Jan Traeger-Synodinos

List of Publications by Citations

162 papers	4,513 citations	36 h-index	61 g-index
176 ext. papers	4,962 ext. citations	4.3 avg, IF	5.17 L-index

#	Paper	IF	Citations
162	The ESHRE PGD Consortium: 10 years of data collection. *Human Reproduction Update*, **2012**, 18, 234-47	15.8	223
161	The causes of misdiagnosis and adverse outcomes in PGD. *Human Reproduction*, **2009**, 24, 1221-8	5.7	175
160	ESHRE PGD consortium best practice guidelines for amplification-based PGD. *Human Reproduction*, **2011**, 26, 33-40	5.7	174
159	What next for preimplantation genetic screening (PGS)? A position statement from the ESHRE PGD Consortium Steering Committee. *Human Reproduction*, **2010**, 25, 821-3	5.7	140
158	Blastocyst biopsy versus cleavage stage biopsy and blastocyst transfer for preimplantation genetic diagnosis of beta-thalassaemia: a pilot study. *Human Reproduction*, **2007**, 22, 1443-9	5.7	127
157	Mutations in the chromatin-associated protein ATRX. *Human Mutation*, **2008**, 29, 796-802	4.7	127
156	Systematic documentation and analysis of human genetic variation in hemoglobinopathies using the microattribution approach. *Nature Genetics*, **2011**, 43, 295-301	36.3	125
155	ESHRE PGD consortium best practice guidelines for organization of a PGD centre for PGD/preimplantation genetic screening. *Human Reproduction*, **2011**, 26, 14-24	5.7	117
154	ESHRE PGD Consortium data collection X: cycles from January to December 2007 with pregnancy follow-up to October 2008. *Human Reproduction*, **2010**, 25, 2685-707	5.7	113
153	ESHRE PGD Consortium data collection XIII: cycles from January to December 2010 with pregnancy follow-up to October 2011. *Human Reproduction*, **2015**, 30, 1763-89	5.7	107
152	ESHRE PGD Consortium data collection IX: cycles from January to December 2006 with pregnancy follow-up to October 2007. *Human Reproduction*, **2009**, 24, 1786-810	5.7	103
151	Unstable and thalassemic alpha chain hemoglobin variants: a cause of Hb H disease and thalassemia intermedia. *Hemoglobin*, **2008**, 32, 327-49	0.6	101
150	ESHRE PGD Consortium data collection VI: cycles from January to December 2003 with pregnancy follow-up to October 2004. *Human Reproduction*, **2007**, 22, 323-36	5.7	98
149	Defective synthesis of HbE is due to reduced levels of beta E mRNA. *Nature*, **1980**, 288, 497-9	50.4	96
148	ESHRE PGD Consortium data collection V: cycles from January to December 2002 with pregnancy follow-up to October 2003. *Human Reproduction*, **2006**, 21, 3-21	5.7	94
147	ESHRE PGD consortium data collection VII: cycles from January to December 2004 with pregnancy follow-up to October 2005. *Human Reproduction*, **2008**, 23, 741-55	5.7	79
146	Unravelling the Genetic Basis of ACTH-Mediated Aldosterone Hypersecretion in Hypertensive Patients Without Primary Aldosteronism. *Journal of the Endocrine Society*, **2021**, 5, A73-A74	0.4	78

145	ESHRE PGD Consortium data collection XII: cycles from January to December 2009 with pregnancy follow-up to October 2010. *Human Reproduction*, **2014**, 29, 880-903	5.7	74
144	ESHRE PGD Consortium data collection VIII: cycles from January to December 2005 with pregnancy follow-up to October 2006. *Human Reproduction*, **2008**, 23, 2629-45	5.7	72
143	Rapid screening of multiple beta-globin gene mutations by real-time PCR on the LightCycler: application to carrier screening and prenatal diagnosis of thalassemia syndromes. *Clinical Chemistry* , **2003**, 49, 769-76	5.5	67
142	The triplicated alpha-globin gene locus in beta-thalassaemia heterozygotes: clinical, haematological, biosynthetic and molecular studies. *British Journal of Haematology*, **1996**, 95, 467-71	4.5	67
141	ESHRE PGD Consortium data collection XI: cycles from January to December 2008 with pregnancy follow-up to October 2009. *Human Reproduction*, **2012**, 27, 1887-911	5.7	66
