List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	Atypical presentation of Dent disease in a patient with interstitial Xp11.22 deletion. Journal of Nephrology, 2021, 34, 2111-2115.	0.9	1
2	Neurodevelopmental Disorders in Patients With Complex Phenotypes and Potential Complex Genetic Basis Involving Non-Coding Genes, and Double CNVs. Frontiers in Genetics, 2021, 12, 732002.	1.1	12
3	Small interstitial 9p24.3 deletions principally involving KANK1 are likely benign copy number variants. European Journal of Medical Genetics, 2020, 63, 103618.	0.7	2
4	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.	0.7	3
5	17q23.3 de novo microdeletion involving only TANC2 gene: A new case. European Journal of Medical Genetics, 2020, 63, 104094.	0.7	3
6	Prenatal Diagnosis of an Uncommon 48, XX ,+18+21 Karyotype in a Fetus With Malformations Typical of Both Trisomies. Journal of Ultrasound in Medicine, 2020, 39, 2277-2279.	0.8	0
7	1p31.1 microdeletion including only NEGR1 gene in two patients. European Journal of Medical Genetics, 2020, 63, 103919.	0.7	5
8	â€~Distal 16p12.2 microdeletion' in a patient with autosomal recessive deafness-22. Journal of Genetics, 2019, 98, 1.	0.4	8
9	Intragenic duplication of KCNQ5 gene results in aberrant splicing leading to a premature termination codon in a patient with intellectual disability. European Journal of Medical Genetics, 2019, 62, 103555.	0.7	22
10	Familial ROBO1 deletion associated with ectopic posterior pituitary, duplication of the pituitary stalk and anterior pituitary hypoplasia. Journal of Pediatric Endocrinology and Metabolism, 2019, 32, 95-99.	0.4	11
11	3q29 microduplication syndrome: Description of two new cases and delineation of the minimal critical region. European Journal of Medical Genetics, 2018, 61, 428-433.	0.7	13
12	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	3.5	86
13	Clinical and Molecular Characterization of Two Patients with CNTN6 Copy Number Variations. Cytogenetic and Genome Research, 2018, 156, 144-149.	0.6	6
14	Intragenic Microdeletion of <i>ULK4</i> and Partial Microduplication of <i>BRWD3</i> in Siblings with Neuropsychiatric Features and Obesity. Cytogenetic and Genome Research, 2018, 156, 14-21.	0.6	9
15	Characterization of the Phenotype Associated with Microduplication Reciprocal to NF1 Microdeletion Syndrome. Cytogenetic and Genome Research, 2017, 152, 22-28.	0.6	0
16	Assessment of copy number variations in 120 patients with Poland syndrome. BMC Medical Genetics, 2016, 17, 89.	2.1	20
17	Interstitial de novo 18q22.3q23 deletion: clinical, neuroradiological and molecular characterization of a new case and review of the literature. Molecular Cytogenetics, 2016, 9, 78.	0.4	8
18	Interstitial 11q24 deletion: a new case and review of the literature. Journal of Applied Genetics, 2016, 57, 357-362.	1.0	8

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19	Interstitial 9p24.3 deletion involving only DOCK8 and KANK1 genes in two patients with non-overlapping phenotypic traits. European Journal of Medical Genetics, 2016, 59, 20-25.	0.7	17
20	Clinical and molecular characterization of a patient with interstitial 6q21q22.1 deletion. Molecular Cytogenetics, 2015, 8, 31.	0.4	17
21	Congenital aural atresia associated with agenesis of internal carotid artery in a girl with a <i>FOXI3</i> deletion. American Journal of Medical Genetics, Part A, 2015, 167, 537-544.	0.7	27
22	Association of achondroplasia with sagittal synostosis and scaphocephaly in two patients, an underestimated condition?. American Journal of Medical Genetics, Part A, 2015, 167, 646-652.	0.7	15
23	Concomitant deletion of chromosome 16p13.11 and triplication of chromosome 19p13.3 in a child with developmental disorders, intellectual disability, and epilepsy. Molecular Cytogenetics, 2015, 8, 9.	0.4	4
24	Clinical and molecular delineation of a 16p13.2p13.13 microduplication. European Journal of Medical Genetics, 2015, 58, 194-198.	0.7	6
25	Clinical and Molecular Cytogenetic Characterization of a de novo Interstitial 1p31.1p31.3 Deletion in a Boy with Moderate Intellectual Disability and Severe Language Impairment. Cytogenetic and Genome Research, 2015, 146, 39-43.	0.6	16
26	Clinico-radiological and molecular characterization of a child with ring chromosome 2 presenting growth failure, microcephaly, kidney and brain malformations. Molecular Cytogenetics, 2015, 8, 17.	0.4	7
27	Thrombocytopenia-absent radius (TAR) syndrome due to compound inheritance for a 1q21.1 microdeletion and a low-frequency noncoding RBM8A SNP: a new familial case. Molecular Cytogenetics, 2015, 8, 87.	0.4	16
28	Pituitary deficiency and congenital infiltrating lipomatosis of the face in a girl with deletion of chromosome 1q24.3q31.1. American Journal of Medical Genetics, Part A, 2014, 164, 495-499.	0.7	16
29	Heterozygous deletion of CHL1 gene: Detailed array-CCH and clinical characterization of a new case and review of the literature. European Journal of Medical Genetics, 2014, 57, 626-629.	0.7	23
30	Phenotypic and genetic characterization of a patient with a de novo interstitial 14q24.1q24.3 deletion. Molecular Cytogenetics, 2014, 7, 49.	0.4	3
31	Recurrent microdeletion 2q21.1: Report on a new patient with neurological disorders. American Journal of Medical Genetics, Part A, 2014, 164, 801-805.	0.7	6
32	Interstitial 7q31.1 copy number variations disrupting IMMP2L gene are associated with a wide spectrum of neurodevelopmental disorders. Molecular Cytogenetics, 2014, 7, 54.	0.4	29
33	De novo deletion of chromosome 11q12.3 in monozygotic twins affected by Poland Syndrome. BMC Medical Genetics, 2014, 15, 63.	2.1	32
34	Constitutional chromosomal events at 22q11 and 15q26 in a child with a pilocytic astrocytoma of the spinal cord. Molecular Cytogenetics, 2014, 7, 31.	0.4	2
35	RORB gene and 9q21.13 microdeletion: Report on a patient with epilepsy and mild intellectual disability. European Journal of Medical Genetics, 2014, 57, 44-46.	0.7	26
36	Interstitial deletion 14q31.1q31.3 transmitted from a mother to her daughter, both with features of hemifacial microsomia. Journal of Applied Genetics, 2013, 54, 361-365.	1.0	7

