

**Publikationen von Mitgliedern des ZSEA,
die in Pubmed für den Zeitraum (01.10.2023 - 30.09.2024) unter Aachener Adresse
registriert sind und einen Bezug zu Seltenen Erkrankungen aufweisen**

(jede Publikation wird nur ein Mal aufgeführt, unterstrichen sind die Autoren, die Sprecher/innen und stv. Sprecher/innen von Behandlungs- und Forschungszentren sind oder i.d.R. anderweitig als Ansprechpartner/in im ZSEA-Internet gelistet sind)

Seltene neurologische Erkrankungen

1. Beijer D, Dohrn MF, Rebelo A, Danzi MC, Grosz BR, Ellis M, Kumar KR, Vucic S, Vais H, Weissenrieder JS, Lunko O, Paudel U, Simpson LC, Raposo J, Saporta M, Arcia Y, Xu I, Feely S, Record CJ, Blake J, Reilly MM, Scherer S, Kennerson M, Lee YC, Foskett JK, Shy M, Zuchner S. A recurrent missense variant in ITPR3 causes demyelinating Charcot-Marie-Tooth with variable severity. *Brain*. 2024 Jun 28:awae206. doi: 10.1093/brain/awae206. Epub ahead of print. PMID: 38938188.
2. Bremer J, Meinhardt A, Katona I, Senderek J, Kämmerer-Gassler EK, Roos A, Ferbert A, Schröder JM, Nikolin S, Nolte K, Sellhaus B, Popzhelyazkova K, Tacke F, Schara-Schmidt U, Neuen-Jacob E, de Groote CC, de Jonghe P, Timmerman V, Baets J, Weis J. Myelin protein zero mutation-related hereditary neuropathies: Neuropathological insight from a new nerve biopsy cohort. *Brain Pathol*. 2024 Jan;34(1):e13200. doi: 10.1111/bpa.13200. Epub 2023 Aug 15. PMID: 37581289; PMCID: PMC10711263.
3. Cortese A, Currò R, Ronco R, Blake J, Rossor AM, Bugiardini E, Laurà M, Warner T, Yousry T, Poh R, Polke J, Rebelo A, Dohrn MF, Saporta M, Houlden H, Zuchner S, Reilly MM. Mutations in alpha-B-crystallin cause autosomal dominant axonal Charcot-Marie-Tooth disease with congenital cataracts. *Eur J Neurol*. 2024 Jan;31(1):e16063. doi: 10.1111/ene.16063. Epub 2023 Sep 29. PMID: 37772343; PMCID: PMC10872581.
4. Danzi MC, Powell E, Rebelo AP, Dohrn MF, Beijer D, Fazal S, Xu IRL, Medina J, Chen S, Arcia de Jesus Y, Schatzman J, Hershberger RE, Saporta M, Baets J, Falk M, Herrmann DN, Scherer SS, Reilly MM, Cortese A, Marques W, Cornejo-Olivas MR, Sanmaneechai O, Kennerson ML, Jordanova A, Silva TYT, Pedroso JL, Schierbaum L, Ebrahimi-Fakhari D, Peric S, Lee YC, Synofzik M, Tekin M, Ravenscroft G, Shy M, Basak N, Schule R, Zuchner S. The GENESIS database and tools: A decade of discovery in Mendelian genomics. *Exp Neurol*. 2024 Dec;382:114978. doi: 10.1016/j.expneurol.2024.114978. Epub 2024 Sep 30. PMID: 39357594.

5. Dohrn MF, Beijer D, Lone MA, Bayraktar E, Oflazer P, Orbach R, Donkervoort S, Foley AR, Rose A, Lyons M, Louie RJ, Gable K, Dunn T, Chen S, Danzi MC, Synofzik M, Bönnemann CG, Nazlı Başak A, Hornemann T, Zuchner S. Recurrent *de-novo* gain-of-function mutation in *SPTLC2* confirms dysregulated sphingolipid production to cause juvenile amyotrophic lateral sclerosis. *J Neurol Neurosurg Psychiatry*. 2024 Feb 14;95(3):201-205. doi: 10.1136/jnnp-2023-332130. PMID: 38041684; PMCID: PMC10922288.
6. Ferreira M, Schaprian T, Kügler D, Reuter M, Deike-Hoffmann K, Timmann D, Ernst TM, Giunti P, Garcia-Moreno H, van de Warrenburg B, van Gaalen J, de Vries J, Jacobi H, Steiner KM, Öz G, Joers JM, Onyike C, Povazan M, Reetz K, Romanzetti S, Klockgether T, Faber J. Cerebellar volumetry in ataxias: Relation to ataxia severity and duration. *Res Sq [Preprint]*. 2023 Nov 16:rs.3.rs-3605029. doi: 10.21203/rs.3.rs-3605029/v1. Update in: *Cerebellum*. 2024 Aug;23(4):1521-1529. doi: 10.1007/s12311-024-01659-0. PMID: 38014351; PMCID: PMC10680914.
7. Ferreira M, Schaprian T, Kügler D, Reuter M, Deike-Hoffmann K, Timmann D, Ernst TM, Giunti P, Garcia-Moreno H, van de Warrenburg B, van Gaalen J, de Vries J, Jacobi H, Steiner KM, Öz G, Joers JM, Onyike C, Povazan M, Reetz K, Romanzetti S, Klockgether T, Faber J. Cerebellar Volumetry in Ataxias: Relation to Ataxia Severity and Duration. *Cerebellum*. 2024 Aug;23(4):1521-1529. doi: 10.1007/s12311-024-01659-0. Epub 2024 Feb 16. Erratum in: *Cerebellum*. 2024 Aug;23(4):1530-1531. doi: 10.1007/s12311-024-01681-2. PMID: 38363498; PMCID: PMC11269395.
8. Ferreira M, Schaprian T, Kügler D, Reuter M, Deike-Hoffmann K, Timmann D, Ernst TM, Giunti P, Garcia-Moreno H, van de Warrenburg B, van Gaalen J, de Vries J, Jacobi H, Steiner KM, Öz G, Joers JM, Onyike C, Povazan M, Reetz K, Romanzetti S, Klockgether T, Faber J. Correction: Cerebellar Volumetry in Ataxias: Relation to Ataxia Severity and Duration. *Cerebellum*. 2024 Aug;23(4):1530-1531. doi: 10.1007/s12311-024-01681-2. Erratum for: *Cerebellum*. 2024 Aug;23(4):1521-1529. doi: 10.1007/s12311-024-01659-0. PMID: 38446346; PMCID: PMC11269408.
9. Fischer F, Dohrn MF, Kapfenberger R, Igharo D, Seeber D, de Moya Rubio E, Pitarokilli K, Börsch N, Mücke M, Rolke R, Schulz JB, Maier A. Neuropathische Schmerzen als Symptom bei autonomen Neuropathien und anderen seltenen Erkrankungen : Kleinfaserneuropathie erkennen, diagnostizieren und behandeln [Neuropathic pain as a symptom in autonomic neuropathies and other rare diseases : Small fiber neuropathy: its recognition, diagnosis, and treatment]. *Schmerz*. 2024 Feb;38(1):33-40. German. doi: 10.1007/s00482-023-00783-w. Epub 2024 Jan 10. PMID: 38197939.
10. Godbole S, Voß H, Gocke A, Schlumbohm S, Schumann Y, Peng B, Mynarek M, Rutkowski S, Dottermusch M, Dorostkar MM, Korshunov A, Mair T, Pfister SM, Kwiatkowski M, Hotze M, Neumann P, Hartmann C, Weis J, Liesche-Starnecker F, Guan Y, Moritz M, Siebels B, Struve N, Schlüter H, Schüller U, Krisp C, Neumann JE. Multiomic profiling of medulloblastoma reveals subtype-specific targetable alterations at the proteome and N-glycan level. *Nat Commun*. 2024 Jul 24;15(1):6237. doi: 10.1038/s41467-024-50554-z. PMID: 39043693; PMCID: PMC11266559.
11. Graessner H, Reinhard C, Bäumer T, Baumgärtner A, Brockmann K, Brüggemann N, Bültmann E, Erdmann J, Heise K, Höglinger G, Hüning I, Kaiser FJ, Klein C, Klopstock T, Krägeloh-Mann I, Kraemer M, Luedtke K, Mücke M, Musacchio T, Nadke A, Osmanovic A, Ritter G, Röse K, Schippers C, Schöls L, Schüle R, Schulz JB, Sproß J, Stasch E, Wunderlich G, Münchau A.

Recommendations for optimal interdisciplinary management and healthcare settings for patients with rare neurological diseases. *Orphanet J Rare Dis.* 2024 Feb 13;19(1):62. doi: 10.1186/s13023-024-03023-1. PMID: 38347616; PMCID: PMC10863275.

12. Hengel H, Martus P, Faber J, Giunti P, Garcia-Moreno H, Solanky N, Klockgether T, Reetz K, van de Warrenburg BP, Santana MM, Silva P, Cunha I, de Almeida LP, Timmann D, Infante J, de Vries J, Lima M, Pires P, Bushara K, Jacobi H, Onyike C, Schmahmann JD, Hübener-Schmid J, Synofzik M; European Spinocerebellar Ataxia Type-3/Machado-Joseph Disease Initiative (ESMI) Study Group; Schöls L. Correction to: The frequency of non-motor symptoms in SCA3 and their association with disease severity and lifestyle factors. *J Neurol.* 2024 Jan;271(1):628-629. doi: 10.1007/s00415-023-12064-8. Erratum for: *J Neurol.* 2023 Feb;270(2):944-952. doi: 10.1007/s00415-022-11441-z. PMID: 37979094; PMCID: PMC10769956.
13. Holzer MT, Uruha A, Roos A, Hentschel A, Schänzer A, Weis J, Claeys KG, Schoser B, Montagnese F, Goebel HH, Huber M, Léonard-Louis S, Kötter I, Streichenberger N, Gally L, Benveniste O, Schneider U, Preusse C, Krusche M, Stenzel W. Anti-Ku + myositis: an acquired inflammatory protein-aggregate myopathy. *Acta Neuropathol.* 2024 Jul 16;148(1):6. doi: 10.1007/s00401-024-02765-3. PMID: 39012547; PMCID: PMC11252205.
14. Indelicato E, Reetz K, Maier S, Nachbauer W, Amprosi M, Giunti P, Mariotti C, Durr A, de Rivera Garrido FJR, Klopstock T, Schöls L, Klockgether T, Bürk K, Pandolfo M, Didszun C, Grobe-Einsler M, Nanetti L, Nenning L, Kiechl S, Dichtl W, Ulmer H, Schulz JB, Boesch S; European Friedreich's Ataxia Consortium for Translational Studies (EFACTS). Predictors of Survival in Friedreich's Ataxia: A Prospective Cohort Study. *Mov Disord.* 2024 Mar;39(3):510-518. doi: 10.1002/mds.29687. Epub 2023 Dec 23. PMID: 38140802.
15. Jing Y, Dogan I, Reetz K, Romanzetti S. Neurochemical changes in the progression of Huntington's disease: A meta-analysis of in vivo ¹H-MRS studies. *Neurobiol Dis.* 2024 Sep;199:106574. doi: 10.1016/j.nbd.2024.106574. Epub 2024 Jun 22. PMID: 38914172.
16. Lavorgna L, Maida E, Reinhard C, Cras P, Reetz K, Molnar MJ, Nonnekes J, Medijainen K, Summa S, Diserens K, Petrarca M, Albanese A, Leocani L, Delussi M, Vinciguerra C, Pagliano E, Kubica J, Lallemand P, Wenning G, Sival D, Groleger Srsen K, Bertini ES, Lopane G, Boesch S, Bonavita S, Crosiers D, Muresanu D, Timmann D, Federico A; European Reference Network on Rare Neurological Disease (ERN-RND) Telerehabilitation Working Group. The Growing Role of Telerehabilitation and Teleassessment in the Management of Movement Disorders in Rare Neurological Diseases: A Scoping Review. *Telemed J E Health.* 2024 Sep;30(9):2419-2430. doi: 10.1089/tmj.2023.0702. Epub 2024 Jul 1. PMID: 38946606.
17. Linse K, Weber C, Reilich P, Schöberl F, Boentert M, Petri S, Rödiger A, Posa A, Otto M, Wolf J, Zeller D, Brunckhorst R, Koch J, Hermann A, Großkreutz J, Schröter C, Groß M, Lingor P, Machetanz G, Semmler L, Dorst J, Lulé D, Ludolph A, Meyer T, Maier A, Metelmann M, Regensburger M, Winkler J, Schrank B, Kohl Z, Hagenacker T, Brakemeier S, Weyen U, Weiler M, Lorenzl S, Bublitz S, Weydt P, Grehl T, Kotterba S, Lapp HS, Freigang M, Vidovic M, Aust E, Günther R. Patients' and caregivers' perception of multidimensional and palliative care in amyotrophic lateral sclerosis - protocol of a German multicentre study. *Neurol Res Pract.* 2024 Jul 4;6(1):34. doi: 10.1186/s42466-024-00328-1. PMID: 38961496; PMCID: PMC11223310.

