

**Journalbeiträge**

1. Atili A, Lübke J, Shoukier M, Schittkowski MP (2012) [Hermansky-Pudlak syndrome]. OPHTHALMOLOGE, 110(2): 160-3.
2. Batsukh T, Schulz Y, Wolf S, Rabe TI, Oellerich T, Urlaub H, Schaefer IM, Pauli S (2012) Identification and characterization of FAM124B as a novel component of a CHD7 and CHD8 containing complex. PLOS ONE, 7(12): e52640.
3. Böckle BC, Shoukier M, Kaulfuß S, Sepp NT (2012) Mucocutaneous telangiectasia - it's the tip of the iceberg. J EUR ACAD DERMATOL, :-:.
4. Bremmer F, Hemmerlein B, Strauss A, Burfeind P, Thelen P, Radzun HJ, Behnes CL (2012) N-cadherin expression in malignant germ cell tumours of the testis. BMC Clin Pathol, 12(1): 19.
5. Buhl T, Shoukier M, Grzmil P, Revencu N, Schön MP, Seitz CS (2012) Multifocal Capillary Malformations Due to RASA1 Mutation Misdiagnosed as Cutaneous Mastocytosis. ARCH DERMATOL, 148(11): 1334-5.
6. Burnicka-Turek O, Mohamed BA, Shirneshan K, Thanasupawat T, Hombach-Klonisch S, Klonisch T, Adham IM (2012) INSL5-deficient mice display an alteration in glucose homeostasis and an impaired fertility. ENDOCRINOLOGY, 153(10): 4655-65.
7. Khromov T, Dressel R, Siamishi I, Nolte J, Opitz L, Engel W, Pantakani K (2012) Apoptosis-related gene expression profiles of mouse ESCs and maGSCs: role of Fgf4 and MnDA in pluripotent cell responses to genotoxicity. PLOS ONE, 7(11): e48869.
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9. Marklein D, Graab U, Naumann I, Yan T, Ridzewski R, Nitzki F, Rosenberger A, Dittmann K, Wienands J, Wojnowski L, Fulda S, Hahn H (2012) PI3K inhibition enhances doxorubicin-induced apoptosis in sarcoma cells. PLOS ONE, 7(12): e52898.
10. Mohamed BA, Barakat AZ, Zimmermann WH, Bittner RE, Mühlfeld C, Hünlich M, Engel W, Maier LS, Adham IM (2012) Targeted disruption of Hspa4 gene leads to cardiac hypertrophy and fibrosis. J MOL CELL CARDIOL, 53(4): 459-68.
11. Nitzki F, Becker M, Frommhold A, Schulz-Schaeffer W, Hahn H (2012) Patched knockout mouse models of Basal cell carcinoma. J Skin Cancer, 2012: 907543.
12. Pauli S, von Velsen N, Burfeind P, Steckel M, Mänz J, Buchholz A, Borozdin W, Kohlhase J (2012) CHD7 mutations causing CHARGE syndrome are predominantly of paternal origin. CLIN GENET, 81(3): 234-9.
13. Pauli S, Schmidt T, Funke R, Zoll B, Burfeind P, Dybowski U, Shoukier M, Bartels I (2012) Discordant phenotype in monozygotic twins with mosaic trisomy 12p in lymphocytes. EUR J MED GENET, 55(8-9): 480-4.
14. Pauli S, Steinemann D, Dittmann K, Wienands J, Shoukier M, Möschner M, Burfeind P, Manukyan G, Göhring G, Escherich G (2012) Occurrence of acute lymphoblastic leukemia and juvenile myelomonocytic leukemia in a patient with Noonan syndrome carrying the germline PTPN11 mutation p.E139D. AM J MED GENET A, 158A(3): 652-8.
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17. Shoukier M, Klein N, Auber B, Wickert J, Schröder J, Zoll B, Burfeind P, Bartels I, Alsat E, Lingen M, Grzmil P, Schulze S, Keyser J, Weise D, Borchers M, Hobbiebrunken E, Röbl M, Gärtner J, Brockmann K, Zirn B (2012) Array CGH in patients with developmental delay or intellectual disability: are there phenotypic clues to pathogenic copy number variants? CLIN GENET, 83(1): 53-65.
18. Shoukier M, Schröder J, Zoll B, Burfeind P, Freiberg C, Klinge L, Kriebel T, Lingen M, Mohr A, Brockmann K (2012) A de novo interstitial deletion of 2p23.3-24.3 in a boy presenting with intellectual disability, overgrowth, dysmorphic features, skeletal myopathy, dilated cardiomyopathy. AM J MED GENET A, 158A(2): 429-33.
19. Smorag L, Zheng Y, Nolte J, Zechner U, Engel W, Pantakani DVK (2012) MicroRNA signature in various cell types of mouse spermatogenesis: evidence for stage-specifically expressed miRNA-221, -203 and -34b-5p mediated spermatogenesis regulation. BIOL CELL, 104(11): 677-92.
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Publikationen und Hochschulschriften 2012  
**Humangenetik**

