's syndrome Published: 2016 in Neurological Sciences DOI: 10.1007/S10072-016-2554-5	3
Recurrent Duplications of 17q12 Associated with Variable Phenotypes Published: Dec 2015 in American Journal of Medical Genetics Part A DOI: 10.1002/AJMG.A.37351	10
Target sequencing approach intended to discover new mutations in non-syndromic intellectual disability Published: Nov 2015 in Mutation Research: Fundamental and Molecular Mechanisms of Mutagenesis DOI: 10.1016/J.MRFMMM.2015.09.002	

A peculiar VNTR in the cystathionine β-synthase gene is a risk factor for Down Syndrome Published: Oct 2015 in Cellular and Molecular Biology DOI: 10.14715/CMB/2015.61.5.8	
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 Published: May 2015 in American Journal of Medical Genetics Part A DOI: 10.1002/AJMG.A.36894	0
NF-kB1 gene expression in Down syndrome patients Published: 2015 in Neurological Sciences DOI: 10.1007/S10072-014-1981-4	3
In utero gene therapy rescues microcephaly caused by Pqbp1-hypofunction in neural stem progenitor cells Published: 2015 in Molecular Psychiatry DOI: 10.1038/MP.2014.69	11
Targeted Next-Generation Sequencing Analysis of 1,000 Individuals with Intellectual Disability Published: 2015 in Human Mutation DOI: 10.1002/HUMU.22901	83
Letter to Editor: Cytochrome b gene expression in down syndrome subjects Published: 2015 in Cellular and Molecular Biology DOI: 10.14715/CMB/2015.61.3.2	1
A peculiar VNTR in the cystathionine beta-synthase gene is a risk factor for Down Syndrome Published: 2015 in Cellular and Molecular Biology	2
Excess of runs of homozygosity is associated with severe cognitive impairment in intellectual disability Published: 2015 in Genetics in Medicine DOI: 10.1038/GIM.2014.118	10
LDOC1 expression in fibroblasts of patients with Down syndrome Published: 2015 in Open Life Sciences DOI: 10.1515/BIOL-2015-0015	0
Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling Published: 2015 in The American Journal of Human Genetics DOI: 10.1016/J.AJHG.2015.07.004	81

Definition of 5q11.2 microdeletion syndrome reveals overlap with CHARGE syndrome and 22q11 deletion syndrome phenotypes Published: Sep 2014 in American Journal of Medical Genetics Part A DOI: 10.1002/AJMG.A.36680	5
The transcriptional regulator ADNP links the BAF (SWI/SNF) complexes with autism Published: Aug 2014 in American Journal of Medical Genetics Part C: Seminars in Medical Genetics DOI: 10.1002/AJMG.C.31413	36
Disruption of the ASTN2/TRIM32 locus at 9q33.1 is a risk factor in males for autism spectrum disorders, ADHD and other neurodevelopmental phenotypes Published: May 2014 in Human Molecular Genetics DOI: 10.1093/HMG/DDT669	63
A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP Published: Feb 2014 in Nature Genetics DOI: 10.1038/NG.2899	142
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Increased FGF3 and FGF4 gene dosage is a risk factor for craniosynostosis Published: 2014 in Gene DOI: 10.1016/J.GENE.2013.09.120	5
Interstitial 22q13 deletions not involving SHANK3 gene: A new contiguous gene syndrome Published: 2014 in American Journal of Medical Genetics Part A DOI: 10.1002/AJMG.A.36513	19
Refining analyses of copy number variation identifies specific genes associated with developmental delay Published: 2014 in Nature Genetics DOI: 10.1038/NG.3092	264
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 Published: 2014 in Molecular Biology Reports DOI: 10.1007/S11033-014-3462-5	11

CASP3 protein expression by flow cytometry in Down's syndrome subjects Published: 2014 in Human Cell DOI: 10.1007/S13577-013-0071-X	1
Disruptive CHD8 Mutations Define a Subtype of Autism Early in Development Published: 2014 in Cell DOI: 10.1016/J.CELL.2014.06.017	309
Identification of pathogenic gene variants in small families with intellectually disabled siblings by exome sequencing Published: Dec 2013 in Journal of Medical Genetics DOI: 10.1136/JMEDGENET-2013-101644	46
The Duplication 17p13.3 Phenotype: Analysis of 21 Families Delineates Developmental, Behvioral and Families Dellineates Developmental, Behavioral and Brain Abnormalities, and Rare Variant Phenotypes Published: Aug 2013 in American Journal of Medical Genetics Part A DOI: 10.1002/AJMG.A.35996	32
Rapid and accurate large-scale genotyping of duplicated genes and discovery of interlocus gene conversions Published: Jul 2013 in Nature Methods DOI: 10.1038/NMETH.2572	25
Pericentrin expression in Down's syndrome Published: 2013 in Neurological Sciences DOI: 10.1007/S10072-013-1529-Z	2
KIF21A mRNA expression in patients with Down syndrome Published: 2013 in Neurological Sciences DOI: 10.1007/S10072-012-1183-X	6
DNMT3B promoter polymorphisms and maternal risk of birth of a child with Down syndrome Published: 2013 in Human Reproduction DOI: 10.1093/HUMREP/DES376	18
3q29 Microdeletion Syndrome: Cognitive and Behavioral Phenotype in Four Patients Published: 2013 in American Journal of Medical Genetics Part A DOI: 10.1002/AJMG.A.36142	12
SPAG5 mRNA is over-expressed in peripheral blood leukocytes of patients with Down's syndrome and cryptorchidism Published: 2013 in Neurological Sciences DOI: 10.1007/S10072-012-1152-4	5

Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders Published: 2013 in European Journal of Human Genetics DOI: 10.1038/EJHG.2012.305	31
The MTR 2756A > G polymorphism and maternal risk of birth of a child with Down syndrome: a case-control study and a meta-analysis Published: 2013 in Molecular Biology Reports DOI: 10.1007/S11033-013-2810-1	9
Poly (ADP-ribose) polymerase 1 expression in fibroblasts of Down syndrome subjects Published: 2013 in Central European Journal of Medicine DOI: 10.2478/S11536-013-0225-Y	0
Multiplex ligation-dependent probe amplification detection of an unknown large deletion of the CREB-binding protein gene in a patient with Rubinstein-Taybi syndrome Published: 2013 in Genetics and Molecular Research DOI: 10.4238/2013.JANUARY.7.2	4
Gene expression profiling and qRT-PCR expression of RRP1B, PCNT, KIF21A and ADRB2 in leucocytes of Down’s syndrome subjects Published: Mar 2012 in Journal of Genetics DOI: 10.1007/S12041-012-0132-Z	
Expression of STRBP mRNA in patients with cryptorchidism and Down's syndrome Published: Jan 2012 in Journal of Endocrinological Investigation DOI: 10.1007/BF03345414	9
6p22.3 deletion: report of a patient with autism, severe intellectual disability and electroencephalographic anomalies Published: 2012 in Molecular Cytogenetics DOI: 10.1186/1755-8166-6-4	11
Communicative and cognitive functioning in Angelman syndrome with UBE3A mutation: a case report Published: 2012 in Life Span and Disability	1
Gene expression profiling and qRT-PCR expression of RRP1B, PCNT, KIF21A and ADRB2 in leucocytes of Down's syndrome subjects Published: 2012 in Journal of Genetics	5
Expression of LDOC1 mRNA in leucocytes of patients with Down's syndrome Published: 2012 in Journal of Genetics DOI: 10.1007/S12041-012-0134-X	1

PTEN Gene: A Model for Genetic Diseases in Dermatology Published: 2012 in The Scientific World Journal DOI: 10.1100/2012/252457	7
Assessment of 2q23.1 Microdeletion Syndrome Implicates MBD5 as a Single Causal Locus of Intellectual Disability, Epilepsy, and Autism Spectrum Disorder Published: Oct 2011 in The American Journal of Human Genetics DOI: 10.1016/J.AJHG.2011.09.011	127
Molecular Mechanisms Generating and Stabilizing Terminal 22q13 Deletions in 44 Subjects with Phelan/McDermid Syndrome Published: 2011 in PLOS Genetics DOI: 10.1371/JOURNAL.PGEN.1002173	100
The Pitt-Hopkins Syndrome: Report of 16 New Patients and Clinical Diagnostic Criteria Published: 2011 in American Journal of Medical Genetics Part A DOI: 10.1002/AJMG.A.34070	44
Relative Burden of Large CNVs on a Range of Neurodevelopmental Phenotypes Published: 2011 in PLOS Genetics DOI: 10.1371/JOURNAL.PGEN.1002334	179
Definition of the neurological phenotype associated with dup (X) (p11.22-p11.23) Published: 2011 in Epileptic Disorders DOI: 10.1684/EPD.2011.0462	3
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Decreased expression of GRAF1/OPHN-1-L in the X-linked alpha thalassemia mental retardation syndrome Published: Jul 2010 in BMC Medical Genomics DOI: 10.1186/1755-8794-3-28	9
Novel TMEM67 Mutations and Genotype-phenotype Correlates in Meckelin-related Ciliopathies Published: 2010 in Human Mutation DOI: 10.1002/HUMU.21239	45

A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay Published: Mar 2010 in Nature Genetics DOI: 10.1038/NG.534	394
Erratum: Corrigendum to: The 2q23.1 microdeletion syndrome: clinical and behavioural phenotype Published: Jan 2010 in European Journal of Human Genetics DOI: 10.1038/EJHG.2009.200	
The 2q23.1 microdeletion syndrome: clinical and behavioural phenotype Published: 2010 in European Journal of Human Genetics DOI: 10.1038/EJHG.2009.152	56
The 2q23.1 microdeletion syndrome: clinical and behavioural phenotype (vol 18, pg 163, 2010) Published: 2010 in European Journal of Human Genetics DOI: 10.1038/EJHG.2010.103	0
A recurrent 16p12. 1 microdeletion supports a two-hit model for severe developmental delay Published: 2010 in Nature Genetics	
An unusual presentation of Becker Nevus Published: 2010 in European Journal of Dermatology DOI: 10.1684/EJD.2010.0971	1
The Clinical Evaluation of Patients with Mental Retardation/ Intellectual Disability Published: 2010 in Genetics of Mental Retardation DOI: 10.1159/000287596	4
Common Pathological Mutations in PQBP1 Induce Nonsense-Mediated mRNA Decay and Enhance Exclusion of the Mutant Exon Published: 2010 in Human Mutation DOI: 10.1002/HUMU.21146	11
Familial 1.1 Mb deletion in chromosome Xq22.1 associated with mental retardation and behavioural disorders in female patients Published: 2010 in European Journal of Medical Genetics DOI: 10.1016/J.EJMG.2010.01.001	15
Expanding CEP290 Mutational Spectrum in Ciliopathies Published: Oct 2009 in American Journal of Medical Genetics Part A DOI: 10.1002/AJMG.A.33025	22

Further delineation of the 15q13 microdeletion and duplication syndromes: a clinical spectrum varying from non-pathogenic to a severe outcome Published: Aug 2009 in Journal of Medical Genetics DOI: 10.1136/JMG.2008.063412	181
Genome rearrangements in patients with blepharophimosis, mental retardation and hypothyroidism, so-called Young-Simpson syndrome Published: Aug 2009 in Clinical Genetics DOI: 10.1111/J.1399-0004.2009.01235.X	3
The molecular landscape of ASPM mutations in primary microcephaly Published: Apr 2009 in Journal of Medical Genetics DOI: 10.1136/JMG.2008.062380	61
Fourteen new cases contribute to the characterization of the 7q11.23 microduplication syndrome Published: Mar 2009 in European Journal of Medical Genetics DOI: 10.1016/J.EJMG.2009.02.006	111
SPANX-B and SPANX-C (Xq27 region) gene dosage analysis in Down's syndrome subjects with undescended testes Published: 2009 in Journal of Genetics DOI: 10.1007/S12041-009-0013-2	1
A controversial case of pigmentary mosaicism along Blaschko's lines with neurological impairment Published: 2009 in European Journal of Pediatric Dermatology	
MKS3/TMEM67 Mutations Are a Major Cause of COACH Syndrome, a Joubert Syndrome Related Disorder with Liver Involvement Published: 2009 in Human Mutation DOI: 10.1002/HUMU.20924	59
[Correction] 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 (vol 85, pg 394, 2009) Published: 2009 in The American Journal of Human Genetics DOI: 10.1016/J.AJHG.2009.08.014	1
Fourteen new cases contribute to the characterization of the 7q11.23 microduplication syndrome Published: 2009 in European Journal of Medical Genetics	

