Maria Tzetis

List of Publications by Year
in descending order

120 papers	4,872 citations	117453 34 h-index	102304 66 g-index
122 all docs	122 docs citations	122 times ranked	6494 citing authors

#	Article	IF	Citations
1	Dating the Origin of the CCR5-Î”32 AIDS-Resistance Allele by the Coalescence of Haplotypes. American Journal of Human Genetics, 1998, 62, 1507-1515.	2.6	507
2	Consensus on the use and interpretation of cystic fibrosis mutation analysis in clinical practice. Journal of Cystic Fibrosis, 2008, 7, 179-196.	0.3	493
3	Recommendations for the classification of diseases as CFTR-related disorders. Journal of Cystic Fibrosis, 2011, 10, S86-S102.	0.3	339
4	Chronic p53-independent p21 expression causes genomic instability by deregulating replication licensing. Nature Cell Biology, 2016, 18, 777-789.	4.6	244
5	Variation in a Repeat Sequence Determines Whether a Common Variant of the Cystic Fibrosis Transmembrane Conductance Regulator Gene Is Pathogenic or Benign. American Journal of Human Genetics, 2004, 74, 176-179.	2.6	227
6	New type of disease causing mutations: the example of the composite exonic regulatory elements of splicing in CFTR exon 12. Human Molecular Genetics, 2003, 12, 1111-1120.	1.4	171
7	CFTR gene mutations - including three novel nucleotide substitutions - and haplotype background in patients with asthma, disseminated bronchiectasis and chronic obstructive pulmonary disease. Human Genetics, 2001, 108, 216-221.	1.8	132
8	Characterization of a novel 21-kb deletion, CFTRdele2,3(21 kb), in the CFTR gene: a cystic fibrosis mutation of Slavic origin common in Central and East Europe. Human Genetics, 2000, 106, 259-268.	1.8	129
9	Serum microRNA array analysis identifies miR-140-3p, miR-33b-3p and miR-671-3p as potential osteoarthritis biomarkers involved in metabolic processes. Clinical Epigenetics, 2017, 9, 127.	1.8	114
10	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. American Journal of Human Genetics, 2015, 97, 922-932.	2.6	101
11	The lysineâ€specific methyltransferase <scp>KMT</scp> 2C/ <scp>MLL</scp> 3 regulates <scp>DNA</scp> repair components in cancer. EMBO Reports, 2019, 20, .	2.0	93
12	Genotype-phenotype correlations for a wide spectrum of mutations in the Wilson disease gene (ATP7B). American Journal of Medical Genetics Part A, 2004, 131A, 168-173.	2.4	91
13	Meta-analysis of association between the ASPN D-repeat and osteoarthritis. Human Molecular Genetics, 2007, 16, 1676-1681.	1.4	78
14	Rapid Screening of Multiple Î²-Globin Gene Mutations by Real-Time PCR on the LightCycler: Application to Carrier Screening and Prenatal Diagnosis of Thalassemia Syndromes. Clinical Chemistry, 2003, 49, 769-776.	1.5	73
15	Wilson Disease in Children: Analysis of 57 Cases. Journal of Pediatric Gastroenterology and Nutrition, 2009, 48, 72-77.	0.9	70
16	Maternal epigenetics and fetal and neonatal growth. Current Opinion in Endocrinology, Diabetes and Obesity, 2017, 24, 43-46.	1.2	65
17	Asporin and knee osteoarthritis in patients of Greek origin. Osteoarthritis and Cartilage, 2006, 14, 609-611.	0.6	64
18	Real-time PCR for single-cell genotyping in sickle cell and thalassemia syndromes as a rapid, accurate, reliable, and widely applicable protocol for preimplantation genetic diagnosis. Human Mutation, 2004, 23, 513-521.	1.1	63

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19	Prenatal diagnosis of the thalassaemia syndromes by rapid DNA analytical methods. Molecular Human Reproduction, 1997, 3, 523-528.	1.3	57
20	Cystic fibrosis mutation screening in CBAVD patients and men with obstructive azoospermia or severe oligozoospermia. Molecular Human Reproduction, 1998, 4, 333-337.	1.3	57
21	Preimplantation genetic diagnosis in 10 couples at risk for transmitting Î²-thalassaemia major: clinical experience including the initiation of six singleton pregnancies. , 1999, 19, 1217-1222.		52
22	A Substitution Involving the NLGN4 Gene Associated with Autistic Behavior in the Greek Population. Genetic Testing and Molecular Biomarkers, 2009, 13, 611-615.	0.3	50
23	Dysregulated placental microRNAs in Early and Late onset Preeclampsia. Placenta, 2018, 61, 24-32.	0.7	49
24	Characterization of nondeletion Î±-thalassemia mutations in the Greek population. American Journal of Hematology, 1993, 44, 162-167.	2.0	47
25	Haplotype and mutation analysis in Greek patients with Wilson disease. European Journal of Human Genetics, 1998, 6, 487-491.	1.4	47
26	Estimating the age of p.(Phe508del) with family studies of geographically distinct European populations and the early spread of cystic fibrosis. European Journal of Human Genetics, 2018, 26, 1832-1839.	1.4	45
27	CFTR Localization in Native Airway Cells and Cell Lines Expressing Wild-type or F508del-CFTR by a Panel of Different Antibodies. Journal of Histochemistry and Cytochemistry, 2004, 52, 193-203.	1.3	44
28	A widely applicable strategy for single cell genotyping of Î²-thalassaemia mutations using DGGE analysis: application to preimplantation genetic diagnosis. , 1999, 19, 1209-1216.		40
29	Cystic fibrosis in Greece: molecular diagnosis, haplotypes, prenatal diagnosis and carrier identification amongst high-risk individuals. Clinical Genetics, 2003, 63, 400-409.	1.0	40
30	Localization studies of rare missense mutations in cystic fibrosis transmembrane conductance regulator (CFTR) facilitate interpretation of genotype-phenotype relationships. Human Mutation, 2008, 29, 1364-1372.	1.1	39
31	Characterization of more than 85% of cystic fibrosis alleles in the Greek population, including five novel mutations. Human Genetics, 1996, 99, 121-125.	1.8	38
32	Inducible nitric oxide synthase as a target for osteoarthritis treatment. Expert Opinion on Therapeutic Targets, 2018, 22, 299-318.	1.5	38
33	Molecular, haematological and clinical studies of the â^'101 Câ€ƒâ†'â€ƒT substitution of the Î²-globin gene promoter in 25 Î²-thalassaemia intermedia patients and 45 heterozygotes. British Journal of Haematology, 1999, 107, 699-706.	1.2	37
34	Contribution of the CFTR gene, the pancreatic secretory trypsin inhibitor gene (SPINK1) and the cationic trypsinogen gene (PRSS1) to the etiology of recurrent pancreatitis. Clinical Genetics, 2007, 71, 451-457.	1.0	37
35	Qualitative and quantitative analysis of mRNA associated with four putative splicing mutations (621+3Aâ†'G, 2751+2Tâ†'A, 296+1Gâ†'C, 1717â€“9Tâ†'C-D565G) and one nonsense mutation (E822X) in the CFTR gene. Human Genetics, 2001, 109, 592-601.		36
36	Methods for RNA extraction, cDNA preparation and analysis of CFTR transcripts. Journal of Cystic Fibrosis, 2004, 3, 11-15.	0.3	36

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37	Evidence for a Common Ethnic Origin of Cystic Fibrosis Mutation 3120+1Gât'A in Diverse Populations. American Journal of Human Genetics, 1998, 63, 656-662.	2.6	34
38	De novo interstitial duplication of the 15q11.2â€q14 PWS/AS region of maternal origin: Clinical description, array CGH analysis, and review of the literature. American Journal of Medical Genetics, Part A, 2010, 152A, 1925-1932.	0.7	34
39	Abnormal mRNA splicing resulting from consensus sequence splicing mutations of ATP7B. Human Mutation, 2002, 20, 260-266.	1.1	32
40	Association of repeat polymorphisms in the estrogen receptors alpha, beta (ESR1, ESR2) and androgen receptor (AR) genes with the occurrence of breast cancer. Breast, 2008, 17, 159-166.	0.9	31
41	Cystic fibrosis patients with the 3272-26A>G splicing mutation have milder disease than F508del homozygotes: a large European study. Journal of Medical Genetics, 2001, 38, 777-783.	1.5	30
42	Array comparative genomic hybridization as a clinical diagnostic tool in syndromic and nonsyndromic congenital heart disease. Pediatric Research, 2013, 73, 772-776.	1.1	29
43	Recurrent copy number variations as risk factors for autism spectrum disorders: analysis of the clinical implications. Clinical Genetics, 2016, 89, 708-718.	1.0	29
44	Phenotypic expression of a spectrum of Neurofibromatosis Type 1 (NF1) mutations identified through NGS and MLPA. Journal of the Neurological Sciences, 2018, 395, 95-105.	0.3	29
45	Microdeletion and microduplication 17q21.31 plus an additional CNV, in patients with intellectual disability, identified by array-CGH. Gene, 2012, 492, 319-324.	1.0	28
46	The improvement of the best practice guidelines for preimplantation genetic diagnosis of cystic fibrosis: toward an international consensus. European Journal of Human Genetics, 2016, 24, 469-478.	1.4	27
47	An evaluation of PGD in clinical genetic services through 3 years application for prevention of beta-thalassaemia major and sickle cell thalassaemia. Molecular Human Reproduction, 2003, 9, 301-307.	1.3	26
48	Further delineation of novel 1p36 rearrangements by array-CGH analysis: Narrowing the breakpoints and clarifying the â€œextendedâ€•phenotype. Gene, 2012, 506, 360-368.	1.0	26
49	Delineation of the Spectrum of Wilson Disease Mutations in the Greek Population and the Identification of Six Novel Mutations. Genetic Testing and Molecular Biomarkers, 2000, 4, 399-402.	1.7	25
50	Gilbert Syndrome as a Predisposing Factor for Cholelithiasis Risk in the Greek Adult Population. Genetic Testing and Molecular Biomarkers, 2009, 13, 143-146.	0.3	25
51	miR-15a and miR-24-1 as putative prognostic microRNA signatures for pediatric pilocytic astrocytomas and ependymomas. Tumor Biology, 2016, 37, 9887-9897.	0.8	25
52	Therapeutic Effects of Mesenchymal Stem Cells Derived From Bone Marrow, Umbilical Cord Blood, and Pluripotent Stem Cells in a Mouse Model of Chemically Induced Inflammatory Bowel Disease. Inflammation, 2019, 42, 1730-1740.	1.7	25
53	Mutation analysis of ten exons of the CFTR gene in Greek cystic fibrosis patients: characterization of 74.5% of CF alleles including one novel mutation. Human Genetics, 1995, 96, 364-6.	1.8	21
54	Development of a novel microarray methodology for the study of SNPs in the promoter region of the TNF-Î± geneâ€"Their association with obstructive pulmonary disease in Greek patients. Clinical Biochemistry, 2007, 40, 843-850.	0.8	21

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55	Potential sperm contributions to the murine zygote predicted by in silico analysis. Reproduction, 2017, 154, 777-788.	1.1	21
56	Spectrum of Cystic Fibrosis Mutations in Serbia and Montenegro and Strategy for Prenatal Diagnosis. Genetic Testing and Molecular Biomarkers, 2004, 8, 276-280.	1.7	20
57	Association of the CALM1 Core Promoter Polymorphism with Knee Osteoarthritis in Patients of Greek Origin. Genetic Testing and Molecular Biomarkers, 2008, 12, 263-265.	1.7	20
58	Development of novel microarray methodology for the study of mutations in the SERPINA1 and ADRB2 genesâ€"Their association with Obstructive Pulmonary Disease and Disseminated Bronchiectasis in Greek patients. Clinical Biochemistry, 2010, 43, 43-50.	0.8	20
59	Association of MMP-1 -1607 1G/2G (rs1799750) polymorphism with primary knee osteoarthritis in the Greek population. Journal of Orthopaedic Research, 2014, 32, 1155-1160.	1.2	20
