FILADELFIA

ANNUAL RESEARCH REPORT 2025

Danish Epilepsy Centre Filadelfia

Content

4 Professors

2 Ass. Professors

2 post docs

14 Ph.D. students

86 Publications *)

122 Oral Presentations

**) papers published in peer reviewed journals including leading journals, such as Annals of Neurology, Brain, EBioMedicine, Neurology, American Journal of Human Genetics and Epilepsia.*

1. Introduction – Filadelfia

Filadelfia, The Danish Epilepsy Centre, is a non-profit foundation with an independent Board of Directors. In addition to the Epilepsy Hospital, Filadelfia comprises a centre for neurorehabilitation, specialized institutions for mentally handicapped persons, and the only special school for children and young people with epilepsy in Denmark. Filadelfia is publicly funded and an integral part of the Danish Healthcare System.

Filadelfia - The Danish Epilepsy Centre - offers highly specialized healthcare services to epilepsy patients of all ages and plays an integral part of the Danish Healthcare System. There is a strong emphasis on addressing the diverse needs many epilepsy patients have, social, psychological and medical, and believe that primary involvement of patients and relatives and a strong coloration between various professionals and sectors is the key to success.

As the sole specialized epilepsy hospital in Denmark, we receive referrals for rare and complex epilepsies. Our focus is on delivering high-quality comprehensive clinical care, research and development, and our achievements are internationally recognized. In 2025, our team, consisting of four professors, two associate professors, two postdocs, and 14 PhD students, published 4 book chapters and 86 research papers, including leading journals in the field of clinical neuroscience, and delivered 122 oral presentations at international medical conferences.

Our researchers and clinical experts are affiliated with the University of Copenhagen and the University of Southern Denmark, Odense. They hold important positions of trust in national and international scientific societies and serve as key hubs in numerous international epilepsy research networks.

As the directors of Filadelfia, we want to express our gratitude to all the collaborators in the research teams for their efforts and great achievements in 2025, and to Professor Sándor Beniczky for his leading role in producing this research report.

Lisbeth Nielsen
CEO
Danish Epilepsy Centre, Filadelfia

Steffen Birk
Medical Director, MD, PhD
Danish Epilepsy Centre, Filadelfia

2. Core Research Team 2025

Sándor Beniczky MD, Ph.D.

Professor, Head of Department

- Editor-in-Chief, Epileptic Disorders
- Guest editor, Epilepsia (Special Issues on AI and wearables)
- Vice-chair, ILAE Publication Council
- Chair - Virtual Epilepsy Academy (VIREPA) and member of the ILAE Education Council
- Executive committee member, European Reference Network EpiCARE
- Founding Co-Chair, ILAE Neurotechnology Section (2022-2023)
- Past-chair, Joint Taskforce on EEG of the International Federation of Clinical Neurophysiology (IFCN) and the International League Against Epilepsy (ILAE)
- Council member, ILAE Congress Council
- Council member, ILAE Publication Council
- Board member, IFCN Guidelines Committee

Rikke Steensbjerre Møller, Ph.D.

Professor, Head of Department

- Member, ILAE Task Force, “Rare Epilepsy and Genetics Big Data”
- Mentor in the ILAE Mentoring program
- Mentor in the EpiCARE Mentor-Mentee program
- Associate Editor: Epilepsia
- Mentor in the REFRESH mentorship scheme at DRCMR
- SAB member: CURE GABAA Variants
- Speaker of the Neurodevelopment Column in Neuroscience Academy Denmark
- SAB member: KCNA2 Foundation
- SAB and CAB member: KCNT1 Epilepsy Foundation
- Co-chair of WG2 (clinical genetics) in EpiCARE
- Member of the management team at Department of Regional Health Research, University of Southern Denmark, Odense, Denmark

Guido Rubboli MD, Ph.D.

Professor, Senior Consultant

- Member, ILAE Task Force, “Transition in care from Childhood into Adulthood”, member
- Director/tutor, Virtual Epilepsy Academy ILAE, Advanced EEG Course,
- Member, ILAE Task Force “SNOMED-CT”
- Member, Executive Committee, European Reference Network EpiCARE,
- Member of the EAN/EPNS, Task Force on the transition of Care
- SAB member, “Epilessia- Fuori dall’ombra”
- SAB member, KCNA2 Foundation
- Co-Chair, Coordinating Panel Rare Neurological Diseases, European Academy of Neurology
- Associate Editor: Epileptic Disorder
- Section Editor, Behavioural Neurology
- Associate Editor, Frontiers in Neurology (Associate Editor)
- Member, Editorial Board, Epilepsy and Behavior Reports Board
- Visiting Professor, Department of Child Neurology, University of Verona, Verona (Italy), 7-18 April, 2025

Elena Gardella MD, