Alina T Midro

List of Publications by Year
in descending order

46 papers	1,214 citations	516710 16 h-index	377865 34 g-index
49 all docs	49 docs citations	49 times ranked	1575 citing authors

#	Article	IF	Citations
1	Genotypic and Phenotypic Spectrum in Tricho-Rhino-Phalangeal Syndrome Types I and III. American Journal of Human Genetics, 2001, 68, 81-91.	6.2	205
2	Genotype-phenotype correlation in 21 patients with Wolf-Hirschhorn syndrome using high resolution array comparative genome hybridisation (CGH). Journal of Medical Genetics, 2007, 45, 71-80.	3.2	111
3	Immortalization and characterization of Nijmegen Breakage Syndrome fibroblasts. Mutation Research DNA Repair, 1999, 434, 17-27.	3.7	98
4	Further delineation of Pitt-Hopkins syndrome: phenotypic and genotypic description of 16 novel patients. Journal of Medical Genetics, 2008, 45, 738-744.	3.2	86
5	Three novel SRY mutations in XY gonadal dysgenesis and the enigma of XY gonadal dysgenesis cases without SRY mutations. Cytogenetic and Genome Research, 1998, 80, 188-192.	1.1	65
6	Rett syndrome in females with CTS hot spot deletions: A disorder profile. American Journal of Medical Genetics, Part A, 2005, 132A, 117-120.	1.2	65
7	Second observation of Silverâ€Russel syndrome in a carrier of a reciprocal translocation with one breakpoint at site 17q25. Clinical Genetics, 1993, 44, 53-55.	2.0	51
8	Experiences with risk estimates for carriers of chromosomal reciprocal translocations. Clinical Genetics, 1992, 41, 113-122.	2.0	50
9	Genetic counseling in Robertsonian translocations der(13;14): Frequencies of reproductive outcomes and infertility in 101 pedigrees. American Journal of Medical Genetics, Part A, 2008, 146A, 2611-2616.	1.2	45
10	ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. Human Mutation, 2016, 37, 653-660.	2.5	40
11	Construction of a Detailed Physical and Transcript Map of the Candidate Region for Russellâ€“Silver Syndrome on Chromosome 17q23â€“q24. Genomics, 2001, 71, 174-181.	2.9	38
12	Interstitial deletion 9q22.32â€q33.2 associated with additional familial translocation t(9;17)(q34.11;p11.2) in a patient with Gorlinâ€“Goltz syndrome and features of Nailâ€Patella syndrome. American Journal of Medical Genetics, Part A, 2004, 124A, 179-191.	1.2	38
13	A molecular epidemiology study in women from Upper Silesia, Poland. Toxicology Letters, 1998, 96-97, 195-202.	0.8	35
14	Distortion products otoacoustic emissions in diagnosis of hearing loss in Down syndrome. International Journal of Pediatric Otorhinolaryngology, 1998, 45, 199-206.	1.0	32
15	The Analysis of Meiotic Segregation Patterns and Aneuploidy in the Spermatozoa of Father and Son With Translocation t(4;5)(p15.1;p12) and the Prediction of the Individual Probability Rate for Unbalanced Progeny at Birth. Journal of Andrology, 2006, 28, 262-272.	2.0	25
16	Risk evaluation of carriers with chromosome reciprocal translocation t(7;13)(q34;q13) and concomitant meiotic segregation analyzed by FISH on ejaculated spermatozoa. American Journal of Medical Genetics, Part A, 2006, 140A, 245-256.	1.2	19
17	MTRNR2L12: A Candidate Blood Marker of Early Alzheimerâ€™s Disease-Like Dementia in Adults with Down Syndrome. Journal of Alzheimer's Disease, 2015, 46, 145-150.	2.6	17
18	Genetic counselling in carriers of reciprocal chromosomal translocations involving short arm of chromosome X. Annales De GÃ©nÃ©tique, 2004, 47, 11-28.	0.4	16

