Douglas Higgs

List of Publications by Year in descending order

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Version: 2024-02-01

166 papers	10,210 citations	31976 53 h-index	40979 93 g-index
236 all docs	236 docs citations	236 times ranked	11156 citing authors

#	Article	IF	Citations
1	Î±-thalassaemia. Orphanet Journal of Rare Diseases, 2010, 5, 13.	2.7	417
2	Analysis of hundreds of cis-regulatory landscapes at high resolution in a single, high-throughput experiment. Nature Genetics, 2014, 46, 205-212.	21.4	417
3	Thalassaemia. Lancet, The, 2012, 379, 373-383.	13.7	371
4	Considerations when investigating lncRNA function in vivo. ELife, 2014, 3, e03058.	6.0	309
5	Genetic dissection of the Î±-globin super-enhancer in vivo. Nature Genetics, 2016, 48, 895-903.	21.4	308
6	A truncated human chromosome 16 associated with Î± thalassaemia is stabilized by addition of telomeric repeat (TTAGGG)n. Nature, 1990, 346, 868-871.	27.8	300
7	Transcription factors LRF and BCL11A independently repress expression of fetal hemoglobin. Science, 2016, 351, 285-289.	12.6	260
8	A Regulatory SNP Causes a Human Genetic Disease by Creating a New Transcriptional Promoter. Science, 2006, 312, 1215-1217.	12.6	254
9	Intragenic Enhancers Act as Alternative Promoters. Molecular Cell, 2012, 45, 447-458.	9.7	237
10	Association between active genes occurs at nuclear speckles and is modulated by chromatin environment. Journal of Cell Biology, 2008, 182, 1083-1097.	5.2	231
11	Multiplexed analysis of chromosome conformation at vastly improved sensitivity. Nature Methods, 2016, 13, 74-80.	19.0	225
12	Long-range chromosomal interactions regulate the timing of the transition between poised and active gene expression. EMBO Journal, 2007, 26, 2041-2051.	7.8	224
13	Suppression of the alternative lengthening of telomere pathway by the chromatin remodelling factor ATRX. Nature Communications, 2015, 6, 7538.	12.8	219
14	Mutations in transcriptional regulator ATRX establish the functional significance of a PHD-like domain. Nature Genetics, 1997, 17, 146-148.	21.4	196
15	Globin gene activation during haemopoiesis is driven by protein complexes nucleated by GATA-1 and GATA-2. EMBO Journal, 2004, 23, 2841-2852.	7.8	193
16	Coregulated human globin genes are frequently in spatial proximity when active. Journal of Cell Biology, 2006, 172, 177-187.	5.2	192
17	Chromatin signatures at transcriptional start sites separate two equally populated yet distinct classes of intergenic long noncoding RNAs. Genome Biology, 2013, 14, R131.	9.6	183
18	Tissue-specific CTCFâ€“cohesin-mediated chromatin architecture delimits enhancer interactions and function in vivo. Nature Cell Biology, 2017, 19, 952-961.	10.3	179

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19	The relationship between genome structure and function. Nature Reviews Genetics, 2021, 22, 154-168.	16.3	160
20	ATRX Plays a Key Role in Maintaining Silencing at Interstitial Heterochromatic Loci and Imprinted Genes. Cell Reports, 2015, 11, 405-418.	6.4	152
21	Comparative genome analysis delimits a chromosomal domain and identifies key regulatory elements in the alpha globin cluster. Human Molecular Genetics, 2001, 10, 371-382.	2.9	151
22	Manipulating the Mouse Genome to Engineer Precise Functional Syntenic ReplacementsÂwith Human Sequence. Cell, 2007, 128, 197-209.	28.9	150
23	Single-allele chromatin interactions identify regulatory hubs in dynamic compartmentalized domains. Nature Genetics, 2018, 50, 1744-1751.	21.4	150
24	The relationship between chromosome structure and function at a human telomeric region. Nature Genetics, 1997, 15, 252-257.	21.4	143
25	Systematic documentation and analysis of human genetic variation in hemoglobinopathies using the microattribution approach. Nature Genetics, 2011, 43, 295-301.	21.4	142
26	Expression of Î±- and Î²-globin genes occurs within different nuclear domains in haemopoietic cells. Nature Cell Biology, 2001, 3, 602-606.	10.3	139
27	Annotation of cis-regulatory elements by identification, subclassification, and functional assessment of multispecies conserved sequences. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 9830-9835.	7.1	133
28	DNA methylation of intragenic CpG islands depends on their transcriptional activity during differentiation and disease. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E7526-E7535.	7.1	125
29	KrÃ¼ppeling erythropoiesis: an unexpected broad spectrum of human red blood cell disorders due to KLF1 variants. Blood, 2016, 127, 1856-1862.	1.4	124
30	How best to identify chromosomal interactions: a comparison of approaches. Nature Methods, 2017, 14, 125-134.	19.0	124
31	Defining genome architecture at base-pair resolution. Nature, 2021, 595, 125-129.	27.8	107
32	The Molecular Basis of Â-Thalassemia. Cold Spring Harbor Perspectives in Medicine, 2013, 3, a011718-a011718.	6.2	106
33	Î±-Globin as a molecular target in the treatment of Î²-thalassemia. Blood, 2015, 125, 3694-3701.	1.4	102
34	Loss of Extreme Long-Range Enhancers in Human Neural Crest Drives a Craniofacial Disorder. Cell Stem Cell, 2020, 27, 765-783.e14.	11.1	101
35	The chromatin remodelling factor <scp>ATRX</scp> suppresses Râ€loops in transcribed telomeric repeats. EMBO Reports, 2017, 18, 914-928.	4.5	99
36	Single-Cell Proteomics Reveal that Quantitative Changes in Co-expressed Lineage-Specific Transcription Factors Determine Cell Fate. Cell Stem Cell, 2019, 24, 812-820.e5.	11.1	99

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37	Global gene expression analysis of human erythroid progenitors. Blood, 2011, 117, e96-e108.	1.4	95
38	5 Î±-Thalassaemia. Best Practice and Research: Clinical Haematology, 1993, 6, 117-150.	1.1	94
39	Molecular Basis and Genetic Modifiers of Thalassemia. Hematology/Oncology Clinics of North America, 2018, 32, 177-191.	2.2	93
40	Editing an Î±-globin enhancer in primary human hematopoietic stem cells as a treatment for Î²-thalassemia. Nature Communications, 2017, 8, 424.	12.8	85
41	ATRX Dysfunction Induces Replication Defects in Primary Mouse Cells. PLoS ONE, 2014, 9, e92915.	2.5	84
42	Sequence, structure and pathology of the fully annotated terminal 2 Mb of the short arm of human chromosome 16. Human Molecular Genetics, 2001, 10, 339-352.	2.9	81
43	Chromosome looping at the human Î±-globin locus is mediated via the major upstream regulatory element (HS â^'40). Blood, 2009, 114, 4253-4260.	1.4	79
44	Dynamics of the 4D genome during in vivo lineage specification and differentiation. Nature Communications, 2020, 11, 2722.	12.8	79
45	Adventitious changes in long-range gene expression caused by polymorphic structural variation and promoter competition. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 21771-21776.	7.1	77
46	Mutations in KrÃ¼ppel-like factor 1 cause transfusion-dependent hemolytic anemia and persistence of embryonic globin gene expression. Blood, 2014, 123, 1586-1595.	1.4	76
47	A nonsense mutation of theATRX gene causing mild mental retardation and epilepsy. Annals of Neurology, 2000, 47, 117-121.	5.3	72
48	Individual specific DNA fingerprints from a hypervariable region probe: alpha-globin 3?HVR. Human Genetics, 1988, 79, 142-146.	3.8	71
49	Homozygous mutations in a predicted endonuclease are a novel cause of congenital dyserythropoietic anemia type I. Haematologica, 2013, 98, 1383-1387.	3.5	71
50	Tissue-specific histone modification and transcription factor binding in Î± globin gene expression. Blood, 2007, 110, 4503-4510.	1.4	69
51	Long-range regulation of ?? globin gene expression during erythropoiesis. Current Opinion in Hematology, 2008, 15, 176-183.	2.5	66
52	Comparison of the human and murine ATRX gene identifies highly conserved, functionally important domains. Mammalian Genome, 1998, 9, 400-403.	2.2	64
53	A tissue-specific self-interacting chromatin domain forms independently of enhancer-promoter interactions. Nature Communications, 2018, 9, 3849.	12.8	62
54	The bipartite TAD organization of the X-inactivation center ensures opposing developmental regulation of Tsix and Xist. Nature Genetics, 2019, 51, 1024-1034.	21.4	60