140	Birth of a healthy infant following trophectoderm biopsy from blastocysts for PGD of beta-thalassaemia major. *Human Reproduction*, **2005**, 20, 1855-9	5.7	66
139	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, 513-21	4.7	57
138	Phenotypic and molecular diversity of haemoglobin H disease: a Greek experience. *British Journal of Haematology*, **2000**, 111, 915-23	4.5	57
137	Genotyping of single-nucleotide polymorphisms by primer extension reaction in a dry-reagent dipstick format. *Analytical Chemistry*, **2007**, 79, 395-402	7.8	55
136	Successful long-term immunologic reconstitution by allogeneic hematopoietic stem cell transplantation cures patients with autosomal dominant hyper-IgE syndrome. *Journal of Allergy and Clinical Immunology*, **2010**, 126, 392-4	11.5	54
135	Evaluation of PCR-based preimplantation genetic diagnosis applied to monogenic diseases: a collaborative ESHRE PGD consortium study. *European Journal of Human Genetics*, **2014**, 22, 1012-8	5.3	51
134	EMQN Best Practice Guidelines for molecular and haematology methods for carrier identification and prenatal diagnosis of the haemoglobinopathies. *European Journal of Human Genetics*, **2015**, 23, 426-37	5.3	50
133	Sensitive Monogenic Noninvasive Prenatal Diagnosis by Targeted Haplotyping. *American Journal of Human Genetics*, **2017**, 101, 326-339	11	50
132	Prenatal diagnosis of the thalassaemia syndromes by rapid DNA analytical methods. *Molecular Human Reproduction*, **1997**, 3, 523-8	4.4	48
131	Multianalyte, dipstick-type, nanoparticle-based DNA biosensor for visual genotyping of single-nucleotide polymorphisms. *Biosensors and Bioelectronics*, **2009**, 24, 3135-9	11.8	45
130	Characterization of nondeletion alpha-thalassemia mutations in the Greek population. *American Journal of Hematology*, **1993**, 44, 162-7	7.1	44
129	A base substitution (T-->C) in codon 29 of the alpha 2-globin gene causes alpha thalassaemia. *British Journal of Haematology*, **1993**, 85, 546-52	4.5	40
128	Cystic fibrosis in Greece: molecular diagnosis, haplotypes, prenatal diagnosis and carrier identification amongst high-risk individuals. *Clinical Genetics*, **2003**, 63, 400-9	4	38

127	Preimplantation genetic diagnosis in 10 couples at risk for transmitting beta-thalassaemia major: clinical experience including the initiation of six singleton pregnancies. *Prenatal Diagnosis*, **1999**, 19, 1217-22	3.2	38
126	Advances in technologies for screening and diagnosis of hemoglobinopathies. *Biomarkers in Medicine*, **2014**, 8, 119-31	2.3	35
125	Low total antioxidant status is implicated with high 8-hydroxy-2-deoxyguanosine serum concentrations in phenylketonuria. *Clinical Biochemistry*, **2005**, 38, 239-42	3.5	35
124	Pre-implantation genetic diagnosis. *Best Practice and Research in Clinical Obstetrics and Gynaecology* , **2017**, 39, 74-88	4.6	34
123	Cell-free DNA levels in acute myocardial infarction patients during hospitalization. *Acta Cardiologica*, **2009**, 64, 51-7	0.9	31
122	An overview of current microarray-based human globin gene mutation detection methods. *Hemoglobin*, **2007**, 31, 289-311	0.6	31
121	Preimplantation genetic diagnosis in clinical practice. *Journal of Medical Genetics*, **2002**, 39, 6-11	5.8	30
120	Erythroid bone marrow activity and red cell hemoglobinization in iron sufficient beta-thalassemia heterozygotes as reflected by soluble transferrin receptor and reticulocyte hemoglobin in content. Correlation with genotypes and Hb A(2) levels. *Haematologica*, **2003**, 88, 631-6	6.6	30
119	Molecular, haematological and clinical studies of the -101 C --> T substitution of the beta-globin gene promoter in 25 beta-thalassaemia intermedia patients and 45 heterozygotes. *British Journal of Haematology*, **1999**, 107, 699-706	4.5	28
118	A widely applicable strategy for single cell genotyping of □thalassaemia mutations using DGGE analysis: application to preimplantation genetic diagnosis **1999**, 19, 1209-1216		27
