

BIOGRAPHICAL SKETCH

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| NAME Tanner, Stephan M. | | POSITION TITLE Research Assistant Professor | |
|--|---------------------------|---|-----------------------|
| eRA COMMONS USER NAME tanner74 | | | |
| EDUCATION/TRAINING (Begin with baccalaureate or other initial professional education, such as nursing, and include postdoctoral training.) | | | |
| INSTITUTION AND LOCATION | DEGREE (if applicable) | YEAR(s) | FIELD OF STUDY |
| Freies Gymnasium Berne, Switzerland | Matura | 1989 | Baccalaureate |
| University of Berne, Switzerland | Undergraduate | 1990-1993 | Biology |
| University of Berne, Switzerland | Lic.Phil.Nat. | 1993-1995 | Molecular Ecology |
| University of Berne, Switzerland | Ph.D. | 1995-1998 | Human Genetics |
| The Ohio State University, Columbus, OH | Post. Doc. | 1998-2002 | Human Cancer Genetics |

A Positions and Honors.**updated: January 1, 2009****Positions and Employment:**

- 1993-1995 Biology diploma thesis (Lic.Phil.Nat.), Institute of Cell Biology, University of Berne, Switzerland
Advisor: Daniel Schümperli, PhD, DVM
- 1995-1998 Graduate Student, Department of Clinical Research, Children's Hospital, University of Berne, Switzerland
Co-advisors: Sabina Gallati, PhD and Daniel Schümperli, PhD, DVM
- 1998-2002 Postdoctoral Fellow, Human Cancer Genetics Program, OSUCCC, Columbus, OH
Mentor: Albert de la Chapelle, MD, PhD
- 2002-2005 Research Scientist, Human Cancer Genetics Program, OSUCCC, Columbus, OH
- 2005-present Research Assistant Professor, Human Cancer Genetics Program, OSUCCC, Columbus, OH

Honors and awards:

- 1998 Ph.D. summa cum laude, Faculty of Science, University of Berne, Switzerland.
- 1998 Recipient of Swiss National Science Foundation junior postdoctoral fellowship No. 81BE-53485 from April, 1, 1998 to March 31, 1999.
- 1999 Faculty prize in Biology awarded for best PhD thesis presented in 1998, Faculty of Science, University of Berne, Switzerland.
- 2000 Recipient of Swiss Foundation for Medical-Biological Grants (SSMBS) advanced postdoctoral fellowship No. 965/2336 from July, 1, 2000 to June 30, 2001.

Other Experience:

- 1999-present Reviewer for *Neuromuscular Disorders*, *Clinical Genetics*, *Journal of Medical Genetics*, *Human Genetics*, *Cancer Research*, *Nature Genetics*, *Journal of Experimental Medicine*

B Selected peer-reviewed publications (32/32) #=first author contribution.

- Laporte J, Guiraud-Chaumeil C, Vincent MC, Mandel JL, **Tanner SM#**, Liechti-Gallati S, Wallgren-Pettersson C, Dahl N, Kress W, Bolhuis PA, Fardeau M, Samson F, Bertini E: Mutations in the MTM1 gene implicated in X-linked myotubular myopathy. ENMC International Consortium on Myotubular Myopathy. European Neuro-Muscular Center. *Hum Mol Genet* 1997, 6(9):1505-1511.
- Tanner SM**, Laporte J, Guiraud-Chaumeil C, Liechti-Gallati S: Confirmation of prenatal diagnosis results of X-linked recessive myotubular myopathy by mutational screening, and description of three new mutations in the MTM1 gene. *Hum Mutat* 1998, 11(1):62-68.
- Laporte J, Guiraud-Chaumeil C, **Tanner SM**, Blondeau F, Hu LJ, Vicaire S, Liechti-Gallati S, Mandel JL: Genomic organization of the MTM1 gene implicated in X-linked myotubular myopathy. *Eur J Hum Genet* 1998, 6(4):325-330.
- Tanner SM**, Schneider V, Thomas NS, Clarke A, Lazarou L, Liechti-Gallati S: Characterization of 34 novel and six known MTM1 gene mutations in 47 unrelated X-linked myotubular myopathy patients. *Neuromuscul Disord* 1999, 9(1):41-49.