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55	A revised model for promoter competition based on multi-way chromatin interactions at the Î±-globin locus. Nature Communications, 2019, 10, 5412.	12.8	60
56	An international registry of survivors with Hb Bart's hydrops fetalis syndrome. Blood, 2017, 129, 1251-1259.	1.4	59
57	Codanin-1 mutations in congenital dyserythropoietic anemia type 1 affect HP1Î± localization in erythroblasts. Blood, 2011, 117, 6928-6938.	1.4	58
58	Structure and expression of the human Î¸l globin gene. Nature, 1988, 331, 94-96.	27.8	53
59	Deletion of the mouse Î±-globin regulatory element (HS â^'26) has an unexpectedly mild phenotype. Blood, 2002, 100, 3450-3456.	1.4	53
60	Functional and comparative analysis of globin loci in pufferfish and humans. Blood, 2003, 101, 2842-2849.	1.4	53
61	Testing the super-enhancer concept. Nature Reviews Genetics, 2021, 22, 749-755.	16.3	53
62	X-linked Î±-thalassemia/mental retardation (ATR-X) syndrome: A new kindred with severe genital anomalies and mild hematologic expression. American Journal of Medical Genetics Part A, 1995, 55, 302-306.	2.4	50
63	Monosomy for the most telomeric, gene-rich region of the short arm of human chromosome 16 causes minimal phenotypic effects. European Journal of Human Genetics, 2001, 9, 217-225.	2.8	47
64	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
65	The haematology of homozygous sickle cell disease after the age of 40 years. British Journal of Haematology, 1991, 77, 382-385.	2.5	44
66	Understanding Î±â€globin gene regulation and implications for the treatment of Î²â€thalassemia. Annals of the New York Academy of Sciences, 2016, 1368, 16-24.	3.8	44
67	Genetic complexity in sickle cell disease. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 11595-11596.	7.1	43
68	Nprl3 is required for normal development of the cardiovascular system. Mammalian Genome, 2012, 23, 404-415.	2.2	38
69	An integrative view of the regulatory and transcriptional landscapes in mouse hematopoiesis. Genome Research, 2020, 30, 472-484.	5.5	38
70	A Dynamic Folded Hairpin Conformation Is Associated with Î±-Globin Activation in Erythroid Cells. Cell Reports, 2020, 30, 2125-2135.e5.	6.4	38
71	De novo deletion within the telomeric region flanking the human Î± globin locus as a cause of Î± thalassaemia. British Journal of Haematology, 2003, 120, 867-875.	2.5	36
72	The Molecular Basis of Î±-Thalassemia: A Model for Understanding Human Molecular Genetics. Hematology/Oncology Clinics of North America, 2010, 24, 1033-1054.	2.2	36

