Stephan Menzel

List of Publications by Year in descending order

77 papers	7,591 citations	147566 31 h-index	76769 74 g-index
80 all docs	80 docs citations	80 times ranked	7619 citing authors

#	Article	IF	Citations
1	Mutations in the hepatocyte nuclear factor-1Î± gene in maturity-onset diabetes of the young (MODY3). Nature, 1996, 384, 455-458.	13.7	1,240
2	Mutations in the hepatocyte nuclear factor-4Î± gene in maturity-onset diabetes of the young (MODY1). Nature, 1996, 384, 458-460.	13.7	1,114
3	A genomeâ€“wide search for human nonâ€“insulinâ€“dependent (type 2) diabetes genes reveals a major susceptibility locus on chromosome 2. Nature Genetics, 1996, 13, 161-166.	9.4	580
4	A QTL influencing F cell production maps to a gene encoding a zinc-finger protein on chromosome 2p15. Nature Genetics, 2007, 39, 1197-1199.	9.4	491
5	A genome-wide meta-analysis identifies 22 loci associated with eight hematological parameters in the HaemGen consortium. Nature Genetics, 2009, 41, 1182-1190.	9.4	481
6	A Genomewide Scan for Loci Predisposing to Type 2 Diabetes in a U.K. Population (The Diabetes UK) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5 Locus on Chromosome 1q. American Journal of Human Genetics, 2001, 69, 553-569.	2.6	300
7	Intergenic variants of HBS1L-MYB are responsible for a major quantitative trait locus on chromosome 6q23 influencing fetal hemoglobin levels in adults. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 11346-11351.	3.3	286
8	Control of fetal hemoglobin: new insights emerging from genomics and clinical implications. Human Molecular Genetics, 2009, 18, R216-R223.	1.4	213
9	Studies of Association between the Gene for Calpain-10 and Type 2 Diabetes Mellitus in the United Kingdom. American Journal of Human Genetics, 2001, 69, 544-552.	2.6	171
10	Discovering the genetics underlying foetal haemoglobin production in adults. British Journal of Haematology, 2009, 145, 455-467.	1.2	171
11	cMYB is involved in the regulation of fetal hemoglobin production in adults. Blood, 2006, 108, 1077-1083.	0.6	163
12	HBS1L-MYB intergenic variants modulate fetal hemoglobin via long-range MYB enhancers. Journal of Clinical Investigation, 2014, 124, 1699-1710.	3.9	157
13	Systematic documentation and analysis of human genetic variation in hemoglobinopathies using the microattribution approach. Nature Genetics, 2011, 43, 295-301.	9.4	142
14	A novel variant on chromosome 7q22.3 associated with mean platelet volume, counts, and function. Blood, 2009, 113, 3831-3837.	0.6	117
15	A low renal threshold for glucose in diabetic patients with a mutation in the hepatocyte nuclear factor-1Î± (HNF-1Î±) gene. , 1998, 15, 816-820.		109
16	Multiple Loci Are Associated with White Blood Cell Phenotypes. PLoS Genetics, 2011, 7, e1002113.	1.5	106
17	Genetics of fetal hemoglobin in Tanzanian and British patients with sickle cell anemia. Blood, 2011, 117, 1390-1392.	0.6	104
18	Polymorphisms in Type II SH2 Domain-Containing Inositol 5-Phosphatase (INPPL1, SHIP2) Are Associated With Physiological Abnormalities of the Metabolic Syndrome. Diabetes, 2004, 53, 1900-1904.	0.3	91

