

Publikationen Prof. Thorsten Rosenbaum

1. DeSouza, C. E., Menezes, R. A., Menezes, C., Nagvekar, S. S., Heiming, R., **Rosenbaum, T.**, DeSasouza S. (1988) Mucocoeles, proptosis and transorbital frontoethmoidectomy. *Orbit*, 7, No. 3, 167-173.
2. **Rosenbaum, T.**, Rammos, S., Kniemeyer, H.-W., Göbel, U. (1993) Extended deep vein and inferior vena cava thrombosis in a 15-year-old boy: successful lysis with a recombinant tissue-type plasminogen activator 2 weeks after onset of symptoms. *Eur J Pediatr* 152, 978-980.
3. Guénard, V., **Rosenbaum, T.**, Gwynn, L. A., Doetschmann, T., Ratner, N., Wood, P. M. (1995) Effect of transforming growth factor β 1 and $-\beta$ 2 on Schwann cell proliferation on neurites. *Glia* 13, 309-318.
4. Kim, H. A., **Rosenbaum, T.**, Marchionni, M. A., Ratner, N., DeClue J. E. (1995) Schwann cells from neurofibromin deficient mice exhibit activation of p21ras, inhibition of cell proliferation and morphological changes. *Oncogene* 11, 325-335.
5. **Rosenbaum, T.**, Boissy, Y. L., Kombrinck, K., Brannan, C.I., Jenkins, N. A., Copeland, N. G., Ratner, N. (1995) Neurofibromin-deficient fibroblasts fail to form perineurium in vitro. *Development* 121, 3583-3592.
6. **Rosenbaum, T.**, Lenard, H.-G. (1997) Die Neurofibromatosen. *Kinderarzt*, 28 (2),154-164.
7. **Rosenbaum, T.**, Patrie, K., Ratner, N. (1997) Genetic and cellular mechanisms of tumor formation in neurofibromatosis type 1. *Neuroscientist*, 3 (6), 412-420.
8. **Rosenbaum, T.**, Gärtner, J., Körholz, D., Janßen, G., Schneider, D., Engelbrecht, V., Göbel, U., Lenard, H.-G. (1998) Paraneoplastic limbic encephalitis in two teenage girls. *Neuropediatrics*,29, 159-162.
9. Reifenberger, J.; Kruse, R.; Schön, M.P.; **Rosenbaum, T.**; Ruzicka, T. (1999) Incontinentia pigmenti bei einem männlichen Neugeborenen. *Zeitschr f Hautkrankh* 74:578-579.
10. **Rosenbaum, T.**; Engelbrecht, V.; Krölls, W.; van Dorsten, F. A.; Hoehn-Berlage, M.; Lenard, H.-G. (1999) MRI abnormalities in neurofibromatosis type 1 (NF1). A study of men and mice. *Brain Dev*, 21, 268-273.
11. **Rosenbaum, T.**; Kim, H. A.; Boissy, Y. L.; Bo, L., Ratner, N. (1999) Neurofibromin, the neurofibromatosis type 1 (NF1) Ras-GAP, is required for appropriate P0 expression and myelination. *Ann NY Acad Sci*, 883, 203-215.
12. **Rosenbaum, T.**; Rosenbaum, C.; Winner, U.; Müller, H.W.; Lenard, H.-G.; Hanemann, C. O. (2000) Long-term culture and characterization of human neurofibroma-derived Schwann cells. *J Neurosci Res*, 61,524-532.
13. Sherman, L. S.; Atit, R.; **Rosenbaum, T.**; Cox, A. D.; Ratner, N. (2000) Single cell Ras-GTP analysis reveals altered Ras activity in a subpopulation of neurofibroma Schwann cells but not fibroblasts. *J Biol Chem*, 275 (39),30740-30745.

