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	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
2	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
3	NFB-17. "Optic Pathway findings in children with Neurofibromatosis type-1 (NF-1). Neuro-Oncology, 2022, 24, i131-i131.	0.6	0
4	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
5	Ophthalmologic manifestations of adult patients with cystic fibrosis. European Journal of Ophthalmology, 2021, , 112067212110087.	0.7	1
6	The effects of aging on molecular modulators of human embryo implantation. IScience, 2021, 24, 102751.	1.9	7
7	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
8	Proliferative and chondrogenic potential of mesenchymal stromal cells from pluripotent and bone marrow cells. Histology and Histopathology, 2020, 35, 1415-1426.	0.5	4
9	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
10	Can trophectoderm RNA analysis predict human blastocyst competency?. Systems Biology in Reproductive Medicine, 2019, 65, 312-325.	1.0	16
11	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
12	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
13	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
14	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
15	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
16	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
17	Inducible nitric oxide synthase as a target for osteoarthritis treatment. Expert Opinion on Therapeutic Targets, 2018, 22, 299-318.	1.5	38
18	Dysregulated placental microRNAs in Early and Late onset Preeclampsia. Placenta, 2018, 61, 24-32.	0.7	49

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19	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
20	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
21	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
22	Maternal epigenetics and fetal and neonatal growth. Current Opinion in Endocrinology, Diabetes and Obesity, 2017, 24, 43-46.	1.2	65
23	Congenital Cataracts, Facial Dysmorphism, and Neuropathy Syndrome: Additional Clinical Features. Pediatric Neurology, 2017, 67, e5-e6.	1.0	2
24	Application of high-resolution array comparative genomic hybridization in children with unknown syndromic microcephaly. Pediatric Research, 2017, 82, 253-260.	1.1	2
25	Potential sperm contributions to the murine zygote predicted by in silico analysis. Reproduction, 2017, 154, 777-788.	1.1	21
26	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
27	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
28	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
29	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
30	Chronic p53-independent p21 expression causes genomic instability by deregulating replication licensing. Nature Cell Biology, 2016, 18, 777-789.	4.6	244
31	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
32	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
33	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
34	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
35	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
36	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

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37	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. American Journal of Human Genetics, 2015, 97, 922-932.	2.6	101
38	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
39	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
40	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
41	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
42	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
43	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
44	Are ALOX5AP gene SNPs a risk or protective factor for stroke?. Gene, 2014, 548, 56-60.	1.0	4
45	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
46	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
47	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
48	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
49	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
50	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
51	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
52	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
53	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
54	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

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55	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
56	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
57	Recommendations for the classification of diseases as CFTR-related disorders. Journal of Cystic Fibrosis, 2011, 10, S86-S102.	0.3	339
58	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
59	VPA-induced recurrent pancreatitis in a cystic fibrosis carrier. European Journal of Paediatric Neurology, 2011, 15, 453-455.	0.7	8
60	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
61	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
62	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
63	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
64	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
65	Wilson Disease in Children: Analysis of 57 Cases. Journal of Pediatric Gastroenterology and Nutrition, 2009, 48, 72-77.	0.9	70
66	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
67	Association of TLR4 Single-Nucleotide Polymorphisms and Sarcoidosis in Greek Patients. Genetic Testing and Molecular Biomarkers, 2009, 13, 849-853.	0.3	7
68	Gilbert Syndrome as a Predisposing Factor for Cholelithiasis Risk in the Greek Adult Population. Genetic Testing and Molecular Biomarkers, 2009, .	1.7	0
69	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
70	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
71	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
72	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

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73	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
74	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
75	Meta-analysis of association between the ASPN D-repeat and osteoarthritis. Human Molecular Genetics, 2007, 16, 1676-1681.	1.4	78
76	Genetic Polymorphisms in theUGT1A1Gene and Breast Cancer Risk in Greek Women. Genetic Testing and Molecular Biomarkers, 2007, 11, 303-306.	1.7	11
77	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
78	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
79	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
80	Steroid hormones polymorphisms and cholelithiasis in Greek population. Liver International, 2007, 27, 61-8.	1.9	8
81	Asporin and knee osteoarthritis in patients of Greek origin. Osteoarthritis and Cartilage, 2006, 14, 609-611.	0.6	64
82	Trigonocephaly and Wilson??s disease in two siblings. Clinical Dysmorphology, 2005, 14, 161-164.	0.1	0
83	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
84	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
85	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
86	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
87	Methods for RNA extraction, cDNA preparation and analysis of CFTR transcripts. Journal of Cystic Fibrosis, 2004, 3, 11-15.	0.3	36
88	Quantitative methods for the analysis of CFTR transcripts/splicing variants. Journal of Cystic Fibrosis, 2004, 3, 17-23.	0.3	13
89	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
90	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

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91	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
92	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
93	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
94	Gilbert's syndrome as a predisposing factor for idiopathic cholelithiasis in children. Haematologica, 2003, 88, 1193-4.	1.7	11
95	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
96	Abnormal mRNA splicing resulting from consensus sequence splicing mutations of ATP7B. Human Mutation, 2002, 20, 260-266.	1.1	32
97	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.	1.8	36
98	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
99	GILBERT SYNDROME ASSOCIATED WITH Î²-THALASSEMIA. Pediatric Hematology and Oncology, 2001, 18, 477-484.	0.3	16
100	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
101	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
102	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
103	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
104	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
105	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
106	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
107	Haplotype and mutation analysis in Greek patients with Wilson disease. European Journal of Human Genetics, 1998, 6, 487-491.	1.4	47
108	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

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109	Evidence for a Common Ethnic Origin of Cystic Fibrosis Mutation 3120+1Gâ†'A in Diverse Populations. American Journal of Human Genetics, 1998, 63, 656-662.	2.6	34
110	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
111	Prenatal diagnosis of the thalassaemia syndromes by rapid DNA analytical methods. Molecular Human Reproduction, 1997, 3, 523-528.	1.3	57
112	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
113	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
114	Molecular Characterization of Homozygous (High HbA2) Î²-Thalassemia Intermedia in Greece. Pediatric Hematology and Oncology, 1995, 12, 37-45.	0.3	13
115	Mild cystic fibrosis phenotype in patients with the 3272-26A > G mutation.. Journal of Medical Genetics, 1995, 32, 406-407.	1.5	6
116	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
117	Hematologic Phenotype of the Mutations Ivs1-n6 (T â†'. C), lVS1-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
118	Characterization of nondeletion Î±-thalassemia mutations in the Greek population. American Journal of Hematology, 1993, 44, 162-167.	2.0	47
119	The Corfu ÎÎ² thalassaemia mutation in Greece: haematological phenotype and prevalence. British Journal of Haematology, 1991, 79, 302-305.	1.2	18
120	Clinical, haematological, and genetic studies of type 2 normal Hb A2 beta thalassaemia.. Journal of Medical Genetics, 1988, 25, 195-199.	1.5	4