John S Waye

List of Publications by Year
in descending order

55 papers	1,079 citations	706676 14 h-index	759306 22 g-index
55 all docs	55 docs citations	55 times ranked	2367 citing authors

#	Article	IF	Citations
1	Adapting the ACMG/AMP variant classification framework: A perspective from the ClinGen Hemoglobinopathy Variant Curation Expert Panel. Human Mutation, 2022, 43, 1089-1096.	1.1	20
2	Outcomes of haemoglobin Bartâ€™s hydrops fetalis following intrauterine transfusion in Ontario, Canada. Archives of Disease in Childhood: Fetal and Neonatal Edition, 2021, 106, 51-56.	1.4	9
3	Microcytosis in patients with haemoglobin C trait: is Î±â€thalassaemia trait to blame?. British Journal of Haematology, 2020, 191, e129-e131.	1.2	0
4	Novel High Oxygen Affinity Hemoglobin Variant in a Patient with Polycythemia: Hb Kennisis [Î²85(F1)Pheâ†'Leu (TTT>TTG); HBB: c.258T>G]. Hemoglobin, 2020, 44, 10-12.	0.4	2
5	A Novel Human Î²-Globin Gene Variant [Hb London-Ontario,HBB: c.332T>G] is Associated with Transfusion-Dependent Anemia in a Patient with a Hemoglobin Electrophoresis Pattern Consistent with Î²-Thalassemia Trait. Hemoglobin, 2019, 43, 129-131.	0.4	2
6	An evaluation of genetic causes and environmental risks for bilateral optic atrophy. PLoS ONE, 2019, 14, e0225656.	1.1	9
7	Hepatoblastoma in a Child With Early-onset Cirrhosis. Journal of Pediatric Hematology/Oncology, 2019, 41, e30-e33.	0.3	3
8	An evaluation of genetic causes and environmental risks for bilateral optic atrophy. , 2019, 14, e0225656.		0
9	An evaluation of genetic causes and environmental risks for bilateral optic atrophy. , 2019, 14, e0225656.		0
10	An evaluation of genetic causes and environmental risks for bilateral optic atrophy. , 2019, 14, e0225656.		0
11	An evaluation of genetic causes and environmental risks for bilateral optic atrophy. , 2019, 14, e0225656.		0
12	Data sharing as a national quality improvement program: reporting on BRCA1 and BRCA2 variant-interpretation comparisons through the Canadian Open Genetics Repository (COGR). Genetics in Medicine, 2018, 20, 294-302.	1.1	27
13	Multiplex Allele-Specific PCR for Simultaneous Detection of H63D and C282Y HFE Mutations in Hereditary Hemochromatosis. journal of applied laboratory medicine, The, 2018, 3, 10-17.	0.6	3
14	Characterization of Two Novel Deletions Involving the 5â€² Region of the Î²-Globin Gene. Hemoglobin, 2017, 41, 239-242.	0.4	2
15	Clinical evaluation of a hemochromatosis nextâ€generation sequencing gene panel. European Journal of Haematology, 2017, 98, 228-234.	1.1	20
16	Î±0-Thalassemia Due to a 90.7â€‰kb Deletion (â€“ â€“NFLD). Hemoglobin, 2017, 41, 218-219.	0.4	1
17	The duplication mutation of Quebec platelet disorder dysregulates PLAU, but not C10orf55, selectively increasing production of normal PLAU transcripts by megakaryocytes but not granulocytes. PLoS ONE, 2017, 12, e0173991.	1.1	18
18	Clinical Next-Generation Sequencing Pipeline Outperforms a Combined Approach Using Sanger Sequencing and Multiplex Ligation-Dependent Probe Amplification in Targeted Gene Panel Analysis. Journal of Molecular Diagnostics, 2016, 18, 657-667.	1.2	47

