# Publikationen 2025, an denen das ZESE und seine Typ B-Zentren mitgearbeitet haben

**Typ A-Zentrum**

Brandstetter LS, Ungethüm K, Heuschmann PU, Pfister L, Deckert J, de Zwaan M, Tüscher O, Witt S, Krauth C, Berger A, Berneburg M, Deibele A, Dieris-Hirche J, Heuft G, Imhof C, Schulz JB, Zipfel S, Hebestreit H, Haas K. Feasibility of the development and psychometric properties of a standardized screening instrument for mental disorders in patients with suspected rare diseases: results of the ZSE-DUO study. Front Psychiatry. 2025 Nov 10;16:1624474. doi: 10.3389/fpsyt.2025.1624474. PMID: 41293202; PMCID: PMC12641394.

Dieris-Hirche J, Herpertz S, Hebestreit H, Bârlescu L, Berger A, Brandstetter L, Deckert J, de Greck M, Gündel H, Haas K, Heinrich I, Herrmann-Werner A, Hett J, Heuft G, Koschitzki K, Krauth C, Lücke T, Maisch T, Mundlos C, Nöhre M, Pfister L, Rutsch F, Schippers C, Schubert K, Schulz JB, Tüscher O, Vogel M, Witt S, Zipfel S, de Zwaan M. Mental disorders in people with undiagnosed diseases presenting to German centres for rare diseases - prevalence, type and relevance. Psychiatry Res. 2025 Jul;349:116509. doi: 10.1016/j.psychres.2025.116509. Epub 2025 Apr 19. PMID: 40286780.

Sauerbruch T, Aretz S, Hebestreit H, Kaemmerer H, Nährlich L, Siegmund B, Ertl G; Deutsche Gesellschaft für Innere Medizin (DGIM). Seltene Erkrankungen – die Rolle der Inneren Medizin [Rare diseases: the role of internal medicine]. Inn Med (Heidelb). 2025 May;66(5):533-539. German. doi: 10.1007/s00108-025-01892-7. Epub 2025 Apr 24. PMID: 40272470; PMCID: PMC12053190.

**Christiane Herzog-Zentrum für Mukoviszidose Unterfranken CHZ**

Rumpf CH, Janssen T, Hait RJ, Romme K, Decker C, Peters J, Czowalla C, Deiwick S, Görlich D, Dübbers A, Große-Onnebrink J, Kessler C, Küster P, Schültingkemper H, Hebestreit H, Graepler-Mainka U, van Koningsbruggen-Rietschel S, Schildberg T, Renner S, Nährig S, Wollschläger B, Schlegtendal A, Sutharsan S, Stehling F, Junge S, Ballmann M, Schwartbeck B, Kahl BC. Mucoid Staphylococcus aureus: A Clinical Trial of Its Prevalence and Association with Lung Function in People with Cystic Fibrosis. Am J Respir Crit Care Med. 2025 May;211(5):854-865. doi: 10.1164/rccm.202407-1474OC. PMID: 39918841.

Urquhart DS, Jamieson P, Burns P, Braun J, Hebestreit H, Radtke T. Feasibility and inter-reporter variability of submaximal outcomes derived from cardiopulmonary exercise testing in people with advanced cystic fibrosis lung disease. ERJ Open Res. 2025 Jun 30;11(3):01001-2024. doi: 10.1183/23120541.01001-2024. PMID: 40589908; PMCID: PMC12208563.

Williams CA, Button B, Dwyer TJ, Hatziagorou E, Mackintosh KA, McNarry MA, Paris D, Shelley J, Hebestreit H, Bradley J, Silveira Bianchim M; Exercise Working Group European CF Society. The 2025 European Cystic Fibrosis Society position statement on physical activity assessment in cystic fibrosis. Eur Respir Rev. 2025 Jul 9;34(177):240279. doi: 10.1183/16000617.0279-2024. PMID: 40633976; PMCID: PMC12249234.

**Craniofaciales Centrum Würzburg (CFC)**

Borst A, Schweitzer T, Horn D, Kunstmann E, König EM, Pluta N, Klopocki E. Functional analyses of splice site variants in TCF12. Hum Genomics. 2025 Apr 26;19(1):45. doi: 10.1186/s40246-025-00758-1. PMID: 40287710; PMCID: PMC12034182.

**Fabry Zentrum für interdisziplinäre Therapie Würzburg (FAZiT)**

Hahn P, Lechner W, Siefen RG, Lampe C, Nordbeck P, Grigull L, Lücke T. Improving a data mining based diagnostic support tool for rare diseases on the example of M. Fabry: Gender differences need to be taken into account. PLoS One. 2025 Jun 30;20(6):e0326372. doi: 10.1371/journal.pone.0326372. PMID: 40587431; PMCID: PMC12208464.

Hessenauer F, Nauroth-Kreß AC, Spitzel M, Kemmerzell S, Weiner S, Schindehütte M, Sommer C, Üçeyler N, Pham M, Kampf T. In vivo imaging of dorsal root ganglia in the mouse: from ex vivo MR-microscopy towards quantitative in vivo MRI. Brain Res. 2025 Oct 15;1865:149872. doi: 10.1016/j.brainres.2025.149872. Epub 2025 Aug 8. PMID: 40784619.

Hughes DA, Sunder-Plassmann G, Jovanovic A, Brand E, West ML, Bichet DG, Pisani A, Nowak A, Torra R, Khan A, Azevedo O, Lehman A, Linhart A, Rutecki J, Giuliano JD, Krusinska E, Nordbeck P. Renal and multisystem effectiveness of 3.9 years of migalastat in a global real-world cohort: Results from the followME Fabry Pathfinders registry. J Inherit Metab Dis. 2025 Jan;48(1):e12771. doi: 10.1002/jimd.12771. Epub 2024 Jul 19. PMID: 39031114; PMCID: PMC11730455.

Kallish S, Camporeale A, Hopkin RJ, Jovanovic A, Nordbeck P, Veleva-Rotse BO, Krusinska E, Torra R. Long-term efficacy of migalastat in females with Fabry disease. J Med Genet. 2025 Nov 21;62(12):798-807. doi: 10.1136/jmg-2024-110596. PMID: 40897525; PMCID: PMC12703307.

Karl-Schöller F, Breyer M, Klopocki E, Üçeyler N. Generation of a gene-corrected human isogenic iPSC line from a patient with Fabry disease carrying the GLA variant c.1069C>T using CRISPR/Cas9-mediated homology directed repair. Stem Cell Res. 2025 Aug;86:103711. doi: 10.1016/j.scr.2025.103711. Epub 2025 Apr 10. PMID: 40222082.

Kmoch S, Živná M, Dvela-Levitt M, Braun F, Rozenfeld P, Wanner C, Hughes D, Schiffmann R, Warnock DG. Expanded View of the Pathophysiology of Fabry Disease. Nephron. 2025;149(10):620-624. doi: 10.1159/000546555. Epub 2025 Jun 30. PMID: 40587960.

Weiner S, Perleth S, Kampf T, Lau K, Hessenauer F, Homola G, Nordbeck P, Üçeyler N, Sommer C, Pham M, Schindehütte M. MRI T2 Mapping of Dorsal Root Ganglia Reveals Increased T2 Relaxation Time in Classical Fabry Disease. Biomedicines. 2025 Feb 28;13(3):592. doi: 10.3390/biomedicines13030592. PMID: 40149569; PMCID: PMC11940213.

**Interdisziplinäres Amyloidosezentrum Nordbayern**

Cejka V, Hosp C, Steinhardt M, Papagianni A, Ihne-Schubert S, Scholz N, Chen M, Schäfer J, Adrah A, Kortüm M, Sommer C, Einsele H, Frantz S, Störk S, Morbach C. Cardiovascular determinants of the 6-minute walk distance in cardiac transthyretin amyloidosis. BMC Cardiovasc Disord. 2025 Nov 14;25(1):810. doi: 10.1186/s12872-025-05215-4. PMID: 41239245; PMCID: PMC12619313.

Fontana M, Berk JL, Gillmore JD, Witteles RM, Grogan M, Drachman B, Damy T, Garcia-Pavia P, Taubel J, Solomon SD, Sheikh FH, Tahara N, González-Costello J, Tsujita K, Morbach C, Pozsonyi Z, Petrie MC, Delgado D, Van der Meer P, Jabbour A, Bondue A, Kim D, Azevedo O, Hvitfeldt Poulsen S, Yilmaz A, Jankowska EA, Algalarrondo V, Slugg A, Garg PP, Boyle KL, Yureneva E, Silliman N, Yang L, Chen J, Eraly SA, Vest J, Maurer MS; HELIOS-B Trial Investigators. Vutrisiran in Patients with Transthyretin Amyloidosis with Cardiomyopathy. N Engl J Med. 2025 Jan 2;392(1):33-44. doi: 10.1056/NEJMoa2409134. Epub 2024 Aug 30. PMID: 39213194.

Jering KS, Fontana M, Lairez O, Longhi S, Azevedo O, Morbach C, Bender S, Jay PY, Vest J, Bulwer BE, Prasad N, Solomon SD, Skali H. Effects of vutrisiran on cardiac structure and function in patients with transthyretin amyloidosis with cardiomyopathy: secondary outcomes of the HELIOS-B trial. Nat Med. 2025 Oct;31(10):3560-3568. doi: 10.1038/s41591-025-03851-z. Epub 2025 Aug 6. PMID: 40770082; PMCID: PMC12532587.

Maurer MS, Berk JL, Damy T, Sheikh FH, González-Costello J, Morbach C, Delgado D, Bondue A, Azevedo O, Poulsen SH, Jankowska EA, Yang L, Bender S, Eraly SA, Jay PY, Vest J, Fontana M. Impact of Vutrisiran on Cardiac Biomarkers in Patients With Transthyretin Amyloidosis With Cardiomyopathy From HELIOS-B. J Am Coll Cardiol. 2025 Aug 12;86(6):459-475. doi: 10.1016/j.jacc.2025.04.055. PMID: 40769675.

Maurer MS, Witteles RM, Garcia-Pavia P, Sheikh FH, Morbach C, Rodriguez Duque D, Aldinc E, Eraly SA, Gillmore JD. Impact of Heart Failure Severity on Vutrisiran Efficacy in Transthyretin Amyloidosis With Cardiomyopathy. J Am Coll Cardiol. 2025 May 27;85(20):1927-1939. doi: 10.1016/j.jacc.2025.03.477. Epub 2025 Mar 17. PMID: 40099776.

Nies RJ, Ney S, Kindermann I, Bewarder Y, Zimmer A, Knebel F, Hahn K, Spethmann S, Luedike P, Michel L, Rassaf T, Papathanasiou M, Störk S, Cejka V, Polzin A, Voss F, Kelm M, Unsöld B, Meindl C, Paulus M, Yilmaz A, Chamling B, Morbach C, Pfister R. Real-world characteristics and treatment of cardiac transthyretin amyloidosis: A multicentre, observational study. ESC Heart Fail. 2025 Apr;12(2):1203-1216. doi: 10.1002/ehf2.15126. Epub 2024 Nov 6. PMID: 39505353; PMCID: PMC11911619.