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73	The Molecular Basis of Î² Thalassemia, ÎÎ² Thalassemia, and Hereditary Persistence of Fetal Hemoglobin. , 2009, , 323-356.		34
74	Using Genomics to Study How Chromatin Influences Gene Expression. Annual Review of Genomics and Human Genetics, 2007, 8, 299-325.	6.2	33
75	Selective silencing of Î±-globin by the histone demethylase inhibitor IOX1: a potentially new pathway for treatment of Î²-thalassemia. Haematologica, 2017, 102, e80-e84.	3.5	33
76	Sasquatch: predicting the impact of regulatory SNPs on transcription factor binding from cell- and tissue-specific DNase footprints. Genome Research, 2017, 27, 1730-1742.	5.5	33
77	Functional characterisation of cis-regulatory elements governing dynamic Eomes expression in the early mouse embryo. Development (Cambridge), 2017, 144, 1249-1260.	2.5	32
78	Chapter 5 Longâ€Range Regulation of Î±â€Globin Gene Expression. Advances in Genetics, 2008, 61, 143-173.	1.8	30
79	Robust detection of chromosomal interactions from small numbers of cells using low-input Capture-C. Nucleic Acids Research, 2017, 45, e184-e184.	14.5	27
80	Enhancers predominantly regulate gene expression during differentiation via transcription initiation. Molecular Cell, 2021, 81, 983-997.e7.	9.7	27
81	A large deletion in the human Â-globin cluster caused by a replication error is associated with an unexpectedly mild phenotype. Human Molecular Genetics, 2008, 17, 3084-3093.	2.9	26
82	Ham-Wasserman Lecture. Hematology American Society of Hematology Education Program, 2004, 2004, 1-13.	2.5	25
83	Potential new approaches to the management of the Hb Bartâ€™s hydrops fetalis syndrome: the most severe form of Î±-thalassemia. Hematology American Society of Hematology Education Program, 2018, 2018, 353-360.	2.5	25
84	MicroRNAs of the miR-290â€“295 Family Maintain Bivalency in Mouse Embryonic Stem Cells. Stem Cell Reports, 2016, 6, 635-642.	4.8	24
85	The mouse alpha-globin cluster: a paradigm for studying genome regulation and organization. Current Opinion in Genetics and Development, 2021, 67, 18-24.	3.3	21
86	Between form and function: the complexity of genome folding. Human Molecular Genetics, 2017, 26, R208-R215.	2.9	20
87	The chromatin remodeller ATRX facilitates diverse nuclear processes, in a stochastic manner, in both heterochromatin and euchromatin. Nature Communications, 2022, 13, .	12.8	20
88	Beta thalassaemia intermedia due to coâ€inheritance of three unique alpha globin cluster duplications characterised by next generation sequencing analysis. British Journal of Haematology, 2018, 180, 160-164.	2.5	19
89	Reactivation of a developmentally silenced embryonic globin gene. Nature Communications, 2021, 12, 4439.	12.8	19
90	Causes and Consequences of Chromatin Variation between Inbred Mice. PLoS Genetics, 2013, 9, e1003570.	3.5	18

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91	A gain-of-function single nucleotide variant creates a new promoter which acts as an orientation-dependent enhancer-blocker. Nature Communications, 2021, 12, 3806.	12.8	18
92	Anaemia among females in child-bearing age: Relative contributions, effects and interactions of Î±- and Î²-thalassaemia. PLoS ONE, 2018, 13, e0206928.	2.5	17
93	Robust CRISPR/Cas9 Genome Editing of the HUDEP-2 Erythroid Precursor Line Using Plasmids and Single-Stranded Oligonucleotide Donors. Methods and Protocols, 2018, 1, 28.	2.0	17
94	An evolutionarily ancient mechanism for regulation of hemoglobin expression in vertebrate red cells. Blood, 2020, 136, 269-278.	1.4	16
95	Enhancerâ€“promoter interactions and transcription. Nature Genetics, 2020, 52, 470-471.	21.4	15
96	The Loss of ATRX Increases Susceptibility to Pancreatic Injury and Oncogenic KRAS in Female But Not Male Mice. Cellular and Molecular Gastroenterology and Hepatology, 2019, 7, 93-113.	4.5	14
97	A New Dawn for Stem-Cell Therapy. New England Journal of Medicine, 2008, 358, 964-966.	27.0	12
98	High-resolution analysis of cis -acting regulatory networks at the Î±-globin locus. Philosophical Transactions of the Royal Society B: Biological Sciences, 2013, 368, 20120361.	4.0	12
99	Dyshemoglobinemias. , 2009, , 607-622.		11
100	Dynamics and Mechanics Of KLF1 Regulation In Erythropoiesis. Blood, 2013, 122, 2176-2176.	1.4	11
101	Clinical Aspects of Î² Thalassemia and Related Disorders. , 2009, , 357-416.		10
102	Hemoglobins of the Embryo, Fetus, and Adult. , 2009, , 119-136.		10
103	Hemoglobin E Disorders. , 2009, , 417-434.		10
104	Analysis of Sequence Variation Underlying Tissue-specific Transcription Factor Binding and Gene Expression. Human Mutation, 2013, 34, 1140-1148.	2.5	10
105	Haemoglobin Variant Screening in Jamaica: Meeting Student's Request. British Journal of Haematology, 2016, 172, 634-636.	2.5	10
106	Recapitulation of erythropoiesis in congenital dyserythropoietic anemia type I (CDA-I) identifies defects in differentiation and nucleolar abnormalities. Haematologica, 2021, 106, 2960-2970.	3.5	10
107	Mechanisms and Clinical Complications of Hemolysis in Sickle Cell Disease and Thalassemia. , 2009, , 201-224.		9
108	Genetic and functional insights into CDA-I prevalence and pathogenesis. Journal of Medical Genetics, 2021, 58, 185-195.	3.2	9