5. **Tanner SM**, Orstavik KH, Kristiansen M, Lev D, Lerman-Sagie T, Sadeh M, Liechti-Gallati S: Skewed X-inactivation in a manifesting carrier of X-linked myotubular myopathy and in her non-manifesting carrier mother. *Hum Genet* 1999, 104(3):249-253.
6. Laporte J, Biancalana V, **Tanner SM**, Kress W, Schneider V, Wallgren-Pettersson C, Herger F, Buj-Bello A, Blondeau F, Liechti-Gallati S, Mandel JL: MTM1 mutations in X-linked myotubular myopathy. *Hum Mutat* 2000, 15(5):393-409.
7. Frühwald MC, O'Dorisio MS, Dai Z, **Tanner SM**, Balster DA, Gao X, Wright FA, Plass C: Aberrant promoter methylation of previously unidentified target genes is a common abnormality in medulloblastomas-implications for tumor biology and potential clinical utility. *Oncogene* 2001, 20(36):5033-5042.
8. Virtaneva K, Wright FA, **Tanner SM**, Yuan B, Lemon WJ, Caligiuri MA, Bloomfield CD, de La Chapelle A, Krahe R: Expression profiling reveals fundamental biological differences in acute myeloid leukemia with isolated trisomy 8 and normal cytogenetics. *Proc Natl Acad Sci U S A* 2001, 98(3):1124-1129.
9. **Tanner SM**, Austin JL, Leone G, Rush LJ, Plass C, Heinonen K, Mrozek K, Sill H, Knuutila S, Kolitz JE, Archer KJ, Caligiuri MA, Bloomfield CD, de la Chapelle A: BAALC, the human member of a novel mammalian neuroectoderm gene lineage, is implicated in hematopoiesis and acute leukemia. *Proc Natl Acad Sci U S A* 2001, 98(24):13901-13906.
10. Kristiansen M, Knudsen GP, **Tanner SM**, McEntagart M, Jungbluth H, Muntoni F, Sewry C, Gallati S, Orstavik KH, Wallgren-Pettersson C: X-inactivation patterns in carriers of X-linked myotubular myopathy. *Neuromuscul Disord* 2003, 13(6):468-471.
11. **Tanner SM**, Aminoff M, Wright FA, Liyanarachchi S, Kuronen M, Saarinen A, Massika O, Mandel H, Broch H, de la Chapelle A: Amnionless, essential for mouse gastrulation, is mutated in recessive hereditary megaloblastic anemia. *Nat Genet* 2003, 33(3):426-429.
12. Baldus CD, **Tanner SM**, Kusewitt DF, Liyanarachchi S, Choi C, Caligiuri MA, Bloomfield CD, de la Chapelle A: BAALC, a novel marker of human hematopoietic progenitor cells. *Exp Hematol* 2003, 31(11):1051-1056.
13. Mrozek K, **Tanner SM**, Heinonen K, Bloomfield CD: Molecular cytogenetic characterization of the KG-1 and KG-1a acute myeloid leukemia cell lines by use of spectral karyotyping and fluorescence in situ hybridization. *Genes Chromosomes Cancer* 2003, 38(3):249-252.
14. Baldus CD, **Tanner SM**, Ruppert AS, Whitman SP, Archer KJ, Marcucci G, Caligiuri MA, Carroll AJ, Vardiman JW, Powell BL, Allen SL, Moore JO, Larson RA, Kolitz JE, de la Chapelle A, Bloomfield CD: BAALC expression predicts clinical outcome of de novo acute myeloid leukemia patients with normal cytogenetics: a Cancer and Leukemia Group B Study. *Blood* 2003, 102(5):1613-1618.
15. **Tanner SM**, Li Z, Bisson R, Acar C, Oner C, Oner R, Cetin M, Abdelaal MA, Ismail EA, Lissens W, Krahe R, Broch H, Grasbeck R, de la Chapelle A: Genetically heterogeneous selective intestinal malabsorption of vitamin B12: founder effects, consanguinity, and high clinical awareness explain aggregations in Scandinavia and the Middle East. *Hum Mutat* 2004, 23(4):327-333.
16. Fyfe JC, Madsen M, Hojrup P, Christensen EI, **Tanner SM**, de la Chapelle A, He Q, Moestrup SK: The functional cobalamin (vitamin B12)-intrinsic factor receptor is a novel complex of cubilin and amnionless. *Blood* 2004, 103(5):1573-1579.
17. Baldus CD, Liyanarachchi S, Mrozek K, Auer H, **Tanner SM**, Guimond M, Ruppert AS, Mohamed N, Davuluri RV, Caligiuri MA, Bloomfield CD, de la Chapelle A: Acute myeloid leukemia with complex karyotypes and abnormal chromosome 21: Amplification discloses overexpression of APP, ETS2, and ERG genes. *Proc Natl Acad Sci U S A* 2004, 101(11):3915-3920.
18. Satoskar AA, **Tanner SM**, Weinstein M, Qualman SJ, de la Chapelle A: Baalc, a marker of mesoderm and muscle. *Gene Expr Patterns* 2005, 5(4):463-473.
19. He Q, Madsen M, Kilkenney A, Gregory B, Christensen EI, Vorum H, Hojrup P, Schaffer AA, Kirkness EF, **Tanner SM**, de la Chapelle A, Giger U, Moestrup SK, Fyfe JC: Amnionless function is required for cubilin brush-border expression and intrinsic factor-cobalamin (vitamin B12) absorption in vivo. *Blood* 2005, 106(4):1447-1453.
20. Al-Alami JR, **Tanner SM**, Tayeh MK, de la Chapelle A, El-Shanti H: Homozygous AMN mutation in hereditary selective intestinal malabsorption of vitamin B12 in Jordan. *Saudi Med J* 2005, 26(7):1061-1064.
21. **Tanner SM**, Li Z, Perko JD, Oner C, Cetin M, Altay C, Yurtsever Z, David KL, Faivre L, Ismail EA, Grasbeck R, de la Chapelle A: Hereditary juvenile cobalamin deficiency caused by mutations in the intrinsic factor gene. *Proc Natl Acad Sci U S A* 2005, 102(11):4130-4133.