14. Serra, E.; **Rosenbaum, T.**; Winner, U.; Aledo, R., Ars, E., Estivill, X.; Lenard, H.-G.; Lázaro, C. (2000) Schwann cells are the NF1 (-/-) cells in neurofibromas: evidence of two different Schwann cell subpopulations. *Hum Mol Genet*, 9(20),3055-3064.
15. Serra, E.; Ars, E.; Ravella, A.; Sánchez, A.; Puig, S.; **Rosenbaum, T.**; Estivill, X.; Lázaro, C. (2001) Somatic mRNA splice defects are common within the NF1 mutational spectrum in neurofibromas. *Hum Genet*, 108:416-429.
16. Serra, E.; **Rosenbaum, T.**; Nadal, M.; Winner, U.; Ars, E.; Estivill, X.; Lázaro, C. (2001) Mitotic recombination effects homozygosity for NF1 germline mutations in neurofibromas. *Nature Genet*, 28:294-296
17. **Rosenbaum, T.**; Richter-Werkle; R.; Köller, H.; Lenard, H.-G. (2002) Psychogenic contractures: the magic side of medicine. *Neuropediatrics*, 33:166-167.
18. **Rosenbaum, T.**; (2002) NF1-Mutationen in Schwannzellen führen zur Neurofibrom-Entstehung. Ein kombinierter genetischer und zellbiologischer Ansatz zur Erforschung der Tumorentstehung bei Neurofibromatose Typ 1. *Neuropaediatric*, 3:93-96.
19. Knorr, M.; Schaper, J.; Harjes, M.; Mayatepek, E.; **Rosenbaum, T.** (2004) Valproat-induced Fanconi-syndrome. *Epilepsia*, 45(7):868-871.
20. Holtkamp, N.; Reuß, D. E.; Atallah, I.; Kuban R.-J.; Hartmann C.; Mautner V.-F.; Frahm, S.; Friedrich, R.E.; Algermissen, B.; Pham V.-A.; Prietz, S.; **Rosenbaum, T.**; Estevez-Schwarz, L.; von Deimling A. (2004) Subclassification of Nerve Sheath Tumors by Gene Expression Profiling. *Brain Pathology*, 14(3):258-264.
21. Groninger, A.; Schaper, J.; Messing-Juenger, M.; Mayatepek, E.; **Rosenbaum, T.** (2005) Subdural Hematoma as Clinical Presentation of Osteogenesis Imperfecta. *Pediat Neurology*, 32:140-142.
22. Groninger, A.; **Rosenbaum, T.** (2005) Neurokutane Syndrome. *Kinderärztl Prax*, 1:24-31.
23. Spiegel, M.; Horn, D.; Oexle, K.; Windt, E.; Buske, A.; Albrecht, B.; Prott, E.-C.; Seemanová, E.; Seidel, J.; **Rosenbaum, T.**; Schmidt, G.; Rauch, A.; Jenne, D.; Kehrer-Sawatzki, H.; Tinschert, S.; (2005) Childhood overgrowth in patients with common *NF1* microdeletions, *Eur J Hum Genet*, 13(7):883-888.
24. Overdiek, A.; **Rosenbaum, T.** (2005) Neurofibromatose Typ 1(NF1) – Empfehlungen zur Diagnose und Langzeitbetreuung. *Kinder- und Jugendarzt*, 36(8):519-526
25. Bernbeck, B.; Schwabe J.; Groninger, A.; Schaper, J.; Messing-Jünger, M.; Mayatepek, E.; **Rosenbaum, T.** (2005) Aplasia congenita of the scalp: How much therapy is necessary in large defects? *Acta paed*, 94:758-765.
26. Wimmer, K.; Decker, M.; Mayatepek, E.; Beiglböck, H.; Eggermann, T.; Kehrer-Sawatzki, H.; Fonatsch, C.; **Rosenbaum, T.** (2005) Silver-Russell-Syndrome-like features in a patient carrying a novel *NF1*-mutation. *Ped Research*, 58(6):1265-1268.