117	A rare example that coinheritance of a severe form of beta-thalassemia and alpha-thalassemia interact in a "synergistic" manner to balance the phenotype of classic thalassemic syndromes. *Blood Cells, Molecules, and Diseases*, **2004**, 32, 319-24	2.1	26
116	Rapid and accurate quantitation of Hb Barts and Hb H using weak cation exchange high performance liquid chromatography: correlation with the alpha-thalassemia genotype. *Hemoglobin*, **1999**, 23, 203-11	0.6	26
115	Preimplantation genetic diagnosis (PGD), a collaborative activity of clinical genetic departments and IVF centres. *Prenatal Diagnosis*, **2001**, 21, 1086-92	3.2	25
114	Phenotypic and molecular diversity of haemoglobin H disease: a Greek experience. *British Journal of Haematology*, **2000**, 111, 915-923	4.5	25
113	Analysis of low density lipoprotein receptor gene mutations and microsatellite haplotypes in Greek FH heterozygous children: six independent ancestors account for 60% of probands. *Human Genetics* , **1998**, 102, 343-7	6.3	24
112	Noninvasive prenatal diagnosis of beta-thalassaemia using individual fetal erythroblasts isolated from maternal blood after enrichment. *Prenatal Diagnosis*, **2007**, 27, 1228-32	3.2	23
111	Instability of beta E-messenger RNA during erythroid cell maturation in hemoglobin E homozygotes. *Journal of Clinical Investigation*, **1982**, 69, 1050-3	15.9	23
110	Photoprotein aequorin as a novel reporter for SNP genotyping by primer extension-application to the variants of mannose-binding lectin gene. *Human Mutation*, **2006**, 27, 279-85	4.7	22

109	Real-time PCR for prenatal and preimplantation genetic diagnosis of monogenic diseases. *Molecular Aspects of Medicine*, **2006**, 27, 176-91	16.7	22
108	Phenotypic and genotypic variability in four males with MECP2 gene sequence aberrations including a novel deletion. *Pediatric Research*, **2010**, 67, 551-6	3.2	21
107	A novel p.Arg970X mutation in the last exon of the CDKL5 gene resulting in late-onset seizure disorder. *European Journal of Paediatric Neurology*, **2010**, 14, 188-91	3.8	20
106	Distinct phenotypic expression associated with a new hyperunstable alpha globin variant (Hb heraklion, alpha1cd37(C2)Pro>0): comparison to other alpha-thalassemic hemoglobinopathies. *Blood Cells, Molecules, and Diseases*, **2000**, 26, 276-84	2.1	20
105	Erythroid Marrow Activity and Hemoglobin H Levels in Hemoglobin H Disease. *Journal of Pediatric Hematology/Oncology*, **1998**, 20, 539-544	1.2	20
104	An electronic infrastructure for research and treatment of the thalassemias and other hemoglobinopathies: the Euro-mediterranean ITHANET project. *Hemoglobin*, **2009**, 33, 163-76	0.6	19
103	High-throughput microtiter well-based chemiluminometric genotyping of 15 HBB gene mutations in a dry-reagent format. *Clinical Chemistry*, **2007**, 53, 384-91	5.5	19
102	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, 301-7	4.4	19
101	Preconception carrier screening and prenatal diagnosis in thalassemia and hemoglobinopathies: challenges and future perspectives. *Expert Review of Molecular Diagnostics*, **2017**, 17, 281-291	3.8	18
100	Prenatal diagnosis of hemoglobin disorders: present and future strategies. *Clinical Biochemistry*, **2009**, 42, 1767-79	3.5	18
99	A rare thalassemic syndrome caused by interaction of Hb Adana [alpha59(E8)Gly-->Asp] with an alpha+-thalassemia deletion: clinical aspects in two cases. *Hemoglobin*, **2008**, 32, 361-9	0.6	18
98	Pregnancies following blastocyst stage transfer in PGD cycles at risk for beta-thalassaemic haemoglobinopathies. *Human Reproduction*, **2002**, 17, 25-31	5.7	18
97	Interaction of an alpha(+)-thalassemia deletion with either a highly unstable alpha-globin variant (alpha2, codon 59, GGC-->GAC) or a nondeletional alpha-thalassemia mutation (AATAAA-->AATAAG): comparison of phenotypes illustrating "dominant" alpha-thalassemia. *Hemoglobin*, **1999**, 23, 325-37	0.6	18
