Manuela Germeshausen

List of Publications by Citations

45 papers	2,700 citations	23 h-index	45 g-index
45 ext. papers	2,965 ext. citations	6.3 avg, IF	4.45 L-index

#	Paper	IF	Citations
45	HAX1 deficiency causes autosomal recessive severe congenital neutropenia (Kostmann disease). *Nature Genetics*, **2007**, 39, 86-92	36.3	381
44	c-mpl mutations are the cause of congenital amegakaryocytic thrombocytopenia. *Blood*, **2001**, 97, 139-46	10.2	294
43	A syndrome with congenital neutropenia and mutations in G6PC3. *New England Journal of Medicine*, **2009**, 360, 32-43	59.2	276
42	Incidence of CSF3R mutations in severe congenital neutropenia and relevance for leukemogenesis: Results of a long-term survey. *Blood*, **2007**, 109, 93-9	2.2	177
41	Mutations in neutrophil elastase causing congenital neutropenia lead to cytoplasmic protein accumulation and induction of the unfolded protein response. *Blood*, **2006**, 108, 493-500	2.2	166
40	LEF-1 is crucial for neutrophil granulocytopoiesis and its expression is severely reduced in congenital neutropenia. *Nature Medicine*, **2006**, 12, 1191-7	50.5	161
39	A high-throughput sequencing test for diagnosing inherited bleeding, thrombotic, and platelet disorders. *Blood*, **2016**, 127, 2791-803	2.2	135
38	Clinical implications of ELA2-, HAX1-, and G-CSF-receptor (CSF3R) mutations in severe congenital neutropenia. *British Journal of Haematology*, **2009**, 144, 459-67	4.5	109
37	Congenital amegakaryocytic thrombocytopenia: clinical presentation, diagnosis, and treatment. *Seminars in Thrombosis and Hemostasis*, **2011**, 37, 673-81	5.3	106
36	Novel HAX1 mutations in patients with severe congenital neutropenia reveal isoform-dependent genotype-phenotype associations. *Blood*, **2008**, 111, 4954-7	2.2	102
35	Congenital amegakaryocytic thrombocytopenia: a retrospective clinical analysis of 20 patients. *British Journal of Haematology*, **2005**, 131, 636-44	4.5	94
34	MPL mutations in 23 patients suffering from congenital amegakaryocytic thrombocytopenia: the type of mutation predicts the course of the disease. *Human Mutation*, **2006**, 27, 296	4.7	79
33	Advances in the understanding of congenital amegakaryocytic thrombocytopenia. *British Journal of Haematology*, **2009**, 146, 3-16	4.5	64
32	The spectrum of ELANE mutations and their implications in severe congenital and cyclic neutropenia. *Human Mutation*, **2013**, 34, 905-14	4.7	60
31	Familial thrombocytosis caused by the novel germ-line mutation p.Pro106Leu in the MPL gene. *British Journal of Haematology*, **2009**, 144, 185-94	4.5	60
30	Granulocyte colony-stimulating factor (G-CSF) treatment of childhood acute myeloid leukemias that overexpress the differentiation-defective G-CSF receptor isoform IV is associated with a higher incidence of relapse. *Journal of Clinical Oncology*, **2010**, 28, 2591-7	2.2	54
29	Mutations in the gene encoding neutrophil elastase (ELA2) are not sufficient to cause the phenotype of congenital neutropenia. *British Journal of Haematology*, **2001**, 115, 222-4	4.5	48

