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Research interests

Neonatal haematology
Regulation of haemoglobin F
Pathophysiology and treatment of sickle cell disease
Primary haemostasis in neonates and children
Megakaryocyte development
Pathophysiology of immune thrombocytopenia. Assessment of bleeding risk in neonates and children.

Current projects

Platelet apoptosis in ITP is ameliorated by IVIg
(Sabine Kroiss, Oliver Speer, Jeannine Winkler, Annie Bang, Margaret L. Rand, Markus Schmugge)
Aspirin resistance in children after cardiac catheterisation
(Markus Schmugge, Oliver Speer, Walter Knirsch, Oliver Kretschmar, Marlis Schmid, Manuela Albisetti)
Thrombin-generation and thrombelastography in children with normal and abnormal primary and secondary haemostasis
(Oliver Speer, Manuela Albisetti, Sabine Kroiss, Margaret L. Rand, Markus Schmugge)
The Swiss sickle cell registry
(Hulya Ozsahin, Gabriele Martin, Karin Zurbriggen, Oliver Speer, Manuela Albisetti, Markus Schmugge)

Publications

Peer reviewed articles

Ruan J, **Schmugge M**, Clemetson K, Cazes E, Bourre F, Nurden AT.
Homozygous CYS542 → ARG mutation of the GPIIb gene in a Swiss patient with type I Glanzmann's thrombasthenia.
Br J Haematol 1999; 105: 523-531. (**Impact factor: 3.2**)

Schmugge M, Plüss HJ, Boltshauser E, Niggli FK.
Long term follow-up and residual sequelae after treatment for intracerebral germ cell tumour in children and adolescents.
Ann Oncol 2000; 11: 527-533. (*Impact factor: 4.34*)

Politopoulou G, Seebach JD, **Schmugge M**, Schwarz HP, Aguzzi A.
Age-related expression of the cellular prion protein in human peripheral blood leukocytes.
Haematologica 2000; 85: 580-587. (*Impact factor: 4.19*)

Neuhaus T, Goetschel P, **Schmugge M**, Leumann E.
Heparin-induced thrombocytopenia type II on hemodialysis: switch to danaparoid.
Pediatr Nephrol 2000; 14: 713-716. (*Impact factor: 1.44*)

Schmugge M, Frischknecht H, Yonekawa Y, Baumgartner RW, Boltshauser E, Humbert J.
Stroke in hemoglobin SD disease with moyamoya: Successful hydroxyurea treatment after cerebro-vascular bypass surgery.
Blood 2001; 97: 2165-2167. (*Impact factor: 9.78*)

Schmugge M, Risch L, Benn A, Huber AR, Fischer JE.
Heparin-induced thrombocytopenia associated thrombosis in pediatric intensive care patients.
Pediatrics 2002; 109: e10. (*Impact factor: 3.9*)

Risch L, Fischer JE, **Schmugge M**, Huber AR.
Association of anti-heparin platelet factor 4 antibody levels and thrombosis in pediatric intensive care patients without thrombocytopenia.
Blood Coag Fibrinolysis 2003; 14: 113-116. (*Impact factor: 1.51*)

Revel-Vilk S, **Schmugge M**, Carcao M, Blanchette P, Rand ML, Blanchette VS.
Desmopressin responsiveness in children with von Willebrand disease.
J Pediatr Hematol Oncol 2003; 25: 874-879. (*Impact factor: 1.16*)

Schmugge M, Rand ML, Bang A, Mody M, Dunn MS, Amankwah KS, Blanchette VS, Freedman J.
The relationship of von Willebrand factor binding to activated platelets from healthy neonates and adults.
Pediatr Res 2003; 54: 474-479. (*Impact factor: 2.88*)

Schmugge M, Revel-Vilk S, Hiraki L, Rand ML, Blanchette VS, Silverman ED.
Thrombocytopenia and thromboembolism in pediatric systemic lupus erythematosus.
J Pediatr 2003; 143: 666-669. (*Impact factor: 3.12*)

