

# Swee Lay Thein

## List of Publications by Year in descending order

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132  
papers

9,926  
citations

71102

41  
h-index

37204

96  
g-index

137  
all docs

137  
docs citations

137  
times ranked

8569  
citing authors

#	ARTICLE	IF	CITATIONS
1	Hypervariable $\alpha$ -minisatellite <sup>TM</sup> regions in human DNA. <i>Nature</i> , 1985, 314, 67-73.	27.8	3,495
2	A QTL influencing F cell production maps to a gene encoding a zinc-finger protein on chromosome 2p15. <i>Nature Genetics</i> , 2007, 39, 1197-1199.	21.4	491
3	A genome-wide meta-analysis identifies 22 loci associated with eight hematological parameters in the HaemGen consortium. <i>Nature Genetics</i> , 2009, 41, 1182-1190.	21.4	481
4	Intergenic variants of HBS1L-MYB are responsible for a major quantitative trait locus on chromosome 6q23 influencing fetal hemoglobin levels in adults. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2007, 104, 11346-11351.	7.1	286
5	The Molecular Basis of $\alpha$ -Thalassemia. <i>Cold Spring Harbor Perspectives in Medicine</i> , 2013, 3, a011700-a011700.	6.2	268
6	American Society of Hematology 2020 guidelines for sickle cell disease: transfusion support. <i>Blood Advances</i> , 2020, 4, 327-355.	5.2	241
7	Control of fetal hemoglobin: new insights emerging from genomics and clinical implications. <i>Human Molecular Genetics</i> , 2009, 18, R216-R223.	2.9	213
8	Discovering the genetics underlying foetal haemoglobin production in adults. <i>British Journal of Haematology</i> , 2009, 145, 455-467.	2.5	171
9	cMYB is involved in the regulation of fetal hemoglobin production in adults. <i>Blood</i> , 2006, 108, 1077-1083.	1.4	163
10	HBS1L-MYB intergenic variants modulate fetal hemoglobin via long-range MYB enhancers. <i>Journal of Clinical Investigation</i> , 2014, 124, 1699-1710.	8.2	157
11	Survival in adults with sickle cell disease in a high-income setting. <i>Blood</i> , 2016, 128, 1436-1438.	1.4	153
12	Systematic documentation and analysis of human genetic variation in hemoglobinopathies using the microattribution approach. <i>Nature Genetics</i> , 2011, 43, 295-301.	21.4	142
13	Molecular basis of $\beta^0$ thalassemia and potential therapeutic targets. <i>Blood Cells, Molecules, and Diseases</i> , 2018, 70, 54-65.	1.4	138
14	Genetic modifiers of the $\beta^0$ -haemoglobinopathies. <i>British Journal of Haematology</i> , 2008, 141, 357-366.	2.5	117
15	Genetics of fetal hemoglobin in Tanzanian and British patients with sickle cell anemia. <i>Blood</i> , 2011, 117, 1390-1392.	1.4	104
16	Association between hemolysis and albuminuria in adults with sickle cell anemia. <i>Haematologica</i> , 2012, 97, 201-205.	3.5	97
17	The HBS1L-MYB intergenic interval associated with elevated HbF levels shows characteristics of a distal regulatory region in erythroid cells. <i>Blood</i> , 2009, 114, 1254-1262.	1.4	95
18	DOMINANT $\beta^0$ THALASSAEMIA: MOLECULAR BASIS AND PATHOPHYSIOLOGY. <i>British Journal of Haematology</i> , 1992, 80, 273-277.	2.5	89