18. Paul MS, Michener SL, Pan H, Chan H, Pfliger JM, Rosenfeld JA, Lerma VC, Tran A, Longley MA, Lewis RA, Weisz-Hubshman M, Bekheirnia MR, Bekheirnia N, Massingham L, Zech M, Wagner M, Engels H, Cremer K, Mangold E, Peters S, Trautmann J, Mester JL, Guillen Sacoto MJ, Person R, McDonnell PP, Cohen SR, Lusk L, Cohen ASA, Le Pichon JB, Pastinen T, Zhou D, Engleman K, Racine C, Faivre L, Moutton S, Denommé-Pichon AS, Koh HY, Poduri A, Bolton J, Knopp C, Julia Suh DS, Maier A, Toosi MB, Karimiani EG, Maroofian R, Schaefer GB, Ramakumaran V, Vasudevan P, Prasad C, Osmond M, Schuhmann S, Vasileiou G, Russ-Hall S, Scheffer IE, Carvill GL, Mefford H; Undiagnosed Diseases Network; Bacino CA, Lee BH, Chao HT. A syndromic neurodevelopmental disorder caused by rare variants in PPFIA3. *Am J Hum Genet.* 2024 Jan 4;111(1):96-118. doi: 10.1016/j.ajhg.2023.12.004. Erratum in: *Am J Hum Genet.* 2024 Apr 4;111(4):805. doi: 10.1016/j.ajhg.2024.03.009. Erratum in: *Am J Hum Genet.* 2024 Jun 6;111(6):1239. doi: 10.1016/j.ajhg.2024.04.022. PMID: 38181735; PMCID: PMC10806447.
19. Porcu L, Fichera M, Nanetti L, Rulli E, Giunti P, Parkinson MH, Durr A, Ewencyk C, Boesch S, Nachbauer W, Indelicato E, Klopstock T, Stendel C, Rodríguez de Rivera FJ, Schöls L, Fleszar Z, Giordano I, Didszun C, Castaldo A, Rai M, Klockgether T, Pandolfo M, Schulz JB, Reetz K, Mariotti C; EFACTS Study Group. Longitudinal changes of SARA scale in Friedreich ataxia: Strong influence of baseline score and age at onset. *Ann Clin Transl Neurol.* 2023 Nov;10(11):2000-2012. doi: 10.1002/acn3.51886. Epub 2023 Aug 28. PMID: 37641437; PMCID: PMC10647003.
20. Rebelo AP, Abad C, Dohrn MF, Li JJ, Tieu EK, Medina J, Yanick C, Huang J, Zotter B, Young JI, Saporta M, Scherer SS, Walz K, Zuchner S. SORD-deficient rats develop a motor-predominant peripheral neuropathy unveiling novel pathophysiological insights. *Brain.* 2024 Sep 3;147(9):3131-3143. doi: 10.1093/brain/awae079. PMID: 38538210.
21. Rebelo AP, Tomaselli PJ, Medina J, Wang Y, Dohrn MF, Nyvltova E, Danzi MC, Garrett M, Smith SE, Pestronk A, Li C, Ruiz A, Jacobs E, Feely SME, França MC, Gomes MV, Santos DF, Kumar S, Lombard DB, Saporta M, Hekimi S, Barrientos A, Wehl C, Shy ME, Marques W, Zuchner S. Biallelic variants in COQ7 cause distal hereditary motor neuropathy with upper motor neuron signs. *Brain.* 2023 Oct 3;146(10):4191-4199. doi: 10.1093/brain/awad158. PMID: 37170631; PMCID: PMC10545612.
22. Rezende TJR, Adanyaguh I, Barsottini OGP, Bender B, Cendes F, Coutinho L, Deistung A, Dogan I, Durr A, Fernandez-Ruiz J, Göricke SL, Grisoli M, Hernandez-Castillo CR, Lenglet C, Mariotti C, Martinez ARM, Massuyama BK, Mochel F, Nanetti L, Nigri A, Ono SE, Öz G, Pedroso JL, Reetz K, Synofzik M, Teive H, Thomopoulos SI, Thompson PM, Timmann D, van de Warrenburg BPC, van Gaalen J, França MC Jr, Harding IH. Genotype-specific spinal cord damage in spinocerebellar ataxias: an ENIGMA-Ataxia study. *J Neurol Neurosurg Psychiatry.* 2024 Jun 17;95(7):682-690. doi: 10.1136/jnnp-2023-332696. PMID: 38383154; PMCID: PMC11187354.

23. Robertson JW, Adanyeguh I, Bender B, Boesch S, Brunetti A, Coccozza S, Coutinho L, Deistung A, Diciotti S, Dogan I, Durr A, Fernandez-Ruiz J, Göricke SL, Grisoli M, Han S, Mariotti C, Marzi C, Mascalchi M, Mochel F, Nachbauer W, Nanetti L, Nigri A, Ono SE, Onyike CU, Prince JL, Reetz K, Romanzetti S, Saccà F, Synofzik M, Ghizoni Teive HA, Thomopoulos SI, Thompson PM, Timmann D, Ying SH, Harding IH, Hernandez-Castillo CR. The Pattern and Staging of Brain Atrophy in Spinocerebellar Ataxia Type 2 (SCA2): MRI Volumetrics from ENIGMA-Ataxia. *bioRxiv [Preprint]*. 2024 Sep 17:2024.09.16.613281. doi: 10.1101/2024.09.16.613281. PMID: 39345594; PMCID: PMC11429976.
24. Rodríguez-Labrada R, Canales-Ochoa N, Galicia-Polo ML, Cruz-Rivas E, Romanzetti S, Peña-Acosta A, Estupiñán-Rodríguez A, Vázquez-Mojena Y, Dogan I, Auburger G, Reetz K, Velázquez-Pérez L. Structural Brain Correlates of Sleep Microstructure in Spinocerebellar Ataxia Type 2 and its Role on Clinical Phenotype. *Cerebellum*. 2024 Oct;23(5):1839-1847. doi: 10.1007/s12311-024-01674-1. Epub 2024 Mar 4. PMID: 38438827.
25. Roos A, Häusler M, Kollipara L, Topf A, Preusse C, Stucka R, Nolte K, Strom T, Berutti R, Jiang X, Koll R, Lochmüller H, Schacht SM, Zahedi RP, Weis J, Senderek J. HNRNPA1 de novo Variant Associated with Early Childhood Onset, Rapidly Progressive Generalized Myopathy. *J Neuromuscul Dis*. 2024;11(5):1131-1137. doi: 10.3233/JND-240050. PMID: 39121134; PMCID: PMC11380306.
26. Roos A, van der Ven PFM, Alrohaif H, Kölbel H, Heil L, Della Marina A, Weis J, Aßent M, Beck-Wödl S, Barresi R, Töpf A, O'Connor K, Sickmann A, Kohlschmidt N, El Gizouli M, Meyer N, Daya N, Grande V, Bois K, Kaiser FJ, Vorgerd M, Schröder C, Schara-Schmidt U, Gangfuss A, Evangelista T, Röbisch L, Hentschel A, Grüneboom A, Fuerst DO, Kuechler A, Tzschach A, Depienne C, Lochmüller H. Bi-allelic variants of FILIP1 cause congenital myopathy, dysmorphism and neurological defects. *Brain*. 2023 Oct 3;146(10):4200-4216. doi: 10.1093/brain/awad152. PMID: 37163662; PMCID: PMC10545528.
27. Saft C, Burgunder JM, Dose M, Jung HH, Katzenschlager R, Priller J, Nguyen HP, Reetz K, Reilmann R, Seppi K, Landwehrmeyer GB. Symptomatic treatment options for Huntington's disease (guidelines of the German Neurological Society). *Neurol Res Pract*. 2023 Nov 16;5(1):61. doi: 10.1186/s42466-023-00285-1. PMID: 37968732; PMCID: PMC10652593.
28. Saft C, Burgunder JM, Dose M, Jung HH, Katzenschlager R, Priller J, Nguyen HP, Reetz K, Reilmann R, Seppi K, Landwehrmeyer GB. Differential diagnosis of chorea (guidelines of the German Neurological Society). *Neurol Res Pract*. 2023 Nov 23;5(1):63. doi: 10.1186/s42466-023-00292-2. PMID: 37993913; PMCID: PMC10666412.
29. Xu IRL, Danzi MC, Ruiz A, Raposo J, De Jesus YA, Reilly MM, Cortese A, Shy ME, Scherer SS, Herrmann DN, Fridman V, Baets J, Saporta M, Seyedsadjadi R, Stojkovic T, Claeys KG, Patel P, Feely S, Rebelo AP; Inherited Neuropathy Consortium; Dohrn MF, Züchner S. A study concept of expeditious clinical enrollment for genetic modifier studies in Charcot-Marie-Tooth neuropathy 1A. *J Peripher Nerv Syst*. 2024 Jun;29(2):202-212. doi: 10.1111/jns.12621. Epub 2024 Apr 5. PMID: 38581130; PMCID: PMC11209807.
30. Neu: Costa AS, Albrecht M, Reich A, Nikoubashman O, Schulz JB, Reetz K, Pinho J. Non-hemorrhagic imaging markers of cerebral amyloid angiopathy in memory clinic patients. *Alzheimers Dement*. 2024 Jul;20(7):4792-4802. doi: 10.1002/alz.13920. Epub 2024 Jun 12. PMID: 38865440; PMCID: PMC11247708.

Seltene hämatologische Erkrankungen

31. Ben Hamza A, Welters C, Stadler S, Brüggemann M, Dietze K, Brauns O, Brümmendorf TH, Winkler T, Bullinger L, Blankenstein T, Rosenberger L, Leisegang M, Kammertöns T, Herr W, Moosmann A, Strobel J, Hackstein H, Dornmair K, Beier F, Hansmann L. Virus-reactive T cells expanded in aplastic anemia eliminate hematopoietic progenitor cells by molecular mimicry. *Blood*. 2024 Apr 4;143(14):1365-1378. doi: 10.1182/blood.2023023142. PMID: 38277625.
32. Bermes M, Rodriguez MJ, de Toledo MAS, Ernst S, Müller-Newen G, Brümmendorf TH, Chatain N, Koschmieder S, Baumeister J. Exploiting Synthetic Lethality between Germline BRCA1 Haploinsufficiency and PARP Inhibition in JAK2V617F-Positive Myeloproliferative Neoplasms. *Int J Mol Sci*. 2023 Dec 16;24(24):17560. doi: 10.3390/ijms242417560. PMID: 38139386; PMCID: PMC10743753.
33. Dingli D, De Castro Iii C, Koprivnikar J, Kulasekararaj A, Maciejewski J, Mulherin B, Panse J, Pullarkat V, Röth A, Shammo J, Terriou L, Weitz I, Yermilov I, Gibbs S, Broder M, Beenhouwer D, Kuter D. Expert consensus on the management of pharmacodynamic breakthrough-hemolysis in treated paroxysmal nocturnal hemoglobinuria. *Hematology*. 2024 Dec;29(1):2329030. doi: 10.1080/16078454.2024.2329030. Epub 2024 Mar 21. PMID: 39665683.
34. Enßle JC, Wolf S, Scheich S, Weber S, Kramer M, Ruhnke L, Schliemann C, Mikesch JH, Krause S, Sauer T, Hanoun M, Reinhardt HC, Kraus S, Kaufmann M, Hänel M, Fransecky L, Burchert A, Neubauer A, Crysandt M, Jost E, Niemann D, Schäfer-Eckart K, Held G, Kaiser U, Wass M, Schaich M, Müller-Tidow C, Platzbecker U, Baldus CD, Bornhäuser M, Röllig C, Serve H; Study Alliance Leukemia (SAL); Steffen B. Impact of BMI on patient outcome in acute myeloid leukaemia patients receiving intensive induction therapy: a real-world registry experience. *Br J Cancer*. 2023 Oct;129(7):1126-1133. doi: 10.1038/s41416-023-02362-3. Epub 2023 Aug 4. PMID: 37542108; PMCID: PMC10539505.
35. Gambacorti-Passerini C, Brümmendorf TH, Abruzzese E, Kelly KR, Oehler VG, García-Gutiérrez V, Hjorth-Hansen H, Ernst T, Leip E, Purcell S, Luscan G, Viqueira A, Giles FJ, Hochhaus A. Efficacy and safety of bosutinib in previously treated patients with chronic myeloid leukemia: final results from the BYOND trial. *Leukemia*. 2024 Oct;38(10):2162-2170. doi: 10.1038/s41375-024-02372-x. Epub 2024 Aug 20. PMID: 39164407; PMCID: PMC11436368.
36. Grasshoff M, Kalmer M, Chatain N, Kricheldorf K, Maurer A, Weiskirchen R, Koschmieder S, Costa IG. Single cell level Genotyping Using scRNA Data (SIGURD). *Brief Bioinform*. 2024 Sep 23;25(6):bbae604. doi: 10.1093/bib/bbae604. PMID: 39559832; PMCID: PMC11574290.
37. Griffin M, Eikema DJ, Verheggen I, Kulagin A, Tjon JM, Fattizzo B, Ingram W, Zaidi U, Desnica L, Giammarco S, Drozd-Sokolowska J, Xicoy B, Patriarca A, Loschi M, Szmigielska-Kaplon A, Beier F, Cignetti A, Drexler B, Gavriilaki E, Lanza F, Orvain C, Risitano AM, De la Camara R, De Latour RP. SARS-CoV-2 vaccination in 361 non-transplanted patients with aplastic anemia and/or paroxysmal nocturnal hemoglobinuria. *Haematologica*. 2024 Jan 1;109(1):283-286. doi: 10.3324/haematol.2023.283863. PMID: 37584297; PMCID: PMC10772488.