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37	Interstitial 2q24.3 deletion including SCN2A and SCN3A genes in a patient with autistic features, psychomotor delay, microcephaly and no history of seizures. Gene, 2013, 532, 294-296.	1.0	13
38	A rare 3q13.31 microdeletion including GAP43 and LSAMP genes. Molecular Cytogenetics, 2013, 6, 52.	0.4	7
39	De novo 13q31.1–q32.1 interstitial deletion encompassing the <i>miRâ€17â€92</i> cluster in a patient with Feingold syndromeâ€2. American Journal of Medical Genetics, Part A, 2013, 161, 894-896.	0.7	19
40	Parental Imbalances Involving Chromosomes 15q and 22q May Predispose to the Formation of De Novo Pathogenic Microdeletions and Microduplications in the Offspring. PLoS ONE, 2013, 8, e57910.	1.1	7
41	Identification of an Interstitial 18p11.32-p11.31 Duplication Including the EMILIN2 Gene in a Family with Porokeratosis of Mibelli. PLoS ONE, 2013, 8, e61311.	1.1	12
42	Genotype-Phenotype Correlation of 2q37 Deletions Including NPPC Gene Associated with Skeletal Malformations. PLoS ONE, 2013, 8, e66048.	1.1	32
43	New recurrent chromosome change in pediatric therapy-related myelodysplastic syndrome: unbalanced translocation 1/6 with cryptic duplication of short arm of chromosome 6. Leukemia and Lymphoma, 2012, 53, 2434-2438.	0.6	2
44	Identification of a rare 17p13.3 duplication including the BHLHA9 and YWHAEgenes in a family with developmental delay and behavioural problems. BMC Medical Genetics, 2012, 13, 93.	2.1	31
45	Molecular cytogenetic characterization of the first reported case of an inv dup (4p)(p15.1-pter) with a concomitant 4q35.1-qter deletion and normal parents. Gene, 2012, 511, 338-340.	1.0	2
46	Monosomal complex karyotype in pediatric mixed phenotype acute leukemia. Cancer Genetics, 2011, 204, 507-511.	0.2	2
47	Refractory cytopenia of childhood with monosomy 7 presenting asÂisolated neutropenia in a patient with fragile site at 16q22. Cancer Genetics and Cytogenetics, 2010, 201, 70-71.	1.0	1
48	Cytogenetic characterization of a fibrous hamartoma of infancy with complex translocations. Cancer Genetics and Cytogenetics, 2010, 201, 66-69.	1.0	28
49	<i>MicroRNAâ€125bâ€1</i> and <i>BLID</i> upregulation resulting from a novel <i>IGH</i> translocation in childhood Bâ€Cell precursor acute lymphoblastic leukemia. Genes Chromosomes and Cancer, 2010, 49, 682-687.	1.5	28
50	Trisomy 17 in congenital plexiform (multinodular) cellular schwannoma. Cancer Genetics and Cytogenetics, 2010, 203, 313-315.	1.0	5
51	Differential diagnosis of lipomaâ€like lipoblastoma. Pediatric Blood and Cancer, 2009, 52, 132-134.	0.8	23
52	Clonal chromosome anomalies and propensity to myeloid malignancies in congenital amegakaryocytic thrombocytopenia (OMIM 604498). Haematologica, 2008, 93, 1271-1273.	1.7	34
53	Gain of 1q in pediatric myelodysplastic syndromes. Leukemia Research, 2006, 30, 1437-1441.	0.4	9
54	Inversion (11)(p15q22) with NUP98–DDX10 fusion gene in pediatric acute myeloid leukemia. Cancer Genetics and Cytogenetics, 2006, 171, 122-125.	1.0	15

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55	t(9;11)(p22;p15) with NUP98-LEDCF fusion gene in pediatric acute myeloid leukemia. Leukemia Research, 2005, 29, 467-470.	0.4	26
56	MLL–MLLT10 fusion gene in pediatric acute megakaryoblastic leukemia. Leukemia Research, 2005, 29, 1223-1226.	0.4	15
57	PLAG1-HAS2 fusion in lipoblastoma with masked 8q intrachromosomal rearrangement. Cancer Genetics and Cytogenetics, 2005, 156, 183-184.	1.0	45
58	Differential diagnosis of congenital fibrosarcoma. Cancer Genetics and Cytogenetics, 2004, 152, 167-168.	1.0	6