27. Heckel, M.; Kieseier, B. C.; Schaper, J., Mayatepek, E., **Rosenbaum, T.** (2005)

Mitoxantrone as escalating therapy in two children with worsening multiple sclerosis, *J Ped Neurol*, 3(4):237-242.

28. Tibussek, D.; Mayatepek, E.; Distelmaier, F.; **Rosenbaum, T.** (2006) Status epilepticus due to attempted suicide with isoniazide. *Eur J Ped*, 165(2):136-137.
29. Distelmaier, F.; Sengler, U.; Messing-Juenger, M.; Assmann, B.; Mayatepek, E.; **Rosenbaum, T.** (2006) Pseudotumor cerebri as an important differential diagnosis of papilledema in children. *Brain Dev*, 28(3):190-195.
30. Assmann, B.; van Kuilenburg, A.B.P.; Distelmaier, F.; Abeling, N.G.G.M.; **Rosenbaum, T.**; Duran, M.; Mayatepek, E. (2006) β -Ureidopropionase deficiency presenting as febrile status epilepticus, *Epilepsia*, 47(1):215-217.
31. Overdiek, A.; Feifel, H.; Schaper, J.; Mayatepek, E.; **Rosenbaum, T.** (2006) Diagnostic delay in hemifacial hypertrophy due to plexiform neurofibromas. *Brain Dev*, 28(5):275-280.
32. Harjes, M.; Mayatepek, E.; Ruzicka, T.; **Rosenbaum, T.** (2006) Retikuläres Exanthem des Abdomens. *Monatsschr Kinderheilkunde*, 154(3):269-270.
33. Distelmaier, F.; Janssen, G.; Mayatepek, E.; Schaper, J.; Göbel, U.; **Rosenbaum, T.** (2006) Disseminated pilocytic astrocytoma involving brain stem and diencephalon: a history of atypical eating disorder and loss of time. *NeuroOncology*, 79(2):197-201.
34. Harjes, M.; Mayatepek, E.; Rübo, J.; **Rosenbaum, T.** (2006) Miller-Fisher Syndrom: Wenn sich die Augen nicht bewegen und der Körper nicht stillstehen will. *Neuropädiatrie*, 5(4):150-153.
35. Maertens, O.; Brems, H.; De Raedt, T.; Heyns, I.; **Rosenbaum, T.**; De Schepper, S.; De Paepe, A.; Janssens, S.; Speleman, F.; Legius, E.; Messiaen, L. (2006) Comprehensive *NF1* Screening on Cultured Schwann Cells from Neurofibromas. *Human Mutation*, 27(10):1030-1040.
36. Distelmaier, F.; Fahsold, R.; Reifenberger, G.; Messing-Juenger, M.; Schaper, J.; Schneider, D.T.; Göbel, U.; Mayatepek, E.; **Rosenbaum, T.** (2007) Fatal glioblastoma multiforme in a patient with neurofibromatosis type I: the dilemma of systematic medical follow-up. *Childs Nerv Syst*, 23(3):343-347.
37. Distelmaier, F.; Göbel, U.; Vandemeulebroecke, N.; Mayatepek, E.; **Rosenbaum, T.**; Laws, H. J. (2007) Secondary pseudotumor cerebri in pediatric oncology and hematology: an unpredictable condition of varying etiology. *Pediatr Blood Cancer*, 49(7):1029-1033.
38. Distelmaier, F.; Richter-Werkle, R.; Schaper, J.; Messing-Jünger, M.; Mayatepek, E.; **Rosenbaum, T.** (2007) How much brain is really necessary? A case of complex cerebral malformation and its clinical course. *J Child Neurology*, 22(6):750-760.
39. Overdiek, A.; Winner, U.; Mayatepek, E.; **Rosenbaum, T.** (2008) Schwann cells from human neurofibromas show increased proliferation rates under the influence of progesterone. *Ped Research*, 64(1):40-43.