38. Griffin M, Kelly RJ, Panse J, de Castro C, Szer J, Horneff R, Tan L, Yeh M, Peffault de Latour R. Management of acute breakthrough hemolysis with intensive pegcetacoplan dosing in patients with PNH. *Blood Adv*. 2024 Apr 9;8(7):1776-1786. doi: 10.1182/bloodadvances.2023011691. PMID: 38315872; PMCID: PMC10985803.
39. Harrison CN, Kiladjian JJ, Koschmieder S, Passamonti F. Myelofibrosis: Current unmet needs, emerging treatments, and future perspectives. *Cancer*. 2024 Jun 15;130(12):2091-2097. doi: 10.1002/cncr.35244. Epub 2024 Feb 19. PMID: 38373144.
40. Holl K, Chatain N, Krapp S, Baumeister J, Maié T, Schmitz S, Scheufen A, Brock N, Koschmieder S, Moreno-Andrés D. Calreticulin and JAK2V617F driver mutations induce distinct mitotic defects in myeloproliferative neoplasms. *Sci Rep*. 2024 Feb 2;14(1):2810. doi: 10.1038/s41598-024-53240-8. PMID: 38308077; PMCID: PMC10837458.
41. Isfort S, Manz K, Teichmann LL, Crysandt M, Burchert A, Hochhaus A, Saussele S, Kiani A, Göthert JR, Illmer T, Schafhausen P, Al-Ali HK, Stegelmann F, Hänel M, Pfeiffer T, Giagounidis A, Franke GN, Koschmieder S, Fabarius A, Ernst T, Warnken-Uhlich M, Wolber U, Kohn D, Pfirrmann M, Wolf D, Brümmendorf TH; German CML study group. Step-in dosing of bosutinib in pts with chronic phase chronic myeloid leukemia (CML) after second-generation tyrosine kinase inhibitor (TKI) therapy: results of the Bosutinib Dose Optimization (BODO) Study. *Ann Hematol*. 2023 Oct;102(10):2741-2752. doi: 10.1007/s00277-023-05394-0. Epub 2023 Aug 18. PMID: 37592092; PMCID: PMC10492675.
42. Kantarjian HM, Jabbour EJ, Lipton JH, Castagnetti F, Brümmendorf TH. A Review of the Therapeutic Role of Bosutinib in Chronic Myeloid Leukemia. *Clin Lymphoma Myeloma Leuk*. 2024 May;24(5):285-297. doi: 10.1016/j.clml.2024.01.005. Epub 2024 Jan 12. PMID: 38278737.
43. Koschmieder S, Isfort S, Schulte C, Jacobasch L, Geer T, Reiser M, Koenigsmann M, Heinrich B, Wehmeyer J, von der Heyde E, Tesch H, Gröschl B, Bachhuber P, Großer S, Pahl HL. Real-world analysis of ruxolitinib in myelofibrosis: interim results focusing on patients who were naïve to JAK inhibitor therapy treated within the JAKoMo non-interventional, phase IV trial. *Ann Hematol*. 2023 Dec;102(12):3383-3399. doi: 10.1007/s00277-023-05458-1. Epub 2023 Oct 4. PMID: 37792065; PMCID: PMC10640411.
44. Lipton JH, Brümmendorf TH, Sweet K, Apperley JF, Cortes JE. Practical considerations in the management of patients treated with bosutinib for chronic myeloid leukemia. *Ann Hematol*. 2024 Sep;103(9):3429-3442. doi: 10.1007/s00277-024-05851-4. Epub 2024 Jul 18. PMID: 39023573; PMCID: PMC11358173.
45. Lommerse IN, Hinnen C, van Vliet LM, Schubert B, Panse J, Halkes CJM, Tjon JM. Quality of life after immune suppressive therapy in aplastic anemia. *Ann Hematol*. 2024 Jun;103(6):2113-2121. doi: 10.1007/s00277-024-05731-x. Epub 2024 Apr 5. PMID: 38578507; PMCID: PMC11090919.

46. Lübke J, Schmid A, Christen D, Oude Elberink HNG, Span LFR, Nidoszytko M, Gorska A, Lange M, Gleixner KV, Hadzijusufovic E, Stefan A, Angelova-Fischer I, Zanotti R, Bonifacio M, Bonadonna P, Shoumariyeh K, von Bubnoff N, Müller S, Perkins C, Elena C, Malcovati L, Hagglund H, Mattsson M, Parente R, Varkonyi J, Fortina AB, Caroppo F, Brockow K, Zink A, Breynaert C, Leven T, Yavuz AS, Doubek M, Sabato V, Schug T, Hartmann K, Triggiani M, Gotlib J, Hermine O, Arock M, Kluin-Nelemans HC, Panse J, Sperr WR, Valent P, Reiter A, Schwaab J. Serum chemistry profiling and prognostication in systemic mastocytosis: a registry-based study of the ECNM and GREM. *Blood Adv*. 2024 Jun 11;8(11):2890-2900. doi: 10.1182/bloodadvances.2024012756. PMID: 38593217; PMCID: PMC11214361.
47. Machova Polakova K, Albeer A, Polivkova V, Krutka M, Vicanova K, Curik N, Fabarius A, Klamova H, Spiess B, Waller CF, Brümmendorf TH, Dengler J, Kunzmann V, Burchert A, Belohlavkova P, Mustjoki S, Faber E, Mayer J, Zackova D, Panayiotidis P, Richter J, Hjorth-Hansen H, Kamińska M, Płonka M, Szczepanek E, Szarejko M, Bober G, Hus I, Grzybowska-Izydorzyc O, Wasilewska E, Paczkowska E, Niesiobędzka-Krężel J, Giannopoulos K, Mahon FX, Sacha T, Sauße S, Pfirrmann M. The SNP rs460089 in the gene promoter of the drug transporter OCTN1 has prognostic value for treatment-free remission in chronic myeloid leukemia patients treated with imatinib. *Leukemia*. 2024 Feb;38(2):318-325. doi: 10.1038/s41375-023-02109-2. Epub 2023 Dec 21. PMID: 38129513; PMCID: PMC10844071.
48. Mahon FX, Pfirrmann M, Dulucq S, Hochhaus A, Panayiotidis P, Almeida A, Mayer J, Hjorth-Hansen H, Janssen JJWM, Mustjoki S, Martinez-Lopez J, Vestergaard H, Ehrencrona H, Machová Poláková K, Olsson-Strömberg U, Ossenkuppele G, Berger MG, Etienne G, Dengler J, Brümmendorf TH, Burchert A, Réa D, Rousselot P, Nicolini FE, Hofmann WK, Richter J, Saussele S; EURO-SKI Investigators; Investigators. European Stop Tyrosine Kinase Inhibitor Trial (EURO-SKI) in Chronic Myeloid Leukemia: Final Analysis and Novel Prognostic Factors for Treatment-Free Remission. *J Clin Oncol*. 2024 Jun 1;42(16):1875-1880. doi: 10.1200/JCO.23.01647. Epub 2024 Mar 12. PMID: 38471049.
49. Naumann N, Rudelius M, Lübke J, Christen D, Bresser J, Sotlar K, Metzgeroth G, Fabarius A, Hofmann WK, Panse J, Horny HP, Cross NCP, Reiter A, Schwaab J. Poor Applicability of Currently Available Prognostic Scoring Systems for Prediction of Outcome in *KIT* D816V-Negative Advanced Systemic Mastocytosis. *Cancers (Basel)*. 2024 Jan 30;16(3):593. doi: 10.3390/cancers16030593. PMID: 38339343; PMCID: PMC10854835.
50. Nidoszytko M, Gorska A, Brockow K, Bonadonna P, Lange M, Kluin-Nelemans H, Oude-Elberink H, Sabato V, Shoumariyeh K, von Bubnoff D, Müller S, Illerhaus A, Doubek M, Angelova-Fischer I, Hermine O, Arock M, Elena C, Malcovati L, Yavuz AS, Schug TD, Fortina AB, Judit V, Gotlib J, Panse J, Vucinic V, Reiter A, Schwaab J, Triggiani M, Mattsson M, Breynaert C, Romantowski J, Zanotti R, Olivieri E, Zink A, van de Ven A, Stefan A, Barete S, Caroppo F, Perkins C, Kennedy V, Christen D, Jawhar M, Luebke J, Parente R, Levedahl K, Hadzijusufovic E, Hartmann K, Nidoszytko B, Sperr WR, Valent P. Prevalence of hypersensitivity reactions in various forms of mastocytosis: A pilot study of 2485 adult patients with mastocytosis collected in the ECNM registry. *Allergy*. 2024 Sep;79(9):2470-2481. doi: 10.1111/all.16132. Epub 2024 Apr 23. PMID: 38651829.
51. Niewisch MR, Beier F, Savage SA. Clinical manifestations of telomere biology disorders in adults. *Hematology Am Soc Hematol Educ Program*. 2023 Dec 8;2023(1):563-572. doi: 10.1182/hematology.2023000490. PMID: 38066848; PMCID: PMC10726987.

52. Olschok K, Altenburg B, de Toledo MAS, Maurer A, Abels A, [Beier F](#), [Gezer D](#), Isfort S, Paeschke K, [Brümmendorf TH](#), Zenke M, Chatain N, [Koschmieder S](#). The telomerase inhibitor imetelstat differentially targets JAK2V617F versus CALR mutant myeloproliferative neoplasm cells and inhibits JAK-STAT signaling. *Front Oncol*. 2023 Oct 24;13:1277453. doi: 10.3389/fonc.2023.1277453. PMID: 37941547; PMCID: PMC10628476.
53. [Panse J](#), Daguindau N, Okuyama S, Peffault de Latour R, Schafhausen P, Straetmans N, Al-Adhami M, Persson E, Wong RSM. Improvements in hematologic markers and decreases in fatigue with pegcetacoplan for patients with paroxysmal nocturnal hemoglobinuria and mild or moderate anemia (hemoglobin ≥ 10 g/dL) who had received eculizumab or were naive to complement inhibitors. *PLoS One*. 2024 Jul 29;19(7):e0306407. doi: 10.1371/journal.pone.0306407. PMID: 39079163; PMCID: PMC11285951.
54. [Panse JP](#), Höchsmann B, Schubert J. Paroxysmal Nocturnal Hemoglobinuria, Pathophysiology, Diagnostics, and Treatment. *Transfus Med Hemother*. 2024 Aug 21;51(5):310-320. doi: 10.1159/000540474. PMID: 39371251; PMCID: PMC11452172.
55. Patriquin CJ, Bogdanovic A, Griffin M, Kelly RJ, Maciejewski JP, Mulherin B, Peffault de Latour R, Röth A, Selvaratnam V, Szer J, Al-Adhami M, Horneff R, Tan L, Yeh M, [Panse J](#). Safety and Efficacy of Pegcetacoplan in Adult Patients with Paroxysmal Nocturnal Hemoglobinuria over 48 Weeks: 307 Open-Label Extension Study. *Adv Ther*. 2024 May;41(5):2050-2069. doi: 10.1007/s12325-024-02827-8. Epub 2024 Apr 4. PMID: 38573482; PMCID: PMC11052769.
56. Peffault de Latour R, Griffin M, Kelly RJ, Szer J, de Castro C, Horneff R, Tan L, Yeh M, [Panse J](#). Hemolysis events in the phase 3 PEGASUS study of pegcetacoplan in patients with paroxysmal nocturnal hemoglobinuria. *Blood Adv*. 2024 Jun 11;8(11):2718-2725. doi: 10.1182/bloodadvances.2024012672. PMID: 38593241; PMCID: PMC11170155.
57. Peffault de Latour R, Röth A, Kulasekararaj AG, Han B, Scheinberg P, Maciejewski JP, Ueda Y, de Castro CM, Di Bona E, Fu R, Zhang L, Griffin M, Langemeijer SMC, [Panse J](#), Schrezenmeier H, Barcellini W, Mauad VAQ, Schafhausen P, Tavitian S, Beggiato E, Chew LP, Gaya A, Huang WH, Jang JH, Kitawaki T, Kutlar A, Notaro R, Pullarkat V, Schubert J, Terriou L, Uchiyama M, Wong Lee Lee L, Yap ES, Sicre de Fontbrune F, Marano L, Alashkar F, Gandhi S, Trikha R, Yang C, Liu H, Kelly RJ, Höchsmann B, Kerloeguen C, Banerjee P, Levitch R, Kumar R, Wang Z, Thorburn C, Maitra S, Li S, Verles A, Dahlke M, Risitano AM. Oral lptacopan Monotherapy in Paroxysmal Nocturnal Hemoglobinuria. *N Engl J Med*. 2024 Mar 14;390(11):994-1008. doi: 10.1056/NEJMoa2308695. PMID: 38477987.
58. Pritchard JE, Pearce JE, Snoeren IAM, Fuchs SNR, Götz K, Peisker F, Wagner S, Benabid A, Lutterbach N, Klöker V, Nagai JS, Hannani MT, Galyga AK, Sistemich E, Banjanin B, Flosdorf N, Bindels E, Olschok K, Biaisch K, Chatain N, Bhagwat N, Dunbar A, Sarkis R, Naveiras O, Berres ML, [Koschmieder S](#), Levine RL, Costa IG, Gleitz HFE, Kramann R, Schneider RK. Non-canonical Hedgehog signaling mediates profibrotic hematopoiesis-stroma crosstalk in myeloproliferative neoplasms. *Cell Rep*. 2024 Jan 23;43(1):113608. doi: 10.1016/j.celrep.2023.113608. Epub 2023 Dec 20. PMID: 38117649; PMCID: PMC10828549.