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109	The Normal Structure and Regulation of Human Globin Gene Clusters. , 0, , 46-61.		8
110	Systematic integration of GATA transcription factors and epigenomes via IDEAS paints the regulatory landscape of hematopoietic cells. IUBMB Life, 2020, 72, 27-38.	3.4	8
111	Scalable in vitro production of defined mouse erythroblasts. PLoS ONE, 2022, 17, e0261950.	2.5	8
112	The Pathophysiology and Clinical Features of Î± Thalassaemia. , 2009, , 266-295.		7
113	An international effort to cure a global health problem: A report on the 19th Hemoglobin Switching Conference. Experimental Hematology, 2015, 43, 821-837.	0.4	7
114	How to Tackle Challenging ChIP-Seq, with Long-Range Cross-Linking, Using ATRX as an Example. Methods in Molecular Biology, 2018, 1832, 105-130.	0.9	7
115	How transcriptional and epigenetic programmes are played out on an individual mammalian gene cluster during lineage commitment and differentiation. Biochemical Society Symposia, 2006, 73, 11-22.	2.7	7
116	ATR-16 syndrome: mechanisms linking monosomy to phenotype. Journal of Medical Genetics, 2020, 57, 414-421.	3.2	7
117	Î± THALASSEMIA. , 0, , 239-240.		6
118	Induction of Fetal Hemoglobin in the Treatment of Sickle Cell Disease and Î² Thalassemia. , 0, , 745-754.		6
119	Unstable Hemoglobins, Hemoglobins with Altered Oxygen Affinity, Hemoglobin M, and Other Variants of Clinical and Biological Interest. , 0, , 589-606.		6
120	Phenotypic and molecular characterization of a serum-free miniature erythroid differentiation system suitable for high-throughput screening and single-cell assays. Experimental Hematology, 2018, 60, 10-20.	0.4	6
121	A remarkable case of HbH disease illustrates the relative contributions of the Î±-globin enhancers to gene expression. Blood, 2021, 137, 572-575.	1.4	6
122	Sickle Cell Pain: Biology, Etiology, and Treatment. , 0, , 497-524.		5
123	Hemoglobin SC Disease and Hemoglobin C Disorders. , 0, , 525-548.		4
124	Genetic Modulation of Sickle Cell Disease and Thalassemia. , 2009, , 638-657.		4
125	Other Sickle Hemoglobinopathies. , 0, , 564-586.		4
126	An Entirely Novel Form of Î± Thalassemia in Patients from the South Pacific Linked to Chromosome 16.. Blood, 2005, 106, 2688-2688.	1.4	4