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19	Molecular therapies in $\beta$ -thalassaemia. British Journal of Haematology, 2007, 136, 353-365.	2.5	83
20	Hepatic Dysfunction in Sickle Cell Disease: A New System of Classification Based on Global Assessment. Clinical Gastroenterology and Hepatology, 2007, 5, 1469-1476.	4.4	79
21	Genome Wide Association Study of Fetal Hemoglobin in Sickle Cell Anemia in Tanzania. PLoS ONE, 2014, 9, e111464.	2.5	78
22	The linear effects of $\alpha$ -thalassaemia, the UGT1A1 and HMOX1 polymorphisms on cholelithiasis in sickle cell disease. British Journal of Haematology, 2007, 138, 263-270.	2.5	77
23	Pathophysiology of $\beta^0$ Thalassemia – A Guide to Molecular Therapies. Hematology American Society of Hematology Education Program, 2005, 2005, 31-37.	2.5	76
24	Spectral domain optical coherence tomography in patients with sickle cell disease. British Journal of Ophthalmology, 2015, 99, 967-972.	3.9	74
25	The HBS1L-MYB intergenic region on chromosome 6q23.3 influences erythrocyte, platelet, and monocyte counts in humans. Blood, 2007, 110, 3624-3626.	1.4	71
26	How we treat sickle hepatopathy and liver transplantation in adults. Blood, 2014, 123, 2302-2307.	1.4	71
27	Genome-wide association analyses based on whole-genome sequencing in Sardinia provide insights into regulation of hemoglobin levels. Nature Genetics, 2015, 47, 1264-1271.	21.4	66
28	Sickle Cell Anemia and Its Phenotypes. Annual Review of Genomics and Human Genetics, 2018, 19, 113-147.	6.2	66
29	Recombination Breakpoints in the Human $\beta^0$ -Globin Gene Cluster. Blood, 1998, 92, 4415-4421.	1.4	64
30	Genetic association studies in $\beta^0$ -hemoglobinopathies. Hematology American Society of Hematology Education Program, 2013, 2013, 354-361.	2.5	62
31	Anemia in the elderly: clinical implications and new therapeutic concepts. Haematologica, 2014, 99, 1127-1130.	3.5	62
32	How I treat renal complications in sickle cell disease. Blood, 2014, 123, 3720-3726.	1.4	62
33	IS IT DOMINANTLY INHERITED $\beta^0$ THALASSAEMIA OR JUST A $\beta^0$ -CHAIN VARIANT THAT IS HIGHLY UNSTABLE?. British Journal of Haematology, 1999, 107, 12-21.	2.5	60
34	The platelet NLRP3 inflammasome is upregulated in sickle cell disease via HMGB1/TLR4 and Bruton tyrosine kinase. Blood Advances, 2018, 2, 2672-2680.	5.2	56
35	Blood transfusion usage among adults with sickle cell disease – a single institution experience over ten years. British Journal of Haematology, 2011, 152, 766-770.	2.5	53
36	Binding patterns of BCL11A in the globin and GATA1 loci and characterization of the BCL11A fetal hemoglobin locus. Blood Cells, Molecules, and Diseases, 2010, 45, 140-146.	1.4	52