96	Prenatal and preimplantation diagnosis of hemoglobinopathies. *International Journal of Laboratory Hematology*, **2018**, 40 Suppl 1, 74-82	2.5	17
95	Molecular studies of beta-thalassemia heterozygotes with raised Hb F levels. *Hemoglobin*, **2000**, 24, 203-26	2.6	17
94	The interaction of alpha zero thalassaemia with Hb Icaria: three unusual cases of haemoglobinopathy H. *British Journal of Haematology*, **1996**, 92, 332-5	4.5	16
93	The clinical utility of PGD with HLA matching: a collaborative multi-centre ESHRE study. *Human Reproduction*, **2018**, 33, 520-530	5.7	15
92	MECP2 mutations and clinical correlations in Greek children with Rett syndrome and associated neurodevelopmental disorders. *Brain and Development*, **2012**, 34, 487-95	2.2	15

91	Novel and known microsatellite markers within the β-globin cluster to support robust preimplantation genetic diagnosis of β-thalassemia and sickle cell syndromes. *Hemoglobin*, **2011**, 35, 56-66	0.6	15
90	Prenatal, noninvasive and preimplantation genetic diagnosis of inherited disorders: hemoglobinopathies. *Expert Review of Molecular Diagnostics*, **2011**, 11, 299-312	3.8	15
89	Scanning of beta-globin gene for identification of beta-thalassemia mutation in Romanian population. *Journal of Cellular and Molecular Medicine*, **2004**, 8, 232-40	5.6	15
88	The Corfu delta beta thalassaemia mutation in Greece: haematological phenotype and prevalence. *British Journal of Haematology*, **1991**, 79, 302-5	4.5	15
87	The molecular basis of normal HbA2 (type 2) beta-thalassemia in Greece. *Hematologic Pathology*, **1994**, 8, 25-34		15
86	Pre-implantation HLA matching: The production of a Saviour Child. *Best Practice and Research in Clinical Obstetrics and Gynaecology*, **2017**, 44, 76-89	4.6	14
85	A high incidence of mutations in exon 6 of the low-density lipoprotein receptor gene in Greek familial hypercholesterolemia patients, including a novel mutation. *Human Mutation*, **1997**, 9, 274-6	4.7	14
84	Hb Aghia Sophia [alpha62(E11)Val-->0 (alpha1)], an "in-frame" deletion causing alpha-thalassemia. *Hemoglobin*, **1999**, 23, 317-24	0.6	14
83	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	6.3	14
82	A minimal set of SNPs for the noninvasive prenatal diagnosis of β-thalassaemia. *Annals of Human Genetics*, **2013**, 77, 115-24	2.2	13
81	Nucleotide variations in the NPHS2 gene in Greek children with steroid-resistant nephrotic syndrome. *Genetic Testing and Molecular Biomarkers*, **2009**, 13, 249-56	1.6	13
80	An alpha-thalassemic hemoglobinopathy: homozygosity for the HB Agrinio alpha 2-globin chain variant. *Hemoglobin*, **1998**, 22, 209-15	0.6	13
79	Severe hypertriglyceridaemia in diabetic ketoacidosis: clinical and genetic study. *Diabetic Medicine*, **2004**, 21, 380-2	3.5	13
78	Erythroid marrow activity and functional anemia in patients with the rare interaction of a single functional a-globin and beta-globin gene. *Haematologica*, **2001**, 86, 363-7	6.6	13
77	Psychomotor development of children born after preimplantation genetic diagnosis and parental stress evaluation. *World Journal of Pediatrics*, **2012**, 8, 309-16	4.6	12
76	Preliminary mutation analysis in the phenylalanine hydroxylase gene in Greek PKU and HPA patients. *Human Genetics*, **1994**, 94, 573-5	6.3	12
75	Abnormal DLK1/MEG3 imprinting correlates with decreased HERV-K methylation after assisted reproduction and preimplantation genetic diagnosis. *Stress*, **2013**, 16, 689-97	3	11
74	Broad and unexpected phenotypic expression in Greek children with steroid-resistant nephrotic syndrome due to mutations in the Wilms Tumor 1 (WT1) gene. *European Journal of Pediatrics*, **2011**, 170, 1529-34	4.1	11

73	Molecular, haematological and clinical studies of a silent beta-gene C-->G mutation at 6 bp 3Ν to the termination codon (+1480 C-->G) in twelve Greek families. *British Journal of Haematology*, **1998**, 103, 45-51	4.5	11