40. Wortmann, M. u. **Rosenbaum, T.** (2008) Phakomatosen. *Paediatr Prax*, 72:285-299.

41. Wüller, D.; Kämmerer, F.; Alfke, H; **Rosenbaum, T.** (2009) Zervikale Myelitis transversa bei einem 15 Monate alten Kind. *Neuropaediatric*, 8(4):121-124.
42. Tibussek, D.; Hübsch S.; Berger, K.; Schaper., J.; **Rosenbaum, T.**; Mayatepek, E. (2009) Infantile onset neurofibromatosis type 2 presenting with peripheral facial palsy, skin patches, retinal hamartoma and foot drop. *Klin Pädiatr*, 221(4):247-250.
43. Harder, A.; Titze, S.; Herbst, L.; Harder, T.; Guse, K.; Tinschert, S.; Kaufmann, D.; **Rosenbaum, T.**; Mautner, V.F.; Windt, E.; Wahlländer-Danek, U.; Wimmer, K.; Mundlos, S.; Peters, H. (2010) Monozygotic twins with neurofibromatosis type 1 (NF1) display differences in methylation of NF1 gene promoter elements, 5' untranslated region, exon and intron 1. *Twin Res Hum Genet*, 13(6):582-594.
44. Linden, K.; Moser, O.; Simon, A.; Eis-Hübinger, A.-M.; Fleischhack, G.; Born, M.; Tschampa, H.; **Rosenbaum, T.**; Köster, B.; Lentze, M. (2011) Transiente Spleniumläsion bei Influenza-A-H1N1-09-Infektion. *Radiologe* 51:220-222.
45. Thimm, A., Kämmerer, F.; Mayatepek, E., **Rosenbaum, T.** (2011) Optikusgliome bei Kindern mit Neurofibromatose Typ 1: Eine Langzeit-Untersuchung an 38 Patienten. *Neuropaediatric*, 10 (2):40-49.
46. Finetti C., Utz N., Krüger S., **Rosenbaum T.** (2011) Bewegungsstörung, Mikrozephalie und Myelinisierungsverzögerung. *Monatsschrift Kinderheilk*, 159:1071-1075.
47. Czeschik JC, Hehr U, Hartmann B, Lüdecke HJ, **Rosenbaum T**, Schweiger B, Wiczorek D (2013) 160 kb deletion in ISPD unmasking a recessive mutation in a patient with Walker-Warburg syndrome. *Eur J Med Genet*, 56:689-94
48. Vogt J, Bengesser K, Claes KB, Wimmer K, Mautner VF, van Minkelen R, Legius E, Brems H, Upadhyaya M, Högel J, Lazaro C, **Rosenbaum T**, Bammert S, Messiaen L, Cooper DN, Kehrer-Sawatzki H (2014) SVA retrotransposon insertion-associated deletion represents a novel mutational mechanism underlying large genomic copy number changes with non-recurrent breakpoints. *Genome Biol* 15(6):R80. doi: 10.1186/gb-2014-15-6-r80
49. **Rosenbaum T**, Borusiak P, Schweitzer T, Berweck S, Sprinz A, Strassburg H, Klepper J (2014) Dynamische Kopforthesen („Helmtherapie“). Stellungnahme der gemeinsamen Therapiekommission der Gesellschaft für Neuropädiatrie und der Deutschen Gesellschaft für Sozialpädiatrie und Jugendmedizin (2012). *Neuropaediatric* 13:4-9.
50. Maslehaty H, Petridis AK, Schreiber L, Brassel F, **Rosenbaum T**, Scholz M. (2014) Management and treatment of paediatric neurovascular diseases in an interdisciplinary setting. *J Neurol Disord* 2:181-184.
51. **Rosenbaum, T**; Wimmer K (2014) Neurofibromatosis type 1 (NF1) and associated tumors. *Klin Paediatric* 226:309-315
52. Niklewski F, Petridis AK, Al Hourani J, Blaeser K, Ntoulis G, Bitter A, **Rosenbaum T**, Scholz M (2015) Pediatric parafalcine empyemas. *J Surg Case Reports*, doi:10.1093/jscr/rjt067
53. Vaassen, P.; **Rosenbaum, T.** (2016) Nevus anemicus as additional diagnostic

marker of neurofibromatosis type 1 in childhood. *Neuropediatrics* 47:190-193.

