



## Publikationen Prof. Schweiger

Institut für Humangenetik / Zentrum für Seltene Erkrankungen

### Originalarbeiten:

1. Stroh A, **Schweiger S**, Ramirez JM, Tüscher O (2024) The selfish network: how the brain preserves behavioral function through shifts in neuronal network state (2024) Trends Neurosci Apr;47(4):246-258. doi: 10.1016/j.tins.2024.02.005. Epub 2024 Mar 13.
2. Frank S\*, Gabessi E\*, Käseberg S, Bertin M, Zografidou L, Pfeiffer D, Brennenstuhl H, Falk S\*, Karow M\*, **Schweiger S\*** (2024) Absence of the RING domain in MID1 results in patterning defects in the developing human brain. *LSA*: <http://doi.org/10.26508/lsa.202302288>
3. Käseberg S\*, Bertin M\*, Menon R\*, Gabassi E\*, Todorov H\*, Frank S, Brennenstuhl H, Lohrer B, Winter J, Krummeich J, Winkler J, Winner B, Weis E, Hartwich D, Diederich S, Luck K, Gerber S, Lunt P, Berninger B, Falk S\*, **Schweiger S\***, Karow M\* (2023). Dynamic X-chromosomal reactivation enhances female brain resilience. *bioRxiv* doi: <https://doi.org/10.1101/2023.06.17.545424>
4. Hebestreit H, Lapstich AM, Brandstetter L, Krauth C, Deckert J, Haas K, Pfister L, Witt S, Schippers C, Dieris-Hirche J, Maisch T, Tüscher O, Bârlescu L, Berger A, Berneburg M, Britz V, Deibele A, Graeßner H, Gündel H, Heuft G, Lücke T, Mundlos C, Quitmann J, Rutsch F, Schubert K, Schulz JB, **Schweiger S**, Zeidler C, Zeltner L, de Zwaan M; ZSE-DUO Working Group. (2023) Effect of the addition of a mental health specialist for evaluation of undiagnosed patients in centres for rare diseases (ZSE-DUO): a prospective, controlled trial with a two-phase cohort design. *EClinicalMedicine*. Oct 6;65:102260.
5. Hoffmann EM, Aghayeva F, Schuster AK, Pfeiffer N, Karsten M, **Schweiger S**, Pirlich N, Wagner FM, Chronopoulos P, Grehn F. (2022) Results of childhood glaucoma surgery over a long-term period. *Acta Ophthalmol*. 100(2):e448-e454. doi: 10.1111/aos.14985.
6. O'Leary A, Fernández-Castillo N, Gan G, Yang Y, Yotova AY, Kranz TM, Grünewald L, Freudenberg F, Antón-Galindo E, Cabana-Domínguez J, Harneit A, Schweiger JI, Schwarz K, Ma R, Chen J, Schwarz E, Rietschel M, Tost H, Meyer-Lindenberg A, Pané-Farré CA, Kircher T, Hamm AO, Burguera D, Mota NR, Franke B, **Schweiger S**, Winter J, Heinz A, Erk S, Romanczuk-Seiferth N, Walter H, Ströhle A, Fehm L, Fydrich T, Lueken U, Weber H, Lang T, Gerlach AL, Nöthen MM, Alpers GW, Arolt V, Witt S, Richter J, Straube B, Cormand B, Slattery DA, Reif A. (2022) Behavioural