59. Rahn K, Abdallah AT, Gan L, Herbrich S, Sonntag R, Benitez O, Malaney P, Zhang X, Rodriguez AG, Brottem J, Marx G, Brümmendorf TH, Ostareck DH, Ostareck- Lederer A, Crysanndt M, Post SM, Naarmann-de Vries IS. Insight into the mechanism of AML del(9q) progression: hnRNP K targets the myeloid master regulators CEBPA (C/EBP α) and SPI1 (PU.1). *Biochim Biophys Acta Gene Regul Mech.* 2024 Mar;1867(1):195004. doi: 10.1016/j.bbagr.2023.195004. Epub 2023 Nov 25. PMID: 38008244.
60. Rolles B, Caballero-Oteyza A, Proietti M, Goldacker S, Warnatz K, Camacho- Ordonez N, Prader S, Schmid JP, Vieri M, Isfort S, Meyer R, Kirschner M, Brümmendorf TH, Beier F, Grimbacher B. Telomere biology disorders may manifest as common variable immunodeficiency (CVID). *Clin Immunol.* 2023 Dec;257:109837. doi: 10.1016/j.clim.2023.109837. Epub 2023 Nov 8. PMID: 37944684.
61. Rolles B, Tometten M, Meyer R, Kirschner M, Beier F, Brümmendorf TH. Inherited Telomere Biology Disorders: Pathophysiology, Clinical Presentation, Diagnostics, and Treatment. *Transfus Med Hemother.* 2024 Jul 30;51(5):292-309. doi: 10.1159/000540109. PMID: 39371255; PMCID: PMC11452174.
62. Röllig C, Steffen B, Schliemann C, Mikesch JH, Alakel N, Herbst R, Hänel M, Noppeney R, Hanoun M, Kaufmann M, Weinbergerova B, Schäfer-Eckart K, Sauer T, Neubauer A, Burchert A, Baldus CD, Mertová J, Jost E, Niemann D, Novák J, Krause SW, Scholl S, Hochhaus A, Held G, Szotkowski T, Rank A, Schmid C, Fransecky L, Kayser S, Schaich M, Kramer M, Fiebig F, Haake A, Schetelig J, Middeke JM, Stölzel F, Platzbecker U, Thiede C, Müller-Tidow C, Berdel WE, Ehninger G, Mayer J, Serve H, Bornhäuser M. Single or Double Induction With 7 + 3 Containing Standard or High-Dose Daunorubicin for Newly Diagnosed AML: The Randomized DaunoDouble Trial by the Study Alliance Leukemia. *J Clin Oncol.* 2025 Jan;43(1):65-74. doi: 10.1200/JCO.24.00235. Epub 2024 Sep 16. PMID: 39284116.
63. Rosti G, Brümmendorf TH, Gjertsen BT, Giraldo-Castellano P, Castagnetti F, Gambacorti-Passerini C, Ernst T, Zhao H, Kuttschreuter L, Purcell S, Giles FJ, Hochhaus A. Impact of age and comorbidities on the efficacy and tolerability of bosutinib in previously treated patients with chronic myeloid leukemia: results from the phase 4 BYOND study. *Leukemia.* 2024 Jan;38(1):126-135. doi: 10.1038/s41375-023-02080-y. Epub 2023 Nov 25. PMID: 38007586; PMCID: PMC10776383.
64. Salimi A, Schemionek-Reinders M, Huber M, Vieri M, Patterson JB, Alten J, Brümmendorf TH, Kharabi Masouleh B, Appelmann I. XBP1 promotes NRAS^{G12D} pre-B acute lymphoblastic leukaemia through IL-7 receptor signalling and provides a therapeutic vulnerability for oncogenic RAS. *J Cell Mol Med.* 2023 Nov;27(21):3363-3377. doi: 10.1111/jcmm.17904. Epub 2023 Sep 27. PMID: 37753803; PMCID: PMC10623536.
65. Scheinberg P, Clé DV, Kim JS, Nur E, Yenerel MN, Barcellini W, Bonito D, Giai V, Hus M, Lee Y, Lekue CB, Panse J, Ueda Y, Buatois S, Gentile B, Kiialainen A, Patel H, Sreckovic S, Uguen M, Edwards J, Nagy Z, Kulasekararaj AG. Phase 3 randomized COMMODORE 1 trial: Crovalimab versus eculizumab in complement inhibitor-experienced patients with paroxysmal nocturnal hemoglobinuria. *Am J Hematol.* 2024 Sep;99(9):1757-1767. doi: 10.1002/ajh.27413. Epub 2024 Jun 25. PMID: 38924124.
66. Stelljes M, Middeke JM, Bug G, Wagner-Drouet EM, Müller LP, Schmid C, Krause SW, Bethge W, Jost E, Platzbecker U, Klein SA, Schubert J, Niederland J, Kaufmann M, Schäfer-Eckart K, Schaich M, Baldauf H, Stölzel F, Petzold C, Röllig C, Alakel N, Steffen B, Hauptrock B,

Schliemann C, Sockel K, Lang F, Kriege O, Schaffrath J, Reicherts C, Berdel WE, Serve H, Ehninger G, Schmidt AH, Bornhäuser M, Mikesch JH, Schetelig J; Study Alliance Leukemia and the German Cooperative Transplant Study Group. Remission induction versus immediate allogeneic haematopoietic stem cell transplantation for patients with relapsed or poor responsive acute myeloid leukaemia (ASAP): a randomised, open-label, phase 3, non-inferiority trial. *Lancet Haematol*. 2024 May;11(5):e324-e335. doi: 10.1016/S2352-3026(24)00065-6. Epub 2024 Apr 4. PMID: 38583455.

67. Szer J, Panse J, Kulasekararaj A, Oliver M, Fattizzo B, Nishimura JI, Horneff R, Szamosi J, Peffault de Latour R. Moving toward Individual Treatment Goals with Pegcetacoplan in Patients with PNH and Impaired Bone Marrow Function. *Int J Mol Sci*. 2024 Aug 6;25(16):8591. doi: 10.3390/ijms25168591. PMID: 39201278; PMCID: PMC11354612.
68. Tilmont R, Yakoub-Agha I, Eikema DJ, Zinger N, Haenel M, Schaap N, Arroyo CH, Schuermans C, Besemer B, Engelhardt M, Kuball J, Michieli M, Schub N, Wilson KMO, Bourhis JH, Mateos MV, Rabin N, Jost E, Kröger N, Moraleda JM, Za T, Hayden PJ, Beksac M, Mclornan D, Schönland S, Manier S. Carfilzomib, lenalidomide and dexamethasone followed by a second ASCT is an effective strategy in first relapse multiple myeloma: a study on behalf of the Chronic malignancies working party of the EBMT. *Bone Marrow Transplant*. 2023 Nov;58(11):1182-1188. doi: 10.1038/s41409-023-02048-7. Epub 2023 Aug 5. PMID: 37543712; PMCID: PMC10622318.
69. Vieri M, Tharmapalan V, Kalmer M, Baumeister J, Nikolić M, Schnitker M, Kirschner M, Flosdorf N, de Toledo MAS, Zenke M, Koschmieder S, Brümmendorf TH, Beier F, Wagner W. Cellular aging is accelerated in the malignant clone of myeloproliferative neoplasms. *Blood Cancer J*. 2023 Nov 6;13(1):164. doi: 10.1038/s41408-023-00936-1. PMID: 37926720; PMCID: PMC10625927.
70. Wang YM, Kaj-Carbaidwala B, Lane A, Agarwal S, Beier F, Bertuch A, Borovsky KA, Brennan SK, Calado RT, Catto LFB, Dufour C, Ebens CL, Fioredda F, Giri N, Gloude N, Goldman F, Hertel PM, Himes R, Keel SB, Koura DT, Kratz CP, Kulkarni S, Liou I, Nakano TA, Nastasio S, Niewisch MR, Penrice DD, Sasa GS, Savage SA, Simonetto DA, Ziegler DS, Miethke AG, Myers KC; Clinical Care Consortium for Telomere-associated Ailments (CCCTAA). Liver disease and transplantation in telomere biology disorders: An international multicenter cohort. *Hepatol Commun*. 2024 Jun 19;8(7):e0462. doi: 10.1097/HC9.0000000000000462. PMID: 38896081; PMCID: PMC11186813.
71. Zeremski V, Adolph L, Beer S, Berisha M, Jacobs B, Kahl C, Koenecke C, Kropf S, Panse J, Petersen J, Schmidt-Hieber M, Schneider J, Vucinic V, Walter J, Weigert O, Witte HM, Mougiakakos D. Relevance of different prognostic scores in primary CNS lymphoma in the era of intensified treatment regimens: A retrospective, multicenter analysis of 174 patients. *Eur J Haematol*. 2024 Apr;112(4):641-649. doi: 10.1111/ejh.14159. Epub 2024 Jan 2. PMID: 38164819.
72. Häm: Lübke J, Christen D, Schwaab J, Kaiser A, Naumann N, Shoumariyeh K, Jentzsch M, Sockel K, Schaffrath J, Ayuk FA, Stelljes M, Hilgendorf I, Sala E, Kaivers J, Schönland S, Wittke C, Hertenstein B, Radsak M, Kaiser U, Brückl V, Kröger N, Brümmendorf TH, Hofmann WK, Klein S, Jost E, Reiter A, Panse J. Allogeneic Hematopoietic Cell Transplantation in Advanced Systemic Mastocytosis: A retrospective analysis of the DRST and GREM registries. *Leukemia*. 2024 Apr;38(4):810-821. doi: 10.1038/s41375-024-02186-x. Epub 2024 Mar 6. PMID: 38448757; PMCID: PMC10997505.

Seltene Lebererkrankungen und gastrointestinale Erkrankungen

73. Bednarsch J, Lang SA, Heise D, Strnad P, Neumann UP, Ulmer TF. Laparoscopic Living donor liver transplantation in irresectable intrahepatic cholangiocarcinoma in primary sclerosing cholangitis associated liver cirrhosis. *Z Gastroenterol.* 2024 Jan;62(1):50-55. English. doi: 10.1055/a-2221-6126. Epub 2024 Jan 9. PMID: 38195108.
74. Berg T, Aehling NF, Bruns T, Welker MW, Weismüller T, Trebicka J, Tacke F, Strnad P, Sterneck M, Settmacher U, Seehofer D, Schott E, Schnitzbauer AA, Schmidt HH, Schlitt HJ, Pratschke J, Pascher A, Neumann U, Manekeller S, Lammert F, Klein I, Kirchner G, Guba M, Glanemann M, Engelman C, Canbay AE, Braun F, Berg CP, Bechstein WO, Becker T, Trautwein C; Collaborators: S2k-Leitlinie Lebertransplantation der Deutschen Gesellschaft für Gastroenterologie, Verdauungs- und Stoffwechselkrankheiten (DGVS) und der Deutschen Gesellschaft für Allgemein- und Viszeralchirurgie (DGAV). *Z Gastroenterol.* 2024 Sep;62(9):1397-1573. German. doi: 10.1055/a-2255-7246. Epub 2024 Sep 9. PMID: 39250961.
75. Calderaro J, Ghaffari Laleh N, Zeng Q, Maille P, Favre L, Pujals A, Klein C, Bazille C, Heij LR, Uguen A, Luedde T, Di Tommaso L, Beaufrère A, Chatain A, Gastineau D, Nguyen CT, Nguyen-Canh H, Thi KN, Gnemmi V, Graham RP, Charlotte F, Wendum D, Vij M, Allende DS, Aucejo F, Diaz A, Rivière B, Herrero A, Evert K, Calvisi DF, Augustin J, Leow WQ, Leung HHW, Boleslawski E, Rela M, François A, Cha AW, Forner A, Reig M, Allaire M, Scatton O, Chatelain D, Boulagnon-Rombi C, Sturm N, Menahem B, Frouin E, Tougeron D, Tournigand C, Kempf E, Kim H, Ningarhari M, Michalak-Provost S, Gopal P, Brustia R, Vibert E, Schulze K, Rütther DF, Weidemann SA, Rhaïem R, Pawlotsky JM, Zhang X, Luciani A, Mulé S, Laurent A, Amaddeo G, Regnault H, De Martin E, Sempoux C, Navale P, Westerhoff M, Lo RC, Bednarsch J, Gouw A, Guettier C, Lequoy M, Harada K, Sripongpun P, Wetwittayaklang P, Loménie N, Tantipisit J, Kaewdech A, Shen J, Paradis V, Caruso S, Kather JN. Deep learning-based phenotyping reclassifies combined hepatocellular-cholangiocarcinoma. *Nat Commun.* 2023 Dec 14;14(1):8290. doi: 10.1038/s41467-023-43749-3. PMID: 38092727; PMCID: PMC10719304.
76. Chen L, Elizalde M, Dubois LJ, Roeth AA, Neumann UP, Olde Damink SWM, Schaap FG, Alvarez-Sola G. GAL3ST1 Deficiency Reduces Epithelial-Mesenchymal Transition and Tumorigenic Capacity in a Cholangiocarcinoma Cell Line. *Int J Mol Sci.* 2024 Jul 2;25(13):7279. doi: 10.3390/ijms25137279. PMID: 39000386; PMCID: PMC11242791.
77. Clark VC, Strange C, Strnad P, Sanchez AJ, Kwo P, Pereira VM, van Hoek B, Barjaktarevic I, Corsico AG, Pons M, Goldklang M, Gray M, Kuhn B, Vargas HE, Vierling JM, Vuppalanchi R, Brantly M, Kappe N, Chang T, Schlupe T, Zhou R, Hamilton J, San Martin J, Loomba R. Fazirsiran for Adults With Alpha-1 Antitrypsin Deficiency Liver Disease: A Phase 2 Placebo Controlled Trial (SEQUOIA). *Gastroenterology.* 2024 Oct;167(5):1008-1018.e5. doi: 10.1053/j.gastro.2024.06.028. Epub 2024 Jul 2. PMID: 38964420.

78. Fromme M, Hamesch K, Schneider CV, Mandorfer M, Pons M, Thorhauge KH, Pereira V, Sperl J, Frankova S, Reichert MC, Benini F, Burbaum B, Kleinjans M, Amzou S, Rademacher L, Bewersdorf L, Verbeek J, Nevens F, Genesca J, Miravittles M, Nuñez A, Schaefer B, Zoller H, Janciauskiene S, Waern J, Oliveira A, Maia L, Simões C, Mahadeva R, Fraughen DD, Trauner M, Krag A, Lammert F, Bals R, Gaisa NT, Aigner E, Griffiths WJ, Denk H, Teumer A, McElvaney NG, Turner AM, Trautwein C, Strnad P. Alpha-1 Antitrypsin Augmentation and the Liver Phenotype of Adults With Alpha-1 Antitrypsin Deficiency (Genotype Pi*ZZ). *Clin Gastroenterol Hepatol*. 2024 Feb;22(2):283-294.e5. doi: 10.1016/j.cgh.2023.08.038. Epub 2023 Sep 15. PMID: 37716616.
79. Fromme M, Rademacher L, Amzou S, Cook CD, Zacharias I, Zhang L, Ripollone JE, Strnad P. Association of circulating Z-polymer with adverse clinical outcomes and liver fibrosis in adults with alpha-1 antitrypsin deficiency. *United European Gastroenterol J*. 2024 Oct;12(8):1091-1101. doi: 10.1002/ueg2.12629. Epub 2024 Jul 18. PMID: 39024029; PMCID: PMC11485299.
80. Fromme M, Schneider CV, Guldiken N, Amzou S, Luo Y, Pons M, Genesca J, Miravittles M, Thorhauge KH, Mandorfer M, Waern J, Schneider KM, Sperl J, Frankova S, Bartel M, Zimmer H, Zorn M, Krag A, Turner A, Trautwein C, Strnad P. Alcohol consumption and liver phenotype of individuals with alpha-1 antitrypsin deficiency. *Liver Int*. 2024 Oct;44(10):2660-2671. doi: 10.1111/liv.16044. Epub 2024 Jul 19. PMID: 39031304.
81. Hohlstein P, Salvarcioglu C, Pollmanns MR, Adams JK, Abu Jhaisha S, Kabak E, Eisert A, Hamesch K, Weiskirchen R, Koch A, Wirtz TH. Diagnostic and Prognostic Value of Serum Leptin in Critically Ill Patients with Acute versus Acute-on-Chronic Liver Failure. *Biomedicines*. 2024 May 24;12(6):1170. doi: 10.3390/biomedicines12061170. PMID: 38927377; PMCID: PMC11200812.
82. Kabak E, Clusmann J, Abu Jhaisha S, Hohlstein P, Adams J, Kernbach J, Drexler S, Schneider CV, Schwenger C, Wirtz TH, Hamesch K, Saritas T, Trautwein C, Pollmanns MR, Koch A. An unusual case of intracerebral hemorrhage: exploring the link with Sneddon's syndrome. *Med Klin Intensivmed Notfmed*. 2024 Feb;119(1):66-68. English. doi: 10.1007/s00063-023-01059-y. Epub 2023 Sep 13. PMID: 37702782.
83. Koelfat KVK, Schaap FG, van Mierlo KMC, Leníček M, Sauer I, van der Kroft G, Röth AAJ, Bednarsch J, Amygdalos I, Lurje G, Dewulf MJL, Lang SA, Neumann UP, Olde Damink SWM. Partial liver resection alters the bile salt-FGF19 axis in patients with perihilar cholangiocarcinoma: Implications for liver regeneration. *Hepatol Commun*. 2024 Jun 5;8(6):e0445. doi: 10.1097/HC9.0000000000000445. PMID: 38836805; PMCID: PMC11155560.
84. Koop PH, Schwenger C, Clusmann J, Vell MS, Jaeger J, Gui W, Trautwein C, Koch A, Bruns T, Schneider CV, Schneider KM. Comorbidities, mortality and metabolic profile in individuals with primary biliary cholangitis-A Phenome- Wide-Association-Study. *Liver Int*. 2024 Aug;44(8):2038-2053. doi: 10.1111/liv.15945. Epub 2024 Apr 25. PMID: 38661318.
85. Kroh A, Walter J, Fragoulis A, Möckel D, Lammers T, Kiessling F, Andruszkow J, Preisinger C, Egbert M, Jiao L, Eickhoff RM, Heise D, Berndt N, Cramer T, Neumann UP, Egners A, Ulmer TF. Hepatocellular loss of mTOR aggravates tumor burden in nonalcoholic steatohepatitis-related HCC. *Neoplasia*. 2023 Dec;46:100945. doi: 10.1016/j.neo.2023.100945. Epub 2023 Nov 15. PMID: 37976569; PMCID: PMC10685311.

86. Loomba R, Clark G, Teckman J, Ajmera V, Behling C, Brantly M, Brenner D, D'Armiento J, Fried MW, Iyer JS, Mandorfer M, Rockey DC, Tincopa M, Vuppalanchi R, Younossi Z, Krag A, Turner AM, Strnad P. Review article: New developments in biomarkers and clinical drug development in alpha-1 antitrypsin deficiency- related liver disease. *Aliment Pharmacol Ther*. 2024 May;59(10):1183-1195. doi: 10.1111/apt.17967. Epub 2024 Mar 22. PMID: 38516814.
87. Lorenz P, Aehling N, Bruns T, Bechstein W, Becker T, Berg T, Freudenberger P, Trautwein C, Klug L, Lynen Jansen P. Leitlinienreport der S2k-Leitlinie Lebertransplantation der Deutschen Gesellschaft für Gastroenterologie, Verdauungs- und Stoffwechselkrankheiten (DGVS) und der Deutschen Gesellschaft für Allgemein und Viszeralchirurgie (DGAV). *Z Gastroenterol*. 2024 Sep;62(9):e537-e554. German. doi: 10.1055/a-2255-7355. Epub 2024 Sep 9. PMID: 39250959.
88. Mantas A, Liu D, Otto CC, Heij LR, Heise D, Bruners P, Lang SA, Ulmer TF, Neumann UP, Bednarsch J. Time to surgery is not an oncological risk factor in patients with cholangiocarcinoma undergoing curative-intent liver surgery. *Sci Rep*. 2024 Jan 18;14(1):1644. doi: 10.1038/s41598-023-50842-6. PMID: 38238432; PMCID: PMC10796920.
89. Mantas A, Otto CC, Olthof PB, Heise D, Hoyer DP, Bruners P, Dewulf M, Lang SA, Ulmer TF, Neumann UP, Bednarsch J. Clinical features and prediction of long- term survival after surgery for perihilar cholangiocarcinoma. *PLoS One*. 2024 Jul 1;19(7):e0304838. doi: 10.1371/journal.pone.0304838. PMID: 38950006; PMCID: PMC11216605.
90. Mohamed MR, Haybaeck J, Wu H, Su H, Bartneck M, Lin C, Boeschoten MV, Boor P, Goeppert B, Rupp C, Strnad P, Davis RJ, Cubero FJ, Trautwein C. JNKs protect from cholestatic liver disease progression by modulating Apelin signalling. *JHEP Rep*. 2023 Jul 18;5(11):100854. doi: 10.1016/j.jhepr.2023.100854. PMID: 37791376; PMCID: PMC10543210.
91. Otto CC, Mantas A, Heij LR, Heise D, Dewulf M, Lang SA, Ulmer TF, Dahl E, Bruners P, Neumann UP, Bednarsch J. Preoperative predictors for non- resectability in perihilar cholangiocarcinoma. *World J Surg Oncol*. 2024 Feb 7;22(1):48. doi: 10.1186/s12957-024-03329-1. PMID: 38326854; PMCID: PMC10851609.
92. Ratti F, Marino R, Olthof PB, Pratschke J, Erdmann JI, Neumann UP, Prasad R, Jarnagin WR, Schnitzbauer AA, Cescon M, Guglielmi A, Lang H, Nadalin S, Topal B, Maithel SK, Hoogwater FJH, Alikhanov R, Troisi R, Sparrelid E, Roberts KJ, Malagò M, Hagendoorn J, Malik HZ, Olde Damink SWM, Kazemier G, Schadde E, Charco R, de Reuver PR, Groot Koerkamp B, Aldrighetti L; Perihilar Cholangiocarcinoma Collaboration Group. Predicting futility of upfront surgery in perihilar cholangiocarcinoma: Machine learning analytics model to optimize treatment allocation. *Hepatology*. 2024 Feb 1;79(2):341-354. doi: 10.1097/HEP.0000000000000554. Epub 2023 Aug 3. PMID: 37530544.
93. van Keulen AM, Olthof PB, Buettner S, Bednarsch J, Verheij J, Erdmann JI, Nooijen LE, Porte RJ, Minnee RC, Murad SD, Neumann UP, Heij L, Groot Koerkamp B, Doukas M. The Influence of Hepatic Steatosis and Fibrosis on Postoperative Outcomes After Major Liver Resection of Perihilar Cholangiocarcinoma. *Ann Surg Oncol*. 2024 Jan;31(1):133-141. doi: 10.1245/s10434-023-14419-x. Epub 2023 Oct 29. PMID: 37899413; PMCID: PMC10695871.

94. Volkert I, Fromme M, Schneider C, Candels L, Lindhauer C, Su H, Thorhauge K, Pons M, Mohamed MR, Schneider KM, Strnad P, Trautwein C. Impact of PNPLA3 I148M on alpha-1 antitrypsin deficiency-dependent liver disease progression. *Hepatology*. 2024 Apr 1;79(4):898-911. doi: 10.1097/HEP.0000000000000574. Epub 2023 Aug 25. PMID: 37625151.
95. Wang G, Mantas A, Heij LR, Al-Masri TM, Liu D, Heise D, Schmitz SM, Olde Damink SWM, Luedde T, Lang SA, Ulmer TF, Neumann UP, Bednarsch J. Body composition is associated with postoperative complications in perihilar cholangiocarcinoma. *Cancer Med*. 2024 Jan;13(1):e6878. doi: 10.1002/cam4.6878. Epub 2024 Jan 1. PMID: 38164056; PMCID: PMC10807576.
96. Wang G, Otto CC, Heij LR, Al-Masri TM, Dahl E, Heise D, Olde Damink SWM, Luedde T, Lang SA, Ulmer TF, Neumann UP, Bednarsch J. Impact of Altered Body Composition on Clinical and Oncological Outcomes in Intrahepatic Cholangiocarcinoma. *J Clin Med*. 2023 Dec 18;12(24):7747. doi: 10.3390/jcm12247747. PMID: 38137817; PMCID: PMC10744221.
97. Wiegand J, Franke A, Stein K, Trautwein C, Berg T. Letter to the Editor: Improving access to transient elastography data for real-world prognostic applications in primary biliary cholangitis. *Hepatology*. 2024 May 1;79(5):E136-E137. doi: 10.1097/HEP.0000000000000744. Epub 2023 Dec 27. PMID: 38150002.
98. Xu T, Herkens L, Jia T, Klinkhammer BM, Kant S, Krusche CA, Buhl EM, Hayat S, Floege J, Strnad P, Kramann R, Djurdjaj S, Boor P. The role of desmoglein-2 in kidney disease. *Kidney Int*. 2024 May;105(5):1035-1048. doi: 10.1016/j.kint.2024.01.037. Epub 2024 Feb 21. PMID: 38395410.

Seltene Nieren-Erkrankungen des Erwachsenen

99. Anders HJ, Fernandez-Juarez GM, Vaglio A, Romagnani P, Floege J. CKD therapy to improve outcomes of immune-mediated glomerular diseases. *Nephrol Dial Transplant*. 2023 Nov 8;38(Supplement_2):ii50-ii57. doi: 10.1093/ndt/gfad069. Erratum in: *Nephrol Dial Transplant*. 2024 Apr 08:gfae072. doi: 10.1093/ndt/gfae072. PMID: 37218706.
100. Barratt J, Lafayette RA, Floege J. Therapy of IgA nephropathy: time for a paradigm change. *Front Med (Lausanne)*. 2024 Aug 15;11:1461879. doi: 10.3389/fmed.2024.1461879. PMID: 39211339; PMCID: PMC11358106.
101. Caravaca-Fontán F, Del Vecchio L, Praga M, Floege J, Zoccali C. Sodium glucose co-transporter 2 inhibitors in the treatment of glomerular diseases: a <i>CKJ</i> controversy. *Clin Kidney J*. 2024 Aug 12;17(9):sfaf237. doi: 10.1093/ckj/sfaf237. PMID: 39228996; PMCID: PMC11367167.

102. Caravaca-Fontán F, Stevens K, Padrón M, Huerta A, Montomoli M, Villa J, González F, Vega C, López Mendoza M, Fernández L, Shabaka A, Rodríguez-Moreno A, Martín-Gómez A, Labrador PJ, Molina Andújar A, Prados Soler MC, Martín-Penagos L, Yerovi E, Medina Zahonero L, De La Flor JC, Mon C, Ibernón M, Rodríguez Gómez A, Miquel R, Sierra M, Mascarós V, Luzardo L, Papatotiriou M, Arroyo D, Verdalles Ú, Martínez-Miguel P, Ramírez-Guerrero G, Pampa-Saico S, Moral Berrio E, Canga JLP, Tarragón B, Fraile Gómez P, Regidor D, Relea J, Xipell M, Andrades Gómez C, Navarro M, Álvarez Á, Rivas B, Quintana LF, Gutiérrez E, Pérez-Valdivia MÁ, Odler B, Kronbichler A, Geddes C, Anders HJ, Floege J, Fernández-Juárez G, Praga M. Sodium-glucose cotransporter 2 inhibition in primary and secondary glomerulonephritis. *Nephrol Dial Transplant*. 2024 Jan 31;39(2):328-340. doi: 10.1093/ndt/gfad175. PMID: 37550217.
103. Casal Moura M, Gauckler P, Anders HJ, Bruchfeld A, Fernandez-Juarez GM, Floege J, Frangou E, Goumenos D, Segelmark M, Turkmen K, van Kooten C, Tesar V, Geetha D, Fervenza FC, Jayne DRW, Stevens KI, Kronbichler A. Management of antineutrophil cytoplasmic antibody-associated vasculitis with glomerulonephritis as proposed by the ACR 2021, EULAR 2022 and KDIGO 2021 guidelines/recommendations. *Nephrol Dial Transplant*. 2023 Oct 31;38(11):2637-2651. doi: 10.1093/ndt/gfad090. PMID: 37164940; PMCID: PMC10615627.
104. Cattran DC, Floege J, Coppo R. Evaluating Progression Risk in Patients With Immunoglobulin A Nephropathy. *Kidney Int Rep*. 2023 Sep 22;8(12):2515-2528. doi: 10.1016/j.ekir.2023.09.020. PMID: 38106572; PMCID: PMC10719597.
105. Chia-Gil A, Floege J, Stamellou E, Moeller MJ. Perihilar FSGS lesions originate from flat parietal epithelial cells. *J Nephrol*. 2024 Jun;37(5):1405-1409. doi: 10.1007/s40620-024-01886-y. Epub 2024 Feb 1. PMID: 38300433; PMCID: PMC11405489.
106. Floege J, Jayne DRW, Sanders JF, Tesar V, Balk EM, Gordon CE, Adam G, Tonelli MA, Cheung M, Earley A, Rovin BH. Executive summary of the KDIGO 2024 Clinical Practice Guideline for the Management of ANCA-Associated Vasculitis. *Kidney Int*. 2024 Mar;105(3):447-449. doi: 10.1016/j.kint.2023.10.009. Erratum in: *Kidney Int*. 2024 Jul;106(1):163-164. doi: 10.1016/j.kint.2024.04.004. PMID: 38388147.
107. Floege J, Jayne DRW, Sanders JF, Tesar V, Balk EM, Gordon CE, Adam G, Tonelli MA, Cheung M, Earley A, Rovin BH. Corrigendum to "Executive summary of the KDIGO 2024 Clinical Practice Guideline for the Management of ANCA-Associated Vasculitis." *Kidney International* 2024;105(3):447-449. *Kidney Int*. 2024 Jul;106(1):163-164. doi: 10.1016/j.kint.2024.04.004. Epub 2024 May 13. Erratum for: *Kidney Int*. 2024 Mar;105(3):447-449. doi: 10.1016/j.kint.2023.10.009. PMID: 38739060.
108. Floege J. A new alternative: inhibiting complement activation in patients with IgA nephropathy. *Kidney Int*. 2024 Jan;105(1):28-30. doi: 10.1016/j.kint.2023.10.012. PMID: 38182298.
109. Floege J. IgA-Nephropathie [IgA nephropathy]. *Inn Med (Heidelb)*. 2023 Oct;64(10):961-969. German. doi: 10.1007/s00108-023-01588-w. Epub 2023 Sep 6. PMID: 37672089.
110. Floege J. IgA-Nephropathie [IgA nephropathy]. *Urologie*. 2024 Jan;63(1):103-111. German. doi: 10.1007/s00120-023-02268-1. PMID: 38170257.