Witteles RM, Garcia-Pavia P, Damy T, Grogan M, Sheikh FH, Morbach C, Bender S, Exter J, Eraly SA, Fontana M. Vutrisiran Improves Survival and Reduces Cardiovascular Events in ATTR Amyloid Cardiomyopathy: HELIOS-B. J Am Coll Cardiol. 2025 Apr 30:S0735-1097(25)06170-4. doi: 10.1016/j.jacc.2025.04.008. Epub ahead of print. PMID: 40380962.

Witteles RM, Garcia-Pavia P, Morbach C, Gillmore JD, Taylor MS, Conceição I, White WB, Kwok C, Sweetser MT, Boyle KL, Adams D. Vutrisiran in Transthyretin Amyloidosis: A Pooled Safety Analysis of HELIOS-A and HELIOS-B. JACC Adv. 2025 Sep;4(9):102066. doi: 10.1016/j.jacadv.2025.102066. Epub 2025 Aug 22. PMID: 40848527; PMCID: PMC12397930.

**Würzburger Zentrum für Neurofibromatose (WZNF)**

Meller E, Amann A, Malik RA, Kampik D, Matthies C, Üçeyler N. Clinical characterization of neuropathic pain and small fiber impairment in neurofibromatosis. Pain Rep. 2025 May 6;10(3):e1285. doi: 10.1097/PR9.0000000000001285. PMID: 40342626; PMCID: PMC12058648.

Schindehütte M, Meller E, Kampf T, Hessenauer F, Üçeyler N, Homola G, Rittner HL, Matthies C, Pham M, Weiner S. Quantitative MRI of dorsal root ganglion alterations in neurofibromatosis type 1 patients with or without pain. Eur Radiol Exp. 2025 May 28;9(1):57. doi: 10.1186/s41747-025-00594-x. PMID: 40437318; PMCID: PMC12119455.

**Zentrum für angeborene Blutzellerkrankung**

Althaus K, Hoepner G, Zieger B, Prüller F, Pavlova A, Boeckelmann D, Birschmann I, Müller J, Rühl H, Sachs U, Kehrel B, Streif W, Bugert P, Zaninetti C, Cooper N, Schulze H, Knöfler R, Bakchoul T, Jurk K. The Diagnostic Assessment of Platelet Function Defects - Part 2: Update on Platelet Disorders. Hamostaseologie. 2025 Jun;45(3):243-253. doi: 10.1055/a-2404-0216. Epub 2025 Jan 27. PMID: 39870108.

Gebetsberger J, Knöfler R, Streif W; ThromKidplus study group#; and mandated experts$ from other participating professional societies. Erratum: "Diagnosis of Inherited Platelet Disorders": Update of the Interdisciplinary S2k-Guideline [*] of the Permanent Pediatric Commission of the Society of Thrombosis and Haemostasis Research (GTH e.V.). Hamostaseologie. 2025 Aug;45(4). doi: 10.1055/s-0045-1811974. Epub 2025 Sep 19. Erratum for: Hamostaseologie. 2025 Aug;45(4):347-354. doi: 10.1055/a-2628-5488. PMID: 40973074.

Gebetsberger J, Knöfler R, Streif W; ThromKidplus study group#; and mandated experts$ from other participating professional societies. "Diagnosis of Inherited Platelet Disorders": Update of the Interdisciplinary S2k-Guideline [ ] of the Permanent Pediatric Commission of the Society of Thrombosis and Haemostasis Research (GTH e.V.). Hamostaseologie. 2025 Aug;45(4):347-354. English. doi: 10.1055/a-2628-5488. Epub 2025 Aug 12. Erratum in: Hamostaseologie. 2025 Aug;45(4). doi: 10.1055/s-0045-1811974. PMID: 40795927.

Hoepner G, Althaus K, Müller J, Zieger B, Pavlova A, Boeckelmann D, Knöfler R, Bugert P, Kehrel B, Streif W, Birschmann I, Rühl H, Sachs U, Prüller F, Zaninetti C, Schulze H, Cooper N, Jurk K, Bakchoul T. The Diagnostic Assessment of Inherited Platelet Function Defects - Part 1: An Overview of the Diagnostic Approach and Laboratory Methods. Hamostaseologie. 2025 Jun;45(3):229-242. doi: 10.1055/a-2436-5318. Epub 2025 Jan 27. PMID: 39870109.

**Zentrum für blasenbildende Autoimmundermatosen**

Sigmund AM, Bayerbach FC, Kugelmann D, Butz E, Moztarzadeh S, Schikora ME, Horn AK, Radeva MY, Engelmayer S, Egu DT, Goebeler M, Schmidt E, Waschke J, Vielmuth F. Epac1 contributes to apremilast-mediated rescue of pemphigus autoantibody-induced loss of keratinocyte adhesion. JCI Insight. 2025 Apr 29;10(10):e187481. doi: 10.1172/jci.insight.187481. PMID: 40299565; PMCID: PMC12128971.

**Zentrum für Genetische Herz- und Gefäßerkrankungen (ZGH)**

Gerull B. Genetisch bedingte Formen der Herzinsuffizienz im Erwachsenenalter [Genetic Causes of Heart Failure in Adults]. Dtsch Med Wochenschr. 2025 Jul;150(14):831-844. German. doi: 10.1055/a-2304-7182. Epub 2025 Jun 25. PMID: 40562059.

Lakdawala NK, Hershberger RE, Garcia-Pavia P, Elliott PM, Ginns J, Meder B, Solomon S, Cunningham JW, Gimeno JR, Barriales-Villa R, Adler E, Gerull B, Pereira NL, Halliday BP, Li W, Jarugula P, Maruyama S, Mohran SE, Papadaki M, Anto AR, Anderson RL, Rodriguez HM, Del Rio CL, Edelberg JM, Kurio G, Maya J, Januzzi JL. Danicamtiv, a Selective Agonist of Cardiac Myosin, for Dilated Cardiomyopathy: A Phase 2 Open-Label Trial. J Am Coll Cardiol. 2025 Dec 23;86(25):2598-2612. doi: 10.1016/j.jacc.2025.09.1511. Epub 2025 Sep 29. PMID: 41217321.

Rockel AF, Brunnbauer T, Wagner N, Gerull B, Ergün S, Wörsdörfer P. Tissue-resident macrophages co-develop with myocardial tissue in human induced pluripotent stem cell-derived organoids. Front Cell Dev Biol. 2025 Nov 13;13:1629988. doi: 10.3389/fcell.2025.1629988. PMID: 41323738; PMCID: PMC12657418.

Schanbacher C, Goebeler ME, Gerull B, Lorenz K. Targeting pathological ERK1/2 signaling in cancer and beyond. Trends Mol Med. 2026 Mar;32(3):231-255. doi: 10.1016/j.molmed.2025.08.001. Epub 2025 Sep 5. PMID: 40914707.

Schulz-Menger J, Collini V, Gröschel J, Adler Y, Brucato A, Christian V, Ferreira VM, Gandjbakhch E, Heidecker B, Kerneis M, Klein AL, Klingel K, Lazaros G, Lorusso R, Nesukay EG, Rahimi K, Ristić AD, Rucinski M, Sade LE, Schaubroeck H, Semb AG, Sinagra G, Thune JJ, Imazio M; ESC Scientific Document Group. 2025 ESC Guidelines for the management of myocarditis and pericarditis. Eur Heart J. 2025 Oct 22;46(40):3952-4041. doi: 10.1093/eurheartj/ehaf192. PMID: 40878297.

Stangner K, Dervishi O, Kuhnert J, Wendt C, Pathak S, Shoykhet M, Olivares-Florez S, Moztarzadeh S, Opsteen J, Wohlfarth NLCS, Biller R, Graf E, Westphal DS, Williams T, Gerull B, Šarić T, Yeruva S, Waschke J. Apremilast improves cardiomyocyte cohesion and arrhythmia in different models for arrhythmogenic cardiomyopathy. Stem Cell Res Ther. 2025 Nov 4;16(1):609. doi: 10.1186/s13287-025-04755-y. PMID: 41185038; PMCID: PMC12584335.

Waschke J, Amagai M, Becker C, Delmar M, Duru F, Garrod DR, Gerull B, Green KJ, Hertl M, Kowalczyk AP, Niessen CM, Nusrat A, Schinner C, Schlegel N, Sivasankar S, Vielmuth F, Spindler V. Meeting report - Alpine desmosome disease meeting 2024: advances and emerging topics in desmosomes and related diseases. J Cell Sci. 2025 Jan 15;138(2):JCS263796. doi: 10.1242/jcs.263796. Epub 2025 Jan 22. PMID: 39838950; PMCID: PMC11972074.

Williams T, Groß R, Arias-Loza AP, Nordbeck P, Noerpel M, Cirnu A, Kimmel L, Ashour D, Ramos G, Waschke J, Higuchi T, Gerull B. Illuminating Cardiac Remodeling: Insights From [$^{18}$F]-Fluorodeoxyglucose Positron Emission Tomography Imaging in Plakoglobin-Associated Arrhythmogenic Cardiomyopathy. J Am Heart Assoc. 2025 Mar 4;14(5):e038331. doi: 10.1161/JAHA.124.038331. Epub 2025 Mar 3. PMID: 40028850; PMCID: PMC12132786.

Xu A, Weissman D, Ermer KJ, Bertero E, Federspiel JM, Stadler F, Grünler E, Tangos M, Zervou S, Waddingham MT, Pearson JT, Reil JC, Scholtz S, Dudek J, Kohlhaas M, Nickel AG, Carrier L, Eschenhagen T, Michels M, Dos Remedios C, Lal S, Prates Roma L, Hamdani N, Kuster DWD, Falcão-Pires I, Johnson CN, Lygate CA, van der Velden J, Maack C, Sequeira V. Hypercontractility and Oxidative Stress Drive Creatine Kinase Dysfunction in Hypertrophic Cardiomyopathy. Circulation. 2025 Dec 16;152(24):1705-1727. doi: 10.1161/CIRCULATIONAHA.125.074120. Epub 2025 Oct 20. PMID: 41111389; PMCID: PMC13227915.

**Zentrum für genetische Innenohrstörungen (ZGI)**

Andries E, Lorens A, Skarżyński PH, Skarżyński H, Calvino M, Gavilán J, Lassaletta L, Tavora-Vieira D, Acharya A, Kurz A, Hagen R, Anderson I, Koinig K, Abdelsamad Y, Van de Heyning P, Van Rompaey V, Mertens G. Impact of cochlear implant indications on outcome, assessed and reported using the international classification of functioning, disability and health model. Eur Arch Otorhinolaryngol. 2025 Oct;282(10):5153-5167. doi: 10.1007/s00405-025-09454-1. Epub 2025 Jun 3. PMID: 40456899.

Bengl D, Koparir A, Prastyo WE, Remmele C, Dittrich M, Flandin S, Shehata-Dieler W, Grimm C, Haaf T, Hofrichter MAH. Whole-genome sequencing, as a powerful diagnostic tool in hearing loss, reveals novel variants in PTPRQ missed by whole-exome sequencing. BMC Med Genomics. 2025 Mar 31;18(1):59. doi: 10.1186/s12920-025-02122-7. PMID: 40165225; PMCID: PMC11956499.