- and functional evidence revealing the role of RBFOX1 variation in multiple psychiatric disorders and traits. *Mol Psychiatry*. 27(11):4464-4473. doi: 10.1038/s41380-022-01722-4.
7. Hebestreit H, Zeidler C, Schippers C, de Zwaan M, Deckert J, Heuschmann P, Krauth C, Bullinger M, Berger A, Berneburg M, Brandstetter L, Deibele A, Dieris-Hirche J, Graessner H, Gündel H, Herpertz S, Heuft G, Lapstich AM, Lücke T, Maisch T, Mundlos C, Petermann-Meyer A, Müller S, Ott S, Pfister L, Quitmann J, Romanos M, Rutsch F, Schaubert K, Schubert K, Schulz JB, **Schweiger S**, Tüscher O, Ungethüm K, Wagner TOF, Haas K; ZSE-DUO working group. (2022) Dual guidance structure for evaluation of patients with unclear diagnosis in centers for rare diseases (ZSE-DUO): study protocol for a controlled multi-center cohort study. *Orphanet J Rare Dis*. 17(1):47. doi: 10.1186/s13023-022-02176-1.
  8. Julia V. Stingl,<sup>†</sup> Stefan Diederich,<sup>†</sup> Heidi Diel, Alexander K. Schuster, Felix M. Wagner, Panagiotis Chronopoulos, Fidan Aghayeva, Franz Grehn, Jennifer Winter, **Susann Schweiger**,<sup>‡</sup> and Esther M. Hoffmann<sup>‡</sup> (2022) First Results from the Prospective German Registry for Childhood Glaucoma: Phenotype–Genotype Association. *J Clin Med*. 11(1): 16.
  9. Weißbach S., Sys S., Hewel C., Todorov H., **Schweiger S.**, Winter J., Pfenninger M., Torkamani A., Evans D., Burger J., Everschor-Sitte K., May-Simera H.L., Gerber S. (2021) Reliability of genomic variants across different next-generation sequencing platforms and bioinformatic processing pipelines. *BMC Genomics*. 22(1):62. doi: 10.1186/s12864-020-07362-8.
  10. Gomez R., Hafezi N., Amrani M., **Schweiger S.**, Dewenter M.K., Thomas P., Lieb C., Hasenburg A., Skala C. (2021) Genetic findings in miscarriages and their relation to the number of previous miscarriages. *Arch Gynecol Obstet*. 303(6):1425-1432. doi: 10.1007/s00404-020-05859-x
  11. Cooper A., Butto T., Hammer N., Jagannath S., Fend-Guella D.L., Akhtar J., Radyushkin K., Lesage F., Winter J., Strand S., Roeper J., Zechner U., **Schweiger S.** (2020) Inhibition of histone deacetylation rescues phenotype in a mouse model of Birk-Barel Intellectual Disability syndrome. *Nature communications* 11(1):480. doi: 10.1038/s41467-019-13918-4
  12. Michels S, Buchholz HG, Rosar F, Heinrich I, Hoffmann MA, **Schweiger S**, Tüscher O, Schreckenberger M. (2019) 18F-FDG PET/CT: an unexpected case of Huntington's disease. *BMC Neurol*. 19(1):78. doi: 10.1186/s12883-019-1311-9
  13. Kahrizi K, Huber M, Galetzka D, Dewi S, Schröder J, Weis E, Kariminejad A, Fattahi Z, Ropers HH, **Schweiger S**, Najmabadi H, Winter J. (2019)