#	Article	IF	Citations
19	XYY syndrome and acute myeloblastic leukemia. Cancer Genetics and Cytogenetics, 1987, 24, 363-365.	1.0	15
20	Measurement of cytogenetic endpoints in women environmentally exposed to air pollution. Mutation Research - Genetic Toxicology and Environmental Mutagenesis, 1999, 445, 139-145.	1.7	15
21	L239F founder mutation in GDAP1 is associated with a mild Charcotâ€“Marieâ€“Tooth type 4C4 (CMT4C4) phenotype. Neurogenetics, 2010, 11, 357-366.	1.4	15
22	Chromatin structure analysis of spermatozoa from reciprocal chromosome translocation (RCT) carriers with known meiotic segregation patterns. Reproductive Biology, 2013, 13, 209-220.	1.9	13
23	Haptoglobin glycoforms in a case of carbohydrate-deficient glycoprotein syndrome. Glycoconjugate Journal, 1999, 16, 573-577.	2.7	12
24	Wolfâ€“Hirschhorn syndrome-associated chromosome changes are not mediated by olfactory receptor gene clusters nor by inversion polymorphism on 4p16. Human Genetics, 2007, 122, 423-430.	3.8	12
25	Genetic counseling in carriers of reciprocal chromosomal translocations involving long arm of chromosome 16. Clinical Genetics, 2004, 66, 189-207.	2.0	11
26	Risk estimates for carriers of chromosome reciprocal translocation t(4;9)(p15.2;p13). Clinical Genetics, 2000, 58, 153-155.	2.0	8
27	Clonal chromosomal aberrations in Philadelphia negative cells such as monosomy 7 and trisomy 8 may persist for years with no impact on the long term outcome in patients with chronic myeloid leukemia. Cancer Genetics, 2017, 216-217, 1-9.	0.4	8
28	Phenotypic expansion of the <scp>BPTF</scp>â€related neurodevelopmental disorder with dysmorphic facies and distal limb anomalies. American Journal of Medical Genetics, Part A, 2021, 185, 1366-1378.	1.2	8
29	Characteristic dental pattern with hypodontia and short roots in Fraser syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1681-1689.	1.2	7
30	Familial occurrence of isodicentric X chromosomes with different breakpoints. Clinical Genetics, 1988, 34, 153-160.	2.0	6
31	Complex balanced chromosomal translocation t(2;5;13) (p21;p15;q22) in a woman with four reproductive failures. Molecular Cytogenetics, 2014, 7, 83.	0.9	6
32	Earlier finishing of Xp21.2 subband replication of the inactive X chromosome in Rett syndrome girl but not in her 47,XXX mother. Clinical Genetics, 1997, 52, 120-125.	2.0	5
33	Two unrelated families with variable expression of Fraser syndrome due to the same pathogenic variant in the <scp>FRAS1</scp> gene. American Journal of Medical Genetics, Part A, 2020, 182, 773-779.	1.2	5
34	Wolfâ€“Hirschhorn syndrome due to pure and translocation forms of monosomy 4p16.1â€‰â†’â€‰pter. American Journal of Medical Genetics, Part A, 2011, 155, 1833-1847.	1.2	4
35	Recurrence risks for different pregnancy outcomes and meiotic segregation analysis of spermatozoa in carriers of t(1;11)(p36.22;q12.2). Journal of Human Genetics, 2014, 59, 667-674.	2.3	4
36	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies syndrome due to disruption of BPTF in a 35â€yearâ€old man initially diagnosed with Silverâ€Russell syndrome. Clinical Genetics, 2019, 95, 534-536.	2.0	4

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37	Cytogenetic and molecular analyses of de novo translocation dic(9;13)(p11.2;p12) in an infertile male. Molecular Cytogenetics, 2014, 7, 14.	0.9	3
38	A 23â€year followâ€up of a male with Hajduâ€Cheney syndrome due to NOTCH2 mutation. American Journal of Medical Genetics, Part A, 2018, 176, 2382-2388.	1.2	3
39	Chromosome (re)positioning in spermatozoa of fathers and sons â€“ carriers of reciprocal chromosome translocation (RCT). BMC Medical Genomics, 2019, 12, 30.	1.5	3
40	Reciprocal chromosome translocations involving short arm of chromosome 9 as a risk factor of unfavorable pregnancy outcomes after meiotic malsegregation 2:2. Advances in Medical Sciences, 2009, 54, 203-10.	2.1	3
41	Meiotic and pedigree segregation analyses in carriers of t(4;8)(p16;p23.1) differing in localization of breakpoint positions at 4p subband 4p16.3 and 4p16.1. Journal of Assisted Reproduction and Genetics, 2016, 33, 189-197.	2.5	2
42	Coâ€segregation of Freiberg's infraction with a familial translocation t(5;7)(p13.3;p22.2) ascertained by a child with cri du chat syndrome and brachydactyly type A1B. American Journal of Medical Genetics, Part A, 2015, 167, 445-449.	1.2	1
43	Abnormalities in tooth morphology, structure and dentition in two children with chromosome aberrations. Translocation trisomy 13 and trisomy 21. Advances in Medical Sciences, 2008, 53, 17-20.	2.1	1
44	Sister-chromatid exchanges in patients with reproductive loss. Mutation Research - Environmental Mutagenesis and Related Subjects Including Methodology, 1982, 97, 235.	0.4	0
45	Molecular Analyses of theBORISGene in Children with Silver-Russell Syndrome. International Journal of Human Genetics, 2009, 9, 269-272.	0.1	0
46	Limited survivability of unbalanced progeny of carriers of a unique t(4;19)(p15.32;p13.3): a study in multiple generations. Molecular Cytogenetics, 2017, 10, 29.	0.9	0