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19	Novel Mutation of the Translation Initiation Codon of the Î±1-Globin Gene (ATG>AAG orHBA1:c.2T>A). Hemoglobin, 2016, 40, 369-370.	0.4	1
20	Hb S/Â+-thalassemia due to Hb sickle and a novel deletion of DNase I hypersensitive sites HS3 and HS4 of the Â locus control region. Haematologica, 2015, 100, e166-e168.	1.7	6
21	Sudanese (Î´Î²)⁰-Thalassemia: Identification and Characterization of a Novel 9.6â€‰kb Deletion. Hemoglobin, 2015, 39, 368-370.	0.4	2
22	Î±+-Thalassemia Due to a Frameshift Mutation of theÎ±2-Globin Gene [codons 55/56 (+T) orHBA2: c.168dup]. Hemoglobin, 2015, 39, 209-210.	0.4	1
23	Non-Thalassemic Phenotype Associated With the -83 (Gâ€‰>â€‰A) Mutation of theÎ²-Globin Gene Promoter (HBB:) Tj ETQq1 1 0.784	0.4	0
24	Compound Heterozygosity for Hb S and a Novel Deletion of Dnase I Hypersensitivity Sites HS3 and HS4 of Î²-Globin Locus Control Region Results in Hb S/Î²+-Thalassemia Phenotype. Blood, 2014, 124, 2692-2692.	0.6	0
25	Normal Hb A₂Î²-Thalassemia Trait: Frameshift Mutation (HBB: c.187_251dup) inCiswith the Hb A_{2â€™}Î´-Globin Gene Missense Mutation (HBD: c.49G>C). Hemoglobin, 2013, 37, 201-204.	0.4	5
26	Mild Î²⁺-Thalassemia Associated With Two Linked Sequence Variants: IVS-II-839 (T>C) and IVS-II-844 (C>A). Hemoglobin, 2013, 37, 378-386.	0.4	2
27	Quebec Platelet Disorder Is Associated With Greater Than Expected Increases In Urokinase Plasminogen Activator In Granulocytes and Monocytes. Blood, 2013, 122, 3573-3573.	0.6	0
28	Î±+-Thalassemia Trait Caused by a Frameshift Mutation in Exon 2 of the Î±2-Globin Gene [HBA2 c.244delT]. Hemoglobin, 2012, 36, 205-207.	0.4	1
29	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.	0.4	3
30	Dysregulation of C10orf55 Expression in Megakaryocytic Cell Lineage From Quebec Platelet Disorder Individuals. Blood, 2011, 118, 2274-2274.	0.6	0
31	Hb North York [Î²117(G19)Hisâ†’Asp]: A New Î² Chain Hemoglobin Variant. Hemoglobin, 2009, 33, 51-53.	0.4	1
32	Î±-Thalassemia Caused by Two Novel Splice Mutations of the Î±2-Globin Gene: IVS-I-1 (G>A and G>T). Hemoglobin, 2009, 33, 519-522.	0.4	5
33	Novel 27.9 kb Î±⁰â€thalassemia deletion in a Filipino woman. American Journal of Hematology, 2009, 84, 197-198.	2.0	2
34	Targeted Gene Sequencing to Identify Polymorphisms in the Protein C and EPCR Genes in Patients with Unprovoked Venous Thromboembolism.. Blood, 2009, 114, 454-454.	0.6	1
35	Prenatal diagnosis of Smith-Lemli-Opitz syndrome (SLOS) byDHCR7 mutation analysis. Prenatal Diagnosis, 2007, 27, 638-640.	1.1	18
36	Identification of nine novelDHCR7 missense mutations in patients with Smith-Lemli-Opitz syndrome (SLOS). Human Mutation, 2005, 26, 59-59.	1.1	3

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37	Smith-Lemli-Opitz (RHS) syndrome: holoprosencephaly and homozygous IVS8-1G?C genotype. American Journal of Medical Genetics Part A, 2001, 103, 75-80.	2.4	41
38	Hemoglobin H (Hb H) disease in Canada: Molecular diagnosis and review of 116 cases. American Journal of Hematology, 2001, 68, 11-15.	2.0	27
39	DHCR7 genotypes of cousins with Smith-Lemli-Opitz syndrome. American Journal of Medical Genetics Part A, 2001, 100, 162-163.	2.4	0
40	Frequency and ethnic distribution of the commonDHCR7 mutation in Smith-Lemli-Opitz syndrome. American Journal of Medical Genetics Part A, 2001, 102, 383-386.	2.4	40
41	Rapid molecular prenatal diagnosis of Smith-Lemli-Opitz syndrome. American Journal of Medical Genetics Part A, 2001, 102, 387-388.	2.4	9
42	NOVEL Î²-THALASSEMIA MUTATION IN A Î²-THALASSEMIA INTERMEDIA PATIENT [POLY A (AATAAA â†'GATAAA)]. Hemoglobin, 2001, 25, 103-105.	0.4	8
43	PCR-based diagnosis of the Filipino (??FIL) and Thai (??THAI) ?-thalassemia-1 deletions. , 2000, 63, 54-56.		32
44	A novel and de novo spontaneous point mutation (Glu271STOP) of the antithrombin gene results in a type I deficiency and thrombophilia. , 1999, 60, 126-129.		6
45	Improved DNA extraction from ancient bones using silica-based spin columns. , 1998, 105, 539-543.		501
46	Improved DNA extraction from ancient bones using silica-based spin columns. , 1998, 105, 539.		2
47	Improved DNA extraction from ancient bones using silica-based spin columns. , 1998, 105, 539.		7
48	The prenatal identification of fetal compatibility in neonatal alloimmune thrombocytopenia using amniotic fluid and variable number of tandem repeat (VNTR) analysis. British Journal of Haematology, 1995, 91, 742-746.	1.2	17
49	Allele frequency data for VNTR locus D17S79: Identification of an internalHaeIII polymorphism in the black population. Human Mutation, 1994, 3, 248-253.	1.1	1
50	Dopamine D4 receptor variant, D4GLYCINE194, in Africans, but not in Caucasians: No association with Schizophrenia. American Journal of Medical Genetics Part A, 1994, 54, 384-390.	2.4	63
51	Anomalous Migration of PCR Products Using Nondenaturing Polyacrylamide Gel Electrophoresis: The Amelogenin Sex-Typing System. Journal of Forensic Sciences, 1994, 39, 1356-1359.	0.9	27
52	Complications in the genotypic molecular diagnosis of pseudo arylsulfatase A deficiency. American Journal of Medical Genetics Part A, 1993, 45, 631-637.	2.4	26
53	Identification of an extensive Î¶â€Î± globin gene deletion in a Chinese individual. British Journal of Haematology, 1992, 80, 378-380.	1.2	28
54	Clinical course and molecular characterization of a compound heterozygote for sickle hemoglobin and hemoglobin kenya. American Journal of Hematology, 1992, 41, 289-291.	2.0	7

#	Article	IF	Citations
55	Hb SÎ²Â°-Thalassemia due to the Ëœ1.4-kb deletion is associated with a relatively mild phenotype. American Journal of Hematology, 1991, 38, 108-112.	2.0	23