Kurz A, Herrmann D, Müller-Graff FT, Voelker J, Hackenberg S, Rak K. Anatomy-based fitting improves speech perception in noise for cochlear implant recipients with single-sided deafness. Eur Arch Otorhinolaryngol. 2025 Jan;282(1):467-479. doi: 10.1007/s00405-024-08984-4. Epub 2024 Sep 19. PMID: 39299967; PMCID: PMC11735480.

Lammers MJ, Raine C, Mertens G, van Rompaey V, Hagen R, Kurz A, Skarzynski PH, Lorens A, Opie J, D'Haese P, Grasso P, Lassaletta L, Calvino M, Anderson I. Towards an adult hearing screening procedure. Braz J Otorhinolaryngol. 2025 Jul-Aug;91(4):101616. doi: 10.1016/j.bjorl.2025.101616. Epub 2025 Apr 11. PMID: 40220484; PMCID: PMC12017982.

Lenarz T, Keintzel T, Sprinzl G, Knoelke N, Busch S, Raffelsberger T, Magele A, Schörg P, Köstler C, Rak K. Lifetime Real-World Evidence on Safety and Performance of the Only Active Middle Ear Implant (AMEI) Covering Conductive to Sensorineural Hearing Loss: Results From a Long-Term Retrospective Analysis. Otol Neurotol. 2026 Jan 1;47(1):119-131. doi: 10.1097/MAO.0000000000004678. Epub 2025 Dec 11. PMID: 41373133.

Müller-Graff FT, Herrmann DP, Spahn B, Voelker J, Kurz A, Neun T, Hackenberg S, Rak K. Position Control of Flexible Electrodes With Regard to Intracochlear Structure Preservation and Hearing Outcomes: A Retrospective Study With Implementation of the Electrode Contact View. Otol Neurotol. 2025 Sep 1;46(8):e307-e315. doi: 10.1097/MAO.0000000000004528. PMID: 40570311.

Spahn B, Müller-Graff FT, Rak K, Engert J, Voelker J, Hackenberg S, Hagen R, Neun T, Petritsch B, Bley TA, Grunz JP, Huflage H. Pre- and Postoperative Imaging of Cochlear Implantation in Cadaveric Specimens Using Low-Dose Photon-Counting Detector CT. AJNR Am J Neuroradiol. 2025 Feb 3;46(2):362-371. doi: 10.3174/ajnr.A8533. PMID: 39900459; PMCID: PMC11878974.

Spahn B, Rak K, Herrmann DP, Cebulla M, Engert J, Kurz A, Voelker J, Hackenberg S, Müller-Graff FT. Intraoperative Auditory Brainstem Response (ABR) Measurement During Vibroplasty in Congenital Aural Malformation. Otol Neurotol. 2025 Aug 1;46(7):821-828. doi: 10.1097/MAO.0000000000004529. Epub 2025 Jun 2. PMID: 40466110.

**Zentrum für das Multiple Myelom**

Bolli N, D'Agostino M, Bagratuni T, Boccadoro M, Cavo M, Driessen C, Einsele H, Engelhardt M, Gay F, Gutiérrez NC, Hájek R, Holien T, João C, Kaiser M, Kortüm KM, Leypoldt L, Moreau P, Musto P, Ocio EM, Raab MS, Rasche L, Schjesvold F, Sevcikova T, Terpos E, Touzeau C, Van de Donk NWCJ, van Duin M, Weisel K, Zamagni E, Cupedo T, Sonneveld P, Terragna C. European Myeloma Network Group Consensus Statement on the use of next-generation sequencing for prognostic stratification of newly diagnosed multiple myeloma. Hemasphere. 2025 Oct 13;9(10):e70216. doi: 10.1002/hem3.70216. PMID: 41089883; PMCID: PMC12517052.

Bolli N, D'Agostino M, Bagratuni T, Boccadoro M, Cavo M, Driessen C, Einsele H, Engelhardt M, Gay F, Gutiérrez NC, Hájek R, Holien T, João C, Kaiser M, Kortüm KM, Leypoldt L, Moreau P, Musto P, Ocio EM, Raab MS, Rasche L, Schjesvold F, Sevcikova T, Terpos E, Touzeau C, Van de Donk NWCJ, van Duin M, Weisel K, Zamagni E, Cupedo T, Sonneveld P, Terragna C. European Myeloma Network Group Consensus Statement on the use of next-generation sequencing for prognostic stratification of newly diagnosed multiple myeloma. Hemasphere. 2025 Oct 13;9(10):e70216. doi: 10.1002/hem3.70216. PMID: 41089883; PMCID: PMC12517052.

Chabrun F, Schwartz D, Gentile S, Mai E, Gupta T, Perry J, Santos DCD, Hielscher T, Werly A, Schmidt S, Theodorakakou F, Fotiou D, Liacos C, Kanellias N, Gisbert N, Martin-Sanchez E, Termini R, Waldschmidt J, Chavda S, Ainley L, Và MCD, de Magistris C, Pettine L, Timonian M, Alberge JB, Patel V, Costello P, Tobia C, Phan S, Lamb J, Silverio MT, Davis M, O'Donnell E, Marinac C, Nadeem O, Bolli N, Yong K, Kortüm M, Einsele H, Manteca MVM, Kumar S, Miguel JS, Paiva B, Kastritis E, Dimopoulos M, Raab M, Trippa L, Ghobrial I. Enhanced Risk Stratification of Smoldering Multiple Myeloma with Dynamic Biomarkers: A Multinational, Multicenter Study including 2,270 Participants (PANGEA 2.0). Res Sq [Preprint]. 2025 Jun 3:rs.3.rs-6696313. doi: 10.21203/rs.3.rs-6696313/v1. Update in: Nat Med. 2026 May;32(5):1745-1753. doi: 10.1038/s41591-026-04304-x. PMID: 40502745; PMCID: PMC12155220.

Dhakal B, Einsele H, Schecter JM, Deraedt W, Lendvai N, Slaughter A, Lonardi C, Nair S, Varghese NE, He J, Kharat A, Lee S, Cost P, Potluri R, Koneru M, Patel N, Florendo E, Rodriguez-Otero P, Yong K. Real-World Treatment Patterns and Outcomes in Patients With Relapsed/Refractory Multiple Myeloma and 1-3 Prior Lines of Therapy: Optum Database. Cancer Med. 2025 Aug;14(15):e71093. doi: 10.1002/cam4.71093. PMID: 40719208; PMCID: PMC12301936.

Dhakal B, He J, Schecter JM, Deraedt W, Slaughter A, Lonardi C, Nair S, He J, Lee S, Cost P, Patel N, Koneru M, Einsele H. Real-World Treatment Patterns and Outcomes in Patients With Lenalidomide-Refractory Multiple Myeloma With 1 to 3 Prior Lines: SEER-Medicare Database. Clin Lymphoma Myeloma Leuk. 2025 Jun;25(6):e394-e403. doi: 10.1016/j.clml.2025.01.009. Epub 2025 Jan 23. PMID: 39966019.

Dimopoulos MA, Terpos E, Boccadoro M, Moreau P, Mateos MV, Zweegman S, Cook G, Engelhardt M, Delforge M, Hajek R, Schjesvold F, Gay F, Manier S, Weisel KC, Kaiser M, van de Donk NWCJ, Zamagni E, Rodriguez-Otero P, Perrot A, Driessen C, Bila J, Laane E, Dytfeld D, Touzeau C, Beksac M, Raab MS, Cavo M, Mohty M, Spencer A, Ludwig H, Einsele H, San-Miguel J, Sonneveld P. EHA-

EMN Evidence-Based Guidelines for diagnosis, treatment and follow-up of patients with multiple myeloma. Nat Rev Clin Oncol. 2025 Sep;22(9):680-700. doi: 10.1038/s41571-025-01041-x. Epub 2025 Jul 7. PMID: 40624367.

Dreher N, Dörrler AL, Kraus S, Rasche L, Higuchi T, Samnick S, Lapa C, Einsele H, Serfling SE, Buck AK, Werner RA. Myeloablative Radioligand Therapy Targeting C-X-C Motif Chemokine Receptor 4 in Advanced Multiple Myeloma. Clin Nucl Med. 2025 Jun 1;50(6):495-500. doi: 10.1097/RLU.0000000000005813. Epub 2025 Mar 5. PMID: 40323995; PMCID: PMC12043262.

Einsele H, Moreau P, Bahlis N, Bhutani M, Vincent L, Karlin L, Perrot A, Goldschmidt H, van de Donk NWCJ, Ocio EM, Martínez López J, Rodríguez-Otero P, Dytfeld D, Jakubowiak A, Schinke C, Besemer B, Anguille S, Manier S, Rasche L, Teipel R, Scheid C, Pawlyn C, Cavo M, Diels J, Ghilotti F, Lau BW, Renaud T, Orel O, Ong F, Ramos DF, Ammann E, Parekh T, Albrecht C, Weisel K, Mateos MV. Comparative Efficacy of Talquetamab vs. Real-World Physician's Choice of Treatment in Triple-Class-Exposed Relapsed/Refractory Multiple Myeloma: Updated Analyses of MonumenTAL-1 vs. LocoMMotion/MoMMent. Adv Ther. 2026 Jan;43(1):333-355. doi: 10.1007/s12325-025-03409-y. Epub 2025 Nov 28. PMID: 41313549; PMCID: PMC12858606.

Fleischer A, Roll M, Frenking JH, Panther F, Gelbrich G, Strunz PP, Riedhammer C, Peter J, Mersi J, Waldschmidt J, Kortüm M, Einsele H, Raab MS, Maatouk I, Rasche L. Talquetamab-Related Dysgeusia in Multiple Myeloma Compared to BCMA-Targeted Bispecifics and High-Dose Melphalan. Cancer Med. 2025 Dec;14(23):e71401. doi: 10.1002/cam4.71401. PMID: 41360668; PMCID: PMC12685487.

Frenking JH, Zhou X, Rejeski K, Wagner V, Costello P, Hielscher T, Gatti L, Kauer J, Nadeem O, Mai EK, Michel CS, Friedrich MJ, Sedloev D, Weinhold N, Goldschmidt H, Herfarth K, Schmitt A, Hundemer M, Schmitt M, Müller-Tidow C, Topp M, Einsele H, Dreger P, Munshi NC, Sperling AS, Rasche L, Sauer S, Raab MS. Bridging intensity is associated with impaired hematopoietic recovery after BCMA CAR-T therapy for multiple myeloma. Blood Adv. 2025 Aug 26;9(16):4151-4166. doi: 10.1182/bloodadvances.2024015732. PMID: 40267180; PMCID: PMC12361772.

Gerotziafas G, Fotiou D, Nijhof I, Ay C, Lecumberri R, Laroca A, Cook G, Engelhardt M, Zweegman S, Delforge M, Maraveyas A, Lefkou E, Marchetti M, Van de Donk NWCJ, Gay F, Ludwig H, Einsele H, San Miguel JF, Dimopoulos MA, Boccadoro M, Sonneveld P, Falanga A, Terpos E. Prevention and treatment of venous thromboembolism in patients with multiple myeloma: Clinical practice guidelines on behalf of the European Myeloma Network. Hemasphere. 2025 Aug 26;9(8):e70177. doi: 10.1002/hem3.70177. Erratum in: Hemasphere. 2025 Nov 14;9(11):e70250. doi: 10.1002/hem3.70250. PMID: 40880878; PMCID: PMC12381471.

Kegyes D, Bancos A, Tigu AB, Rus I, Dima D, Cenariu D, Nistor M, Munteanu R, Gulei D, Tanase A, Colita A, Buzoianu A, Iuga C, Zdrenghea M, Terpos E, Ciurea SO, Ciechanover A, Einsele H, Tomuleasa C. Targeting degradation of IKZF1 and IKZF3 through modulation of the E3 ligase substrates in the context of cellular therapies for multiple myeloma. Biomark Res. 2025 Aug 15;13(1):105. doi: 10.1186/s40364-025-00825-8. PMID: 40817326; PMCID: PMC12357425.

Kegyes D, Borlea B, Damian M, Tigu AB, Nistor M, Cenariu D, Munteanu R, Gulei D, Dascalescu A, Antohe I, Tanase A, Terpos E, Einsele H, Tomuleasa C. Adoptive cellular therapies in multiple myeloma. Best Pract Res Clin Haematol. 2025 Sep;38(3):101638. doi: 10.1016/j.beha.2025.101638. Epub 2025 May 27. PMID: 40915705.

Kegyes D, Tat A, Vizitiu AS, Vazar-Tripon D, Ilie R, Tigu AB, Cenariu D, Bancos A, Iluta S, Jitaru C, Nistor M, Tomai R, Gulei D, Zdrenghea M, Einsele H, Ghiaur G, Croce CM, Tomuleasa C. Comprehensive view on chemotherapy-free management of acute myeloid leukemia by using

venetoclax in combination with targeted and/or immune therapies. Cell Death Discov. 2025 Aug 13;11(1):379. doi: 10.1038/s41420-025-02678-4. PMID: 40804253; PMCID: PMC12350645.

Knop S, Einsele H, Dhanda D, Marshall TS, Eliason L, McLoone D, Caisip C, Chen JMH, Boehm D, Dhamane AD, Ramasamy K, Cope S, Towle K. Joint modeling of progression-free survival and patient-reported outcomes to evaluate the association between disease progression and symptoms among patients with relapsed/refractory multiple myeloma. J Patient Rep Outcomes. 2025 Oct 14;9(1):118. doi: 10.1186/s41687-025-00943-9. PMID: 41085804; PMCID: PMC12521705.

Kortüm KM, Einsele H. Guidance on the interpretation of CRBN mutations in myeloma. Blood. 2025 May 29;145(22):2542. doi: 10.1182/blood.2024028272. PMID: 40440041.

Lim SL, Engelhardt M, Terpos E, Gay F, Van de Donk NWCJ, Einsele H, Sonneveld P, Kaiser M, Boccadoro M, Spencer A. European Myeloma Network Consensus Statement on Functional High-Risk Multiple Myeloma. Am J Hematol. 2025 Dec;100(12):2320-2332. doi: 10.1002/ajh.70070. Epub 2025 Sep 9. PMID: 40926508.

Lim SL, Engelhardt M, Terpos E, Gay F, Van de Donk NWCJ, Einsele H, Sonneveld P, Kaiser M, Boccadoro M, Spencer A. European Myeloma Network Consensus Statement on Functional High-Risk Multiple Myeloma. Am J Hematol. 2025 Dec;100(12):2320-2332. doi: 10.1002/ajh.70070. Epub 2025 Sep 9. PMID: 40926508.

Merz M, Gagelmann N, Smaili S, Flossdorf S, Sauer S, Scheid C, von Tresckow B, Wulf G, Weisel K, Blau IW, Engelhardt M, Wäsch R, Schub N, Teipel R, Hecker J, Waldschmidt J, Besemer B, Baermann BN, Call S, Hansmann L, Ayuk FA, Raab MS, Einsele H, Platzbecker U, Kröger N. Remission conversion drives outcomes after CAR T-cell therapy for multiple myeloma: a registry analysis from the DRST. Blood. 2025 Oct 2;146(14):1677-1686. doi: 10.1182/blood.2025028330. PMID: 40504993.

Musto P, Engelhardt M, van de Donk NWCJ, Gay F, Terpos E, Einsele H, Fernández de Larrea C, Sgherza N, Bolli N, Katodritou E, Gentile M, Royer B, Derudas D, Jelinek T, Zamagni E, Rosiñol L, Paiva B, Caers J, Kaiser M, Beksac M, Hájek R, Spencer A, Ludwig H, Cavo M, Bladé J, Moreau P, Mateos MV, San-Miguel JF, Dimopoulos MA, Boccadoro M, Sonneveld P. European Myeloma Network Group review and consensus statement on primary plasma cell leukemia. Ann Oncol. 2025 Apr;36(4):361-374. doi: 10.1016/j.annonc.2025.01.022. Epub 2025 Feb 7. PMID: 39924085.

Musto P, Engelhardt M, van de Donk NWCJ, Gay F, Terpos E, Einsele H, Fernández de Larrea C, Sgherza N, Bolli N, Katodritou E, Gentile M, Royer B, Derudas D, Jelinek T, Zamagni E, Rosiñol L, Paiva B, Caers J, Kaiser M, Beksac M, Hájek R, Spencer A, Ludwig H, Cavo M, Bladé J, Moreau P, Mateos MV, San-Miguel JF, Dimopoulos MA, Boccadoro M, Sonneveld P. European Myeloma Network Group review and consensus statement on primary plasma cell leukemia. Ann Oncol. 2025 Apr;36(4):361-374. doi: 10.1016/j.annonc.2025.01.022. Epub 2025 Feb 7. PMID: 39924085.

Musto P, Engelhardt M, van de Donk NWCJ, Gay F, Terpos E, Einsele H, Fernández de Larrea C, Sgherza N, Bolli N, Katodritou E, Gentile M, Royer B, Derudas D, Jelinek T, Zamagni E, Rosiñol L, Paiva B, Caers J, Kaiser M, Beksac M, Hájek R, Spencer A, Ludwig H, Cavo M, Bladé J, Moreau P, Mateos MV, San-Miguel JF, Dimopoulos MA, Boccadoro M, Sonneveld P. European Myeloma Network Group review and consensus statement on primary plasma cell leukemia. Ann Oncol. 2025 Apr;36(4):361-374. doi: 10.1016/j.annonc.2025.01.022. Epub 2025 Feb 7. PMID: 39924085.

Neubert S, Munawar U, Mersi J, Noderer J, Nerreter S, Kurian S, Han S, Verbruggen C, Besant E, Rein N, Köppel M, Lehmann J, Köhler T, Labinsky H, Häusl S, Tamamushi Y, Zhou X, Pinter J, Herzog AL, Lopau K, Hoxha E, Rummelt C, Gerhard-Hartmann E, Rosenwald A, Steinbrunn T, Nerreter T, Hudecek M, Einsele H, Rasche L, Kortüm KM, Waldschmidt JM. Plasmapheresis facilitates soluble

BCMA clearance and contributes to reversing primary resistance to anti-BCMA immunotherapy in multiple myeloma. Leukemia. 2025 Oct;39(10):2563-2567. doi: 10.1038/s41375-025-02757-6. Epub 2025 Sep 10. PMID: 40931044; PMCID: PMC12463651.

Rade M, Fandrei D, Kreuz M, Seiffert S, Grahnert A, Friedrich M, Wiemers T, Born P, Fischer L, Weidner H, Hofbauer LC, Baber R, Wang SY, Bach E, Hoffmann S, Scolnick J, Friedrich M, Keramati F, Brazda P, Sebestyen Z, Kuball J, Alb M, Scheller L, Hudecek M, Einsele H, Metzeler KH, Herling M, Herling CD, Jentzsch M, Franke GN, Boldt A, Köhl U, Platzbecker U, Vucinic V, Reiche K, Merz M. A longitudinal single-cell atlas to predict outcome and toxicity after BCMA-directed CAR T cell therapy in multiple myeloma. Cancer Cell. 2026 Mar 9;44(3):586-603.e9. doi: 10.1016/j.ccell.2025.10.014. Epub 2025 Dec 4. PMID: 41349540.

Rodriguez-Otero P, Voorhees PM, Boccadoro M, Laubach J, Einsele H, Sborov DW, Dimopoulos MA, Broijl A, Mina R, Spencer A, Schjesvold F, Silbermann R, Gay F, Costa LJ, Perrot A, Liu Y, Wang J, Sitthi-Amorn A, Carson R, Cortoos A, Usmani SZ, Richardson PG, Moreau P, Sonneveld P, Kaufman JL. Daratumumab for newly diagnosed multiple myeloma: Pooled analysis of patients aged ≥65 years from GRIFFIN and PERSEUS. Clin Lymphoma Myeloma Leuk. 2025 Oct;25(10):746-758.e6. doi: 10.1016/j.clml.2025.04.007. Epub 2025 Apr 11. PMID: 40360369.

Steinhardt MJ, Schaefers C, Leypoldt LB, Blau IW, Harzer M, Zhou X, Riedhammer C, Kamili A, Kosch R, Topp LS, Molwitz I, Gross-Fengels NO, Melzer YF, Artzenroth J, Al-Bazaz M, Alsdorf W, Topp MS, Duell J, Mersi J, Waldschmidt J, Bokemeyer C, Einsele H, Kortüm KM, Weisel K, Rasche L. Correction: Activity of CAR-T cells and bispecific antibodies in multiple myeloma with extramedullary involvement. Blood Cancer J. 2025 Oct 31;15(1):187. doi: 10.1038/s41408-025-01403-9. Erratum for: Blood Cancer J. 2025 Jul 30;15(1):126. doi: 10.1038/s41408-025-01330-9. PMID: 41173845; PMCID: PMC12578947.

Steinhardt MJ, Schaefers C, Leypoldt LB, Blau IW, Harzer M, Zhou X, Riedhammer C, Kamili A, Kosch R, Topp LS, Molwitz I, Gross-Fengels NO, Melzer YF, Artzenroth J, Al-Bazaz M, Alsdorf W, Topp MS, Duell J, Mersi J, Waldschmidt J, Bokemeyer C, Einsele H, Kortüm KM, Weisel K, Rasche L. Activity of CAR-T cells and bispecific antibodies in multiple myeloma with extramedullary involvement. Blood Cancer J. 2025 Jul 30;15(1):126. doi: 10.1038/s41408-025-01330-9. Erratum in: Blood Cancer J. 2025 Oct 31;15(1):187. doi: 10.1038/s41408-025-01403-9. PMID: 40738883; PMCID: PMC12311153.

van de Donk NWCJ, Moreau P, San-Miguel JF, Mateos MV, Dimopoulos MA, Zweegman S, Gay F, Engelhardt M, Mina R, Zamagni E, Delforge M, Beksac M, Spencer A, Schjesvold F, Driessen C, Kaiser M, Perrot A, Wäsch R, Korst CLBM, Broijl A, Touzeau C, Manier S, Hajek R, Bila J, Seval GC, O'Dwyer M, Ludwig H, Fernandez de Larrea C, Popat R, Musto P, Rodriguez-Otero P, Yong K, Kortüm M, Rasche L, Terpos E, Raab MS, Boccadoro M, Sonneveld P, Einsele H; EMN Guidelines Committee. Sequencing BCMA- and GPRC5D-targeting immunotherapies in multiple myeloma: Practical guidance from the European Myeloma Network. Hemasphere. 2025 Nov 25;9(11):e70260. doi: 10.1002/hem3.70260. PMID: 41306326; PMCID: PMC12645799.

van de Donk NWCJ, Moreau P, San-Miguel JF, Mateos MV, Dimopoulos MA, Zweegman S, Gay F, Engelhardt M, Mina R, Zamagni E, Delforge M, Beksac M, Spencer A, Schjesvold F, Driessen C, Kaiser M, Perrot A, Wäsch R, Korst CL, Broijl A, Touzeau C, Manier S, Hajek R, Ludwig H, Fernandez de Larrea C, Popat R, Musto P, Rodriguez-Otero P, Yong K, Rasche L, Terpos E, Raab MS, Boccadoro M, Sonneveld P, Einsele H; EMN Guidelines Committee. Optimising T-cell immunotherapy in patients with multiple myeloma: practical considerations from the European Myeloma Network. Lancet Haematol. 2025 Aug;12(8):e635-e649. doi: 10.1016/S2352-3026(25)00117-6. Epub 2025 Jun 25. Erratum in: Lancet Haematol. 2025 Oct;12(10):e772. doi: 10.1016/S2352-3026(25)00257-1. PMID: 40580975.

Waldschmidt JM, Rasche L, Kortüm KM, Einsele H. Comprehensive Review of Bispecific Antibody Constructs In Multiple Myeloma: Affinities, Dosing Strategies and Future Perspectives. Clin Lymphoma Myeloma Leuk. 2025 May;25(5):309-315. doi: 10.1016/j.clml.2024.11.012. Epub 2024 Nov 22. PMID: 39676006.

Weber J, Rade M, Michael J, Tony LT, Freitag F, Spieler P, Müller C, Kalogirou C, Mainz L, Lehmann J, Klopfleisch R, Rosenfeldt MT, Danhof S, Kortüm KM, Köhl U, Einsele H, Rader C, Reiche K, Nerreter T, Hudecek M. ROR2-specific CAR T cells are effective against hematologic and solid tumors and well tolerated in mice. Cell Rep Med. 2025 Oct 21;6(10):102400. doi: 10.1016/j.xcrm.2025.102400. Epub 2025 Oct 6. PMID: 41056951; PMCID: PMC12629790.

Zhou X, Waldschmidt JM, Einsele H. Bispecific antibodies in multiple myeloma: maximizing potential through rational combination therapies. Blood Rev. 2025 Nov;74:101342. doi: 10.1016/j.blre.2025.101342. Epub 2025 Oct 30. PMID: 41177723.

**Zentrum für Deletionssyndrom 22q11.2 (ZEDE22q11)**

Sauter C, Hofbeck M, Franz P, Kettenstock L, Steger K, Linhardt M, Reiter A, Störk S, Romanos M, Radtke F. Congenital heart disease in 22q11.2 deletion syndrome: a meta-analysis and systematic review of the literature. J Med Genet. 2025 Oct 20;62(11):700-708. doi: 10.1136/jmg-2025-110624. PMID: 40744517; PMCID: PMC12573319.

**Zentrum für endokrine Tumore (ZET)**

Detomas M, Altieri B, Nasi-Kordhishti I, Ryba A, Haberbosch L, Chierigo F, Deutschbein T, Fassnacht M, Mortini P, Flitsch J, Honegger J, Losa M. Recovery of hypopituitarism in macroprolactinomas: a comparison of medical vs. surgical treatment. Results from a European multicenter study. J Endocrinol Invest. 2025 Jun;48(6):1363-1370. doi: 10.1007/s40618-025-02559-8. Epub 2025 Mar 4. PMID: 40035956; PMCID: PMC12226669.

Detomas M, Kagan LR, Pittaway JFH, Schauer MP, Kloock S, Geiger N, Kimpel O, Fassnacht M, Altieri B, Dischinger U. Agouti-related peptide: a promising prognostic biomarker in adrenocortical carcinoma. Results from a large single-centre cross-sectional analysis. Eur J Endocrinol. 2025 Oct 30;193(5):604-612. doi: 10.1093/ejendo/lvaf226. PMID: 41172328.

Fassnacht M, Puglisi S, Kimpel O, Terzolo M. Adrenocortical carcinoma: a practical guide for clinicians. Lancet Diabetes Endocrinol. 2025 May;13(5):438-452. doi: 10.1016/S2213-8587(24)00378-4. Epub 2025 Mar 11. PMID: 40086465.

Ganner A, Ferrara AM, Sekula P, Schiavi F, Joo JH, Sanso G, Almeida MQ, Knoblauch AL, Gizaw CJ, Krzystolik K, Astheimer SC, Achatz MI, Vieites A, Donegan D, Hundsberger T, Lubinski J, Yildirim Simsir I, Bandgar T, Hasse-Lazar K, Pawlaczek A, Zandee W, Yu K, Kater CE, Rostomyan L, Qi XP, Deutschbein T, Remde H, Dallagnol TN, Yukina M, Baudrand R, Andreescu CE, Kunavisarut T, Ishak ND, Le Guillou Horn X, Shutler G, Jovanovic M, Pęczkowska M, Calissendorff J, Circosta F, Bugalho MJ, Corssmit EPM, Gimm O, Quinkler M, Goldmann A, Watutantrige Fernando S, Zovato S, Santana LS, Freitas-Castro F, Rothermundt C, Zimmermann J, Durmaz A, Aykut A, Vroonen L, Krauss T, Taschner C, Ruf J, Klingler JH, Gläsker S, Lang S, Bucher F, Agostini H, Jilg C, Schultze-Seemann W, Bausch B, Bergfeld A, Rhein K, Uslar T, Concistrè A, Juhlin CC, Casali-da-Rocha JC, Petramala L, Tsoy U, Grineva E, Fang XD, Kotsis F, Schaefer T, Links TP, Makay Ö, Fagundes GFC, Ngeow J, Shah N, Opocher G, Barontini M, Larsson C, Januszewicz A, Viana Lima J, Wohllk N, Letizia C, Donatini G, Maher ER, Beltsevich D, Bancos I, Cybulski C, Walz MK, Köttgen A, Eng C, Neumann HPH, Neumann-Haefelin E. Genotype-specific neoplastic risk profiles in patients with VHL disease. Endocr Relat Cancer. 2025 Apr 28;32(5):e240260. doi: 10.1530/ERC-24-0260. PMID: 40202835; PMCID: PMC12060576.

Hahner S, Hartrampf P, Beuschlein F, Miederer M, Miehle K, Schlötelburg W, Fuß CT, Pfluger T, Fottner C, Tönjes A, Herrmann K, Amthauer H, Reincke M, Schreckenberger M, Sabri O, Werner J, Reuter M, Kircher S, Arlt W, Fassnacht M, Konrad Buck A, Müller HH, Schirbel A; FAMIAN investigators. Combined [$^{18}$F]Fluorodeoxyglucose PET and [$^{123}$I]Iodometomidate-SPECT for diagnostic evaluation of indeterminate adrenal neoplasias-the cross-sectional diagnostic test accuracy study FAMIAN. EBioMedicine. 2025 Jun;116:105735. doi: 10.1016/j.ebiom.2025.105735. Epub 2025 May 20. PMID: 40398350; PMCID: PMC12148602.

Jimenez C, Andreassen M, Durand A, Moog S, Hendifar A, Welin S, Spada F, Sharma R, Wolin E, Ruether J, Garcia-Carbonero R, Fassnacht M, Capdevila J, Del Rivero J, Iliopoulos O, Huillard O, Jang R, Mai K, Artamonova E, Hallqvist A, Else T, Odeleye-Ajakaye A, Gozman A, Naik GS, Berruti A; LITESPARK-015 Investigators. Belzutifan for Advanced Pheochromocytoma or Paraganglioma. N Engl J Med. 2025 Nov 20;393(20):2012-2022. doi: 10.1056/NEJMoa2504964. Epub 2025 Oct 18. PMID: 41124218.

Maier T, Landwehr LS, Triebig A, Kircher S, Schauer MP, Knösel T, Sbiera S, Schwarzlmueller P, Zimmermann P, Reincke M, Weigand I, Fassnacht M, Kroiss M. Wnt/β-catenin pathway activation is associated with glucocorticoid secretion in adrenocortical carcinoma, but not directly with immune cell infiltration. Front Endocrinol (Lausanne). 2025 Mar 10;16:1502117. doi: 10.3389/fendo.2025.1502117. PMID: 40130164; PMCID: PMC11930824.

Nieman LK, Boscaro M, Scaroni C, Deutschbein T, Mezôsi E, Driessens N, Georgescu CE, Motyka M, Hubalewska-Dydejczyk A, Jarzab B, Maiter D, Reincke M, Loli P, Zampetti B, Atmaca A, Badiu C, Beckers A, Bolanowski M, Cavagnini F, Unger N, Giordano R, Hanzu FA, Terzolo M, Nader MB, Sinaii N, Tőke J, Tóth M. Prospective study of metyrapone in endogenous Cushing's syndrome (PROMPT). Eur J Endocrinol. 2025 Aug 29;193(3):391-402. doi: 10.1093/ejendo/lvaf181. PMID: 40966724; PMCID: PMC13187941.

Pamporaki C, Pommer G, Apostolopoulos ID, Filippatos A, Peitzsch M, Remde H, Constantinescu G, Berends AMA, Nazari MA, Beuschlein F, Fassnacht M, Prejbisz A, Pacak K, Eisenhofer G. Utility of disease probability scores to guide decision-making during screening for phaeochromocytoma and paraganglioma: a machine learning modelling cross sectional study. EClinicalMedicine. 2025 Mar 29;82:103181. doi: 10.1016/j.eclinm.2025.103181. PMID: 40224674; PMCID: PMC11992530.

Riedmeier M, Agarwal S, Antonini SRR, Ekinci S, Fassnacht M, Figueiredo BC, Härtel C, Meena JP, Marks SD, Munarin J, Puglisi S, Tuli G, Yalcin B, Schlegel PG, Wiegering A, Wiegering V. Evaluation of prognostic factors in advanced pediatric ACC. Endocr Relat Cancer. 2025 Apr 2;32(5):e240135. doi: 10.1530/ERC-24-0135. PMID: 40126129.

Riedmeier M, Idkowiak J, Frey H, Antonini SRR, Canali GFL, Classen CF, Domínguez-Pinilla N, Fassnacht M, Fuchs S, Härtel C, Janús D, de Krijger R, Kutluk T, Bui NL, Meena JP, Mezoued M, Munarin J, van Noesel MM, Köse NÖ, Pearce SH, Perwein T, Puglisi S, Del Rivero J, Schlegel PG, Schmid I, Tuli G, Walenciak J, Yalcin B, Wiegering V. Inflammation-based score in pediatric adrenocortical carcinoma. Endocr Relat Cancer. 2025 Mar 28;32(5):e240244. doi: 10.1530/ERC-24-0244. PMID: 40100700; PMCID: PMC12002726.