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37	Treatment of sickle cell disease by increasing oxygen affinity of hemoglobin. Blood, 2021, 138, 1172-1181.	1.4	52
38	Circulating mitochondrial DNA is a proinflammatory DAMP in sickle cell disease. Blood, 2021, 137, 3116-3126.	1.4	51
39	Molecular characterization of a novel 10.3 kb deletion causing $\beta^2$ -thalassaemia with unusually high Hb A <sub>2</sub> . British Journal of Haematology, 1992, 82, 735-744.	2.5	48
40	Sickle cell disease in the older adult. Pathology, 2017, 49, 1-9.	0.6	48
41	MYB – A regulatory factor in hematopoiesis. Gene, 2018, 665, 6-17.	2.2	48
42	Elimination of Transfusions Through Induction of Fetal Hemoglobin Synthesis in Cooley's Anemia <sup>a</sup> . Annals of the New York Academy of Sciences, 1998, 850, 100-109.	3.8	47
43	A base substitution (T→C) in codon 29 of the $\beta^2$ -globin gene causes $\beta^2$ thalassaemia. British Journal of Haematology, 1993, 85, 546-552.	2.5	47
44	Genetic determinants of haemolysis in sickle cell anaemia. British Journal of Haematology, 2013, 161, 270-278.	2.5	45
45	Erythroblastic Inclusions in Dominantly Inherited $\beta^2$ Thalassemias. Blood, 1997, 89, 322-328.	1.4	45
46	Treating sickle cell anemia. Science, 2020, 367, 1198-1199.	12.6	44
47	Hemolytic transfusion reactions in sickle cell disease: underappreciated and potentially fatal. Haematologica, 2020, 105, 539-544.	3.5	44
48	The carrier state for sickle cell disease is not completely harmless. Haematologica, 2019, 104, 1106-1111.	3.5	38
49	Pain and opioid use after reversal of sickle cell disease following <sc>HLA</sc>-matched sibling haematopoietic stem cell transplant. British Journal of Haematology, 2019, 184, 690-693.	2.5	37
50	The Molecular Basis of $\beta^2$ Thalassemia, $\beta^2$ Thalassemia, and Hereditary Persistence of Fetal Hemoglobin. , 2009, , 323-356.		34
51	A phase 1 dose escalation study of the pyruvate kinase activator mitapivat (AG-348) in sickle cell disease. Blood, 2022, 140, 2053-2062.	1.4	34
52	Genetic Basis and Genetic Modifiers of $\beta^2$ -Thalassemia and Sickle Cell Disease. Advances in Experimental Medicine and Biology, 2017, 1013, 27-57.	1.6	33
53	g(HbF): a genetic model of fetal hemoglobin in sickle cell disease. Blood Advances, 2018, 2, 235-239.	5.2	33
54	Pro-inflammatory cytokines associate with NETosis during sickle cell vaso-occlusive crises. Cytokine, 2020, 127, 154933.	3.2	33

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55	$\alpha^2\beta^2$ levels in normal adults are influenced by two distinct genetic mechanisms. British Journal of Haematology, 2013, 160, 101-105.	2.5	32
56	Genetic Modifiers of Fetal Haemoglobin in Sickle Cell Disease. Molecular Diagnosis and Therapy, 2019, 23, 235-244.	3.8	32
57	Cardiovascular complications of sickle cell disease. Trends in Cardiovascular Medicine, 2021, 31, 187-193.	4.9	32
58	Pulmonary function, CT and echocardiographic abnormalities in sickle cell disease. Thorax, 2014, 69, 746-751.	5.6	31
59	How I treat the older adult with sickle cell disease. Blood, 2018, 132, 1750-1760.	1.4	31
60	COVID-19 and sickle cell disease. Haematologica, 2020, 105, 2501-2504.	3.5	30
61	Associations between environmental factors and hospital admissions for sickle cell disease. Haematologica, 2017, 102, 666-675.	3.5	29
62	Platelets at the crossroads of thrombosis, inflammation and haemolysis. British Journal of Haematology, 2018, 180, 761-767.	2.5	28
63	Intracranial Aneurysms in Sickle-Cell Disease Are Associated With the Hemoglobin SS Genotype But Not With Moyamoya Syndrome. Stroke, 2016, 47, 1710-1713.	2.0	27
64	Allosteric control of hemoglobin S fiber formation by oxygen and its relation to the pathophysiology of sickle cell disease. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 15018-15027.	7.1	26
65	Association of sickle avascular necrosis with bone morphogenic protein 6. Annals of Hematology, 2009, 88, 803-805.	1.8	25
66	Reduced rate of sickle cell-related complications in Brazilian patients carrying HbF-promoting alleles at the <i>BCL11A</i> and <i>HMIP-2</i> loci. British Journal of Haematology, 2016, 173, 456-460.	2.5	25
67	Safety and Tolerability Of MP4CO: A Dose Escalation Study In Stable Patients With Sickle Cell Disease. Blood, 2013, 122, 2205-2205.	1.4	25
68	Genetic Modifiers of Sickle Cell Disease. Hemoglobin, 2011, 35, 589-606.	0.8	24
69	Global Genetic Architecture of an Erythroid Quantitative Trait Locus, <i>HMIP-2</i> . Annals of Human Genetics, 2014, 78, 434-451.	0.8	24
70	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
71	Pulmonary Haemodynamics in Sickle Cell Disease Are Driven Predominantly by a High-Output State Rather Than Elevated Pulmonary Vascular Resistance: A Prospective 3-Dimensional Echocardiography/Doppler Study. PLoS ONE, 2015, 10, e0135472.	2.5	24
72	Association of plasma CD40L with acute chest syndrome in sickle cell anemia. Cytokine, 2017, 97, 104-107.	3.2	23