72	Molecular characterization of homozygous (high HbA2) beta-thalassemia intermedia in Greece. *Pediatric Hematology and Oncology*, **1995**, 12, 37-45	1.7	11
71	The experience of 3 years of external quality assessment of preimplantation genetic diagnosis for cystic fibrosis. *European Journal of Human Genetics*, **2013**, 21, 800-6	5.3	10
70	Two new beta-thalassemia deletions compromising prenatal diagnosis in an Italian and a Turkish couple seeking prevention. *Haematologica*, **2009**, 94, 1289-92	6.6	10
69	High-throughput microtiter well-based bioluminometric genotyping of two single-nucleotide polymorphisms in the toll-like receptor-4 gene. *Analytical Biochemistry*, **2008**, 376, 235-41	3.1	10
68	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, 880-6	4.4	10
67	Complex preimplantation genetic diagnosis for beta-thalassaemia, sideroblastic anaemia, and human leukocyte antigen (HLA)-typing. *Systems Biology in Reproductive Medicine*, **2016**, 62, 69-76	2.9	9
66	Visual screening for JAK2V617F mutation by a disposable dipstick. *Analytical and Bioanalytical Chemistry*, **2010**, 397, 1911-6	4.4	9
65	First observation of Hb Taybe [Codons 38/39 (-Acc) Thr-->0 (alpha1)] in Greece: clinical and hematological findings in patients with co-inherited alpha+-thalassemia mutations. *Hemoglobin*, **2008**, 32, 371-8	0.6	9
64	Unusual phenotypic observations associated with a rare HbH disease genotype (--Med/alphaTSaudialpha): implications for clinical management. *British Journal of Haematology*, **2002**, 119, 265-7	4.5	9
63	Hb Mont Saint Aignan [beta128(H6)Ala-->Pro]: a new unstable variant leading to chronic microcytic anemia. *Hemoglobin*, **2001**, 25, 57-65	0.6	9
62	Hematologic phenotype of the mutations IVS1-n6 (T-->C), IVS1-n110 (G-->A), and CD39 (C-->T) in carriers of beta-thalassemia in Greece. *Pediatric Hematology and Oncology*, **1994**, 11, 509-17	1.7	9
61	Rare beta-thalassemia alleles in the Greek and Greek Cypriot populations. *Hemoglobin*, **1998**, 22, 89-94	0.6	8
60	Association of unstable hemoglobin variants and heterozygous beta-thalassemia: example of a new variant Hb Acharnes or [beta53(D4) Ala --> Thr]. *American Journal of Hematology*, **1999**, 62, 186-92	7.1	8
59	Preimplantation genetic diagnosis, an alternative to conventional prenatal diagnosis of the hemoglobinopathies. *International Journal of Laboratory Hematology*, **2013**, 35, 571-9	2.5	7
58	Sertoli cell tumor and gonadoblastoma in an untreated 29-year-old 46,XY phenotypic male with Frasier syndrome carrying a WT1 IVS9+4C>T mutation. *Hormones*, **2012**, 11, 361-7	3.1	7
57	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, 1968-72	6.2	7
56	The c.504T>C (p.Asn168Asn) polymorphism in the ABCB4 gene as a predisposing factor for intrahepatic cholestasis of pregnancy in Greece. *Liver International*, **2010**, 30, 489-91	7.9	7

55	Bioluminometric assay for relative quantification of mutant allele burden: application to the oncogenic somatic point mutation JAK2 V617F. *Analytical Chemistry*, **2009**, 81, 8596-602	7.8	7
54	A rare 33 bp in-frame deletion (alpha63-74 or alpha64-74 or alpha65-75) in the alpha1-globin gene causing alpha(+)-thalassemia: a second observation. *Hemoglobin*, **2004**, 28, 137-43	0.6	7
53	Severe hypertriglyceridaemia in a Greek infant: a clinical, biochemical and genetic study. *European Journal of Pediatrics*, **2004**, 163, 462-6	4.1	7
52	An economic analysis of preimplantation genetic testing for aneuploidy by polar body biopsy in advanced maternal age. *BJOG: an International Journal of Obstetrics and Gynaecology*, **2020**, 127, 710-718	3.7	6
51	A generic, flexible protocol for preimplantation human leukocyte antigen typing alone or in combination with a monogenic disease, for rapid case work-up and application. *Hemoglobin*, **2014**, 38, 49-55	0.6	6
