Krystyna Chrzanowska

List of Publications by Year
in descending order

36	1,145	706676 14	466096 32
papers	citations	h-index	g-index

40	40	40	2454
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Hematopoietic Stem Cell Transplantation Positively Affects the Natural History of Cancer in Nijmegen Breakage Syndrome. Clinical Cancer Research, 2021, 27, 575-584.	3.2	13
2	Variable degree of mosaicism for tetrasomy 18p in phenotypically discordant monozygotic twinsâ€"Diagnostic implications. Molecular Genetics & Genomic Medicine, 2021, 9, e1526.	0.6	7
3	Rare clinical findings in three sporadic cases of Beckwith-Wiedemann syndrome due to novel mutations in the CDKN1C gene. Clinical Dysmorphology, 2020, 29, 28-34.	0.1	5
4	Occurrence of Portal Hypertension and Its Clinical Course in Patients With Molecularly Confirmed Autosomal Recessive Polycystic Kidney Disease (ARPKD). Frontiers in Pediatrics, 2020, 8, 591379.	0.9	2
5	Breakpoint Mapping of Symptomatic Balanced Translocations Links the EPHA6, KLF13 and UBR3 Genes to Novel Disease Phenotype. Journal of Clinical Medicine, 2020, 9, 1245.	1.0	4
6	Telomere attrition and dysfunction: a potential trigger of the progeroid phenotype in nijmegen breakage syndrome. Aging, 2020, 12, 12342-12375.	1.4	6
7	A case of premature ovarian insufficiency in Nijmegen breakage syndrome patient and review of literature. From gene mutation to clinical management. Gynecological Endocrinology, 2019, 35, 999-1002.	0.7	6
8	Small supernumerary marker chromosomes: A legacy of trisomy rescue?. Human Mutation, 2019, 40, 193-200.	1.1	33
9	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€"Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	1.1	80
10	Mapping of breakpoints in balanced chromosomal translocations by shallow whole-genome sequencing points toEFNA5,BAHD1andPPP2R5Eas novel candidates for genes causing human Mendelian disorders. Journal of Medical Genetics, 2019, 56, 104-112.	1.5	13
11	Evidence for a pre-malignant cell line in a skin biopsy from a patient with Nijmegen breakage syndrome. Molecular Cytogenetics, 2018, 11, 17.	0.4	3
12	Transient Elastography for Detection of Liver Fibrosis in Children With Autosomal Recessive Polycystic Kidney Disease. Frontiers in Pediatrics, 2018, 6, 422.	0.9	8
13	Novel pathogenic variant in the HRAS gene with lethal outcome and a broad phenotypic spectrum among Polish patients with Costello syndrome. Clinical Dysmorphology, 2017, 26, 83-90.	0.1	1
14	Constitutional mosaicism of a de novo TP53 mutation in a patient with bilateral choroid plexus carcinoma. Cancer Genetics, 2017, 216-217, 79-85.	0.2	10
15	Anemia in Patients With Resistance to Thyroid Hormone Î±: A Role for Thyroid Hormone Receptor Î± in Human Erythropoiesis. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 3517-3525.	1.8	16
16	A novel IGF2/H19 domain triplication in the 11p15.5 imprinting region causing either Beckwithâ€"Wiedemann or Silverâ€"Russell syndrome in a single family. American Journal of Medical Genetics, Part A, 2017, 173, 72-78.	0.7	11
17	Prenatal molecular testing for Beckwithâ€"Wiedemann and Silverâ€"Russell syndromes: a challenge for molecular analysis and genetic counseling. European Journal of Human Genetics, 2016, 24, 784-793.	1.4	44
18	Nijmegen Breakage Syndrome: Clinical and Immunological Features, Long-Term Outcome and Treatment Options â€" a Retrospective Analysis. Journal of Clinical Immunology, 2015, 35, 538-549.	2.0	73

#	Article	IF	Citations
19	Biliary Atresia in Children With Aberrations Involving Chromosome 11q. Journal of Pediatric Gastroenterology and Nutrition, 2014, 58, e26-9.	0.9	5
20	Gain-of-function mutations in the phosphatidylserine synthase 1 (PTDSS1) gene cause Lenz-Majewski syndrome. Nature Genetics, 2014, 46, 70-76.	9.4	74
21	Spectrum of JAG1 gene mutations in Polish patients with Alagille syndrome. Journal of Applied Genetics, 2014, 55, 329-336.	1.0	24
22	Coffin-Siris Syndrome and the BAF Complex: Genotype-Phenotype Study in 63 Patients. Human Mutation, 2013, 34, 1519-1528.	1.1	178
23	A comprehensive molecular study on Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes identifies a broad molecular and clinical spectrum converging on altered chromatin remodeling. Human Molecular Genetics, 2013, 22, 5121-5135.	1.4	190
24	A girl with two syndromes: Turner syndrome and Costello syndrome. A case history. American Journal of Medical Genetics, Part A, 2012, 158A, 1486-1488.	0.7	5
25	Nijmegen breakage syndrome with macrocephaly, schizencephaly and large CSF spacesâ€”extended spectrum of the condition. Journal of Applied Genetics, 2012, 53, 189-191.	1.0	0
26	Differences between predicted and established diagnoses of Smithâ€Łemliâ€Opitz syndrome in the Polish population: underdiagnosis or loss of affected fetuses?. Journal of Inherited Metabolic Disease, 2010, 33, 241-248.	1.7	29
27	Four novel RSK2 mutations in females with Coffinâ€“Lowry syndrome. European Journal of Medical Genetics, 2010, 53, 268-273.	0.7	14
28	Severe neonatal spondylometaphyseal dysplasia in two siblings. American Journal of Medical Genetics, Part A, 2009, 149A, 2166-2172.	0.7	6
29	Spontaneously immortalized T lymphocytes from Nijmegen Breakage Syndrome patients display phenotypes typical for lymphoma cells. Leukemia Research, 2008, 32, 569-577.	0.4	2
30	Impaired elimination of DNA double-strand break-containing lymphocytes in ataxia telangiectasia and Nijmegen breakage syndrome. DNA Repair, 2006, 5, 904-913.	1.3	43
31	TheARX mutations: A frequent cause of X-linked mental retardation. American Journal of Medical Genetics, Part A, 2006, 140A, 727-732.	0.7	29
32	Syndromic dystelephalangy. Clinical Dysmorphology, 2002, 11, 103-105.	0.1	2
33	Clinical ascertainment of Nijmegen breakage syndrome (NBS) and prevalence of the major mutation, 657del5, in three Slav populations. European Journal of Human Genetics, 2000, 8, 900-902.	1.4	130
34	Syndromic foramina parietalia permagna. , 1998, 78, 401-405.		12
35	Radiation Induction of p53 in Cells from Nijmegen Breakage Syndrome Is Defective but Not Similar to Ataxia-Telangiectasia. Biochemical and Biophysical Research Communications, 1998, 242, 602-607.	1.0	44
36	A further report of Brachmannâ€de Lange syndrome in two sibs with normal parents. Clinical Genetics, 1995, 47, 324-327.	1.0	16