14. Homozygous variants in the gene SCAPER cause syndromic intellectual disability. *Am J Med Genet A* May 9. doi: 10.1002/ajmg.a.61172. [Epub ahead of print]
15. Gucev Z, Tasic V, Bogevska I, Laban N, Saveski A, Polenakovic M, Plaseska-Karanfilska D, Komlosi K, Winter J, **Schweiger S**, Nishimura G, Spranger J, Bartsch O. (2019) Heterotopic ossifications and Charcot joints: Congenital insensitivity to pain with anhidrosis (CIPA) and a novel NTRK1 gene mutation. *Eur J Med Genet*. 2019 Jan 21. pii: S1769-7212(18)30561-5. doi: 10.1016/j.ejmg.2019.01.003. [Epub ahead of print]
16. Arnoux I<sup>#</sup>, Willam M<sup>#</sup>, Griesche N<sup>#</sup>, Krummeich J, Watari H, Offermann N, Weber S, Narayan Dey P, Chen C, Monteiro O, Buettner S, Meyer K, Bano D, Radyushkin K, Langston R, Lambert JJ, Wanker E, Methner A, Krauss S\*, **Schweiger S\***, Stroh A\*. (2018) Metformin reverses early cortical network dysfunction and behavior changes in Huntington's disease. *Elife*. 2018 Sep 4;7. pii: e38744. doi: 10.7554/eLife.38744.
17. Snijders Blok L, Rousseau J, Twist J, Ehresmann S, Takaku M, Venselaar H, Rodan LH, Nowak CB, Douglas J, Swoboda KJ, Steeves MA, Sahai I, Stumpel CTRM, Stegmann APA, Wheeler P, Willing M, Fiala E, Kochhar A, Gibson WT, Cohen ASA, Agbahovbe R, Innes AM, Au PYB, Rankin J, Anderson IJ, Skinner SA, Louie RJ, Warren HE, Afenjar A, Keren B, Nava C, Buratti J, Isapof A, Rodriguez D, Lewandowski R, Propst J, van Essen T, Choi M, Lee S, Chae JH, Price S, Schnur RE, Douglas G, Wentzensen IM, Zweier C, Reis A, Bialer MG, Moore C, Koopmans M, Brilstra EH, Monroe GR, van Gassen KLI, van Binsbergen E, Newbury-Ecob R, Bownass L, Bader I, Mayr JA, Wortmann SB, Jakielski KJ, Strand EA, Kloth K, Bierhals T; **DDD study**, Roberts JD, Petrovich RM, Machida S, Kurumizaka H, Lelieveld S, Pfundt R, Jansen S, Deriziotis P, Faivre L, Thevenon J, Assoum M, Shriberg L, Kleefstra T, Brunner HG, Wade PA, Fisher SE, Campeau PM. (2018) CHD3 helicase domain **mutations** cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. *Nat Commun*. 2018 Nov 5;9(1):4619. doi: 10.1038/s41467-018-06014-6.
18. Monteiro O, Chen C, Bingham R, Argyrou A, Buxton R, Pancevac Jönsson C, Jones E, Bridges A, Gatfield K, Krauß S, Lambert J, Langston R, **Schweiger S**, Uings I. (2018) Pharmacological disruption of the MID1/ $\alpha$ 4 interaction reduces mutant Huntingtin levels in primary neuronal cultures. *Neurosci Lett*. Feb 27;673:44-50. doi: 10.1016/j.neulet.2018.02.061. [Epub ahead of print]
19. Komlosi K, Diederich S, Fend-Guella DL, Bartsch O, Winter J, Zechner U, Beck M, Meyer P, **Schweiger S**. (2018) Targeted next-generation sequencing analysis in couples at increased risk for autosomal recessive disorders. *Orphanet J Rare Dis*. 2018 Jan 26;13(1):23. doi:

- 10.1186/s13023-018-0763-0.
20. Sadleir LG, Mountier EI, Gill D, Davis S, Joshi C, DeVile C, Kurian MA; **DDD Study**, Mandelstam S, Wirrell E, Nickels KC, Murali HR, Carvill G, Myers CT, Mefford HC, Scheffer IE. Not all SCN1A epileptic encephalopathies are Dravet syndrome: Early profound Thr226Met phenotype. (2017) *Neurology*. Sep 5;89(10):1035-1042.
21. **Deciphering Developmental Disorders Study**. Prevalence and architecture of de novo mutations in developmental disorders. (2017) *Nature*. Feb 23;542(7642):433-438.
22. Etzold A, Galetzka D, Weis E, Bartsch O, Haaf T, Spix C, Itzel T, **Schweiger S**, Strand D, Strand S, Zechner U. CAF-like state in primary skin fibroblasts with constitutional BRCA1 epimutation sheds new light on tumor suppressor deficiency-related changes in healthy tissue. (2016) *Epigenetics* 11(2):120-31.
23. Mircsof D, Langouët M, Rio M, Moutton S, Siquier-Pernet K, Bole-Feysot C, Cagnard N, Nitschke P, Gaspar L, Žnidarič M, Alibeu O, Fritz AK, Wolfer DP, Schröter A, Bosshard G, Rudin M, Koester C, Crestani F, Seebeck P, Boddaert N, Prescott K; **DDD Study**, Hines R, Moss SJ, Fritschy JM, Munnich A, Amiel J, Brown SA, Tyagarajan SK, Colleaux L. Mutations in NONO lead to syndromic intellectual disability and inhibitory synaptic defects. (2015) *Nat Neurosci*. 2015 Dec;18(12):1731-6.
24. **Deciphering Developmental Disorders Study**. Large-scale discovery of novel genetic causes of developmental disorders. (2015) *Nature*. Mar 12;519(7542):223-8
25. Wright CF, Fitzgerald TW, Jones WD, Clayton S, McRae JF, van Kogelenberg M, King DA, Ambridge K, Barrett DM, Bayzatinova T, Bevan AP, Bragin E, Chatzimichali EA, Gribble S, Jones P, Krishnappa N, Mason LE, Miller R, Morley KI, Parthiban V, Prigmore E, Rajan D, Sifrim A, Swaminathan GJ, Tivey AR, Middleton A, Parker M, Carter NP, Barrett JC, Hurles ME, FitzPatrick DR, Firth HV; **DDD study** Genetic diagnosis of developmental disorders in the DDD study: a scalable analysis of genome-wide research data (2015) *Lancet*. 2015 Apr 4;385(9975):1305-14. doi: 10.1016/S0140-6736(14)61705-0
26. Reuter MS<sup>1</sup>, Musante L, Hu H, Diederich S, Sticht H, Ekici AB, Uebe S, Wienker TF, Bartsch O, Zechner U, Oppitz C, Keleman K, Jamra RA, Najmabadi H, **Schweiger S**, Reis A, Kahrizi K. NDST1 missense mutations in autosomal recessive intellectual disability (2014) *Am J Med Genet A*. (2014) Nov;164(11):2753-63. doi: 10.1002/ajmg.a.36723. Epub 2014 Aug 14.
27. Krauß S, Griesche N, Jastrzebska E, Chen C, Rutschow D, Achmüller C, Dorn S, Boesch SM, Lalowski M, Wanker E, Schneider R, **Schweiger S**. Translation of HTT mRNA with expanded CAG repeats is regulated by the MID1-PP2A protein complex (2013). *Nature communications*, 4:1511 doi: 10.1038/ncomms2514.