50	Microsatellite markers within the β-globin gene cluster for robust preimplantation genetic diagnosis of severe β-thalassemia syndromes in Mediterranean populations. *Hemoglobin*, **2012**, 36, 253-64	0.6	6
49	Lateral flow dipstick test for genotyping of 15 beta-globin gene (HBB) mutations with naked-eye detection. *Analytica Chimica Acta*, **2012**, 727, 61-6	6.6	6
48	Variable and often severe phenotypic expression in patients with the β-thalassemic variant Hb Agrinio [β29(B10)Leu->Pro (β)]. *Hemoglobin*, **2010**, 34, 430-8	0.6	6
47	Association of TLR4 single-nucleotide polymorphisms and sarcoidosis in Greek patients. *Genetic Testing and Molecular Biomarkers*, **2009**, 13, 849-53	1.6	6
46	The homozygous state for Hb Crete [beta129 (H7) Ala-->Pro] is associated with a complex phenotype including erythrocytosis and functional anemia. *Blood Cells, Molecules, and Diseases*, **2005**, 34, 229-34	2.1	6
45	Critical points for an accurate human genome analysis. *Human Mutation*, **2017**, 38, 912-921	4.7	5
44	A new gene associated with a β-thalassemia phenotype: the observation of variants in SUPT5H. *Blood*, **2020**, 136, 1789-1793	2.2	5
43	Preimplantation genetic diagnosis157-171		5
42	A novel (α) -thalassemia deletion in a Greek patient with HbH disease and β-thalassemia trait. *European Journal of Haematology*, **2012**, 88, 356-62	3.8	5
41	Absolute quantification of the alleles in somatic point mutations by bioluminometric methods based on competitive polymerase chain reaction in the presence of a locked nucleic acid blocker or an allele-specific primer. *Analytical Chemistry*, **2011**, 83, 6545-51	7.8	5
40	Coinheritance of mutated SMN1 and MECP2 genes in a child with phenotypic features of spinal muscular atrophy (SMA) type II and Rett syndrome. *European Journal of Paediatric Neurology*, **2007**, 11, 235-9	3.8	5
39	Rare thalassemic syndrome caused by interaction of Hb Questembert (alpha1 codon 131, TCT>CCT, Ser>Pro) with an alpha-thalassemia-2 deletion: implications for diagnosis and management. *Blood Cells, Molecules, and Diseases*, **2004**, 32, 118-23	2.1	5
38	DNA amplification techniques in pharmacogenomics. *Pharmacogenomics*, **2011**, 12, 845-60	2.6	4

37	PGD for X-linked and gender-dependent disorders using a robust, flexible single-tube PCR protocol. *Reproductive BioMedicine Online*, **2009**, 19, 418-25	4	4
36	Further identification of the hyperunstable alpha-globin chain variant Hb Heraklion [codons 36/37 (-CCC); Pro-->0 (alpha1)] in Greek cases with co-inherited alpha+-thalassemia mutations. *Hemoglobin*, **2008**, 32, 379-85	0.6	4
35	Hb Sitia [beta128(H6)Ala-->Val]: an unstable variant with a substitution in the alpha1beta1 interface. *Hemoglobin*, **2001**, 25, 45-56	0.6	4
34	ITHANET: Information and database community portal for haemoglobinopathies		4
33	Case Report: A Novel Synonymous ARPC1B Gene Mutation Causes a Syndrome of Combined Immunodeficiency, Asthma, and Allergy With Significant Intrafamilial Clinical Heterogeneity. *Frontiers in Immunology*, **2021**, 12, 634313	8.4	4
32	Synthesized allosteric effectors of the hemoglobin molecule: a possible mechanism for improved erythrocyte oxygen release capability in hemoglobinopathy H disease. *Experimental Hematology*, **1998**, 26, 922-6	3.1	4
31	Erythroid marrow activity and hemoglobin H levels in hemoglobin H disease. *Journal of Pediatric Hematology/Oncology*, **1998**, 20, 539-44	1.2	4
30	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	2.1	3
29	Adult-onset beta-thalassaemia intermedia caused by a 5-Mb somatic clonal segmental deletion in haemopoietic stem cells involving the β-globin locus. *British Journal of Haematology*, **2019**, 186, e165-e170	4.5	3
28	Genotyping of β-globin gene mutations in single lymphocytes: a preliminary study for preimplantation genetic diagnosis of monogenic disorders. *Hemoglobin*, **2012**, 36, 230-43	0.6	3