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19	Environmental determinants of severity in sickle cell disease. Haematologica, 2015, 100, 1108-1116.	1.7	90
20	alpha-Haemoglobin stabilising protein is a quantitative trait gene that modifies the phenotype of beta-thalassaemia. British Journal of Haematology, 2006, 133, 675-682.	1.2	79
21	Genome Wide Association Study of Fetal Hemoglobin in Sickle Cell Anemia in Tanzania. PLoS ONE, 2014, 9, e111464.	1.1	78
22	The linear effects ofÎ±-thalassaemia, theUGT1A1andHMOX1polymorphisms on cholelithiasis in sickle cell disease. British Journal of Haematology, 2007, 138, 263-270.	1.2	77
23	Genetic Variation on Chromosome 6 Influences F Cell Levels in Healthy Individuals of African Descent and HbF Levels in Sickle Cell Patients. PLoS ONE, 2009, 4, e4218.	1.1	77
24	The HBS1L-MYB intergenic region on chromosome 6q23.3 influences erythrocyte, platelet, and monocyte counts in humans. Blood, 2007, 110, 3624-3626.	0.6	71
25	Genome-wide association analyses based on whole-genome sequencing in Sardinia provide insights into regulation of hemoglobin levels. Nature Genetics, 2015, 47, 1264-1271.	9.4	66
26	A Genome-Wide Scan in Families With Maturity-Onset Diabetes of the Young: Evidence for Further Genetic Heterogeneity. Diabetes, 2003, 52, 872-881.	0.3	62
27	Quantitative trait locus on chromosome 8q influences the switch from fetal to adult hemoglobin. Blood, 2004, 104, 2184-2186.	0.6	59
28	Evidence for Linkage of Stature to Chromosome 3p26 in a Large U.K. Family Data Set Ascertained for Type 2 Diabetes. American Journal of Human Genetics, 2002, 70, 543-546.	2.6	53
29	Association and Haplotype Analysis of the Insulin-Degrading Enzyme (IDE) Gene, a Strong Positional and Biological Candidate for Type 2 Diabetes Susceptibility. Diabetes, 2003, 52, 1300-1305.	0.3	52
30	Young-Onset Type 2 Diabetes Families Are the Major Contributors to Genetic Loci in the Diabetes UK Warren 2 Genome Scan and Identify Putative Novel Loci on Chromosomes 8q21, 21q22, and 22q11. Diabetes, 2003, 52, 1857-1863.	0.3	43
31	Evidence From a Large U.K. Family Collection That Genes Influencing Age of Onset of Type 2 Diabetes Map to Chromosome 12p and to the MODY3/NIDDM2 Locus on 12q24. Diabetes, 2004, 53, 855-860.	0.3	41
32	g(HbF): a genetic model of fetal hemoglobin in sickle cell disease. Blood Advances, 2018, 2, 235-239.	2.5	33
33	Genetic architecture of hemoglobin F control. Current Opinion in Hematology, 2009, 16, 179-186.	1.2	32
34	<scp>H</scp>b<scp>A</scp>₂ levels in normal adults are influenced by two distinct genetic mechanisms. British Journal of Haematology, 2013, 160, 101-105.	1.2	32
35	Genetic Modifiers of Fetal Haemoglobin in Sickle Cell Disease. Molecular Diagnosis and Therapy, 2019, 23, 235-244.	1.6	32
36	Associations between environmental factors and hospital admissions for sickle cell disease. Haematologica, 2017, 102, 666-675.	1.7	29

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37	Interaction Between Two Quantitative Trait Loci Affects Fetal Haemoglobin Expression. Annals of Human Genetics, 2005, 69, 707-714.	0.3	25
38	Association of sickle avascular necrosis with bone morphogenic protein 6. Annals of Hematology, 2009, 88, 803-805.	0.8	25
39	Reduced rate of sickleâ€related complications in Brazilian patients carrying HbFâ€promoting alleles at the BCL11A and HMIPâ€2 loci. British Journal of Haematology, 2016, 173, 456-460.	1.2	25
40	The genetics of type 2 diabetes. British Journal of Clinical Pharmacology, 2001, 51, 195-199.	1.1	24
41	Global Genetic Architecture of an Erythroid Quantitative Trait Locus,HMIP-2. Annals of Human Genetics, 2014, 78, 434-451.	0.3	24
42	Genetic association of fetal-hemoglobin levels in individuals with sickle cell disease in Tanzania maps to conserved regulatory elements within the MYB core enhancer. BMC Medical Genetics, 2015, 16, 4.	2.1	24
43	Proteomic analysis of plasma from children with sickle cell anemia and silent cerebral infarction. Haematologica, 2018, 103, 1136-1142.	1.7	22
44	Genetic regulation of fetal haemoglobin in inherited bone marrow failure syndromes. British Journal of Haematology, 2013, 162, 542-546.	1.2	21
45	Genetic variants at HbFâ€modifier loci moderate anemia and leukocytosis in sickle cell disease in T anzania. American Journal of Hematology, 2015, 90, E1-4.	2.0	21
46	A gain of function variant in PIEZO1 (E756del) and sickle cell disease. Haematologica, 2019, 104, e91-e93.	1.7	20
47	Localization of the Glucagon Receptor Gene to Human Chromosome Band 17q25. Genomics, 1994, 20, 327-328.	1.3	19
48	Circulating DNA: a potential marker of sickle cell crisis. British Journal of Haematology, 2007, 139, 331-336.	1.2	19
49	Genetic and Molecular Analyses of Complex Metabolic Disorders: Genetic Linkage. Annals of the New York Academy of Sciences, 2002, 967, 249-257.	1.8	18
50	Hydroxyurea therapy lowers circulating DNA levels in sickle cell anemia. American Journal of Hematology, 2008, 83, 714-716.	2.0	18
51	A survey of genetic fetal-haemoglobin modifiers in Nigerian patients with sickle cell anaemia. PLoS ONE, 2018, 13, e0197927.	1.1	18
52	No Evidence for Linkage at Candidate Type 2 Diabetes Susceptibility Loci on Chromosomes 12 and 20 in United Kingdom Caucasians. Journal of Clinical Endocrinology and Metabolism, 2000, 85, 853-857.	1.8	17
53	Prognostic factors of disease severity in infants with sickle cell anemia: A comprehensive longitudinal cohort study. American Journal of Hematology, 2018, 93, 1411-1419.	2.0	17
54	Two candidate genes for low platelet count identified in an Asian Indian kindred by genome-wide linkage analysis: glycoprotein IX and thrombopoietin. European Journal of Human Genetics, 2006, 14, 101-108.	1.4	15