Riedmeier M, Schlötelburg W, Agarwal S, Biswas A, Ekinci S, Fassnacht M, Villares Fragoso MCB, Gonc EN, Gultekin M, Haliloglu M, Jain V, Jana M, Janus D, Meena JP, Munarin J, Orhan D, Del Rivero J, Sharma R, Tuli G, Yalcin B, Wiegering V. Multidisciplinary Tumor Board Evaluation of Pediatric Patients with Adrenocortical Tumors Across Seven International Centers. Cancers (Basel). 2025 Mar 17;17(6):1014. doi: 10.3390/cancers17061014. PMID: 40149347; PMCID: PMC11940591.

Sojat AS, Rance B, Neuraz A, Fassnacht M, Beuschlein F, Robledo M, Luconi M, Vassiliadi D, Stell A, Igaz P, Dugic B, Marina LV, Burgun A, Kastelan D, Assie G; European Network for the Study of Adrenal Tumors (ENSAT)/COST Action Harmonisation (CA 20122) consortium. How ready are endocrine scientists to share retrospective clinical data for research: a perspective from the European Network for the Study of Adrenal Tumors. Eur J Endocrinol. 2025 Mar 27;192(4):491-509. doi: 10.1093/ejendo/lvaf005. PMID: 40289285.

Terzolo M, Fassnacht M, Berruti A, Ronchi C. Navigating Uncertainty in Adjuvant Mitotane Therapy. Endocr Pract. 2025 Jul;31(7):975-976. doi: 10.1016/j.eprac.2025.03.012. Epub 2025 Mar 31. PMID: 40174725.

Wang K, Fischer A, Maccio U, Zitzmann K, Robledo M, Lauseker M, Bauer J, Bechmann N, Gahr S, Maurer J, Peischer L, Reul A, Nieß H, Zimmermann P, Ilmer M, Schilbach K, Knösel T, Kroiss M, Fassnacht M, Müller SA, Morand GB, Huber A, Vetter D, Lehmann K, Kulcsar Z, Mohr H, Pellegata NS, Hantel C, Reincke M, Beuschlein F, Pacak K, Grossman AB, Auernhammer CJ, Nölting S. Impact of sex hormones on pheochromocytomas, paragangliomas, and gastroenteropancreatic neuroendocrine tumors. Eur J Endocrinol. 2025 Jan 6;192(1):46-60. doi: 10.1093/ejendo/lvae163. PMID: 39804847.

Weigand I, Triebig AS, Maier T, Anderlik T, Remde H, Landwehr LS, Kimpel O, Reuter M, Schreiner J, Wedekink F, Scherf-Clavel O, Hoster E, Wollert KC, Budde I, Altieri B, Schwarzlmueller P, Reincke M, Wischhusen J, Fassnacht M, Kroiss M. Mitotane treatment of adrenocortical carcinoma induces tumoural secretion of GDF-15: impact on poor prognosis and impaired responsiveness to immunotherapy. Eur J Endocrinol. 2025 Jun 30;193(1):146-155. doi: 10.1093/ejendo/lvaf135. PMID: 40570159.

**Zentrum für Maligne Hyperthermie**

Glahn KPE, Girard T, Hellblom A, Hopkins PM, Johannsen S, Rüffert H, Snoeck MM, Urwyler A; European Malignant Hyperthermia Group. Recognition and management of a malignant hyperthermia crisis: updated 2024 guideline from the European Malignant Hyperthermia Group. Br J Anaesth. 2025 Jan;134(1):221-223. doi: 10.1016/j.bja.2024.09.022. Epub 2024 Oct 30. PMID: 39482150.

**Zentrum für Primäre Immundefekte und Autoinflammatorische Erkrankungen**

Bogaert DJA, Wolfsberger CH, Attarbaschi A, Gathmann J, Warnatz K, Mueller G, Mukhina A, Rusch S; ESID Registry Working Party; Kindle G, van Montfrans JM, Seidel MG. Epidemiology and management of malignancies in patients with inborn errors of immunity-An ESID registry study of 19,959 patients. J Allergy Clin Immunol. 2026 Mar;157(3):739-753. doi: 10.1016/j.jaci.2025.10.033. Epub 2025 Nov 14. PMID: 41242640.

Dirks J, Fortmann I, Marißen J, Pagel J, Reichert L, Kipke H, Dammann MT, Göpel W, Birkner T, Dahm K, Forslund-Startceva SK, Viemann D, Rupp J, Morbach H, Härtel C. Effect of gestational age on lymphocyte phenotypes in hospitalized preterm infants. J Allergy Clin Immunol. 2026 Feb;157(2):506-516. doi: 10.1016/j.jaci.2025.09.030. Epub 2025 Oct 23. PMID: 41138924.

Hofmann C, Holl-Wieden A, Reiser C, Beer M, Raab P, Morbach H, Girschick HJ. Chronic non-bacterial osteomyelitis in children- five-year standardized follow-up of a prospective observational cohort in the pre-biological era. Pediatr Rheumatol Online J. 2025 May 13;23(1):50. doi: 10.1186/s12969-025-01106-2. PMID: 40361097; PMCID: PMC12076821.

Hospach T, Blankenburg F, Heinkele A, Rücklová K, Müller-Abt P, Weller-Heinemann F, Buchtala L, Maier J, Kümmerle-Deschner J, Reiser C, Janda A, Urban A, Berendes R, Skrabl-Baumgartner A, Brunner J, Trauzeddel R, Haas JP, Holl-Wieden A, Morbach H, Schnabel A, Brück N, Hahn G, Emminger W, Valent I, Borchers R, Klemann C, Tsiflikas I, Klotsche J, Minden K, Hedrich CM, von

Kalle T. Clinical Presentation and Course of Pulmonary Involvement in Chronic Nonbacterial Osteomyelitis. ACR Open Rheumatol. 2025 Jan;7(1):e11799. doi: 10.1002/acr2.11799. PMID: 39876070; PMCID: PMC11774723.

Schwaneck EC, Harasim AS, Tony HP, Gawlik M, Witte T, Joos S, Gernert M, Schmalzing M, Morbach H, Fröhlich M, Krone M. Vaccination status of patients with primary immunodeficiencies in Germany-a multicentric epidemiologic analysis. Z Rheumatol. 2025 Feb;84(1):68-74. English. doi: 10.1007/s00393-024-01549-0. Epub 2024 Aug 22. PMID: 39174715.

Schwitzkowski M, Veeranki SPK, Seidel BN, Kindle G, Rusch S; European Society for Immunodeficiencies Registry Working Party; Kramer D, Seidel MG. Machine learning-assisted diagnosis classification of primary immune dysregulation using IDDA2.1 phenotype profiling. J Allergy Clin Immunol. 2026 Feb;157(2):470-485. doi: 10.1016/j.jaci.2025.10.022. Epub 2025 Nov 5. PMID: 41202990.

Vogele D, Akbulut J, Müller-Reichart F, Janda A, Morbach H, Girschick HJ, Schaal MC, Beer M, Benoit C. The impact of sarcopenia on the progression of chronic non-bacterial osteomyelitis. Insights Imaging. 2025 Jun 4;16(1):117. doi: 10.1186/s13244-025-02001-w. PMID: 40467922; PMCID: PMC12137865.

**Zentrum für Riesenzellarteriitis (ZeRi)**

Guggenberger KV, Riedling L, Kern D, Werner RA, Vogt ML, Fröhlich M, Schmalzing M, Pham M, Bley TA. Isolated inflammatory involvement of the occipital artery in giant cell arteritis and polymyalgia rheumatica: findings from a retrospective analysis and the critical role of MRI in diagnosis. Rheumatol Int. 2025 Jan 13;45(1):27. doi: 10.1007/s00296-024-05765-4. PMID: 39804420; PMCID: PMC11729197.

Guggenberger KV, Vogt ML, Song JW, Fröhlich M, Schmalzing M, Venhoff N, Werner RA, Hillenkamp J, Pham M, Meckel S, Bley TA. High-resolution magnetic resonance imaging visualizes intracranial large artery involvement in giant cell arteritis. Rheumatology (Oxford). 2025 Feb 1;64(2):842-848. doi: 10.1093/rheumatology/keae010. PMID: 38197587.

Kern D, Bley TA, Riedling L, Fröhlich M, Hillenkamp J, Gernert M, Schmalzing M, Werner RA, Guggenberger KV. Vascular MRI also depicts musculoskeletal manifestations in giant-cell arteritis-polymyalgia rheumatica spectrum disease patients. Rheumatology (Oxford). 2025 Aug 1;64(8):4750-4755. doi: 10.1093/rheumatology/keaf169. PMID: 40131396.

Labinsky H, Schmalzing M. Riesenzellarteriitis – Wann sollte der Hausarzt daran denken? [Giant cell arteritis - When should the GP consider it?]. MMW Fortschr Med. 2025 Oct;167(18):65-67. German. doi: 10.1007/s15006-025-5364-y. PMID: 41131256.

Marvisi C, Guggenberger KV, Besutti G, Werner RA, Mancuso P, Serfling SE, Farì R, Gernert M, Durmo R, Fröhlich M, Ricordi C, Labinsky H, Boiardi L, Hillenkamp J, Spaggiari L, Versari A, Pattacini P, Giorgi Rossi P, Bley TA, Salvarani C, Schmalzing M, Muratore F. The influence of baseline aortitis on aortic dilation risk in GCA: a multicenter imaging study. Rheumatology (Oxford). 2025 Dec 9:keaf656. doi: 10.1093/rheumatology/keaf656. Epub ahead of print. PMID: 41365838.

Rhee RL, Bathla G, Rebello R, Kurtz RM, Junek M, Warrington KJ, Khalidi N, Merkel PA, Guggenberger KV, Tamhankar MA, Bley TA; Vasculitis Clinical Research Consortium. Vessel wall MRI in giant cell arteritis: standardized protocol and scoring approach developed by an international working group. Rheumatology (Oxford). 2025 May 1;64(5):2910-2918. doi: 10.1093/rheumatology/keae498. PMID: 39331619.

Zeng R, Rebello R, Guggenberger KV, Baker JF, Banerjee S, Kurtz R, Amudala N, Merkel PA, Rhee RL. Longitudinal changes on cranial magnetic resonance imaging in relapsing giant cell arteritis. Semin Arthritis Rheum. 2025 Oct;74:152815. doi: 10.1016/j.semarthrit.2025.152815. Epub 2025 Aug 14. PMID: 40845732; PMCID: PMC12639502.