27	β+-Thalassemia trait due to a novel mutation in the β-globin gene promoter: -26 (A>C) [HBB c.-76A>C]. *Hemoglobin*, **2011**, 35, 84-6	0.6	3
26	Association of mild and severely unstable alpha chain variants: the first observation of a compound heterozygote with Hb Setif [alpha94(G1)Asp-->Tyr (alpha2)] and Hb Agrinio [alpha29(B10)Leu-->Pro (alpha2)] in a Greek family. *Hemoglobin*, **2008**, 32, 592-5	0.6	3
25	ATR-16 syndrome: mechanisms linking monosomy to phenotype. *Journal of Medical Genetics*, **2020**, 57, 414-421	5.8	3
24	Rapid detection of fetal Mendelian disorders: thalassemia and sickle cell syndromes. *Methods in Molecular Biology*, **2008**, 444, 133-45	1.4	3
23	A widely applicable strategy for single cell genotyping of beta-thalassaemia mutations using DGGE analysis: application to preimplantation genetic diagnosis. *Prenatal Diagnosis*, **1999**, 19, 1209-16	3.2	3
22	Two novel variants in the gene identified in cases with craniosynostosis. *The Application of Clinical Genetics*, **2019**, 12, 19-25	3.1	2
21	Hb Souli, a 6 bp in-frame deletion on the HBA2 gene (HBA2: c.[41-46delCCTGGG]) leads to β-thalassemia intermedia, when in trans to a single β-globin gene deletion. *Hemoglobin*, **2015**, 39, 55-7	0.6	2
20	Multi-allele DNA biosensor for the rapid genotyping of "non-deletional" alpha thalassaemia mutations in HBA1 and HBA2 genes by means of multiplex primer extension reaction. *Clinica Chimica Acta*, **2015**, 446, 241-7	6.2	2

19	Evaluation of Genotypes and Epidemiology of Spinal Muscular Atrophy in Greece: A Nationwide Study Spanning 24 Years. *Journal of Neuromuscular Diseases*, **2020**, 7, 247-256	5	2
18	Screening non-deletion α-thalassaemia mutations in the HBA1 and HBA2 genes by high-resolution melting analysis. *Clinical Chemistry and Laboratory Medicine*, **2015**, 53, 1951-9	5.9	2
17	Phenotypic and molecular diversity of haemoglobin H disease: a Greek experience. *British Journal of Haematology*, **2000**, 111, 915-923	4.5	2
16	Proliferative and chondrogenic potential of mesenchymal stromal cells from pluripotent and bone marrow cells. *Histology and Histopathology*, **2020**, 35, 1415-1426	1.4	2
15	A Novel β-Thalassemia Deletion Associated with Severe Anemia at Birth and a β-Thalassemia Intermedia Phenotype Later in Life in Three Generations of a Greek Family. *Hemoglobin*, **2019**, 1-4	0.6	2
14	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, 163-70	4.1	1
13	A simplified approach for FSHD molecular testing. *Clinica Chimica Acta*, **2014**, 429, 96-103	6.2	1
12	Novel and known nephrin gene (NPHS1) mutations in two Greek cases with congenital nephrotic syndrome including a complex genotype. *Journal of Genetics*, **2013**, 92, 577-81	1.2	1
11	Molecular basis of a-thalassaemia. *Thalassemia Reports*, **2012**, 1,	2	1
10	Phenotype-driven variant filtration strategy in exome sequencing toward a high diagnostic yield and identification of 85 novel variants in 400 patients with rare Mendelian disorders. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2561-2571	2.5	1
9	Identification of two novel mutations (296 + 1G-C and A46D) in exon 2 of the CFTR gene in Greek cystic fibrosis patients. *Molecular and Cellular Probes*, **1995**, 9, 283-5	3.3	0
8	Detection of acquired hemoglobinopathy in children with hematological malignancies at disease onset: results form a national referral centre. *International Journal of Hematology*, **2013**, 98, 563-8	2.3	
7	Diagnosis and molecular characterization of a novel β-thalassemia deletion (-Kozani) found in a Greek child with unexplained microcytic hypochromic anemia. *International Journal of Laboratory Hematology*, **2017**, 39, e124-e126	2.5	
6	PGD for glycogen storage disease type IV: birth of healthy twins following successful clinical application of a mutation-specific protocol. *Prenatal Diagnosis*, **2010**, 30, 180-2	3.2	
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