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73	Switching from fetal to adult hemoglobin. <i>Nature Genetics</i> , 2018, 50, 478-480.	21.4	23
74	Genetic variants at HbF modifier loci moderate anemia and leukocytosis in sickle cell disease in Tanzania. <i>American Journal of Hematology</i> , 2015, 90, E1-4.	4.1	21
75	Serum ferritin and total units transfused for assessing iron overload in adults with sickle cell disease. <i>British Journal of Haematology</i> , 2012, 157, 645-647.	2.5	20
76	Next Generation Sequencing Identifies a Novel Rearrangement in the <i>HBB</i> Cluster Permitting to-the-Base Characterization. <i>Human Mutation</i> , 2015, 36, 142-150.	2.5	20
77	Circulating DNA: a potential marker of sickle cell crisis. <i>British Journal of Haematology</i> , 2007, 139, 331-336.	2.5	19
78	The presence of $\alpha^+$ thalassaemia trait blunts the response to hydroxycarbamide in patients with sickle cell disease. <i>British Journal of Haematology</i> , 2008, 143, 589-592.	2.5	19
79	Beta thalassaemia intermedia due to coinheritance of three unique alpha globin cluster duplications characterised by next generation sequencing analysis. <i>British Journal of Haematology</i> , 2018, 180, 160-164.	2.5	19
80	An in vitro system for expression analysis of mutations of the $\beta^2$ -globin gene: validation and application to two mutations in the 5' UTR. <i>British Journal of Haematology</i> , 1999, 106, 938-947.	2.5	18
81	Hydroxyurea therapy lowers circulating DNA levels in sickle cell anemia. <i>American Journal of Hematology</i> , 2008, 83, 714-716.	4.1	18
82	The clinical significance of K-Cl cotransport activity in red cells of patients with HbSC disease. <i>Haematologica</i> , 2015, 100, 595-600.	3.5	18
83	A survey of genetic fetal-haemoglobin modifiers in Nigerian patients with sickle cell anaemia. <i>PLoS ONE</i> , 2018, 13, e0197927.	2.5	18
84	The investigation of resveratrol and analogs as potential inducers of fetal hemoglobin. <i>Blood Cells, Molecules, and Diseases</i> , 2016, 58, 6-12.	1.4	16
85	Neutrophils remain detrimentally active in hydroxyurea-treated patients with sickle cell disease. <i>PLoS ONE</i> , 2019, 14, e0226583.	2.5	16
86	Research in Sickle Cell Disease: From Bedside to Bench to Bedside. <i>HemaSphere</i> , 2021, 5, e584.	2.7	16
87	Sickle cell disease in a carrier with pyruvate kinase deficiency. <i>Hematology</i> , 2008, 13, 369-372.	1.5	15
88	Response to hydroxyurea among KwaZulu-Natal patients with sickle cell disease and elevated baseline HbF levels. <i>American Journal of Hematology</i> , 2015, 90, E138-9.	4.1	15
89	The spectrum of $\beta^2$ thalassaemia in Burma. <i>British Journal of Haematology</i> , 1992, 81, 574-578.	2.5	14
90	Dietary iron restriction improves markers of disease severity in murine sickle cell anemia. <i>Blood</i> , 2021, 137, 1553-1555.	1.4	14