28	Digenic mutations in severe congenital neutropenia. *Haematologica*, **2010**, 95, 1207-10	6.6	43
27	Time course of increasing numbers of mutations in the granulocyte colony-stimulating factor receptor gene in a patient with congenital neutropenia who developed leukemia. *Blood*, **2001**, 97, 1882-4	2.2	42
26	G-CSF receptor mutations in patients with congenital neutropenia. *Current Opinion in Hematology*, **2008**, 15, 332-7	3.3	40
25	MECOM-associated syndrome: a heterogeneous inherited bone marrow failure syndrome with amegakaryocytic thrombocytopenia. *Blood Advances*, **2018**, 2, 586-596	7.8	36
24	Granulocyte colony-stimulating factor receptor mutations in a patient with acute lymphoblastic leukemia secondary to severe congenital neutropenia. *Blood*, **2001**, 97, 829-30	2.2	27
23	Implications of mutations in hematopoietic growth factor receptor genes in congenital cytopenias. *Annals of the New York Academy of Sciences*, **2001**, 938, 305-20; discussion 320-1	6.5	26
22	In vivo expansion of cells expressing acquired CSF3R mutations in patients with severe congenital neutropenia. *Blood*, **2009**, 113, 668-70	2.2	19
21	Reduced intensity hematopoietic stem-cell transplantation across human leukocyte antigen barriers in a patient with congenital amegakaryocytic thrombocytopenia and monosomy 7. *Pediatric Blood and Cancer*, **2005**, 45, 212-6	3	19
20	Clinic, pathogenic mechanisms and drug testing of two inherited thrombocytopenias, ANKRD26-related Thrombocytopenia and MYH9-related diseases. *European Journal of Medical Genetics*, **2018**, 61, 715-722	2.6	15
19	Congenital amegakaryocytic thrombocytopenia (CAMT) presenting as severe pancytopenia in the first month of life. *Pediatric Blood and Cancer*, **2013**, 60, E94-6	3	13
18	Unrelated cord blood transplantation in children with severe congenital neutropenia. *Pediatric Transplantation*, **2009**, 13, 777-81	1.8	8
17	Eponym. Kostmann disease. *European Journal of Pediatrics*, **2010**, 169, 657-60	4.1	8
16	Kostmann disease with developmental delay in three patients. *European Journal of Pediatrics*, **2010**, 169, 759-62	4.1	8
15	RAS and CSF3R mutations in severe congenital neutropenia. *Blood*, **2009**, 114, 3504-5	2.2	6
14	Flow cytometric detection of MPL (CD110) as a diagnostic tool for differentiation of congenital thrombocytopenias. *Haematologica*, **2015**, 100, e341-4	6.6	5
13	A novel G6PC3 gene mutation in severe congenital neutropenia: pancytopenia and variable bone marrow phenotype can also be part of this syndrome. *European Journal of Haematology*, **2015**, 94, 79-82	3.8	4
12	CAMT-MPL: congenital amegakaryocytic thrombocytopenia caused by MPL mutations - heterogeneity of a monogenic disorder - a comprehensive analysis of 56 patients. *Haematologica*, **2021**, 106, 2439-2448	6.6	4
11	Acute lymphoblastic leukemia following severe congenital neutropenia or de novo ALL?. *Leukemia Research*, **2009**, 33, e139-42	2.7	2

10	TNF and lymphotoxin-alpha polymorphisms in patients with severe chronic neutropenia. *International Journal of Hematology*, **2001**, 74, 477-8	2.3	2
9	RAS and G-CSF Receptor Mutations Are Mutually Exclusive in Leukemogenesis in Severe Congenital Neutropenia.. *Blood*, **2005**, 106, 3073-3073	2.2	2
8	Defective Expression of Neutrophil Serine Proteases in Myeloid Progenitors of Congenital Neutropenia Patients Carrying Either ELA2 or HAX1 Mutations.. *Blood*, **2007**, 110, 3299-3299	2.2	2
7	In Vivo Growth Advantage of Cells Expressing Acquired CSF3R Mutations in Patients with Severe Congenital Neutropenia.. *Blood*, **2007**, 110, 3296-3296	2.2	1
6	GATA1 Mutations in Transient Leukemia and Myeloid Leukemia in Down Syndrome.. *Blood*, **2008**, 112, 923-923	2.2	1
5	Congenital amegakaryocytic thrombocytopenia - Not a single disease. *Best Practice and Research in Clinical Haematology*, **2021**, 34, 101286	4.2	1
4	Mutations in the Gene of Neutrophil Elastase Associated with Congenital Neutropenia Lead to Alteration of Intracellular Trafficking and Proteolytic Function of NE Protein.. *Blood*, **2004**, 104, 780-780	2.2	
3	c-mpl Mutations in Congenital Amegakaryocytic Thrombocytopenia: Residual TPO Receptor Activity in Patients of Group CAMT II.. *Blood*, **2005**, 106, 2157-2157	2.2	
2	Serial Analysis of Hematopoietic Progenitors in Mpl Mice.. *Blood*, **2007**, 110, 2100-2100	2.2	
1	A Novel Clinical Syndrome Associating Severe Congenital Neutropenia and Complex Developmental Aberrations Caused by Deficiency of G6PC3. *Blood*, **2008**, 112, 5-5	2.2	