Jan Traeger-Synodinos

List of Publications by Year
in descending order

168 papers	5,533 citations	87888 38 h-index	102487 66 g-index
176 all docs	176 docs citations	176 times ranked	3877 citing authors

#	Article	IF	Citations
1	ESHRE PGD Consortium data collection XIII: cycles from January to December 2010 with pregnancy follow-up to October 2011. Human Reproduction, 2015, 30, 1763-1789.	0.9	273
2	The ESHRE PGD Consortium: 10 years of data collection. Human Reproduction Update, 2012, 18, 234-247.	10.8	264
3	The causes of misdiagnosis and adverse outcomes in PGD. Human Reproduction, 2009, 24, 1221-1228.	0.9	209
4	ESHRE PGD consortium best practice guidelines for amplification-based PGD. Human Reproduction, 2011, 26, 33-40.	0.9	208
5	What next for preimplantation genetic screening (PGS)? A position statement from the ESHRE PGD Consortium steering committeeÂ. Human Reproduction, 2010, 25, 821-823.	0.9	165
6	Mutations in the chromatin-associated protein ATRX. Human Mutation, 2008, 29, 796-802.	2.5	155
7	Blastocyst biopsy versus cleavage stage biopsy and blastocyst transfer for preimplantation genetic diagnosis of Î²-thalassaemia: a pilot study. Human Reproduction, 2007, 22, 1443-1449.	0.9	146
8	Systematic documentation and analysis of human genetic variation in hemoglobinopathies using the microattribution approach. Nature Genetics, 2011, 43, 295-301.	21.4	142
9	ESHRE PGD consortium best practice guidelines for organization of a PGD centre for PGD/preimplantation genetic screening. Human Reproduction, 2011, 26, 14-24.	0.9	137
10	ESHRE PGD consortium data collection X: cycles from January to December 2007 with pregnancy follow-up to October 2008â€. Human Reproduction, 2010, 25, 2685-2707.	0.9	124
11	Unstable and Thalassemic Î± Chain Hemoglobin Variants: A Cause of Hb H Disease and Thalassemia Intermedia. Hemoglobin, 2008, 32, 327-349.	0.8	117
12	ESHRE PGD Consortium data collection IX: cycles from January to December 2006 with pregnancy follow-up to October 2007. Human Reproduction, 2009, 24, 1786-1810.	0.9	116
13	Defective synthesis of HbE is due to reduced levels of Î²E mRNA. Nature, 1980, 288, 497-499.	27.8	106
14	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.	0.9	106
15	ESHRE PGD Consortium data collection VI: cycles from January to December 2003 with pregnancy follow-up to October 2004. Human Reproduction, 2007, 22, 323-336.	0.9	105
16	Birth of a healthy infant following trophectoderm biopsy from blastocysts for PGD of Î²-thalassaemia major: Case report. Human Reproduction, 2005, 20, 1855-1859.	0.9	94
17	ESHRE PGD consortium data collection VII: cycles from January to December 2004 with pregnancy follow-up to October 2005. Human Reproduction, 2008, 23, 741-755.	0.9	85
18	The triplicated Î±â€globin gene locus in Î²â€thalassaemia heterozygotes: clinical, haematological, biosynthetic and molecular studies. British Journal of Haematology, 1996, 95, 467-471.	2.5	84