60	Cystic fibrosis mutational spectrum and genotypic/phenotypic features in Greek-Cypriots, with emphasis on dehydration as presenting symptom. Clinical Genetics, 2007, 71, 290-292.	1.0	19
61	The Corfu ÎÎ² thalassaemia mutation in Greece: haematological phenotype and prevalence. British Journal of Haematology, 1991, 79, 302-305.	1.2	18
62	Generation of Human Î²-Thalassemia Induced Pluripotent Cell Lines by Reprogramming of Bone Marrowâ€"Derived Mesenchymal Stromal Cells Using Modified mRNA. Cellular Reprogramming, 2014, 16, 447-455.	0.5	17
63	Asthma and pulmonary function abnormalities in heterozygotes for cystic fibrosis transmembrane regulator gene mutations. International Journal of Clinical and Experimental Medicine, 2008, 1, 345-9.	1.3	17
64	GILBERT SYNDROME ASSOCIATED WITH Î²-THALASSEMIA. Pediatric Hematology and Oncology, 2001, 18, 477-484.	0.3	16
65	The clinical utility of molecular karyotyping using high-resolution array-comparative genomic hybridization. Expert Review of Molecular Diagnostics, 2012, 12, 449-457.	1.5	16
66	Can trophectoderm RNA analysis predict human blastocyst competency?. Systems Biology in Reproductive Medicine, 2019, 65, 312-325.	1.0	16
67	Molecular Characterization of Homozygous (High HbA2) Î²-Thalassemia Intermedia in Greece. Pediatric Hematology and Oncology, 1995, 12, 37-45.	0.3	13
68	Quantitative methods for the analysis of CFTR transcripts/splicing variants. Journal of Cystic Fibrosis, 2004, 3, 17-23.	0.3	13
69	Gilbert syndrome: analysis of the promoter region of the uridine diphosphate-glucuronosyltransferase 1 gene in the Greek population. European Journal of Pediatrics, 2000, 159, 873-874.	1.3	12
70	Development of a multidisciplinary clinic of neurofibromatosis type 1 and other neurocutaneous disorders in Greece. A 3-year experience. Postgraduate Medicine, 2019, 131, 445-452.	0.9	12
71	The impact of preimplantation genetic testing for aneuploidies (PGT-A) on clinical outcomes in high risk patients. Journal of Assisted Reproduction and Genetics, 2022, 39, 1341-1349.	1.2	12
72	Multiplex sequence variation detection throughout the CFTR gene appropriate for preimplantation genetic diagnosis in populations with heterogeneity of cystic fibrosis mutations. Molecular Human Reproduction, 2002, 8, 880-886.	1.3	11

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73	Genetic Polymorphisms in theUGT1A1Gene and Breast Cancer Risk in Greek Women. Genetic Testing and Molecular Biomarkers, 2007, 11, 303-306.	1.7	11
74	An interstitial deletion at 8q23.1-q24.12 associated with Langer-Giedion syndrome/ Trichorhinophalangeal syndrome (TRPS) type II and Cornelia de Lange syndrome 4. Molecular Cytogenetics, 2015, 8, 64.	0.4	11
75	Compound heterozygosity of a paternal submicroscopic deletion and a maternal missense mutation in POR gene: Antleyâ€bixler syndrome phenotype in three sibling fetuses. Birth Defects Research Part A: Clinical and Molecular Teratology, 2016, 106, 536-541.	1.6	11
76	Gilbert's syndrome as a predisposing factor for idiopathic cholelithiasis in children. Haematologica, 2003, 88, 1193-4.	1.7	11
77	Hematologic Phenotype of the Mutations Ivs1-n6 (T â†’ C), IVS1-n110 (C â†’ A), AND CD39 (C â†’ T) IN CARRIERS OF P-THALASSEMIA IN GREECE. Pediatric Hematology and Oncology, 1994, 11, 509-517.	0.3	10
78	Rapid clinical-scale propagation of mesenchymal stem cells using cultures initiated with immunoselected bone marrow CD105+ cells. Journal of Cellular and Molecular Medicine, 2011, 15, 1983-1988.	1.6	10
79	Quadruple-allele dipstick test for simultaneous visual genotyping of A896G (Asp299Gly) and C1196T (Thr399Ile) polymorphisms in the toll-like receptor-4 gene. Clinica Chimica Acta, 2011, 412, 1968-1972.	0.5	9