22. Grogan PM, **Tanner SM**, Orstavik KH, Knudsen GP, Saperstein DS, Vogel H, Barohn RJ, Herbelin LL, McVey AL, Katz JS: Myopathy with skeletal asymmetry and hemidiaphragm elevation is caused by myotubularin mutations. *Neurology* 2005, 64(9):1638-1640.
23. Zervos EE, **Tanner SM**, Osborne DA, Bloomston M, Rosemurgy AS, Ellison EC, Melvin WS, de la Chapelle A: Differential Gene Expression in Patients Genetically Predisposed to Pancreatic Cancer. *J Surg Res* 2006, 135(2):317-322.
24. Broides A, Yerushalmi B, Levy R, Hadad N, Kaplun N, **Tanner SM**, de la Chapelle A, Levy J: Imerslund-Gräsbeck syndrome associated with recurrent aphthous stomatitis and defective neutrophil function. *J Pediatr Hematol Oncol* 2006, 28(11):715-719.
25. Raval A, **Tanner SM**, Byrd JC, Angerman EB, Perko JD, Chen SS, Hackanson B, Grever MR, Lucas DM, Matkovic JJ, Lin TS, Kipps TJ, Murray F, Weisenburger D, Sanger W, Lynch J, Watson P, Jansen M, Yoshinaga Y, Rosenquist R, de Jong PJ, Coggill P, Beck S, Lynch H, de la Chapelle A, Plass C: Downregulation of Death-Associated Protein Kinase 1 (DAPK1) in Chronic Lymphocytic Leukemia. *Cell* 2007, 129(5):879-890.
26. Hauck FH, **Tanner SM**#, Henker J, Laass MW: Imerslund-Gräsbeck syndrome in a 15-year old German girl caused by compound heterozygous mutations in CUBN. *Eur J Pediatrics* 2008, 167(6):671-675; Epub Aug 1, 2007.
27. Plass C, Byrd JC, Raval A, **Tanner SM**, de la Chapelle A: Molecular profiling of chronic lymphocytic leukaemia: genetics meets epigenetics to identify predisposing genes. Review *Br J Haematol* 2007, 139(5):744-752.
28. **Tanner SM** & Byrd JC: Advances in genome-wide CLL linkage. Invited commentary. *Blood* 2007, 110(9):3094-3095.
29. Luder AS, **Tanner SM**, de la Chapelle A, Walter JH: Amnionless (AMN) mutations in Imerslund-Gräsbeck syndrome may be associated with disturbed vitamin B₁₂ transport into the CNS. *J Inherit Metab Dis* 2008, Epub Jan 7.
30. Trimboli AJ, Fukino K, de Bruin A, Wei G, Shen L, **Tanner SM**, Creasap N, Rosol TJ, Robinson ML, Eng C, Ostrowski MC, Leone G: Direct evidence for epithelial-mesenchymal transitions in breast cancer. *Cancer Res* 2008, 68(3):937-945.
31. Valle L, Serena-Acedo T, Liyanarachchi S, Hampel H, Comeras I, Li Z, Zeng Q, Zhang H-T, Pennison MJ, Sadim M, Pasche B, **Tanner SM**^, and de la Chapelle A. Germline Allele-specific Expression of TGFBR1 Confers an Increased Risk of Colorectal Cancer. *Science* 2008, 321(5894):1361-1365; Epub August 14, (**senior co-author**)
32. Ament AE, Li Z, Sturm AC, Perko JD, Lawson S, Masterson M, Quadros EV, and **Tanner SM**. Juvenile cobalamin deficiency in individuals of African ancestry is caused by a founder mutation in the intrinsic factor gene GIF. *Br J Haematol*, Epub November 19, 2008.

C Research Support.

Past:

NIH/NCI R01 CA098933 07/01/04-06/30/08

Title: *BAALC* in neurogenesis and hematopoiesis

PI: Albert de la Chapelle

Co-I: Stephan M. Tanner

To specifically determine *BAALC*'s role in neurogenesis and hematopoiesis using conventional and conditional, tissue-specific *Baalc* knock-out mice.

CLL Global Research Foundation 01/01/08-12/31/08

Title: Genetic and epigenetic pathways in CLL

PI: Stephan M. Tanner

To expose *miR16-1* germline mutations and clarify their impact on *DAPK1* regulation, and ultimately explain how they predispose to development of CLL.

ACS Institutional Research Grant 67-003-44 01/01/08-12/31/08

Title: Isolation of monoclonal B-cell lymphocytosis (MBL) cells by fluorescence activated cell sorting

PI: Stephan M. Tanner

To identify monoclonal B-cell lymphocytosis cases for *DAPK1* allele specific expression analysis.

Other accomplishments.

- 1) US Patent application #20030119043 for "*BAALC* expression as a diagnostic marker for acute leukemia".
- 2) US Provisional Patent application #61/088,080 "*Constitutionally altered allele-specific expression and haplotypes of the TGFBR1 gene aid in determining individual constitutional (genetic) risk of colorectal cancer.*"