111. Kronbichler A, Barnini C, Matyjek A, Gauckler P, Bruchfeld A, Caravaca- Fontan F, [Floege J](#), Frangou E, Mirioglou S, Moran SM, Stevens KI, Teng YKO, Steiger S. Antibody-mediated podocytopathies: a disease entity that implies immunotherapy. *Nephrol Dial Transplant*. 2024 Jul 17:gfae166. doi: 10.1093/ndt/gfae166. Epub ahead of print. PMID: 39020250.
112. Lafayette R, Barbour S, Israni R, Wei X, Eren N, [Floege J](#), Jha V, Kim SG, Maes B, Phoon RKS, Singh H, Tesař V, Lin CJF, Barratt J. A phase 2b, randomized, double-blind, placebo-controlled, clinical trial of atacicept for treatment of IgA nephropathy. *Kidney Int*. 2024 Jun;105(6):1306-1315. doi: 10.1016/j.kint.2024.03.012. Epub 2024 Mar 27. PMID: 38552841.
113. [Rauen T](#), Vogt K, Krämer S. Vaskulitiden [Vasculitides]. *Schmerz*. 2024 Feb;38(1):28-32. German. doi: 10.1007/s00482-023-00760-3. Epub 2023 Oct 12. PMID: 37828257.
114. [Rauen T](#), Vogt K, Seikrit C. Corticosteroids in Patients With IgA Nephropathy: Is the Lower the Better? *Kidney Int Rep*. 2024 May 29;9(7):1966-1968. doi: 10.1016/j.ekir.2024.05.031. PMID: 39081733; PMCID: PMC11284487.
115. Rovin BH, Barratt J, Heerspink HJL, Alpers CE, Bieler S, Chae DW, Diva UA, [Floege J](#), Gesualdo L, Inrig JK, Kohan DE, Komers R, Kooienga LA, Lafayette R, Maes B, Matecki R, Mercer A, Noronha IL, Oh SW, Peh CA, Praga M, Preciado P, Radhakrishnan J, Rheault MN, Rote WE, Tang SCW, Tesar V, Trachtman H, Trimarchi H, Tumlin JA, Wong MG, Perkovic V; DUPRO steering committee and PROTECT Investigators. Efficacy and safety of sparsentan versus irbesartan in patients with IgA nephropathy (PROTECT): 2-year results from a randomised, active- controlled, phase 3 trial. *Lancet*. 2023 Dec 2;402(10417):2077-2090. doi: 10.1016/S0140-6736(23)02302-4. Epub 2023 Nov 3. PMID: 37931634.
116. Stamellou E, Nadal J, Hendry B, Mercer A, Bechtel-Walz W, Schiffer M, Eckardt KU, Kramann R, [Moeller MJ](#), [Floege J](#); GCKD study investigators. Long-term outcomes of adults with FSGS in the German Chronic Kidney Disease cohort. *Clin Kidney J*. 2024 Apr 27;17(7):sfae131. doi: 10.1093/ckj/sfae131. PMID: 38989280; PMCID: PMC11234294.
117. Stamellou E, Nadal J, Hendry B, Mercer A, Seikrit C, Bechtel-Walz W, Schmid M, [Moeller MJ](#), Schiffer M, Eckardt KU, Kramann R, [Floege J](#); GCKD study investigators. Long-term outcomes of patients with IgA nephropathy in the German CKD cohort. *Clin Kidney J*. 2024 Jul 22;17(8):sfae230. doi: 10.1093/ckj/sfae230. PMID: 39149090; PMCID: PMC11324945.
118. Stamellou E, Seikrit C, Tang SCW, Boor P, Tesař V, [Floege J](#), Barratt J, Kramann R. IgA nephropathy. *Nat Rev Dis Primers*. 2023 Nov 30;9(1):67. doi: 10.1038/s41572-023-00476-9. PMID: 38036542.
119. Vogt K, Fink CB, Schreibung TM, Krämer S, Reinartz S, [Rauen T](#). Distinct pulmonary patterns in ANCA-associated vasculitides: insights from a retrospective single center cohort study. *Rheumatol Int*. 2024 Nov;44(11):2435-2443. doi: 10.1007/s00296-024-05664-8. Epub 2024 Aug 13. PMID: 39136785.
120. Wimbury D, Muto M, Bhachu JS, Scionti K, Brown J, Molyneux K, Seikrit C, Maixnerová D, Pérez-Alós L, Garred P, [Floege J](#), Tesař V, Fellstrom B, Coppo R, Barratt J. Targeted-release budesonide modifies key pathogenic biomarkers in immunoglobulin A nephropathy: insights from the NEFIGAN trial. *Kidney Int*. 2024 Feb;105(2):381-388. doi: 10.1016/j.kint.2023.11.003. Epub 2023 Nov 25. PMID: 38008160.

Seltene entzündlich vermittelte Erkrankungen des Kindes- und Jugendalters

121. Chen S, Abou-Khalil BW, Afawi Z, Ali QZ, Amadori E, Anderson A, Anderson J, Andrade DM, Annesi G, Arslan M, Auce P, Bahlo M, Baker MD, Balagura G, Balestrini S, Banks E, Barba C, Barboza K, Bartolomei F, Bass N, Baum LW, Baumgartner TH, Baykan B, Bebek N, Becker F, Bennett CA, Beydoun A, Bianchini C, Bisulli F, Blackwood D, Blatt I, Borggräfe I, Bosselmann C, Braatz V, Brand H, Brockmann K, Buono RJ, Busch RM, Caglayan SH, Canafoglia L, Canavati C, Castellotti B, Cavalleri GL, Cerrato F, Chassoux F, Cherian C, Cherny SS, Cheung CL, Chou IJ, Chung SK, Churchhouse C, Ciullo V, Clark PO, Cole AJ, Cosico M, Cossette P, Cotsapas C, Cusick C, Daly MJ, Davis LK, Jonghe P, Delanty N, Dennig D, Depondt C, Derambure P, Devinsky O, Di Vito L, Dickerson F, Dlugos DJ, Doccini V, Doherty CP, El-Naggar H, Ellis CA, Epstein L, Evans M, Faucon A, Feng YA, Ferguson L, Ferraro TN, Da Silva IF, Ferri L, Feucht M, Fields MC, Fitzgerald M, Fonferko-Shadrach B, Fortunato F, Franceschetti S, French JA, Freri E, Fu JM, Gabriel S, Gagliardi M, Gambardella A, Gauthier L, Giangregorio T, Gili T, Glauser TA, Goldberg E, Goldman A, Goldstein DB, Granata T, Grant R, Greenberg DA, Guerrini R, Gundogdu-Eken A, Gupta N, Haas K, Hakonarson H, Haryanyan G, Häusler M, Hegde M, Heinzen EL, Helbig I, Hengsbach C, Heyne H, Hirose S, Hirsch E, Ho CJ, Hoepfer O, Howrigan DP, Hucks D, Hung PC, Iacomino M, Inoue Y, Inuzuka LM, Ishii A, Jehi L, Johnson MR, Johnstone M, Kälviäinen R, Kanaan M, Kara B, Kariuki SM, Kegele J, Kesim Y, Khoueiry-Zgheib N, Khoury J, King C, Klein KM, Kluger G, Knake S, Kok F, Korczyn AD, Korinthenberg R, Koupparis A, Kousiappa I, Krause R, Krenn M, Krestel H, Krey I, Kunz WS, Kurlemann G, Kuzniecky RI, Kwan P, La Vega-Talbott M, Labate A, Lacey A, Lal D, Laššuthová P, Lauxmann S, Lawthom C, Leech SL, Lehesjoki AE, Lemke JR, Lerche H, Lesca G, Leu C, Lewin N, Lewis-Smith D, Li GH, Liao C, Licchetta L, Lin CH, Lin KL, Linnankivi T, Lo W, Lowenstein DH, Lowther C, Lubbers L, Lui CHT, Macedo- Souza LI, Madeleyn R, Madia F, Magri S, Maillard L, Marcuse L, Marques P, Marson AG, Matthews AG, May P, Mayer T, McArdle W, McCarroll SM, McGoldrick P, McGraw CM, McIntosh A, McQuillan A, Meador KJ, Mei D, Michel V, Millichap JJ, Minardi R, Montomoli M, Mostacci B, Muccioli L, Muhle H, Müller-Schlüter K, Najm IM, Nasreddine W, Neaves S, Neubauer BA, Newton CRJC, Noebels JL, Northstone K, Novod S, O'Brien TJ, Owusu-Agyei S, Özkara Ç, Palotie A, Papacostas SS, Parrini E, Pato C, Pato M, Pendziwiat M, Pennell PB, Petrovski S, Pickrell WO, Pinsky R, Pinto D, Pippucci T, Piras F, Piras F, Poduri A, Pondrelli F, Posthuma D, Powell RHW, Privitera M, Rademacher A, Ragona F, Ramirez-Hamouz B, Rau S, Raynes HR, Rees MI, Regan BM, Reif A, Reinthaler E, Rheims S, Ring SM, Riva A, Rojas E, Rosenow F, Ryvlin P, Saarela A, Sadleir LG, Salman B, Salmon A, Salpietro V, Sammarra I, Scala M, Schachter S, Schaller A, Schankin CJ, Scheffer IE, Schneider N, Schubert-Bast S, Schulze-Bonhage A, Scudieri P, Sedláčková L, Shain C, Sham PC, Shiedley BR, Siena SA, Sills GJ, Sisodiya SM, Smoller JW, Solomonson M, Spalletta G, Sparks KR, Sperling MR, Stamberger H, Steinhoff BJ, Stephani U, Štěřbová K, Stewart WC, Stipa C, Striano P, Strzelczyk A, Surges R, Suzuki T, Talarico M, Talkowski ME, Taneja RS, Tanteles GA, Timonen O, Timpson NJ, Tinuper P, Todaro M, Topaloglu P, Tsai MH, Tumiene B, Turkdogan D, Uğur-İşeri S, Utkus A, Vaidiswaran P, Valton L, van Baalen A, Vari MS, Vetro A, Vlčková M, von Brauchitsch S, von Spiczak S, Wagner RG, Watts N, Weber YG, Weckhuysen S, Widdess-Walsh P, Wiebe S, Wolf SM, Wolff M, Wolking S, Wong I, von Wrede R, Wu D, Yamakawa K, Yapıcı Z, Yis U, Yolken R, Yücesan E, Zagaglia S, Zahnert F, Zara F, Zimprich F, Zizovic M, Zsurka G, Neale BM, Berkovic SF. Exome sequencing of 20,979 individuals with epilepsy reveals shared and distinct ultra-rare genetic risk across disorder subtypes. medRxiv [Preprint]. 2024 Sep 20:2023.02.22.23286310. doi:

10.1101/2023.02.22.23286310. Update in: Nat Neurosci. 2024 Oct;27(10):1864-1879. doi: 10.1038/s41593-024-01747-8. PMID: 36865150; PMCID: PMC9980234.

122. Horneff G, Minden K, Foell D, Klotsche J, Tenbrock K; PROKIND- Arbeitsgruppe. Protokolle in der Kinderrheumatologie (PROKIND): Treat-to-Target bei polyartikulärer juveniler idiopathischer Arthritis [Protocols in pediatric rheumatology (PROKIND): treat-to-target in polyarticular juvenile idiopathic arthritis]. Z Rheumatol. 2024 Feb;83(1):15-27. German. doi: 10.1007/s00393-023-01452-0. Epub 2023 Dec 29. PMID: 38157052.

Retinopathien und Retinopathie-Syndrome

123. Lange CA, Ohlmeier C, Kiskämper A, von Schwarzkopf C, Hufnagel H, Gruber M, Schworm B, Brocks U, Reinking F, Schreiner L, Miura Y, Grundel M, Lohmann T, Clemens CR, Gamulescu MA, Eter N, Grisanti S, Priglinger S, Spitzer MS, Walter P, Agostini HA, Stahl A, Pauleikhoff LJB; Retina.net CSC-Registry-Study Group. Clinical Landscape of Central Serous Chorioretinopathy in Germany: Retina.net CSC Registry Report Number 1. Ophthalmologica. 2024;247(2):95-106. doi: 10.1159/000535930. Epub 2024 Feb 16. PMID: 38368867; PMCID: PMC11160426.
124. Pfeil JM, Barth T, Lagrèze WA, Lorenz B, Hufendiek K, Liegl R, Breuss H, Bemme S, Aisenbrey S, Glitz B, Süsskind D, Gabel-Pfisterer A, Skevas C, Krohne TU, Kakkassery V, Bründer MC, Engelmann K, Guthoff R, Walter P, Choritz L, Stahl A; Retina.net ROP Registry Study Group. Treated Cases of Retinopathy of Prematurity in Germany: 10-Year Data from the Retina.net Retinopathy of Prematurity Registry. Ophthalmol Retina. 2024 Jun;8(6):579-589. doi: 10.1016/j.oret.2023.12.002. Epub 2023 Dec 16. PMID: 38104929.