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19	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.	0.9	80
20	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, 426-437.	2.8	79
21	ESHRE PGD Consortium data collection XI: cycles from January to December 2008 with pregnancy follow-up to October 2009. Human Reproduction, 2012, 27, 1887-1911.	0.9	77
22	Sensitive Monogenic Noninvasive Prenatal Diagnosis by Targeted Haplotyping. American Journal of Human Genetics, 2017, 101, 326-339.	6.2	76
23	ESHRE PGD Consortium data collection VIII: cycles from January to December 2005 with pregnancy follow-up to October 2006. Human Reproduction, 2008, 23, 2629-2645.	0.9	75
24	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.	3.2	73
25	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, 392-394.	2.9	69
26	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.	2.5	63
27	Genotyping of Single-Nucleotide Polymorphisms by Primer Extension Reaction in a Dry-Reagent Dipstick Format. Analytical Chemistry, 2007, 79, 395-402.	6.5	60
28	Evaluation of PCR-based preimplantation genetic diagnosis applied to monogenic diseases: a collaborative ESHRE PGD consortium study. European Journal of Human Genetics, 2014, 22, 1012-1018.	2.8	59
29	Phenotypic and molecular diversity of haemoglobin H disease: a Greek experience. British Journal of Haematology, 2000, 111, 915-23.	2.5	59
30	Prenatal diagnosis of the thalassaemia syndromes by rapid DNA analytical methods. Molecular Human Reproduction, 1997, 3, 523-528.	2.8	57
31	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
32	Multianalyte, dipstick-type, nanoparticle-based DNA biosensor for visual genotyping of single-nucleotide polymorphisms. Biosensors and Bioelectronics, 2009, 24, 3135-3139.	10.1	50
33	Characterization of nondeletion Î±-thalassemia mutations in the Greek population. American Journal of Hematology, 1993, 44, 162-167.	4.1	47
34	A base substitution (Tâ†'C) in codon 29 of the Î±2â€globin gene causes Î± thalassaemia. British Journal of Haematology, 1993, 85, 546-552.	2.5	47
35	Low total antioxidant status is implicated with high 8-hydroxy-2-deoxyguanosine serum concentrations in phenylketonuria. Clinical Biochemistry, 2005, 38, 239-242.	1.9	42
36	Preimplantation genetic diagnosis in clinical practice. Journal of Medical Genetics, 2002, 39, 6-11.	3.2	41

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37	Advances in technologies for screening and diagnosis of hemoglobinopathies. Biomarkers in Medicine, 2014, 8, 119-131.	1.4	41
38	Pre-implantation genetic diagnosis. Best Practice and Research in Clinical Obstetrics and Gynaecology, 2017, 39, 74-88.	2.8	41
39	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
40	Cystic fibrosis in Greece: molecular diagnosis, haplotypes, prenatal diagnosis and carrier identification amongst highâ€risk individuals. Clinical Genetics, 2003, 63, 400-409.	2.0	40
41	Cell-free DNA levels in acute myocardial infarction patients during hospitalization. Acta Cardiologica, 2009, 64, 51-57.	0.9	39
42	Molecular, haematological and clinical studies of the â^'101 Câ€ƒâ†'â€ƒT substitution of the Î²â€globin gene promoter in 25 Î²â€thalassaemia intermedia patients and 45 heterozygotes. British Journal of Haematology, 1999, 107, 699-706.	2.5	37
43	An Overview of Current Microarray-Based Human Globin Gene Mutation Detection Methods. Hemoglobin, 2007, 31, 289-311.	0.8	36
44	Phenotypic and molecular diversity of haemoglobin H disease: a Greek experience. British Journal of Haematology, 2000, 111, 915-923.	2.5	33
45	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.	3.5	33
46	A rare example that coinheritance of a severe form of Î²-thalassemia and Î±-thalassemia interact in a â€œsynergisticâ€•manner to balance the phenotype of classic thalassemic syndromes. Blood Cells, Molecules, and Diseases, 2004, 32, 319-324.	1.4	32
47	Preimplantation genetic diagnosis (PGD), a collaborative activity of clinical genetic departments and IVF centres. Prenatal Diagnosis, 2001, 21, 1086-1092.	2.3	31
48	Noninvasive prenatal diagnosis of Î²â€thalassaemia using individual fetal erythroblasts isolated from maternal blood after enrichment. Prenatal Diagnosis, 2007, 27, 1228-1232.	2.3	29
49	Instability of beta E-messenger RNA during erythroid cell maturation in hemoglobin E homozygotes.. Journal of Clinical Investigation, 1982, 69, 1050-1053.	8.2	29
50	Distinct Phenotypic Expression Associated with a New Hyperunstable Alpha Globin Variant (Hb) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 22 Molecules, and Diseases, 2000, 26, 276-284.	1.4	28
51	Real-time PCR for prenatal and preimplantation genetic diagnosis of monogenic diseases. Molecular Aspects of Medicine, 2006, 27, 176-191.	6.4	28
52	Rapid and Accurate Quantitation of Hb Bart's and Hb H Using Weak Cation Exchange High Performance Liquid Chromatography: Correlation with the Î±-Thalassemia Genotype. Hemoglobin, 1999, 23, 203-211.	0.8	27
53	Phenotypic and molecular diversity of haemoglobin H disease: a Greek experience. British Journal of Haematology, 2000, 111, 915-923.	2.5	27
54	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.	2.8	26