28. Banka S, Veeramachaneni R, Reardon W, Howard E, Bunstone S, Ragge N, Parker MJ, Crow YJ, Kerr B, Kingston H, Metcalfe K, Chandler K, Magee A, Stewart F, McConnell VP, Donnelly DE, Berland S, Houge G, Morton JE, Oley C, Revencu N, Park SM, Davies SJ, Fry AE, Lynch SA, Gill H, **Schweiger S**, Lam WW, Tolmie J, Mohammed SN, Hobson E, Smith A, Blyth M, Bennett C, Vasudevan PC, García-Miñaur S, Henderson A, Goodship J, Wright MJ, Fisher R, Gibbons R, Price SM, C de Silva D, Temple IK, Collins AL, Lachlan K, Elmslie F, McEntagart M, Castle B, Clayton-Smith J, Black GC, Donnai D. How genetically heterogeneous is Kabuki syndrome?: MLL2 testing in 116 patients, review and analyses of mutation and phenotypic spectrum. (2011) *Eur. J. Hum. Genet.* 20: 381-388
29. Krauß S., So J., Hambrock M., Köhler A., Kunath M., Scharff C., Wessling M., Grzeschik K.H., Schneider R., **Schweiger S.** (2009) Point mutations in GLI3 lead to misregulation of its subcellular localization. *PLoSone* 4(10): e7471
30. Krauß S., Foerster J., Schneider R., **Schweiger S.** (2008) PP2A and rapamycin regulate the nuclear localization and activity of the transcription factor GLI3. *Cancer Research* 68(12): 4658-65
31. Aranda-Orgillés B., Winter J., Aigner J., Köhler A., Jastrzebska E., Stahl J., Müller E.C., Otto A., Wanker E.E., Schneider R., **Schweiger S.** (2008) The Opitz syndrome gene product MID1 assembles a microtubule-associated ribonucleoprotein complex. (2008) *Hum. Genet* 123(2):163-76
32. So J., Müller I., Kunath M., Herrmann S., Ullmann R., **Schweiger S.** Diagnosis of a terminal deletion of 4p with duplication of Xp22.31 in a patient with findings of Opitz G/BBB syndrome and Wolf-Hirschhorn syndrome. (2008) *Am J Med Genet A.* Jan 1;146A(1):103-9.
33. Winter J., Kunath M., Roepcke S., Krause S., Schneider R., **Schweiger S.** Alternative polyadenylation signals and promoters act in concert to control tissue-specific expression of the Opitz Syndrome gene MID1. (2007) *BMC Mol Biol.* 8:105.
34. Shoichet SA., Duprez L., Hagens O., Waetzig V., Menzel C., Herdegen T., **Schweiger S.**, Dan B., Vamos E., Ropers HH., Kalscheuer VM. (2006) Truncation of the CNS-expressed JNK3 in a patient with a severe developmental epileptic encephalopathy. *Hum. Genet.* 118: 559-567
35. Hagens O., Minina E., **Schweiger S.**, Ropers HH., Kalscheuer V. (2006) Characterization of FBX25, encoding a novel brain-expressed F-box protein. *Biochim. Biophys. Acta.* 1760: 110-118
36. Wieland I., Reardon W., Jakubicska S., Franco B., Kress W., Vincent-Delorme C., Thierry P., Edwards M., König R., Rusu C., **Schweiger S.**, Thompson E., Tinschert S., Stewart F., Wieacker P. (2005) Twenty-six novel EFNB1 mutations in familial and sporadic craniofrontonasal syndrome. *Hum. Mutat.* 26: 113-118
- 37.