Syndromale Erkrankungen und kindliche Atemregulationsstörungen

125. Amin AK, Krause J, Eggermann T. 11p13 microduplication: a differential diagnosis of Silver-Russell syndrome? Mol Cytogenet. 2024 Mar 14;17(1):5. doi: 10.1186/s13039-024-00672-6. PMID: 38486332; PMCID: PMC10941370.
126. Asif M, Khayyat AIA, Alawbathani S, Abdullah U, Sanner A, Georgomanolis T, Haasters J, Becker K, Budde B, Becker C, Thiele H, Baig SM, Isidoro-García M, Winter D, Pogoda HM, Muhammad S, Hammerschmidt M, Kraft F, Kurth I, Martin HG, Wagner M, Nürnberg P, Hussain MS. Biallelic loss-of-function variants of ZFTRAF1 cause neurodevelopmental disorder with microcephaly and hypotonia. Genet Med. 2024 Jul;26(7):101143. doi: 10.1016/j.gim.2024.101143. Epub 2024 Apr 16. PMID: 38641995.
127. Bhasin MA, Knaus A, Incardona P, Schmid A, Holtgrewe M, Elbracht M, Krawitz PM, Hsieh TC. Enhancing Variant Prioritization in VarFish through On-Premise Computational Facial Analysis. Genes (Basel). 2024 Mar 17;15(3):370. doi: 10.3390/genes15030370. PMID: 38540429; PMCID: PMC10969976.
128. Brunet T, Zott B, Lieftüchter V, Lenz D, Schmidt A, Peters P, Kopajtich R, Zaddach M, Zimmermann H, Hüning I, Ballhausen D, Stauffer C, Bianzano A, Hughes J, Taylor RW, McFarland R, Devlin A, Mihaljević M, Barišić N, Rohlf M, Wilfling S, Sondheimer N, Hewson S, Marinakis NM, Kosma K, Traeger-Synodinos J, Elbracht M, Begemann M, Trepels-Kottek S, Hasan D, Scala M, Capra V, Zara F, van der Ven AT, Driemeyer J, Apitz C, Krämer J, Strong A,

- Hakonarson H, Watson D, Mayr JA, Prokisch H, Meitinger T, Borggraefe I, Spiegler J, Baric I, Paolini M, Gerstl L, Wagner M. De novo variants in RNF213 are associated with a clinical spectrum ranging from Leigh syndrome to early-onset stroke. *Genet Med*. 2024 Feb;26(2):101013. doi: 10.1016/j.gim.2023.101013. Epub 2023 Nov 1. PMID: 37924258.
129. Eggermann T. Human Reproduction and Disturbed Genomic Imprinting. *Genes (Basel)*. 2024 Jan 26;15(2):163. doi: 10.3390/genes15020163. PMID: 38397153; PMCID: PMC10888310.
130. Huang X, Henck J, Qiu C, Sreenivasan VKA, Balachandran S, Amarie OV, Hrabě de Angelis M, Behncke RY, Chan WL, Despang A, Dickel DE, Duran M, Feuchtinger A, Fuchs H, Gailus-Durner V, Haag N, Hägerling R, Hansmeier N, Hennig F, Marshall C, Rajderkar S, Ringel A, Robson M, Saunders LM, da Silva-Buttkus P, Spielmann N, Srivatsan SR, Ulferts S, Wittler L, Zhu Y, Kalscheuer VM, Ibrahim DM, Kurth I, Kornak U, Visel A, Pennacchio LA, Beier DR, Trapnell C, Cao J, Shendure J, Spielmann M. Single-cell, whole-embryo phenotyping of mammalian developmental disorders. *Nature*. 2023 Nov;623(7988):772-781. doi: 10.1038/s41586-023-06548-w. Epub 2023 Nov 15. PMID: 37968388; PMCID: PMC10665194.
131. Kastellan S, Kalb R, Sajjad B, McReynolds LJ, Giri N, Samuel D, Milde T, Elbracht M, Holzhauser S, Niewisch MR, Kratz CP. Germline biallelic BRCA2 pathogenic variants and medulloblastoma: an international cohort study. *J Hematol Oncol*. 2024 Apr 29;17(1):26. doi: 10.1186/s13045-024-01547-4. PMID: 38685107; PMCID: PMC11057105.
132. Lischka A, Eggermann K, Record CJ, Dohrn MF, Laššuthová P, Kraft F, Begemann M, Dey D, Eggermann T, Beijer D, Šoukalová J, Laura M, Rossor AM, Mazanec R, Van Lent J, Tomaselli PJ, Ungelenk M, Debus KY, Feely SME, Gläser D, Jagadeesh S, Martin M, Govindaraj GM, Singhi P, Baineni R, Biswal N, Ibarra-Ramírez M, Bonduelle M, Gess B, Romero Sánchez J, Suthar R, Udani V, Nalini A, Unnikrishnan G, Marques W Junior, Mercier S, Procaccio V, Bris C, Suresh B, Reddy V, Skorupinska M, Bonello-Palot N, Mochel F, Dahl G, Sasidharan K, Devassikutty FM, Nampoothiri S, Rodovalho Doriqui MJ, Müller-Felber W, Vill K, Haack TB, Dufke A, Abele M, Stucka R, Siddiqi S, Ullah N, Spranger S, Chiabrando D, Bolgöl BS, Parman Y, Seeman P, Lampert A, Schulz JB, Wood JN, Cox JJ, Auer-Grumbach M, Timmerman V, de Winter J, Themistocleous AC, Shy M, Bennett DL, Baets J, Hübner CA, Leipold E, Züchner S, Elbracht M, Çakar A, Senderek J, Hornemann T, Woods CG, Reilly MM, Kurth I. Genetic landscape of congenital insensitivity to pain and hereditary sensory and autonomic neuropathies. *Brain*. 2023 Dec 1;146(12):4880-4890. doi: 10.1093/brain/awad328. PMID: 37769650; PMCID: PMC10689924.
133. Mackay DJG, Gazdagh G, Monk D, Brioude F, Giabicani E, Krzyzewska IM, Kalish JM, Maas SM, Kagami M, Beygo J, Kahre T, Tenorio-Castano J, Ambrozaitytė L, Burnytė B, Cerrato F, Davies JH, Ferrero GB, Fjodorova O, Manero-Azua A, Pereda A, Russo S, Tannorella P, Temple KI, Ōunap K, Riccio A, de Nancrales GP, Maher ER, Lapunzina P, Netchine I, Eggermann T, Blik J, Tümer Z. Multi-locus imprinting disturbance (MLID): interim joint statement for clinical and molecular diagnosis. *Clin Epigenetics*. 2024 Aug 1;16(1):99. doi: 10.1186/s13148-024-01713-y. PMID: 39090763; PMCID: PMC11295890.
134. Menzel M, Martis-Thiele M, Goldschmid H, Ott A, Romanovsky E, Siemanowski-Hrach J, Seillier L, Bröchle NO, Maurer A, Lehmann KV, Begemann M, Elbracht M, Meyer R, Dintner S, Claus R, Meier-Kolthoff JP, Blanc E, Möbs M, Joosten M, Benary M, Basitta P, Hölscher F, Tischler V, Groß T, Kutz O, Prause R, William D, Horny K, Goering W, Sivalingam S, Borkhardt A, Blank C, Junk SV, Yasin L, Moskalev EA, Carta MG, Ferrazzi F, Tögel L, Wolter S, Adam E, Matysiak U, Rosenthal T, Dönitz J, Lehmann U, Schmidt G, Bartels S, Hofmann W, Hirsch S,

- Dikow N, Göbel K, Banan R, Hamelmann S, Fink A, Ball M, Neumann O, Rehker J, Kloth M, Murtagh J, Hartmann N, Jurmeister P, Mock A, Kumbrink J, Jung A, Mayr EM, Jacob A, Trautmann M, Kirmse S, Falkenberg K, Ruckert C, Hirsch D, Immel A, Dietmaier W, Haack T, Marienfeld R, Fürstberger A, Niewöhner J, Gerstenmaier U, Eberhardt T, Greif PA, Appenzeller S, Maurus K, Doll J, Jelting Y, Jonigk D, Märkl B, Beule D, Horst D, Wulf AL, Aust D, Werner M, Reuter-Jessen K, Ströbel P, Auber B, Sahm F, Merkelbach-Bruse S, Siebolts U, Roth W, Lassmann S, Klauschen F, Gaisa NT, Weichert W, Evert M, Armeanu-Ebinger S, Ossowski S, Schroeder C, Schaaf CP, Malek N, Schirmacher P, Kazdal D, Pfarr N, Budczies J, Stenzinger A. Benchmarking whole exome sequencing in the German network for personalized medicine. *Eur J Cancer*. 2024 Nov;211:114306. doi: 10.1016/j.ejca.2024.114306. Epub 2024 Sep 8. PMID: 39293347.
135. Moch J, Radtke M, Liehr T, Eggermann T, Gilissen C, Pfundt R, Astuti G, Hentschel J, Schumann I. Automated detection of uniparental disomies in a large cohort. *Hum Genet*. 2024 Aug;143(8):955-964. doi: 10.1007/s00439-024-02687-w. Epub 2024 Jul 16. PMID: 39012485; PMCID: PMC11303498.
136. Prawitt D, Eggermann T. Molecular mechanisms of human overgrowth and use of *omics* in its diagnostics: chances and challenges. *Front Genet*. 2024 Jun 4;15:1382371. doi: 10.3389/fgene.2024.1382371. PMID: 38894719; PMCID: PMC11183334.
137. Rigter PMF, de Konink C, Dunn MJ, Proietti Onori M, Humberson JB, Thomas M, Barnes C, Prada CE, Weaver KN, Ryan TD, Caluseriu O, Conway J, Calamaro E, Fong CT, Wuyts W, Meuwissen M, Hordijk E, Jonkers CN, Anderson L, Yuseinova B, Polonia S, Beysen D, Stark Z, Savva E, Poulton C, McKenzie F, Bhoj E, Bupp CP, Bézieau S, Mercier S, Blevins A, Wentzensen IM, Xia F, Rosenfeld JA, Hsieh TC, Krawitz PM, Elbracht M, Veenma DCM, Schulman H, Stratton MM, Küry S, van Woerden GM. Role of CAMK2D in neurodevelopment and associated conditions. *Am J Hum Genet*. 2024 Feb 1;111(2):364-382. doi: 10.1016/j.ajhg.2023.12.016. Epub 2024 Jan 24. PMID: 38272033; PMCID: PMC10870144.
138. Schlaich E, Hubens WHG, Eggermann T. First-time application of droplet digital PCR for methylation testing of the 11p15.5 imprinting regions. *Mol Genet Genomic Med*. 2023 Dec;11(12):e2264. doi: 10.1002/mgg3.2264. Epub 2023 Jul 31. PMID: 37519217; PMCID: PMC10724498.
139. Schmetz A, Lüdecke HJ, Surowy H, Sivalingam S, Bruel AL, Caumes R, Charles P, Chatron N, Chrzanowska K, Codina-Solà M, Colson C, Cuscó I, Denommé-Pichon AS, Edery P, Faivre L, Green A, Heide S, Hsieh TC, Hustinx A, Kleinendorst L, Knopp C, Kraft F, Krawitz PM, Lasa-Aranzasti A, Lesca G, López-González V, Maraval J, Mignot C, Neuhann T, Netzer C, Oehl-Jaschkowitz B, Petit F, Philippe C, Posmyk R, Putoux A, Reis A, Sánchez-Soler MJ, Suh J, Tkemaladze T, Tran Mau Them F, Travessa A, Trujillano L, Valenzuela I, van Haelst MM, Vasileiou G, Vincent-Delorme C, Walther M, Verde P, Bramswig NC, Wieczorek D. Delineation of the adult phenotype of Coffin-Siris syndrome in 35 individuals. *Hum Genet*. 2024 Jan;143(1):71-84. doi: 10.1007/s00439-023-02622-5. Epub 2023 Dec 20. PMID: 38117302.
140. Venger K, Elbracht M, Carlens J, Deutz P, Zeppernick F, Lassay L, Kratz C, Zenker M, Kim J, Stewart DR, Wieland I, Schultz KAP, Schwerk N, Kurth I, Kontny U. Unusual phenotypes in patients with a pathogenic germline variant in DICER1. *Fam Cancer*. 2023 Oct;22(4):475-480. doi: 10.1007/s10689-021-00271-z. Epub 2021 Jul 31. PMID: 34331184; PMCID: PMC9743360.

Seltene Allergien und Hauterkrankungen

141. Didona D, Scarsella L, Hudemann C, Volkmann K, Zimmer CL, Beckert B, Tikkanen R, Korff V, Kühn K, Wienzek-Lischka S, Bein G, Di Zenzo G, Böhme J, Cunha T, Solimani F, Pieper J, Juratli HA, Göbel M, Schmidt T, Borradori L, Yazdi AS, Sitaru C, Garn H, Eming R, Fleischer S, Hertl M. Type 2 T-Cell Responses against Distinct Epitopes of the Desmoglein 3 Ectodomain in Pemphigus Vulgaris. *J Invest Dermatol*. 2024 Feb;144(2):263-272.e8. doi: 10.1016/j.jid.2023.07.025. Epub 2023 Sep 16. PMID: 37717934.