15q13. 3 microdeletions increase risk of idiopathic generalized epilepsy Published: 2009 in Nature Genetics	
15q13.3 microdeletions increase risk of idiopathic generalized epilepsy Published: 2009 in Nature Genetics DOI: 10.1038/NG.292	401
Oligonucleotide-based array-CGH in subjects with mental retardation/developmental delay revealed unexpected findings in subjects both with normal and abnormal karyotype Published: 2009 in Chromosome Research	0
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 Published: 2009 in The American Journal of Human Genetics DOI: 10.1016/J.AJHG.2009.08.001	44
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Further delineation of deletion 1 p36 syndrome in 60 patients: A recognizable phenotype and common cause of developmental delay and mental retardation (vol 121, pg 404, 2008) Published: 2008 in Pediatrics DOI: 10.1542/PEDS.2008-0990	1
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Recurrent rearrangements of chromosome 1q21. 1 and variable pediatric phenotypes Published: 2008 in New England Journal of Medicine	
Three new patients with dup(17)(p11.2p11.2) without autism Published: 2008 in Clinical Genetics DOI: 10.1111/J.1399-0004.2007.00959.X	7

A recurrent 15q13. 3 microdeletion syndrome associated with mental retardation and seizures Published: 2008 in Nature Genetics	
Further delineation of deletion 1p36 syndrome in 60 patients: A recognizable phenotype and common cause of developmental delay and mental retardation Published: 2008 in Pediatrics DOI: 10.1542/PEDS.2007-0929	163
Recurrent rearrangements of chromosome 1q21.1 and variable pediatric phenotypes Published: 2008 in New England Journal of Medicine DOI: 10.1056/NEJMOA0805384	506
A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures Published: 2008 in Nature Genetics DOI: 10.1038/NG.93	402
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The Italian XLMR bank: A clinical and molecular database Published: 2007 in Human Mutation DOI: 10.1002/HUMU.20411	2
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Cryptic deletions are a common finding in “balanced” reciprocal and complex chromosome rearrangements: a study of 59 patients Published: 2007 in Journal of Medical Genetics	
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1.5 Mb de novo 22q11.21 microduplication in a patient with cognitive deficits and dysmorphic facial features Published: 2007 in Clinical Genetics DOI: 10.1111/J.1399-0004.2007.00750.X	44
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Psychopathologic disorder in Prader-Willi syndrome Published: 2007 in American Journal of Medical Genetics Part A	0
A new chromosome 17q21.31 microdeletion syndrome associated with a common inversion polymorphism Published: Aug 2006 in Nature Genetics DOI: 10.1038/NG1853	297
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A new chromosome 17q21. 31 microdeletion syndrome associated with a common inversion polymorphism Published: 2006 in Nature Genetics	
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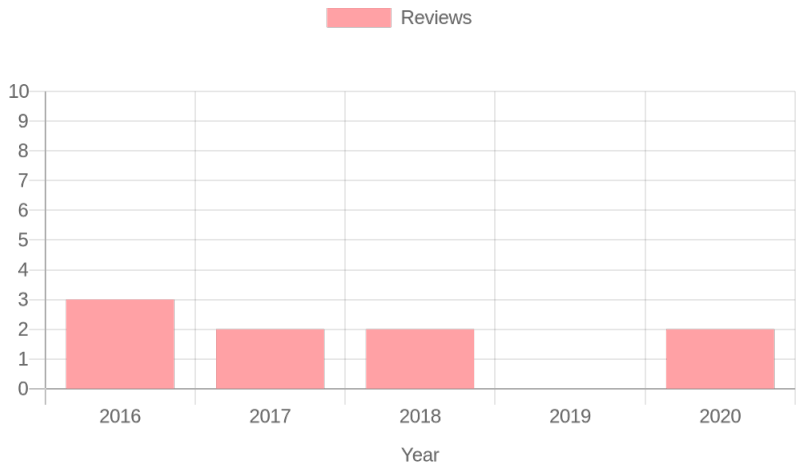
Verified reviews

REVIEW SUMMARY



REVIEWER SUMMARY

(1) Life Sciences WOS	(1) Gene Reports WOS
(2) BMC Medical Genomics WOS	(1) Journal of Community Genetics WOS
(4) Orphanet Journal of Rare Diseases WOS	

MANUSCRIPTS REVIEWED (8)

Reviewed: Jun 2020 for Life Sciences

Reviewed: Jun 2020 for Gene Reports

2 rounds from Aug 2018 to Sep 2018 for BMC Medical Genomics

Reviewed: Sep 2017 for Orphanet Journal of Rare Diseases

Reviewed: Aug 2017 for Journal of Community Genetics

Reviewed: Jul 2016 for Orphanet Journal of Rare Diseases

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