Schmugge M, Dunn MS, Amankwah KS, Blanchette VS, Freedman J, Rand ML.
The activity of the von Willebrand factor cleaving protease ADAMTS-13 in newborn infants.
J Thromb Haemost 2004; 2: 228-233. (*Impact factor: 4.83*)

Nurden AT, Breillat C, Bourre F, Lillicrap D, Wang H, Blanchette VS, **Schmugge M**, Rand ML.
Triple Heterozygosity in the alphaIIb gene of a patient with Glanzmann thrombasthenia and platelets with a low alphaIIb beta3 content.
J Thromb Haemost 2004; 2: 813-819. (*Impact factor: 4.83*)

Kellenberger CJ, **Schmugge M**, Saurenmann T, Eber S, Willi UV, Babyn PS.
Radiographic and MRI Features of Deferiprone (L1) Related Arthropathy of the Knees in Patients with beta-Thalassemia.
AJR 2004; 183: 989-994. (*Impact factor: 2.38*)

Zurbriggen K, **Schmugge M**, Schmid M, Durka S, Kleinert P, Kuster T, Heizmann CW, Troxler H.
Analysis of Minor Hemoglobins with MALDI-TOF Mass Spectrometry.
Clinical Chemistry 2005; 51: 989-996. (*Impact factor: 6.51*)

Frischknecht H, Kiewitz R, **Schmugge M**.

A 4 base pair TGAT insertion at codon 116 of the beta globin gene causes β^0 – Thalassaemia. *Haematologica* 2005; 90: Suppl: ECR20. (**Impact factor: 4.19**)

Albisetti M, **Schmugge M**, Haas R, Eckhardt BP, Bauersfeld U, Baenziger O, Hug MI.

Arterial thromboembolic complications in critically ill children.

J Crit Care 2005; 20: 296-300. (**Impact factor: 2.1**)

***Schmugge M**, Bang A, Blanchette VS, Albisetti M, Connolly BL, Freedman J, Rand ML.
von Willebrand factor binding to platelets and platelet activation in newborn infants with central venous lines.

Acta Haematol, 2007;117:145-148. (**Impact factor: 1.4**)

Albisetti M, Moeller A, Waldvogel K, Bernet-Buettiker V, Cannizzaro V, Anagnostopoulos A, Balmer C, **Schmugge M**.

Congenital prothrombotic disorders in children with peripheral venous and arterial thrombosis.

Acta Haematol, 2007;117:149-155. (**Impact factor: 1.4**)

Cannizzaro E, Albisetti M, Wohlrab G, **Schmugge M**.

Severe Bleeding Complications during Antiepileptic Treatment with Valproic Acid in Children. *Neuropediatrics* 2007;38:42-5. (**Impact factor: 1.4**)

Bontadelli J, Moeller A, **Schmugge M**, Schraner T, Kretschmar O, Bauersfeld U, Bernet-Buettiker V, Albisetti M.

Enoxaparin therapy for arterial thrombosis in infants with congenital heart disease.

Intensive Care Med. 2007;33:1978-1984. (**Impact factor: 4.4**).

Kleinert P, Schmid M, Zurbriggen K, Speer O, **Schmugge M**, Roschitzki B, Durka SS, Leopold U, Kuster T, Heizmann CW, Frischknecht H, Troxler H. Mass Spectrometry: A Tool for Enhanced Detection of Hemoglobin Variants. *Clin Chem*. 2007; 54: 69-76.

(**Impact factor: 5.5**).

Schmugge M, Waye, JS, Basran RK, Zurbriggen K, Frischknecht H.

Hb S/ β^+ -thalassemia phenotype demonstrates that the IVS-I (-2) A→C mutation is a mild β - thalassaemia allele. *Hemoglobin* 2008;32:303-7.