Seltene erbliche Tumorerkrankungen

142. Makowska A, Kontny U, Weiskirchen R. HeLa cells cross-contaminated nasopharyngeal carcinoma cell lines: Still a common problem. *Br J Cancer*. 2024 Jun;130(12):1885-1886. doi: 10.1038/s41416-024-02675-x. Epub 2024 Apr 25. PMID: 38664578; PMCID: PMC11182765.
143. Römer T, Vokuhl C, Staatz G, Mottaghy FM, Christiansen H, Eble MJ, Timmermann B, Klusmann JP, Elbracht M, Calaminus G, Zimmermann M, Brümmendorf TH, Feuchtinger T, Kerp H, Kontny U. Combination of nivolumab with standard induction chemotherapy in children and adults with EBV-positive nasopharyngeal carcinoma : Protocol of a prospective multicenter phase 2 trial. *HNO*. 2024 Jun;72(6):423-439. doi: 10.1007/s00106-023-01404-9. Epub 2024 Jan 12. PMID: 38214716; PMCID: PMC11116201.

Gesichts- und Skelettfehlbildungen

144. Lecker N, Höhn T, Rossaint R, Orlikowsky T, Trepels-Kottek S. Strategien zur Atemwegssicherung bei Neugeborenen : Ergebnisse einer Befragung deutscher Perinatalzentren der Level I und II [Strategies for airway management in neonates : Results of a survey of German perinatal centers levels II and III]. *Anaesthesiologie*. 2023 Oct;72(10):703-709. German. doi: 10.1007/s00101-023-01317-w. Epub 2023 Jul 5. PMID: 37405472.

Arrhythmogene rechtsventrikuläre Kardiomyopathien und Keratinopathien

145. Fabritz L, Fortmueller L, Gehmlich K, Kant S, Kemper M, Kucerova D, Syeda F, Faber C, Leube RE, Kirchhof P, Krusche CA. Endurance Training Provokes Arrhythmogenic Right Ventricular Cardiomyopathy Phenotype in Heterozygous Desmoglein-2 Mutants: Alleviation by Preload Reduction. *Biomedicines*. 2024 Apr 30;12(5):985. doi: 10.3390/biomedicines12050985. PMID: 38790949; PMCID: PMC11117820.

Geschäftsstelle ZSE

146. Börsch N, Mücke M, Maier A, Conrad R, Pantel JT, Sellin J, Mani K, Chopra P. Treating pain in patients with Ehlers-Danlos syndrome : Multidisciplinary management of a multisystemic disease. *Schmerz*. 2024 Feb;38(1):12-18. English. doi: 10.1007/s00482-023-00778-7. Epub 2024 Jan 8. PMID: 38189943.

147. Cuhls H, Hesse M, Heuser G, Radbruch L, Ateş G. Meaning in life of terminally ill parents with minor children compared to palliative care patients – a quantitative analysis using SMiLE. *Palliat Care Soc Pract*. 2024 Jul 31;18:26323524241264883. doi: 10.1177/26323524241264883. PMID: 39086470; PMCID: PMC11289801.
148. 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. 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*. 2023 Oct 6;65:102260. doi: 10.1016/j.eclinm.2023.102260. PMID: 37855024; PMCID: PMC10579280.
149. Ibaldo LC, Witte V, Klawonn F, Conrad R, Mücke M, Sellin J, Teschke M. Suggestion of a new standard in measuring the mandible via MRI and an overview of reference values in young women. *Oral Maxillofac Surg*. 2024 Mar;28(1):373-383. doi: 10.1007/s10006-023-01153-7. Epub 2023 Apr 26. Erratum in: *Oral Maxillofac Surg*. 2024 Mar;28(1):457. doi: 10.1007/s10006-023-01164-4. PMID: 37099046; PMCID: PMC10914874.
150. Ibaldo LC, Witte V, Klawonn F, Conrad R, Mücke M, Sellin J, Teschke M. Correction to: Suggestion of a new standard in measuring the mandible via MRI and an overview of reference values in young women. *Oral Maxillofac Surg*. 2024 Mar;28(1):457. doi: 10.1007/s10006-023-01164-4. Erratum for: *Oral Maxillofac Surg*. 2024 Mar;28(1):373-383. doi: 10.1007/s10006-023-01153-7. PMID: 37296276; PMCID: PMC10914908.
151. La Rocca LA, Frank J, Bentzen HB, Pantel JT, Gerischer K, Bovier A, Krawitz PM. Understanding recessive disease risk in multi-ethnic populations with different degrees of consanguinity. *Am J Med Genet A*. 2024 Mar;194(3):e63452. doi: 10.1002/ajmg.a.63452. Epub 2023 Nov 3. PMID: 37921563.
152. Lesmann H, Hustinx A, Moosa S, Klinkhammer H, Marchi E, Caro P, Abdelrazek IM, Pantel JT, Hagen MT, Thong MK, Binti Mazlan RA, Tae SK, Kamphans T, Meiswinkel W, Li JM, Javanmardi B, Knaus A, Uwineza A, Knopp C, Tkemaladze T, Elbracht M, Mattern L, Jamra RA, Velmans C, Strehlow V, Jacob M, Peron A, Dias C, Nunes BC, Vilella T, Pinheiro IF, Kim CA, Melaragno MI, Weiland H, Kaptain S, Chwiałkowska K, Kwasniewski M, Saad R, Wiethoff S, Goel H, Tang C, Hau A, Barakat TS, Panek P, Nabil A, Suh J, Braun F, Gomy I, Averdunk L, Ekure E, Bergant G, Peterlin B, Graziano C, Gaboon N, Fiesco-Roa M, Spinelli AM, Wilpert NM, Phowthongkum P, Güzel N, Haack TB, Bitar R, Tzschach A, Rodriguez-Palmero A, Brunet T, Rudnik-Schöneborn S, Contreras-Capetillo SN, Oberlack A, Samango-Sprouse C, Sadeghin T, Olaya M, Platzer K, Borovikov A, Schnabel F, Heuft L, Herrmann V, Oegema R, Elkhateeb N, Kumar S, Komlosi K, Mohamed K, Kalantari S, Sirchia F, Martinez-Monseny AF, Höller M, Toutouna L, Mohamed A, Lasa-Aranzasti A, Sayer JA, Ehmke N, Danyel M, Sczakiel H, Schwartzmann S, Boschann F, Zhao M, Adam R, Einicke L, Horn D, Chew KS, Kam CC, Karakoyun M, Pode-Shakked B, Eliyahu A, Rock R, Carrion T, Chorin O, Zarate YA, Conti MM, Karakaya M, Tung ML, Chandra B, Bouman A, Lumaka A, Wasif N, Shinawi M, Blackburn PR, Wang T, Niehues T, Schmidt A, Roth RR, Wiczorek D, Hu P, Waikel RL, Ledgister Hanchard SE, Elmakkawy G, Safwat S, Ebstein F, Krüger E, Küry S, Bézieau S, Arlt A, Olinger E, Marbach F, Li D, Dupuis L, Mendoza-Londono R, Houge SD, Weis D, Chung BH, Mak CCY, Kayserili H, Elcioglu N, Aykut A, Şimşek-Kiper PÖ, Bögershausen N, Wollnik B, Bentzen HB,

- Kurth I, Netzer C, Jezela-Stanek A, Devriendt K, Gripp KW, Mücke M, Verloes A, Schaaf CP, Nellåker C, Solomon BD, Nöthen MM, Abdalla E, Lyon GJ, Krawitz PM, Hsieh TC. GestaltMatcher Database - A global reference for facial phenotypic variability in rare human diseases. Res Sq [Preprint]. 2024 Jun 10;rs.3.rs-4438861. doi: 10.21203/rs.3.rs-4438861/v1. PMID: 38903062; PMCID: PMC11188141.
153. Mangal AL, Mücke M, Rolke R, Appelmann I. Advance directives in amyotrophic lateral sclerosis - a systematic review and meta-analysis. BMC Palliat Care. 2024 Jul 29;23(1):191. doi: 10.1186/s12904-024-01524-1. PMID: 39075493; PMCID: PMC11285133.
154. Mücke M, Börsch N. Seltene Erkrankungen und Schmerzen – ein oft übersehenes Zusammenspiel [Rare diseases and pain-A frequently overlooked interaction]. Schmerz. 2024 Feb;38(1):3-5. German. doi: 10.1007/s00482-024-00789-y. Epub 2024 Feb 2. PMID: 38305884.
155. Reiter AMV, Pantel JT, Danyel M, Horn D, Ott CE, Mensah MA. Validation of 3 Computer-Aided Facial Phenotyping Tools (DeepGestalt, GestaltMatcher, and D-Score): Comparative Diagnostic Accuracy Study. J Med Internet Res. 2024 Mar 13;26:e42904. doi: 10.2196/42904. PMID: 38477981; PMCID: PMC10973953.
156. Schmidt A, Danyel M, Grundmann K, Brunet T, Klinkhammer H, Hsieh TC, Engels H, Peters S, Knaus A, Moosa S, Averdunk L, Boschann F, Sczakiel HL, Schwartzmann S, Mensah MA, Pantel JT, Holtgrewe M, Bösch A, Weiß C, Weinhold N, Suter AA, Stoltenburg C, Neugebauer J, Kallinich T, Kaindl AM, Holzhauer S, Bühner C, Bufler P, Kornak U, Ott CE, Schülke M, Nguyen HHP, Hoffjan S, Grasemann C, Rothoefel T, Brinkmann F, Matar N, Sivalingam S, Perne C, Mangold E, Kreiss M, Cremer K, Betz RC, Mücke M, Grigull L, Klockgether T, Spier I, Heimbach A, Bender T, Brand F, Stieber C, Morawiec AM, Karakostas P, Schäfer VS, Bernsen S, Weydt P, Castro-Gomez S, Aziz A, Grobe-Einsler M, Kimmich O, Kobeleva X, Önder D, Lesmann H, Kumar S, Tacik P, Basin MA, Incardona P, Lee-Kirsch MA, Berner R, Schuetz C, Körholz J, Kretschmer T, Di Donato N, Schröck E, Heinen A, Reuner U, Hanßke AM, Kaiser FJ, Manka E, Munteanu M, Kuechler A, Cordula K, Hirtz R, Schlapakow E, Schlein C, Lisfeld J, Kubisch C, Herget T, Hempel M, Weiler-Normann C, Ullrich K, Schramm C, Rudolph C, Rillig F, Groffmann M, Muntau A, Tibelius A, Schwaibold EMC, Schaaf CP, Zawada M, Kaufmann L, Hinderhofer K, Okun PM, Kotzaeridou U, Hoffmann GF, Choukair D, Bettendorf M, Spielmann M, Ripke A, Pauly M, Münchau A, Lohmann K, Hüning I, Hanker B, Bäumer T, Herzog R, Hellenbroich Y, Westphal DS, Strom T, Kovacs R, Riedhammer KM, Mayerhanser K, Graf E, Brugger M, Hoefele J, Oexle K, Mirza-Schreiber N, Berutti R, Schatz U, Krenn M, Makowski C, Weigand H, Schröder S, Rohlf s M, Vill K, Hauck F, Borggraefe I, Müller-Felber W, Kurth I, Elbracht M, Knopp C, Begemann M, Kraft F, Lemke JR, Hentschel J, Platzer K, Strehlow V, Abou Jamra R, Kehrer M, Demidov G, Beck-Wödl S, Graessner H, Sturm M, Zeltner L, Schöls LJ, Magg J, Bevot A, Kehrer C, Kaiser N, Turro E, Horn D, Grüters-Kieslich A, Klein C, Mundlos S, Nöthen M, Riess O, Meitinger T, Krude H, Krawitz PM, Haack T, Ehmke N, Wagner M. Next-generation phenotyping integrated in a national framework for patients with ultrarare disorders improves genetic diagnostics and yields new molecular findings. Nat Genet. 2024 Aug;56(8):1644-1653. doi: 10.1038/s41588-024-01836-1. Epub 2024 Jul 22. PMID: 39039281; PMCID: PMC11319204.
157. Sellin J, Pantel JT, Börsch N, Conrad R, Mücke M. Kurze Wege zur Diagnose mit künstlicher Intelligenz – systematische Literaturrecherche zu „diagnostic decision support systems“ [Short paths to diagnosis with artificial intelligence: systematic literature review on diagnostic decision

support systems]. Schmerz. 2024 Feb;38(1):19-27. German. doi: 10.1007/s00482-023-00777-8. Epub 2024 Jan 2. PMID: 38165492.

158. Steinfeld E, Schneegans K, Benstoem C, Hesse M, Dohmen S. Bürgerzentrierte Gesundheitsversorgung in Deutschland – eine Analyse von ExpertInneninterviews [Citizen-centred health care in Germany: An analysis of expert interviews]. Z Evid Fortbild Qual Gesundhwes. 2024 Jun;187:34-41. German. doi: 10.1016/j.zefq.2024.03.010. Epub 2024 May 11. PMID: 38735813.
159. Woestemeier A, Semaan A, Block A, Arensmeyer J, Dohmen J, Kania A, Verrel F, Mücke M, Kalff JC, Lingohr P. Prognostic factors for the long term outcome after surgical celiac artery decompression in MALS. Orphanet J Rare Dis. 2023 Oct 23;18(1):334. doi: 10.1186/s13023-023-02952-7. PMID: 37872625; PMCID: PMC10594872.
160. Zoch M, Gierschner C, Andreeff AK, Henke E, Sedlmayr M, Müller G, Tippmann J, Hebestreit H, Choukair D, Hoffmann GF, Fritz-Kebede F, Toepfner N, Berner R, Biergans S, Verbücheln R, Schaaf J, Fleck J, Wirth FN, Schepers J, Prasser F. Secondary use of patient data within decentralized studies using the example of rare diseases in Germany: A data scientist's exploration of process and lessons learned. Digit Health. 2024 Aug 10;10:20552076241265219. doi: 10.1177/20552076241265219. PMID: 39130526; PMCID: PMC11316959.