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55	Phenotypic and Genotypic Variability in Four Males With MECP2 Gene Sequence Aberrations Including a Novel Deletion. Pediatric Research, 2010, 67, 551-556.	2.3	26
56	Preconception carrier screening and prenatal diagnosis in thalassemia and hemoglobinopathies: challenges and future perspectives. Expert Review of Molecular Diagnostics, 2017, 17, 281-291.	3.1	26
57	Prenatal and preimplantation diagnosis of hemoglobinopathies. International Journal of Laboratory Hematology, 2018, 40, 74-82.	1.3	26
58	Photoprotein aequorin as a novel reporter for SNP genotyping by primer extensionâ€“application to the variants of mannose-binding lectin gene. Human Mutation, 2006, 27, 279-285.	2.5	25
59	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, 188-191.	1.6	25
60	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, 343-347.	3.8	24
61	Pregnancies following blastocyst stage transfer in PGD cycles at risk for beta-thalassaemic haemoglobinopathies. Human Reproduction, 2002, 17, 25-31.	0.9	24
62	Prenatal diagnosis of hemoglobin disorders: Present and future strategies. Clinical Biochemistry, 2009, 42, 1767-1779.	1.9	24
63	The clinical utility of PGD with HLA matching: a collaborative multi-centre ESHRE study. Human Reproduction, 2018, 33, 520-530.	0.9	24
64	Phenotypeâ€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.	1.2	24
65	An Electronic Infrastructure for Research and Treatment of the Thalassemias and Other Hemoglobinopathies: The Euro-Mediterranean Ithanet Project. Hemoglobin, 2009, 33, 163-176.	0.8	23
66	Erythroid Marrow Activity and Hemoglobin H Levels in Hemoglobin H Disease. Journal of Pediatric Hematology/Oncology, 1998, 20, 539-544.	0.6	22
67	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.	3.8	21
68	The interaction of Î±Â° thalassaemia with Hb Icaria: three unusual cases of haemoglobinopathy H. British Journal of Haematology, 1996, 92, 332-335.	2.5	20
69	High-Throughput Microtiter Well-Based Chemiluminometric Genotyping of 15 HBB Gene Mutations in a Dry-Reagent Format. Clinical Chemistry, 2007, 53, 384-391.	3.2	20
70	Adapting the ACMG/AMP variant classification framework: A perspective from the ClinGen Hemoglobinopathy Variant Curation Expert Panel. Human Mutation, 2022, 43, 1089-1096.	2.5	20
71	Interaction of an Î±+-Thalassemia Deletion with Either a Highly Unstable Î±-Globin Variant (Î±2, Codon 59,) Tj ETQq1 1 0.784314 rgBT Illustrating â€œDominantâ€•Î±-Thalassemia. Hemoglobin, 1999, 23, 325-337.	0.8	19
72	Scanning of ?-globin gene for identification of ?-thalassemia mutation in Romanian population. Journal of Cellular and Molecular Medicine, 2004, 8, 232-240.	3.6	19