54. Wimmer, K; **Rosenbaum, T.**; Messiaen, L. (2017) Connections between constitutional mismatch repair deficiency syndrome and neurofibromatosis type 1. *Clin Genet* 91(4):507-519.
55. Schaefer, S A; **Rosenbaum, T.**; Vester, U.; Cetiner, M., Rehme, C.; Feldkamp, A. (2017) Dysurische Beschwerden bei einem syrischen Jungen. *Monatsschr Kinderheilk*: DOI 10.1007/s00112-017-0248-6
56. **Rosenbaum T**; Vaassen P. (2017) Comment on: Petersen, D.; Höger, P.: Welche Diagnose wird gestellt? Mosaische Neurofibromatose. *KiJuA* 48(3):115.
57. Altmüller J, Gärtner J, Krätzner R, Nürnberg P, Reinert MC, **Rosenbaum T**, Weissbach S (2017) A new *CUL4B* variant associated with an exceptional pattern of white matter lesions. *Am J Med Genet* 173(10):2803-2807.
58. Tesch VK, Ijspeert H, Raicht A, Rueda D, Dominguez-Pinilla N, Allende L.M., Colas C, **Rosenbaum T**, Ilencikova D, Baris HN, Nathrath M, Suerink M, Januszkiewicz-Lewandowska D, Ragab I, Azizi AA, Wenzel SS, Zschocke J, Schwinger W, Kloor M, Blattmann C, Brugieres L, van der Burg M, Wimmer K, and Seidel MG (2018) No Overt Clinical Immunodeficiency Despite Immune Biological Abnormalities in Patients With Constitutional Mismatch Repair Deficiency. *Front. Immunol.* 9:1506. doi: 10.3389/fimmu.2018.01506
59. Chiu C, Loth S, Kuhlen M, Ginzel S, Schaper J, **Rosenbaum T**, Pietsch T, Borkhardt A, Hoell JI (2018) Mutated SON putatively causes a cancer syndrome comprising high-risk medulloblastoma combined with café-au-lait spots. *Fam Cancer*. 2019 doi: 10.1007/s10689-019-00121-z.
60. Gallon R, Mühlegger B, Wenzel SS, Sheth H, Hayes C, Aretz S, Dahan K, Foulkes W, Kratz CP, Ripperger T, Azizi AA, Baris Feldman H, Chong AL, Demirsoy U, Florkin B, Imschweiler T, Januszkiewicz-Lewandowska D, Lobitz S, Nathrath M, Pander HJ, Perez-Alonso V, Perne C, Ragab I, **Rosenbaum T**, Rueda D, Seidel MG, Suerink M, Taeubner J, Zimmermann SY, Zschocke J, Borthwick GM, Burn J, Jackson MS, Santibanez-Koref M, Wimmer K (2019) A sensitive and scalable microsatellite instability assay to diagnose constitutional mismatch repair deficiency by sequencing of peripheral blood leukocytes *Hum Mutat* ;40(5):649-655.
61. Vaassen P, Dürr N, Röhrig A, Willing R, **Rosenbaum T** (2019) Trametinib induces neurofibroma shrinkage and enables surgery. *Neuropediatrics*; 50(5):300-303.