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91	Super-elevated <scp>LDH</scp> and thrombocytopenia are markers of a severe subtype of vaso-occlusive crisis in sickle cell disease. American Journal of Hematology, 2015, 90, E206-7.	4.1	13
92	The effect of Duffy antigen receptor for chemokines on severity in sickle cell disease. Haematologica, 2013, 98, e87-e89.	3.5	12
93	Heterogeneity of respiratory disease in children and young adults with sickle cell disease. Thorax, 2018, 73, 575-577.	5.6	12
94	NLRP3 inflammasome and bruton tyrosine kinase inhibition interferes with upregulated platelet aggregation and in-vitro thrombus formation in sickle cell mice. Biochemical and Biophysical Research Communications, 2021, 555, 196-201.	2.1	12
95	Airway and alveolar nitric oxide production, lung function, and pulmonary blood flow in sickle cell disease. Pediatric Research, 2016, 79, 313-317.	2.3	11
96	Acute human parvovirus B19 infection and nephrotic syndrome in patients with sickle cell disease. British Journal of Haematology, 2010, 149, 289-291.	2.5	10
97	Prevention of Morbidity in sickle cell disease - qualitative outcomes, pain and quality of life in a randomised cross-over pilot trial of overnight supplementary oxygen and auto-adjusting continuous positive airways pressure (POMS2a): study protocol for a randomised controlled trial. Trials, 2015, 16, 376.	1.6	10
98	Genetic control of erythropoiesis. Current Opinion in Hematology, 2017, 24, 173-182.	2.5	10
99	Voxelotor treatment of a patient with sickle cell disease and very severe anemia. American Journal of Hematology, 2019, 94, E88-E90.	4.1	9
100	A phenotypic risk score for predicting mortality in sickle cell disease. British Journal of Haematology, 2021, 192, 932-941.	2.5	9
101	Mitapivat increases ATP and decreases oxidative stress and erythrocyte mitochondria retention in a SCD mouse model. Blood Cells, Molecules, and Diseases, 2022, 95, 102660.	1.4	9
102	First reported duplication of the entire beta globin gene cluster causing an unusual sickle cell trait phenotype. British Journal of Haematology, 2015, 170, 128-131.	2.5	8
103	Increased prevalence of renal cysts in patients with sickle cell disease. BMC Nephrology, 2017, 18, 298.	1.8	8
104	Detection and Quantification of Histone H4 Citrullination in Early NETosis With Image Flow Cytometry Version 4. Frontiers in Immunology, 2020, 11, 1335.	4.8	8
105	Revisiting anemia in sickle cell disease and finding the balance with therapeutic approaches. Blood, 2022, 139, 3030-3039.	1.4	8
106	Prenatal Diagnosis in Combined Antithrombin and Factor V Gene Mutation. British Journal of Haematology, 1996, 94, 753-755.	2.5	7
107	A Plea for the Newborn Diagnosis of Hb S-Hereditary Persistence of Fetal Hemoglobin. Hemoglobin, 2017, 41, 216-217.	0.8	7
108	Optimal disease management and health monitoring in adults with sickle cell disease. Hematology American Society of Hematology Education Program, 2019, 2019, 505-512.	2.5	7