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73	MECP2 mutations and clinical correlations in Greek children with Rett syndrome and associated neurodevelopmental disorders. Brain and Development, 2012, 34, 487-495.	1.1	19
74	The Corfu ÎÎ² thalassaemia mutation in Greece: haematological phenotype and prevalence. British Journal of Haematology, 1991, 79, 302-305.	2.5	18
75	Molecular Studies of Î²-Thalassemia Heterozygotes with Raised Hb F Levels. Hemoglobin, 2000, 24, 203-220.	0.8	18
76	A Rare Thalassemic Syndrome Caused by Interaction of Hb Adana [Î±59(E8)Glyâ†'Asp] with an Î±⁺-Thalassemia Deletion: Clinical Aspects in Two Cases. Hemoglobin, 2008, 32, 361-369.	0.8	18
77	Prenatal, noninvasive and preimplantation genetic diagnosis of inherited disorders: hemoglobinopathies. Expert Review of Molecular Diagnostics, 2011, 11, 299-312.	3.1	18
78	Pre-implantation HLA matching: The production of a Saviour Child. Best Practice and Research in Clinical Obstetrics and Gynaecology, 2017, 44, 76-89.	2.8	18
79	A high incidence of mutations in exon 6 of the lowâ€density lipoprotein receptor gene in Greek familial hypercholesterolemia patients, including a novel mutation. Human Mutation, 1997, 9, 274-276.	2.5	17
80	Hb Aghia Sophia [Î±62(E11)Valâ†'0 (Î±1)], an â€œIn-Frameâ€•Deletion Causing Î±-Thalassemia. Hemoglobin, 1999, 23, 317-324.	0.8	17
81	Novel and Known Microsatellite Markers Within the Î²-Globin Cluster to Support Robust Preimplantation Genetic Diagnosis of Î²-Thalassemia and Sickle Cell Syndromes. Hemoglobin, 2011, 35, 56-66.	0.8	17
82	Preliminary mutation analysis in the phenylanaline hydroxylase gene in Greek PKU and HPA patients. Human Genetics, 1994, 94, 573-5.	3.8	16
83	Molecular, haematological and clinical studies of a silent Î²-gene Câ€ƒâ†'â€ƒG mutation at 6â€ƒbp 3â€² to the termination codon (+1480 Câ€ƒâ†'â€ƒG) in twelve Greek families. British Journal of Haematology, 1998, 103, 45-51.	2.5	16
84	Severe hypertriglyceridaemia in diabetic ketoacidosis: clinical and genetic study. Diabetic Medicine, 2004, 21, 380-382.	2.3	16
85	Nucleotide Variations in theNPHS2Gene in Greek Children with Steroid-Resistant Nephrotic Syndrome. Genetic Testing and Molecular Biomarkers, 2009, 13, 249-256.	0.7	16
86	The molecular basis of normal HbA2 (type 2) beta-thalassemia in Greece. Hematologic Pathology, 1994, 8, 25-34.	0.2	16
87	A Minimal Set of SNPs for the Noninvasive Prenatal Diagnosis of Î²â€Thalassaemia. Annals of Human Genetics, 2013, 77, 115-124.	0.8	15
88	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.	2.3	15
89	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.	3.5	15
90	An Î±-Thalassemic Hemoglobinopathy: Homozygosity for the Hb Agrinio Î±2-Globin Chain Variant. Hemoglobin, 1998, 22, 209-215.	0.8	14

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91	Broad and unexpected phenotypic expression in Greek children with steroid-resistant nephrotic syndrome due to mutations in the Wilmsâ€™ tumor 1 (WT1) gene. European Journal of Pediatrics, 2011, 170, 1529-1534.	2.7	14
92	AbnormalDLK1/MEG3imprinting correlates with decreased HERV-K methylation after assisted reproduction and preimplantation genetic diagnosis. Stress, 2013, 16, 689-697.	1.8	14
93	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.	4.8	14
94	Molecular Characterization of Homozygous (High HbA2) Î²-Thalassemia Intermedia in Greece. Pediatric Hematology and Oncology, 1995, 12, 37-45.	0.8	13
95	Psychomotor development of children born after preimplantation genetic diagnosis and parental stress evaluation. World Journal of Pediatrics, 2012, 8, 309-316.	1.8	13
96	A new gene associated with a Î²-thalassemia phenotype: the observation of variants in SUPT5H. Blood, 2020, 136, 1789-1793.	1.4	13
97	The experience of 3 years of external quality assessment of preimplantation genetic diagnosis for cystic fibrosis. European Journal of Human Genetics, 2013, 21, 800-806.	2.8	12
98	Association of unstable hemoglobin variants and heterozygous ?-thalassemia: Example of a new variant Hb acharnes or [?53(D4) Ala ? Thr]. , 1999, 62, 186-192.		11
99	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.	2.8	11
100	Unusual phenotypic observations associated with a rare HbH disease genotype (-â€Š-Med /Î±TSaudi Î±): implications for clinical management. British Journal of Haematology, 2002, 119, 265-267.	2.5	11
101	Two new Â-thalassemia deletions compromising prenatal diagnosis in an Italian and a Turkish couple seeking prevention. Haematologica, 2009, 94, 1289-1292.	3.5	11
102	Visual screening for JAK2V617F mutation by a disposable dipstick. Analytical and Bioanalytical Chemistry, 2010, 397, 1911-1916.	3.7	11
103	Preimplantation genetic diagnosis, an alternative to conventional prenatal diagnosis of the hemoglobinopathies. International Journal of Laboratory Hematology, 2013, 35, 571-579.	1.3	11
104	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.	0.8	10
105	The homozygous state for Hb Crete [Î²129 (H7) Alaâ†’Pro] is associated with a complex phenotype including erythrocytosis and functional anemia. Blood Cells, Molecules, and Diseases, 2005, 34, 229-234.	1.4	10
106	High-throughput microtiter well-based bioluminometric genotyping of two single-nucleotide polymorphisms in the toll-like receptor-4 gene. Analytical Biochemistry, 2008, 376, 235-241.	2.4	10
107	Complex preimplantation genetic diagnosis for beta-thalassaemia, sideroblastic anaemia, and human leukocyte antigen (HLA)-typing. Systems Biology in Reproductive Medicine, 2016, 62, 69-76.	2.1	10
108	Hb MONT SAINT AIGNAN [Î²128(H6)Ala â†’ Pro]: A NEW UNSTABLE VARIANT LEADING TO CHRONIC MICROCYTIC ANEMIA. Hemoglobin, 2001, 25, 57-65.	0.8	9