**Zentrum für Seltene Hormonstörungen**

Atila C, Chifu I, Drummond JB, Vogt DR, Nahum U, Fassnacht M, Winzeler B, Refardt J, Christ-Crain M. A novel diagnostic score for diagnosing arginine vasopressin deficiency (central diabetes insipidus) or primary polydipsia with basal laboratory and clinical parameters: results from two international multicentre prospective diagnostic studies. Lancet Diabetes Endocrinol. 2025 Jun;13(6):505-515. doi: 10.1016/S2213-8587(25)00053-1. Epub 2025 Apr 25. Erratum in: Lancet Diabetes Endocrinol. 2025 Jun;13(6):e9. doi: 10.1016/S2213-8587(25)00130-5. Erratum in: Lancet Diabetes Endocrinol. 2025 Jun;13(6):e9. doi: 10.1016/S2213-8587(25)00133-0. PMID: 40294614.
Detomas M, Altieri B, Nasi-Kordhishti I, Ryba A, Haberbosch L, Chierigo F, Deutschbein T, Fassnacht M, Mortini P, Flitsch J, Honegger J, Losa M. Recovery of hypopituitarism in macroprolactinomas: a comparison of medical vs. surgical treatment. Results from a European multicenter study. J Endocrinol Invest. 2025 Jun;48(6):1363-1370. doi: 10.1007/s40618-025-02559-8. Epub 2025 Mar 4. PMID: 40035956; PMCID: PMC12226669.

Atila C, Lustenberger S, Chifu I, Ferrante E, Erlic Z, Drummond JB, Indirli R, Drexhage R, Powlson AS, Gurnell M, Santana Soares B, Hofland J, Beuschlein F, Fassnacht M, Winzeler B, Refardt J, Christ-Crain M. Relationship between plasma urea and copeptin in response to arginine stimulation in healthy adults, patients with vasopressin deficiency and primary polydipsia. Pituitary. 2025 Jan 25;28(1):18. doi: 10.1007/s11102-024-01489-7. PMID: 39863827; PMCID:PMC11762438.

Atila C, Winzeler B, Chifu I, Fassnacht M, Refardt J, Christ-Crain M. A post- hoc internal validation of arginine-stimulated copeptin cut-offs for diagnosing AVP deficiency (central diabetes insipidus). Pituitary. 2025 Apr 26;28(3):53. doi: 10.1007/s11102-025-01523-2. PMID: 40281371; PMCID: PMC12031852.

Detomas M, Deutschbein T, Dolce P, Möhres Y, Fassnacht M, Altieri B. Exploring sex-specific hematological changes and their impact on quality of life in patients with prolactinoma. Pituitary. 2025 Feb 3;28(1):24. doi: 10.1007/s11102-024-01493-x. PMID: 39900728; PMCID: PMC11790753.

Fuss CT, Gronemeyer K, Hermes F, Dörr M, Schmid B, Morbach C, Schmidbauer L, Schlegel N, Fassnacht M, Koschker AC, Nordbeck P, Hannemann A, Hahner S. Cardiovascular status in chronic hypoparathyroidism: a systematic cross- sectional assessment in 168 patients. Eur J Endocrinol. 2025 Mar 27;192(4):373-384. doi: 10.1093/ejendo/lvaf023. PMID: 40172208.

Nikaj A, Atila C, Chifu I, Ferrante E, Erlic Z, Drummond JB, Indirli R, Drexhage R, Powlson AS, Gurnell M, Soares BS, Hofland J, Beuschlein F, Fassnacht M, Winzeler B, Refardt J, Christ-Crain M. Disrupted ACTH and cortisol response to osmotic and non-osmotic stress in patients with arginine vasopressin deficiency. Eur J Endocrinol. 2025 Jun 30;193(1):1-9. doi: 10.1093/ejendo/lvaf119. PMID: 40498919.

Nowak E, Zhang Q, Zhang S, Zhao Y, Ye H, Machado MC, Moises CCS, Tóth M, Stark J, Yuen KCJ, Gurnell M, MacFarlane J, McCormack A, Govinna M, Gilis- Januszewska A, Minasyan M, Bonaventura I, Czepielewski MA, Ferriere A, Gadelha M, Isidori AM, Kastelan D, Maiter D, Tabarin A, Alexandraki KI, Chang J, Frontera ED, Hanzu FA, Matikainen N, Miljic D, Pichler R, Popovic V, Spencer- Segal JL, Tordjman K, Akirov A, Araujo-Castro M, Arvat E, Bancos I, Bioletto F, Burman P, Castinetti F, Detomas M, Fassnacht M, Feelders RA, Fountas A, Igaz P, Ilic S, Isand K, Kaltsas G, Meyer G, Parasiliti-Caprino M, Newell-Price J, Ragnarsson O, Valassi E, Vila G, Wass J, Yoel U, Fleseriu M, Reincke M. Cycle characterisation and clinical complications in patients with cyclic Cushing's syndrome: insights from an international retrospective cohort study. Lancet Diabetes Endocrinol. 2025 Dec;13(12):1030-1040. doi:10.1016/S2213-8587(25)00249-9. Epub 2025 Oct 23. PMID: 41235993.

Shishkov S, Iotova V, Pelsma I, Priego Zurita AL, Biermasz N; Endo-ERN Pituitary Transition of Care Study Group; Ahmed F. Feasibility of transition research in pituitary disease using patient registries: a EuRREB secondary survey. Endocr Connect. 2025 Dec 2;14(12):e250586. doi: 10.1530/EC-25-0586. PMID: 41235838; PMCID: PMC12678848.

Siggelkow H, Peschke KA, Tsourdi E, Hofbauer LC, Berr CM, Hahner S, Lottspeich C, Schmidmaier R, Blaschke M. Switch From rhPTH1-84 to TransCon PTH With Individual Dose Adjustment in Adult Hypoparathyroidism-4-Week Results. J Endocr Soc. 2025 Jun 30;9(9):bvaf113. doi: 10.1210/jendso/bvaf113. PMID: 40709359; PMCID: PMC12287600.

**Zentrum für seltene kindliche Knochenerkrankungen**

Hofmann C, Holl-Wieden A, Reiser C, Beer M, Raab P, Morbach H, Girschick HJ. Chronic non-bacterial osteomyelitis in children- five-year standardized follow-up of a prospective observational cohort in the pre-biological era. Pediatr Rheumatol Online J. 2025 May 13;23(1):50. doi: 10.1186/s12969-025-01106-2. PMID: 40361097; PMCID: PMC12076821.

Seefried L, Genest F, Hofmann C, Brandi ML, Rush E. Diagnosis and Treatment of Hypophosphatasia. Calcif Tissue Int. 2025 Mar 6;116(1):46. doi: 10.1007/s00223-025-01356-y. PMID: 40047955; PMCID: PMC11885340.

**Zentrum für seltene neuromuskuläre Erkrankungen**

Ackermann L, Zeller D, Odorfer T, Homola GA, Kampf T, Pham M, Aster HC, Sommer C. Effect of Repetitive Transcranial Magnetic Stimulation on the Symptoms and Brain Imaging in Patients with Fibromyalgia Syndrome: A Randomized Controlled Pilot Trial. Pain Ther. 2025 Oct;14(5):1547-1572. doi: 10.1007/s40122-025-00770-2. Epub 2025 Aug 30. PMID: 40884738; PMCID: PMC12484483.

Andersen NH, Borghammer P, Doppler K, Gottrup H, Knudsen K, Kuzkina A, Van Den Berge N, Otto M, Terkelsen AJ. Idiopathic polyneuropathy with neurogenic autonomic failure - an early manifestation of Lewy body disease? a case report. BMC Neurol. 2025 Oct 17;25(1):432. doi: 10.1186/s12883-025-04414-y. PMID: 41107742; PMCID: PMC12533446.

Appeltshauser L, Moritz CP, Reinhardt L, Kreß L, Üçeyler N, Lassablière F, Barcic A, Seefried S, Sommer C, Tholance Y, Antoine JC, Camdessanché JP, Doppler K. Inter-laboratory comparison of a serum fibroblast growth factor receptor 3 (FGFR3) antibody test in sensory neuropathies. Front Immunol. 2025 Oct 10;16:1604456. doi: 10.3389/fimmu.2025.1604456. PMID: 41142778; PMCID: PMC12549695.

Breyer M, Lamer S, Schlosser A, Üçeyler N. Human sensory-like neuron surfaceome analysis. PLoS One. 2025 Apr 2;20(4):e0320056. doi: 10.1371/journal.pone.0320056. PMID: 40173182; PMCID: PMC11964241.

da Cruz Neris Geßner V, Linke JT, Peulen TO, Appeltshauser L, Sommer C, Brämer D, Geis C, Heinze KG, Doppler K. Super-resolution of nodal and paranodal disruption in anti-pan-neurofascin-associated autoimmune nodopathy. Front Immunol. 2025 Feb 20;16:1540859. doi: 10.3389/fimmu.2025.1540859. PMID: 40051618; PMCID: PMC11882429.

Habib M, Wiessler AL, Fischer P, Niesner M, Selcho M, Abrante L, Werner C, Sodmann A, Koch M, Zare A, Prüss H, Dargvainiene J, Lewerenz J, Handreka R, Körtvelyessy P, Reinhold D, Thaler FS, Pitarokoili K, Kittel RJ, Briese M, Sendtner M, Rittner H, Leypoldt F, Sommer C, Blum R, Doppler K, Villmann C. Neuropathic Pain and Distinct CASPR2 Autoantibody IgG Subclasses Drive Neuronal Hyperexcitability. Neurol Neuroimmunol Neuroinflamm. 2025 Jul;12(4):e200423. doi: 10.1212/NXI.0000000000200423. Epub 2025 Jun 25. PMID: 40561371; PMCID: PMC12202018.

Heming M, Börsch AL, Wolbert J, Thomas C, Mausberg AK, Szepanowski F, Eggert B, Lu IN, Tietz J, Dienhart F, Meschnark M, Strecker JK, Glatza M, Thomas C, Gmahl N, Dambietz C, Müther M, Uerschels AK, Keyvani K, Minnerup J, Doppler K, Üçeyler N, Aprea J, Dahl A, Stassart R, Fledrich R, Wiendl H, Sommer C, Stettner M, Meyer Zu Hörste G. Multi-omic identification of perineurial hyperplasia and lipid-associated nerve macrophages in human polyneuropathies. Nat Commun. 2025 Aug 23;16(1):7872. doi: 10.1038/s41467-025-62964-8. PMID: 40849297; PMCID: PMC12375038.

Karl-Schöller F, Breyer M, Klopocki E, Üçeyler N. Generation of a gene-corrected human isogenic iPSC line from a patient with Fabry disease carrying the GLA variant c.1069C>T using CRISPR/Cas9-mediated homology directed repair. Stem Cell Res. 2025 Aug;86:103711. doi: 10.1016/j.scr.2025.103711. Epub 2025 Apr 10. PMID: 40222082.

Kreß L, Meyer Zu Altenschildesche C, Egenolf N, Sommer C, Hornemann T, Üçeyler N. Blood 1-Deoxysphingolipid Levels Are Associated With Epidermal Denervation in Small Fiber Neuropathy. J Peripher Nerv Syst. 2025 Dec;30(4):e70089. doi: 10.1111/jns.70089. PMID: 41376410; PMCID: PMC12696513.