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109	Loss of Major DNase I Hypersensitive Sites in Duplicated $\beta^2$ -globin Gene Cluster Incompletely Silences HBB Gene Expression. Human Mutation, 2016, 37, 1153-1156.	2.5	6
110	<sc>ASH</sc> 1L (a histone methyltransferase protein) is a novel candidate globin gene regulator revealed by genetic study of an English family with beta $\alpha$ -thalassaemia unlinked to the beta $\alpha$ -globin locus. British Journal of Haematology, 2016, 175, 525-530.	2.5	6
111	Detection of Hb Rothschild HBB: c.[112T>A or 112T>C], Through High Index of Suspicion on Abnormal Pulse Oximetry. Hemoglobin, 2017, 41, 137-139.	0.8	6
112	Genetic variants of <i>PKLR</i> are associated with acute pain in sickle cell disease. Blood Advances, 2022, 6, 3535-3540.	5.2	6
113	A Growing Population of Older Adults with Sickle Cell Disease. Clinics in Geriatric Medicine, 2019, 35, 349-367.	2.6	5
114	Whole genome sequence-based haplotypes reveal a single origin of the 1393 bp HBB deletion. Journal of Medical Genetics, 2020, 57, 567-570.	3.2	5
115	Evaluation of Hepatic Iron Overload Using a Contemporary 0.<sc>55 $\pm$ %T MRI</sc> System. Journal of Magnetic Resonance Imaging, 2022, 55, 1855-1863.	3.4	4
116	Automated genotyping for accurate assignment of the (AT) $\times$ N $\alpha$ (AT) $\gamma$ motif within the $\beta^2$ -globin locus control region-hypersensitive site 2. British Journal of Haematology, 2001, 112, 488-492.	2.5	3
117	Genetic Factors Modifying Sickle Cell Disease Severity. , 2016, , 371-397.		3
118	An Imaging Flow Cytometry Method to Measure Citrullination of H4 Histone as a Read-out for Neutrophil Extracellular Traps Formation. Bio-protocol, 2021, 11, e3927.	0.4	3
119	Sickle Cell Anaemia in East Africa: Preliminary Results from a Cohort Study.. Blood, 2006, 108, 3802-3802.	1.4	3
120	Sickle cell disease $\alpha$ ”Unanswered questions and future directions in therapy. Seminars in Hematology, 2018, 55, 51-52.	3.4	2
121	Targeting ZNF410 as a potential $\beta^2$ -hemoglobinopathy therapy. Nature Genetics, 2021, 53, 589-590.	21.4	2
122	Deferasirox Efficacy and Safety for the Treatment of Transfusion- Dependent Iron Overload in Patients with a Range of Rare Anemias.. Blood, 2008, 112, 1419-1419.	1.4	2
123	HBS1L-MYB intergenic Variants Modulate Fetal Hemoglobin Via Long-Range MYB Enhancers. Blood, 2013, 122, 43-43.	1.4	1
124	Heterogeneity of the $\beta^0\beta^0$ Thalassemias: Characterization of 3 Novel English Deletions.. Blood, 2004, 104, 3609-3609.	1.4	0
125	Cell Free Fetal and Total DNA Levels in Pregnancies at Risk of Sickle Cell Disease and Significant Ethnic Variation.. Blood, 2006, 108, 3791-3791.	1.4	0
126	Extracranial Internal Carotid Arterial Disease in Children with Sickle Cell Disease.. Blood, 2009, 114, 2560-2560.	1.4	0



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127	Addition of Hydroxyurea to Transfusion Programme to Treat Progressive Cerebral Vasculopathy. Blood, 2010, 116, 4813-4813.	1.4	0
128	Two Distinct Genetic Mechanisms Modify the Level of HbA2 in Peripheral Blood,. Blood, 2011, 118, 3193-3193.	1.4	0
129	Renal Iron Load in Sickle Cell Disease Correlates with Hemolysis and Transfusion History, but Not with Hepatic Iron. Blood, 2011, 118, 2129-2129.	1.4	0
130	European Chromosome 6 Haplotypes Significantly Augment Fetal Hemoglobin Levels in Brazilian Sickle Cell Anemia Patients: Influence of Four HBS1L-MYB Intergenic Region SNPs. Blood, 2012, 120, 1002-1002.	1.4	0
131	Super-Elevated LDH and Thombocytopenia Are Markers Of An Unusual Sickle Phenomenon. Blood, 2013, 122, 2220-2220.	1.4	0
132	Telomere Lengths Correlate With Inflammatory Markers In Sickle Cell Disease. Blood, 2013, 122, 2230-2230.	1.4	0