21. van Dop WA, Rosekrans SL, Uhmann A, Jaks V, Offerhaus GJA, van den Bergh Weerman MA, Kasper M, Heijmans J, Hardwick JCH, Verspaget HW, Hommes DW, Toftgård R, Hahn H, van den Brink GR (2012) Hedgehog signalling stimulates precursor cell accumulation and impairs epithelial maturation in the murine oesophagus. *GUT*, 62(3): 348-57.
22. Yang K, Meinhardt A, Zhang B, Grzmil P, Adham IM, Hoyer-Fender S (2012) The small heat shock protein ODF1/HSPB10 is essential for tight linkage of sperm head to tail and male fertility in mice. *MOL CELL BIOL*, 32(1): 216-25.
23. Zheng Y, Tan X, Pyczek J, Nolte J, Pantakani DVK, Engel W (2012) Generation and characterization of yeast two-hybrid cDNA libraries derived from two distinct mouse pluripotent cell types. *MOL BIOTECHNOL*, 54(2): 228-37.
24. Zirn B, Shoukier M, Bartels I, Arning L, Hoffjan S, Neubauer B, Hahn A (2012) Neurofibromatose Typ II und geistige Behinderung bei Ringchromosom 22: Was ist der genetische Zusammenhang? *Neuropädiatrie in Klinik und Praxis*, 4: 161-164.

**Buchbeiträge**

1. Nolte J, Pantakani DV, Dihazi H, Zechner U (2012) Gene expression and epigenetics signatures of germ cell-derived pluripotent stem cells and embryonic stem cells. In: Hayat, M.A. (Hg.) *Stem Cells and Cancer Stem Cells*, Vol. 6, Therapeutic Applications in Disease and Injury. Springer Verlag, Berlin/Heidelberg/New York, 21 - 34.

**Medizinische Dissertationen**

1. Ellen HL, Dr. med. (2012) Zur Funktion des Bruno14-Gens. Dissertation Universität Göttingen.
2. Lammering I, Dr. med. (2012) 1alpha,25-Dihydroxy-VitaminD3 hemmt das Wachstum von Patched-assoziierten Rhabdomyosarkomen und Basaliomen. Dissertation Universität Göttingen.
3. Mohamed BM, Dr. med. (2012) The molecular role of the heat shock protein family110 (HSP110). Dissertation Universität Göttingen.
4. Warntjen N, Dr. med. (2012) DNA-Strangbrüche in männlichen Keimzellen der Maus. Dissertation Universität Göttingen.

**Naturwiss. u.a. nichtmed. Diss.**

1. Batsukh T, Dr. rer. nat. (2012) Novel interaction partners of the chromatin remodeler CHD7, a protein mutated in CHARGE syndrome. Dissertation Universität Göttingen.
2. Smorag L, Dr. rer. nat. (2012) miRNA functions in pluripotency and spermatogenesis. Dissertation Universität Göttingen.
3. Tan X, Dr. rer. nat. (2012) Functional analysis of Zfp819 in pluripotency and embryonic development. Dissertation Universität Göttingen.
4. Wieczerek K, Dr. rer. nat. (2012) Analysis of coiled coil domain containing 33 protein (CCDC33) and determination of infertility causes in mutant mouse line with the deletion of six germ cell-specific genes. Dissertation Universität Göttingen.
5. Witt D, Dr. rer. nat. (2012) Analyse zur Rolle von pflanzlichen Wirkstoffen und Histondeacetylase-Inhibitoren auf Wachstumsfaktoren und deren Signalwege in Prostatakarzinomzellen. Dissertation Universität Göttingen.
6. Xu X, Dr. rer. nat. (2012) Stage-specific germ cell marker genes function in establishment and germ cell lineage commitment of pluripotent stem cells. Dissertation Universität Göttingen.

**Diplomarbeiten**

1. Hamdaoui A, Dipl.-Biol. (2012) Untersuchungen zur Funktion des Pelota-Gens in der postnatalen Entwicklung. Diplomarbeit Universität Göttingen.

**Masterarbeiten**

1. Can K, MSc (2012) Identification of transgene insertion site in Mecp2R270\_EGFP transgenic mice and functional characterization of MeCP2 R270\_EGFP mutant mouse models. Masterarbeit Universität Göttingen.
2. Kolb S, MSc (2012) Analysis of putative interaction partners of mouse Lrrc34. Masterarbeit Universität Göttingen.
3. Oberthür R, MSc (2012) Analysis of DNA damage after combined radiochemotherapy in combination with receptor tyrosine kinase inhibition. Masterarbeit Universität Göttingen.