Hochuli H, **Zurbriggen K**, Schmid M, Speer O, Rochat P, Frauchiger B, Kleinert P,

Schmugge M, Troxler H. A New-Globin Variant with increased oxygen affinity in a swiss family: Hb Frauenfeld [a2 138(H21)Ser→Phe]. *Hemoglobin* 2009;33:54-8.

Zurbriggen K, Schmid M, **Schmugge M**, Troxler H, Speer O

HbAlperton [b135(H13)Ala →Val] shows decreased oxygen affinity

Hemoglobin 2009 (in press)

Ravel Vilik S, Blanchette VS, **Schmugge M**, Clark D, Lillicrap D, Rand ML. In vitro and in vivo stability of diluted recombinant factor VIII for continuous infusion use in haemophilia A,. *Hemophilia* 2009 (in press).

Speer O, Kroiss S, Winkler J, Azzouzi I, Staubli G, Bang A, Rand M, **Schmugge M**

Platelet apoptosis in paediatric primary immune thrombocytopenia
in preparation

Azzouzi I, Winkler J, Fauchere JC, **Schmugge M**, Speer O

Fetal Hemoglobin is post-transcriptionally regulated by microRNAs
in preparation

Reviews

Schmugge M, Rand ML, Freedman J.
Platelets and von Willebrand Factor.
Transf Aph Science 2003; 28: 269-277. (**Impact factor: 0.98**)

Risch L, Huber AR, **Schmugge M**.
Diagnosis and treatment of heparin-induced thrombocytopenia in neonates and children.
Thromb Res 2006; 118: 123-135. (**Impact factor: 1.54**)

Betts D, **Schmugge M**. Shwachman syndrome.
Atlas Genet Cytogenet Oncol Haematol. November 2004.
URL : <http://atlasgeneticsoncology.org/Kprones/ShwachmanID10058.html>
(**Impact factor: not rated**)

Schmugge M.
Die Beta Thalassaemie, Pathophysiologie, Klinik und Therapie.
Leading Opinions Hämatologie & Onkologie 2006; 2: 5-6. (**Impactfactor: not rated**)

Bechtold A, Kalb R, Neveling K, Friedl R, Gottwald B, Herterich S, **Schmugge-Liner M**,
Heilmann C, Hanenberg H, Schindler D.
Prenatal Diagnosis of Fanconi Anemia: Functional and Molecular Testing. In: Fanconi
Anemia A Paradigmatic Disease for the Understanding of Cancer and Aging.
Monographs in Human Genetics; Vol 15 Editors: Schindler, D., Hoehn, H. Karger Basel
2007. (**Impact factor: not rated**)

Schmugge M, Speer O, Hulya Ozsahin A, Martin G (2008)
Die Sichelzellanämie in der Schweiz. Teil 1: Pathophysiologie, Klinik.
Schweiz Med Forum.;8:582-586

Schmugge M, Speer O, Ozsahin AH, Martin G (2008)
Die Sichelzellerkrankung in der Schweiz. Teil 2: Therapeutische und prophylaktische
Massnahmen. *Schweiz Med Forum.* 8:606-608

Case Reports

Hany M, Betts DR, **Schmugge M**, Plüss HJ, Niggli F.
Childhood fibrolamellar hepatocellular carcinoma with increased aromatase activity and a near
triploid karyotype.
Med Pediatr Oncol 1997; 28: 136-138. (**Impact factor: 1.36**)

Schmugge M, Toelle S, Marbet GA, Laroche P, Meili E.
Gingival bleeding, epistaxis, and diffuse hematoma three days after gastroenteritis: The
haemorrhagic lupus anticoagulant syndrome.
Eur J Pediatr 2001; 160: 43-46. (**Impact factor: 1.37**)

Research Correspondence

Schmugge M, Lauener R, Bossart W, Seger R, Guengoer T.
Chronic enteroviral meningoencephalitis in X-linked agammaglobulinemia: unusual
course and favorable response to anti-enteroviral treatment.
Eur. J. Pediatr 1999; 158: 1010-1011.