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109	Severe hypertriglyceridaemia in a Greek infant: a clinical, biochemical and genetic study. European Journal of Pediatrics, 2004, 163, 462-6.	2.7	9
110	First Observation of Hb Taybe [Codons 38/39 (â^'Acc) Thrâ†'0 (Î±1)] In Greece: Clinical and Hematological Findings in Patients With Co-Inherited Î±⁺-Thalassemia Mutations. Hemoglobin, 2008, 32, 371-378.	0.8	9
111	The c.504T>C (p.Asn168Asn) polymorphism in theABCB4gene as a predisposing factor for intrahepatic cholestasis of pregnancy in Greece. Liver International, 2010, 30, 489-491.	3.9	9
112	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.	1.1	9
113	Microsatellite Markers Within the Î±-Globin Gene Cluster for Robust Preimplantation Genetic Diagnosis of Severe Î±-Thalassemia Syndromes in Mediterranean Populations. Hemoglobin, 2012, 36, 253-264.	0.8	9
114	Rare Î²-Thalassemia Alleles In the Greek and Greek Cypriot Populations. Hemoglobin, 1998, 22, 89-94.	0.8	8
115	Variable and Often Severe Phenotypic Expression in Patients with the Î±-Thalassemic Variant Hb Agrinio [Î±29(B10)Leuâ†'Pro (Î±2)]. Hemoglobin, 2010, 34, 430-438.	0.8	8
116	Lateral flow dipstick test for genotyping of 15 beta-globin gene (HBB) mutations with naked-eye detection. Analytica Chimica Acta, 2012, 727, 61-66.	5.4	8
117	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, 361-367.	1.9	8
118	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.	2.6	8
119	A Rare 33 bp Inâ€Frame Deletion (Î±63â€"74 or Î±64â€"74 or Î±65â€"75) in the Î±1â€Globin Gene Causing Î±+â€Thalassemia: A Second Observation. Hemoglobin, 2004, 28, 137-143.	0.8	7
120	Bioluminometric Assay for Relative Quantification of Mutant Allele Burden: Application to the Oncogenic Somatic Point Mutation JAK2 V617F. Analytical Chemistry, 2009, 81, 8596-8602.	6.5	7
121	Association of TLR4 Single-Nucleotide Polymorphisms and Sarcoidosis in Greek Patients. Genetic Testing and Molecular Biomarkers, 2009, 13, 849-853.	0.7	7
122	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, 6545-6551.	6.5	7
123	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.	0.8	7
124	ATR-16 syndrome: mechanisms linking monosomy to phenotype. Journal of Medical Genetics, 2020, 57, 414-421.	3.2	7
125	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.	0.4	7
126	Rare thalassemic syndrome caused by interaction of Hb Questembert (Î±1 codon 131, TCT>CCT, Ser>Pro) with an Î±-thalassemia-2 deletion: implications for diagnosis and management. Blood Cells, Molecules, and Diseases, 2004, 32, 118-123.	1.4	6