80	Clinical and molecular description of a fetus in prenatal diagnosis with a rare de novo ring 10 and deletions of 12.59Mb in 10p15.3â€“p14 and 4.22Mb in 10q26.3. European Journal of Medical Genetics, 2012, 55, 75-79.	0.7	9
81	Cystic fibrosis genetic counseling difficulties due to the identification of novel mutations in the CFTR gene. Journal of Cystic Fibrosis, 2012, 11, 344-348.	0.3	9
82	Steroid hormones polymorphisms and cholelithiasis in Greek population. Liver International, 2007, 27, 61-8.	1.9	8
83	Combined microdeletions and CHD7 mutation causing severe CHARGE/DiGeorge syndrome: clinical presentation and molecular investigation by array-CGH. Journal of Human Genetics, 2010, 55, 761-763.	1.1	8
84	VPA-induced recurrent pancreatitis in a cystic fibrosis carrier. European Journal of Paediatric Neurology, 2011, 15, 453-455.	0.7	8
85	Association of TLR4 Single-Nucleotide Polymorphisms and Sarcoidosis in Greek Patients. Genetic Testing and Molecular Biomarkers, 2009, 13, 849-853.	0.3	7
86	BTNL2 gene SNPs as a contributing factor to sarcoidosis pathogenesis in a cohort of Greek patients. Meta Gene, 2014, 2, 619-630.	0.3	7
87	Central precocious puberty in a boy with 22q13 deletion syndrome and NOTCH-1 gene duplication. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 1307-1311.	0.4	7
88	The effects of aging on molecular modulators of human embryo implantation. IScience, 2021, 24, 102751.	1.9	7
89	Mild cystic fibrosis phenotype in patients with the 3272-26A > G mutation.. Journal of Medical Genetics, 1995, 32, 406-407.	1.5	6
90	Cystic Fibrosis Conductance Regulator, Tumor Necrosis Factor, Interferon Alpha-10, Interferon Alpha-17, and Interferon Gamma Genotyping as Potential Risk Markers in Pulmonary Sarcoidosis Pathogenesis in Greek Patients. Genetic Testing and Molecular Biomarkers, 2010, 14, 577-584.	0.3	6

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91	Mesenchymal Derivatives of Genetically Unstable Human Embryonic Stem Cells Are Maintained Unstable but Undergo Senescence in Culture As Do Bone Marrowâ€“Derived Mesenchymal Stem Cells. Cellular Reprogramming, 2014, 16, 1-8.	0.5	6
92	High resolution Chromosomal Microarray Analysis (CMA) enhances the genetic profile of pediatric B-cell Acute Lymphoblastic Leukemia patients. Leukemia Research, 2019, 83, 106177.	0.4	6
93	Reprogramming of bone marrow derived mesenchymal stromal cells to human induced pluripotent stem cells from pediatric patients with hematological diseases using a commercial mRNA kit. Blood Cells, Molecules, and Diseases, 2019, 76, 32-39.	0.6	6
94	Microduplication 3q13.2q13.31 identified in a male with dysmorphic features and multiple congenital anomalies. American Journal of Medical Genetics, Part A, 2014, 164, 666-670.	0.7	5
95	Prenatal diagnosis for CF using High Resolution Melting Analysis and simultaneous haplotype analysis through QF-PCR. Journal of Cystic Fibrosis, 2014, 13, 617-622.	0.3	5
96	A boy with conduct disorder (CD), attention deficit hyperactivity disorder (ADHD), borderline intellectual disability, and 47,XXY syndrome in combination with a 7q11.23 duplication, 11p15.5 deletion, and 20q13.33 deletion. Child and Adolescent Psychiatry and Mental Health, 2016, 10, 33.	1.2	5
97	Coffin-Siris Syndrome 4-Related Spectrum in a Young Woman Caused by a Heterozygous <i>SMARCA4</i> Deletion Detected by High-Resolution aCGH. Molecular Syndromology, 2020, 11, 141-145.	0.3	5
98	Clinical, haematological, and genetic studies of type 2 normal Hb A2 beta thalassaemia.. Journal of Medical Genetics, 1988, 25, 195-199.	1.5	4
99	Array-CGH revealed one of the smallest 16q21q22.1 microdeletions in a female patient with psychomotor retardation. European Journal of Paediatric Neurology, 2013, 17, 316-320.	0.7	4