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55	Response to hydroxyurea among <scp>K</scp>uwaiti patients with sickle cell disease and elevated baseline <scp>H</scp>b<scp>F</scp> levels. American Journal of Hematology, 2015, 90, E138-9.	2.0	15
56	Localization of the Gene Encoding a Neutral Amino Acid Transporter-like Protein to Human Chromosome Band 19q13.3 and Characterization of a Simple Sequence Repeat DNA Polymorphism. Genomics, 1994, 23, 490-491.	1.3	13
57	The effect of Duffy antigen receptor for chemokines on severity in sickle cell disease. Haematologica, 2013, 98, e87-e89.	1.7	12
58	Localization of MODY3 to a 5-cM region of human chromosome 12. Diabetes, 1995, 44, 1408-1413.	0.3	12
59	Fetal Hemoglobin is Associated with Peripheral Oxygen Saturation in Sickle Cell Disease in Tanzania. EBioMedicine, 2017, 23, 146-149.	2.7	11
60	F cell numbers are associated with an Xâ€linked genetic polymorphism and correlate with haematological parameters in patients with sickle cell disease. British Journal of Haematology, 2020, 191, 888-896.	1.2	10
61	Genome wide association study of silent cerebral infarction in sickle cell disease (HbSS and HbSC). Haematologica, 2021, 106, 1770-1773.	1.7	10
62	Experimental Generation of SNP Haplotype Signatures in Patients with Sickle Cell Anaemia. PLoS ONE, 2010, 5, e13004.	1.1	9
63	Identifying genetic variants and pathways associated with extreme levels of fetal hemoglobin in sickle cell disease in Tanzania. BMC Medical Genetics, 2020, 21, 125.	2.1	9
64	Evaluation of oxidative stress-related genetic variants for predicting stroke in patients with sickle cell anemia. Journal of the Neurological Sciences, 2020, 414, 116839.	0.3	9
65	Isolation of a cDNA clone encoding a KATP channel-like protein expressed in insulin-secreting cells, localization of the human gene to chromosome band 21q22.1, and linkage studies with NIDDM. Diabetes, 1995, 44, 592-596.	0.3	9
66	Ethnic differences in F cell levels in Jamaica: a potential tool for identifying new genetic loci controlling fetal haemoglobin. British Journal of Haematology, 2009, 144, 954-960.	1.2	8
67	Genetic Analysis of Patients With Sickle Cell Anemia and Stroke Before 4 Years of Age Suggest an Important Role for Apoliprotein E. Circulation Genomic and Precision Medicine, 2020, 13, 531-540.	1.6	8
68	The effects of hydroxycarbamide on the plasma proteome of children with sickle cell anaemia. British Journal of Haematology, 2019, 186, 879-886.	1.2	7
69	<scp>ASH</scp>1L (a histone methyltransferase protein) is a novel candidate globin gene regulator revealed by genetic study of an English family with betaâ€thalassaemia unlinked to the betaâ€globin locus. British Journal of Haematology, 2016, 175, 525-530.	1.2	6
70	Sickle cell disease in Sri Lanka: clinical and molecular basis and the unanswered questions about disease severity. Orphanet Journal of Rare Diseases, 2020, 15, 177.	1.2	6
71	Genetic variants of PKLR are associated with acute pain in sickle cell disease. Blood Advances, 2022, 6, 3535-3540.	2.5	6
72	Variantes genÃ©ticas asociadas con niveles de hemoglobina fetal muestran diversos orÃgenes Ã©tnicos en pacientes colombianos con anemia falciforme. Biomedica, 2015, 35, 437-43.	0.3	5

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73	A Twins Heritability Study on Alpha Hemoglobin Stabilizing Protein (AHSP) Expression Variability. Twin Research and Human Genetics, 2010, 13, 567-572.	0.3	3
74	Fetalâ€haemoglobin enhancing genotype at BCL11A reduces HbA 2 levels in patients with sickle cell anaemia. EJHaem, 2021, 2, 459-461.	0.4	1
75	HBS1L-MYB intergenic Variants Modulate Fetal Hemoglobin Via Long-Range MYB Enhancers. Blood, 2013, 122, 43-43.	0.6	1
76	A Machine Learning Model for Predicting Fetal Hemoglobin Levels in Sickle Cell Disease Patients. Lecture Notes in Networks and Systems, 2022, , 79-91.	0.5	0
77	Searching for NIDDM susceptibility genes: studies of genes with triplet repeats expressed in skeletal muscle. Diabetologia, 1996, 39, 725-730.	2.9	0