38. So J., Suckow V., Kijas Z., Kalscheuer V., Moser B., Baars M., Firth H., Lunt P., Hamel B., Moraine C., Odent S., Schinzel A., van der Smagt J.J., Devriendt K., Passarge E., van der Burgt I., Cox T., Opitz J., Ropers H.H. & **Schweiger S.** (2005) Mild phenotypic spectrum in a series of Opitz BBB/G syndrome patients with MID1 mutations. *Am. J. Med. Genet.* 132A:1-7
39. Kalscheuer VM, Freude K, Musante L\*, Jensen LR\*, Yntema HG, Géczy J, Sefiani A, Hoffmann K, Moser B, Haas S, Gurok U, Haesler S, Aranda B, Nshedjan A, Tzschach A, Hartmann N, Roloff TC, Shoichet S, Hagens O, Tao J, van Bokhoven H, Turner G, Chelly J, Moraine C, Fryns JP, Nuber U, Hoeltzenbein M, Scharff C, Scherthan H, Lenzner S, Hamel BCJ, **Schweiger S** & Ropers HH (2003) Mutations in the polyglutamine-binding protein 1 gene cause X-linked mental retardation. *Nature genetics* 35 (4): 313-5
40. Zeitz C., Scherthan H., Freier S., Feil S., Suckow V., **Schweiger S.**, Berger W. (2003) NYX (nyctalopin on chromosome X), the gene mutated in congenital stationary night blindness encodes a cell surface protein. *IOVD* 44 (10): 4184-91
41. Musante L., Kehl H.G., Majewski F., Meinecke P., **Schweiger S.**, Gillissen-Kaesbach G., Wieczorek D., Hinkel G.K., Tinschert S., Hoeltzenbein M., Ropers H.H. (2003) Kalscheuer V.M. Spectrum of mutations in *PTPN11* and genotype-phenotype correlation in 96 patients with Noonan syndrome and five patients with cardio-facio-cutaneous syndrom. *EJHG* 11, 201-206
42. **Schweiger S.**, Chaoui R., Tennstedt C., Lehmann K., Mundlos S., Tinschert S. (2003) Antenatal onset of cortical hyperostosis (Caffey disease): Case report and review. *Am. J. Med. Genet.*: 120A(4) 547-52
43. Winter J.\*, Lehmann T.\*, Suckow V., Kijas Z., Kulozic A., Hamel B., Opitz J., Lenzner S., Ropers HH., **Schweiger S.** (2003) Duplication of exon 1 of the MID1 gene in a patient with Opitz G/BBB syndrome. *Hum. Genet.* 112, 249-254
44. Trockenbacher A., Suckow V., Foerster J., Winter J., Krauß S., Ropers HH., Schneider R., **Schweiger S.** (2001) MID1, mutated in Opitz syndrome encodes an ubiquitin ligase that targets phosphatase 2A for degradation. *Nature genetics* 29, 287-294
45. **Schweiger S.**, Foerster J., Lehmann T., Suckow V., Muller Y.A., Walter G., Davies T., Porter H., van Bokhoven H., Lunt P.W., Traub P., Ropers H.H. (1999) The Opitz syndrome gene product, MID1, associates with microtubules. *Proc. Natl. Acad. Sci USA* 96, 2794-2799
46. Quaderi N., **Schweiger S.**, Gaudenz K., Franco B., Rugarli E.I., Berger W., Feldmann G.J., Volta M., Andolfi G., Gilgenkrantz S., Marion R.W., Hennekam R.C.M., Opitz J.M., Muenke M., Ropers H.H., Ballabio A.

(1997) Opitz G/BBB syndrome, a defect of midline development, is due to mutations in a new RING finger gene on Xp22. *Nature genetics* 17, 285-291