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127	Molecular and Cellular Basis of Hemoglobin Switching. , 0, , 86-100.		3
128	The Erythrocyte Membrane. , 2009, , 158-184.		3
129	Nuclear Factors That Regulate Erythropoiesis. , 2009, , 62-85.		3
130	Clinical and Pathophysiological Aspects of Sickle Cell Anemia. , 2009, , 437-496.		3
131	Laboratory Methods for Diagnosis and Evaluation of Hemoglobin Disorders. , 0, , 658-686.		3
132	A Novel Splicing Mutation in the Gene Encoding the Chromatin-Associated Factor ATRX Associated with Acquired Hemoglobin H Disease in Myelodysplastic Syndrome (ATMDS).. Blood, 2004, 104, 3606-3606.	1.4	3
133	Transfusion and Iron Chelation Therapy in Thalassemia and Sickle Cell Disease. , 2009, , 689-744.		2
134	Unusual Types of Î± Thalassemia. , 0, , 296-320.		2
135	Studying epigenomics in single cells: what is feasible and what can we learn?. Epigenomics, 2015, 7, 1231-1234.	2.1	2
136	The Molecular Basis of Î± Thalassemia. , 2009, , 241-265.		1
137	Rheology and Vascular Pathobiology in Sickle Cell Disease and Thalassemia. , 2009, , 139-157.		1
138	Population Genetics and Global Health Burden. , 0, , 625-637.		1
139	Erythropoiesis. , 2009, , 24-45.		1
140	Stem Cell Transplantation. , 2015, , 651-675.		1
141	Hereditary Disorders of the Red Cell Membrane and Disorders of Red Cell Metabolism. , 2015, , 114-137.		1
142	Expression of the Human Alpha-Globin Cluster in the Absence of the Major Regulatory Element Mcs-R2. Blood, 2018, 132, 3632-3632.	1.4	1
143	The EHA Research Roadmap: Normal Hematopoiesis. HemaSphere, 2021, 5, e669.	2.7	1
144	A Developmental Approach to Hematopoiesis. , 2009, , 3-23.		0

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145	Prospects for Gene Therapy of Sickle Cell Disease and Thalassemia. , 2009, , 791-814.		0
146	Foreword, by H. Franklin Bunn. , 2009, , xv-xvi.		0
147	Introduction, by David J. Weatherall. , 0, , xix-xx.		0
148	The Biology of Vascular Nitric Oxide. , 0, , 185-200.		0
149	Animal Models of Hemoglobinopathies and Thalassemia. , 0, , 225-238.		0
150	Novel Approaches to Treatment. , 2009, , 755-773.		0
151	Stem Cell Transplantation. , 2009, , 774-790.		0
152	A lack of border guards?. Blood, 2006, 108, 1121-1121.	1.4	0
153	5-Azacytidine treatment of the patient with ATMDS. European Journal of Haematology, 2006, 76, 453-453.	2.2	0
154	THE Î² THALASSEMIAS. , 2009, , 321-322.		0
155	Structure and Function of Hemoglobin and Its Dysfunction in Sickle Cell Disease. , 0, , 101-118.		0
156	THE MOLECULAR, CELLULAR, AND GENETIC BASIS OF HEMOGLOBIN DISORDERS. , 2009, , 1-2.		0
157	PATHOPHYSIOLOGY OF HEMOGLOBIN AND ITS DISORDERS. , 2009, , 137-138.		0
158	Sickle Cell Trait. , 2009, , 549-563.		0
159	SPECIAL TOPICS IN HEMOGLOBINOPATHIES. , 2009, , 623-624.		0
160	NEW APPROACHES TO THE TREATMENT OF HEMOGLOBINOPATHIES AND THALASSEMIA. , 2009, , 687-688.		0
161	OTHER CLINICALLY IMPORTANT DISORDERS OF HEMOGLOBIN. , 2009, , 587-588.		0
162	Switching Genes On and Off During Hematopoiesis.. Blood, 2008, 112, sci-17-sci-17.	1.4	0

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163	Analysis of DNA Methylation at the Human Alpha Globin Cluster during Hematopoiesis.. Blood, 2008, 112, 1861-1861.	1.4	0
164	Regulated RNA Expression and Normal Erythropoiesis. Blood, 2014, 124, SCI-34-SCI-34.	1.4	0
165	Base Editing Repairs the HbE Mutation Restoring the Production of Normal Globin Chains in Severe HbE/Î²-Thalassemia Patient Hematopoietic Stem Cells and Erythroid Cells. Blood, 2021, 138, 2935-2935.	1.4	0
166	Functional impairment of erythropoiesis in Congenital Dyserythropoietic Anaemia type I arises at the progenitor level. British Journal of Haematology, 2022, , .	2.5	0