Lleixà C, Titulaer M, Rohrbacher S, Mgbachi V, Halstead S, Fehmi J, Pascual-Goñi E, Zhu L, Appeltshauser L, Franken S, Paunovic M, Waters P, Willison H, Sommer C, Querol L, Huizinga R, Doppler K, Rinaldi S. Inter-Laboratory Validation of Nodal/Paranodal Antibody Testing. J Peripher Nerv Syst. 2025 Mar;30(1):e70000. doi: 10.1111/jns.70000. PMID: 39887819; PMCID: PMC11780190.

Meller E, Amann A, Malik RA, Kampik D, Matthies C, Üçeyler N. Clinical characterization of neuropathic pain and small fiber impairment in neurofibromatosis. Pain Rep. 2025 May 6;10(3):e1285. doi: 10.1097/PR9.0000000000001285. PMID: 40342626; PMCID: PMC12058648.

Rawat A, Vu DT, Erbacher C, Stigloher C, Üçeyler N, Metousis A, Mann M, Briese M, Sendtner M. Proteomic analysis of isolated nerve terminals from Na V 1.9 knockout mice reveals pathways relevant for pain perception. Pain. 2025 Dec 1;166(12):e770-e787. doi: 10.1097/j.pain.0000000000003743. Epub 2025 Jul 29. PMID: 40719881; PMCID: PMC12617664.

Rittel JC, Hudasch D, Doppler K, Bergh FT, Lesser M, Aktas O, Nagel M, Huttner HB, Rostasy K, Tauber S, Friese MA, Malter M, Madlener M, Kraft A, Hoffmann F, Lewerenz J, Senel M, Wickel J, Geis C, Moser A, Wandinger KP, Bartsch T, Leypoldt F, Thaler F, Kümpfel T, Meuth S, Melzer N, Finke C, Prüss H, Stangel M, Sühs KW; German Network for Research on Autoimmune Encephalitis (GENERATE). Intravenous immunoglobulin as first-line acute treatment in adults with

autoimmune encephalitis caused by antibodies to NMDAR, LGI1 and CASPR2. J Neurol. 2025 Mar 25;272(4):287. doi: 10.1007/s00415-025-13032-0. PMID: 40131535; PMCID: PMC11937155.

Seefried S, Barcic A, Grijalva Yepez MF, Reinhardt L, Appeltshauser L, Doppler K, Üçeyler N, Sommer C. Autoantibodies in patients with fibromyalgia syndrome. Pain. 2025 Feb 5;166(8):1922-1933. doi: 10.1097/j.pain.0000000000003535. PMID: 39907533.

Weigel J, Appeltshauser L, Sommer C, Doppler K. Proprioceptive endings at muscle spindles as a possible target of autoantibodies. J Neuroimmunol. 2025 Aug 15;405:578648. doi: 10.1016/j.jneuroim.2025.578648. Epub 2025 May 21. PMID: 40424942.

Wurster CD, Stolte B, Kessler T, Freigang M, Bjelica B, Ilse B, Koch JC, Cordts I, Mensch A, Zeller D, Uzelac Z, Sam G, Lapp HS, Wohnrade C, Rödiger A, Muhandes MT, Schneider I, Bellut J, Nentwich J, Dorst J, Schuster J, Schreiber-Katz O, Osmanovic A, Totzeck A, Thimm A, Steinbach R, Grosskreutz J, Kleinschnitz C, Ludolph AC, Deschauer M, Kirschner J, Dreyhaupt J, Wollinsky K, Petri S, Weiler M, Hagenacker T, Günther R. Respiratory function in 192 adult patients with spinal muscular atrophy (SMA) treated with nusinersen - a multicenter observational study. Orphanet J Rare Dis. 2025 Sep 8;20(1):476. doi: 10.1186/s13023-025-04009-3. PMID: 40922023; PMCID: PMC12418672.

**Zentrum für seltene Bewegungsstörungen**

Ganoza CA, Westenberger A, Paul JJ, Curado F, Rennecke J, Mannepalli S, Zonic E, Saravanakumar D, Paknia O, Al-Ali R, Laabs BH, Csoti I, Valzania F, Vandenberghe W, Reetz K, Afshari M, Hassin-Baer S, Fonoff ET, Gruber D, de Rosa A, Musacchio T, de Carvalho Aguiar P, Negrotti A, Tumas V, Gomez-Esteban JC, Gurevich T, Pavese N, Kulisevsky J, Sammler E, Klein C, Bauer P, Beetz C. Pathogenic or Likely Pathogenic GRN Variants Are Found in 0.1% of Parkinson's Disease Patients. Mov Disord. 2026 Apr;41(4):1020-1027. doi: 10.1002/mds.70161. Epub 2025 Dec 24. PMID: 41439479; PMCID: PMC13067306.

Krause P, Mahlknecht P, Skogseid IM, Steigerwald F, Deuschl G, Erasmi R, Schnitzler A, Warnecke T, Müller J, Poewe W, Schneider GH, Vesper J, Warneke N, Eisner W, Prokop T, Müller JU, Volkmann J, Kühn AA; Deep-Brain Stimulation for Dystonia Study Group. Long-Term Outcomes on Pallidal Neurostimulation for Dystonia: A Controlled, Prospective 10-Year Follow-Up. Mov Disord. 2025 Jun;40(6):1098-1111. doi: 10.1002/mds.30130. Epub 2025 Feb 5. PMID: 39907392; PMCID: PMC12160999.

Lange F, Guarin DL, Mosert S, Karrasch B, Roothans J, Weigl B, Navratil P, Daniels C, Odorfer T, Brandt G, Mahlknecht P, Krauss JK, Runge J, Kühn AA, Deuschl G, Volkmann J, Peach R, Reich MM. Bradykinesia induced by pallidal neurostimulation in dystonia: clinical risk factors and anatomical mapping. NPJ Parkinsons Dis. 2025 Oct 23;11(1):308. doi: 10.1038/s41531-025-01177-8. PMID: 41131044; PMCID: PMC12550046.

McFleder RL, Musacchio T, Keller J, Knorr S, Petschner T, Chen JZ, Muthuraman M, Badr M, Harder-Rauschenberger L, Kremer F, Asci S, Steinhauser S, Karl AK, Brotchie JM, Koprich JB, Volkmann J, Ip CW. Deep brain stimulation halts Parkinson's disease-related immune dysregulation in the brain and peripheral blood. Brain Behav Immun. 2025 Jan;123:851-862. doi: 10.1016/j.bbi.2024.10.039. Epub 2024 Oct 29. PMID: 39481497.

Odorfer TM, Junge A, Volkmann J, Zeller D. Temporal discrimination at the neck region is associated with severity of cervical dystonia. Sci Rep. 2025 Dec 3;15(1):43113. doi: 10.1038/s41598-025-31139-2. PMID: 41339696; PMCID: PMC12678771.

Reinhold C, Knorr S, McFleder RL, Harder-Rauschenberger L, Gräfenhan T, Schlosser A, Sendtner M, Volkmann J, Ip CW. Peripheral nerve injury induces dystonia-like movements and dysregulation in the energy metabolism: A multi-omics descriptive study in Thap1$^{+/-}$ mice. Neurobiol Dis. 2025 Feb;205:106783. doi: 10.1016/j.nbd.2024.106783. Epub 2024 Dec 26. PMID: 39732371.

Svorenova T, Romito LM, Kaymak A, Mulroy E, Cif L, Moro E, Zeuner KE, Zittel S, Petry-Schmelzer JN, Gruber D, Centen L, Albanese A, Ostrozovicova M, Han V, Magocova V, Knorovsky K, Kollova A, Garavaglia B, Golfrè-Andreasi N, Reale C, Mazzoni A, Zorzi G, Eleopra R, Levi V, Foltynie T, Limousin P, Akram H, Zrinzo L, Magrinelli F, Murphy D, Houlden H, Kurian MA, Baiata C, Paschen S, Lohmann K, Volkmann J, Hamel W, Barbe MT, van Egmond ME, Tijssen M, Ambro L, Jurkova V, Jech R, Havrankova P, Winkelmann J, Zech M, Skorvanek M. Deep Brain Stimulation for VPS16-Related Dystonia: A Multicenter Study. Ann Neurol. 2025 Oct;98(4):711-725. doi: 10.1002/ana.27290. Epub 2025 Jun 20. PMID: 40539388; PMCID: PMC12542321.

Yang J, Burrello J, Goi J, Reincke M, Adolf C, Asbach E, Brűdgam D, Li Q, Song Y, Hu J, Yang S, Satoh F, Ono Y, Libianto R, Stowasser M, Li N, Zhu Q, Hong N, Nayak D, Puar TH, Wu VC, Vaidya A, Araujo-Castro M, Kocjan T, O'Toole SM, Hundemer GL, Ragnarsson O, Lacroix A, Larose S, Nakai K, Nishikawa T, Ladygina D, Turcu AF, Sholinyan J, Fardella CE, Uslar T, Quinkler M, Mulatero P, Pintus G, Rossi GP, Hahner S, Amar L, Drake WM, Varsani C, Brown MJ, Wu X, Deinum J, Freel EM, Kline G, Naruse M, Prejbisz A, Young WF Jr, Williams TA, Fuller PJ. Outcomes after medical treatment for primary aldosteronism: an international consensus and analysis of treatment response in an international cohort. Lancet Diabetes Endocrinol. 2025 Feb;13(2):119-133. doi: 10.1016/S2213-8587(24)00308-5. Epub 2025 Jan 14. Erratum in: Lancet Diabetes Endocrinol. 2025 Mar;13(3):e6. doi: 10.1016/S2213-8587(25)00021-X. PMID: 39824204.

Zech M, Dzinovic I, Skorvanek M, Harrer P, Necpal J, Kopajtich R, Kittke V, Tilch E, Zhao C, Tsoma E, Sorrentino U, Indelicato E, Stehr A, Saparov A, Abela L, Adamovicova M, Afenjar A, Assmann B, Baloghova J, Baumann M, Berutti R, Brezna Z, Brugger M, Brunet T, Cogne B, Colangelo I, Conboy E, Distelmaier F, Eckenweiler M, Garavaglia B, Geerlof A, Graf E, Hackenberg A, Harvanova D, Haslinger B, Havrankova P, Hoffmann GF, Janzarik WG, Keren B, Kolnikova M, Kolokotronis K, Kosutzka Z, Koy A, Krenn M, Krygier M, Kusikova K, Maier O, Meitinger T, Mertes C, Milenkovic I, Monfrini E, Santos Dias Mourao A, Musacchio T, Nizon M, Ostrozovicova M, Pavlov M, Prihodova I, Rektorova I, Romito LM, Rybanska B, Sadr-Nabavi A, Schwenger S, Shoeibi A, Sitzberger A, Smirnov D, Svantnerova J, Tautanova R, Toelle SP, Ulmanova O, Vetrini F, Vill K, Wagner M, Weise D, Zorzi G, Di Fonzo A, Oexle K, Berweck S, Mall V, Boesch S, Schormair B, Prokisch H, Jech R, Winkelmann J. Combined genomics and proteomics unveils elusive variants and vast aetiologic heterogeneity in dystonia. Brain. 2025 Aug 1;148(8):2827-2846. doi: 10.1093/brain/awaf059. PMID: 39937650; PMCID: PMC12316014