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127	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, 235-239.	1.6	6
128	Further Identification of The Hyperunstable Î±-Globin Chain Variant Hb Heraklion [codons 36/37 (â€“CCC); Proâ†'0 (Î±1)] in Greek Cases With Co-Inherited Î±⁺-Thalassemia Mutations. Hemoglobin, 2008, 32, 379-385.	0.8	6
129	DNA amplification techniques in pharmacogenomics. Pharmacogenomics, 2011, 12, 845-860.	1.3	6
130	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.	1.4	6
131	PGD for X-linked and gender-dependent disorders using a robust, flexible single-tube PCR protocol. Reproductive BioMedicine Online, 2009, 19, 418-425.	2.4	5
132	Genotyping of Î²-Globin Gene Mutations in Single Lymphocytes: A Preliminary Study for Preimplantation Genetic Diagnosis of Monogenic Disorders. Hemoglobin, 2012, 36, 230-243.	0.8	5
133	A novel Î±⁰â€thalassemia deletion in a Greek patient with HbH disease and Î²â€thalassemia trait. European Journal of Haematology, 2012, 88, 356-362.	2.2	5
134	Preimplantation genetic diagnosis. , 0, , 157-171.		5
135	Critical points for an accurate human genome analysis. Human Mutation, 2017, 38, 912-921.	2.5	5
136	Rapid Detection of Fetal Mendelian Disorders: Thalassemia and Sickle Cell Syndromes. Methods in Molecular Biology, 2008, 444, 133-145.	0.9	5
137	Erythroid marrow activity and hemoglobin H levels in hemoglobin H disease. Journal of Pediatric Hematology/Oncology, 1998, 20, 539-44.	0.6	5
138	Hb SITIA [Î²128(H6)Alaâ†'Val]: AN UNSTABLE VARIANT WITH A SUBSTITUTION IN THE Î±1Î²1 INTERFACE. Hemoglobin, 2001, 25, 45-56.	0.8	4
139	Association of Mild and Severely Unstable Î± Chain Variants: The First Observation of a Compound Heterozygote with Hb Setif [Î±94(G1)Aspâ†'Tyr (Î±2)] and Hb Agrinio [Î±29(B10)Leuâ†'Pro (Î±2)] in a Greek Family. Hemoglobin, 2008, 32, 592-595.	0.8	4
140	Proliferative and chondrogenic potential of mesenchymal stromal cells from pluripotent and bone marrow cells. Histology and Histopathology, 2020, 35, 1415-1426.	0.7	4
141	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.	2.3	4
142	Î²⁺-Thalassemia Trait Due to a Novel Mutation in the Î²-Globin Gene Promoter: â^'26 (A>C) [HBB c.â^'76A>C]. Hemoglobin, 2011, 35, 84-86.	0.8	3
143	A simplified approach for FSHD molecular testing. Clinica Chimica Acta, 2014, 429, 96-103.	1.1	3
144	Multi-allele DNA biosensor for the rapid genotyping of â€˜nondeletionâ€™ alpha thalassaemia mutations in HBA1 and HBA2 genes by means of multiplex primer extension reaction. Clinica Chimica Acta, 2015, 446, 241-247.	1.1	3

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145	Adultâ€onset betaâ€thalassaemia intermedia caused by a 5â€Mb somatic clonal segmental deletion in haemopoietic stem cells involving the Î²â€globin locus. British Journal of Haematology, 2019, 186, e165-e170.	2.5	3
146	A Novel ÎµÎ³ÎγÎ²-Thalassemia Deletion Associated with Severe Anemia at Birth and a Î²-Thalassemia Intermedia Phenotype Later in Life in Three Generations of a Greek Family. Hemoglobin, 2021, 45, 351-354.	0.8	3
147	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.	2.1	2
148	Molecular Basis of Î±-thalassaemia. Thalassemia Reports, 2011, 1, e13.	0.5	2
149	Eleven years of preimplantation genetic diagnosis for human leukocyte antigen matching: is there room for improvement?. Expert Review of Hematology, 2013, 6, 215-217.	2.2	2
150	Session 09: ESHRE data reporting on PGD cycles and oocyte donation. Human Reproduction, 2013, 28, i18-i19.	0.9	2
151	Screening non-deletion Î±-thalassaemia mutations in the HBA1 and HBA2 genes by high-resolution melting analysis. Clinical Chemistry and Laboratory Medicine, 2015, 53, 1951-9.	2.3	2
152	Hb Souli, a 6â€‰bp In-Frame Deletion on theHBA2Gene (HBA2: c.[41-46delCCTGGG]) Leads toÎ±-Thalassemia Intermedia, When inTransto a SingleÎ±-Globin Gene Deletion. Hemoglobin, 2015, 39, 55-57.	0.8	2
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