62. González-Acosta M, Marín F, Puliafito B, Bonifaci N, Fernández A, Navarro M, Salvador H, Balaguer F, Iglesias S, Velasco A, Grau Garces E, Moreno V, Gonzalez-Granado LI, Guerra-García P, Ayala R, Florkin B, Kratz C, Ripperger T, **Rosenbaum T**, Januszkiewicz-Lewandowska D, Azizi AA, Ragab I, Nathrath M, Pander HJ, Lobitz S, Suerink M, Dahan K, Imschweiler T, Demirsoy U, Brunet J, Lázaro C, Rueda D, Wimmer K, Capellá G, Pineda M (2020) High-sensitivity Microsatellite Instability Assessment for the Detection of Mismatch Repair Defects in Normal Tissue of Biallelic Germline Mismatch Repair Mutation Carriers. *J Med Genet* 57(4):269-273
63. Heidelberg F, **Rosenbaum T**, Abicht A, Valcheva D (2020) Autismus-Spektrum-Störung als Erstsymptom einer neuronalen Ceroid-Lipoduzinose Typ 7 (NCL7).

64. Kehrer-Sawatzki H, Kluwe L, Salamon J, Well L, Farschtschi S, **Rosenbaum T**, Mautner VF (2020) Clinical characterization of children and adolescents with NF1 microdeletions. *Child's Nervous System*. doi:10.1007/s00381-020-047170
65. Friedrich RE, Zustin J, Luebke AM, **Rosenbaum T**, Gosau M, Hagel C, Kohlrusch FK, Wieland I, Zenker M (2021) Neurofibromatosis type 1 with cherubism-like phenotype, multiple osteolytic bone lesions of lower extremities, and Alagille-syndrome: case report with literature survey. *In Vivo*;35(3):1711-1736.
66. Vaassen P, Dürr NR, **Rosenbaum T** (2022) Treatment of plexiform neurofibromas with MEK inhibitors: First results with a new therapeutic option. *Neuropediatrics*; 53:52-60.
67. Vaassen P, Dürr NR, **Rosenbaum T** (2022) Neue Therapieoptionen bei Neurofibromatose Typ 1 (NF1). *Neuropaediatric*; 21:4-11.
68. Vaassen P, Dürr NR, **Rosenbaum T** (2022) Neurofibromatose Typ 1 (NF1) – Klinisches Bild und neue Therapieansätze. *Päd*; 28:282-288.
69. Schneider DT, Blessing T, Graf N, Abele M, Brecht IB, **Rosenbaum T** (2022) Tumorerkrankungen im Kontext von Störungen von Wachstum und Entwicklung. *Monatsschr Kinderheilkd*; 170:730-737.
70. Kaindl AM, Hennermann JB, Niller HH, Hehr U, von Bernuth H, Chaoui R, Landwehr-Kenzel S, Hahn G, Mundlos C, Thomale UW, **Rosenbaum T**, Moog U, Horn D, von der Hagen M (2022) Handlungsempfehlungen nach der Leitlinie Klassifikation und Diagnostik der Mikrozephalie. *Monatsschr Kinderheilkd*; 170:929-933.
71. Gallon R, Phelps R, Hayes C, Brugieres L, Guerrini-Rousseau L, Colas C, Muleris M, Ryan N, D. Evans GD, Grice H, Jessop E, Kunzemann-Martinez A, Marshall L, Oberhuber K, Azizi A, Feldman HB, Beilken A, Dahan K, Demirsoy U, Florkin B, Foulkes W, Imschweiler T, Januszkiewicz-Lewandowska D, Jones KJ, Kratz C, Lobitz S, Meade J, Nathrath M, Pander HJ, Perne C, Ragab I, Ripperger I, **Rosenbaum T**, Rueda D, Sehested A, Suerink M, Taeubner J, Sarosiek Tomasz S, Zimmermann SY, Zschocke J, Borthwick GM, Wimmer K, Burn J, Jackson MS, Santibanez-Koref M (2022) Constitutional microsatellite instability, genotype, and phenotype correlations in Constitutional Mismatch Repair Deficiency, *Gastroenterology*: S0016 5085(22) 01444-5
72. Carton, C et al. on behalf of the ERN GENTURIS NF1 Tumour Management Guideline Group (2023). ERN GENTURIS tumour surveillance guidelines for individuals with neurofibromatosis type 1. *eClinicalMedicine* 56: 101818. <https://doi.org/10.1016/j.eclinm.2022.101818>