100	Are ALOX5AP gene SNPs a risk or protective factor for stroke?. Gene, 2014, 548, 56-60.	1.0	4
101	Multi-allele genotyping platform for the simultaneous detection of mutations in the Wilson disease related ATP7B gene. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2015, 1006, 201-208.	1.2	4
102	Genomic screening of ABCA4 and array CGH analysis underline the genetic variability of Greek patients with inherited retinal diseases. Meta Gene, 2016, 8, 37-43.	0.3	4
103	Proliferative and chondrogenic potential of mesenchymal stromal cells from pluripotent and bone marrow cells. Histology and Histopathology, 2020, 35, 1415-1426.	0.5	4
104	Familial Pelizaeusâ€“Merzbacher disease caused by a 320.6â€kb Xq22.2 duplication and the pathological findings of a male fetus. Birth Defects Research Part A: Clinical and Molecular Teratology, 2012, 94, 494-498.	1.6	3
105	A Female Patient with Xq28 Microduplication Presenting with Myotubular Myopathy, Confirmed with a Custom-Designed X-array. Neuropediatrics, 2019, 50, 061-063.	0.3	3
106	Identification of two novel mutations (296 + 1G â^' C and A46D) in exon 2 of the CFTR gene in Greek cystic fibrosis patients. Molecular and Cellular Probes, 1995, 9, 283-285.	0.9	2
107	Genotyping Efficiency of 2 Primer Sets and an Unlabeled Oligonucleotide Probe for the p.Phe508del in Exon 10 of the CFTR Gene as Determined with High-Resolution Melting Analysis. Clinical Chemistry, 2012, 58, 1490-1492.	1.5	2
108	An unusual case of catâ€eye syndrome phenotype and extragonadal mature teratoma: Review of the literature. Birth Defects Research Part A: Clinical and Molecular Teratology, 2012, 94, 561-566.	1.6	2

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109	Congenital Cataracts, Facial Dysmorphism, and Neuropathy Syndrome: Additional Clinical Features. Pediatric Neurology, 2017, 67, e5-e6.	1.0	2
110	Application of high-resolution array comparative genomic hybridization in children with unknown syndromic microcephaly. Pediatric Research, 2017, 82, 253-260.	1.1	2
111	Association of Polymorphisms in the Promoter Region of NOS2A Gene with Primary Knee Osteoarthritis in the Greek Population. Cureus, 2020, 12, e6780.	0.2	2
112	Single-cell high resolution melting analysis: A novel, generic, pre-implantation genetic diagnosis (PGD) method applied to cystic fibrosis (HRMA CF-PGD). Journal of Cystic Fibrosis, 2016, 15, 163-170.	0.3	1
113	Ophthalmologic manifestations of adult patients with cystic fibrosis. European Journal of Ophthalmology, 2021, , 112067212110087.	0.7	1
114	Severe Hemophilia A and Moyamoya Syndrome in a 19-Year-Old Boy Caused by Xq28 Microdeletion. Case Reports in Neurology, 2022, 14, 261-267.	0.3	1
115	Trigonocephaly and Wilson??s disease in two siblings. Clinical Dysmorphology, 2005, 14, 161-164.	0.1	0
116	MULTIDISCIPLINARY MEDICAL EVALUATION OF CHILDREN YOUNGER THAN 7.5 YEARS BORN AFTER PREIMPLANTATION GENETIC DIAGNOSIS FOR MONOGENIC DISEASES. Pediatrics, 2008, 121, S102-S102.	1.0	0
117	239-kb Microdeletion Spanning KMT2E in a Child with Developmental Delay: Further Delineation of the Phenotype. Molecular Syndromology, 2021, 12, 321-326.	0.3	0
118	Gilbert Syndrome as a Predisposing Factor for Cholelithiasis Risk in the Greek Adult Population. Genetic Testing and Molecular Biomarkers, 2009, .	1.7	0
119	A novel de novo paracentric inversion [inv(20)(q13.1q13.3)] accompanied by an 11q14.3-q21 microdeletion in a pediatric patient with an intellectual disability. Balkan Journal of Medical Genetics, 2018, 21, 63-67.	0.5	0
120	NFB-17. "Optic Pathway findings in children with Neurofibromatosis type-1 (NF-1). Neuro-Oncology, 2022, 24, i131-i131.	0.6	0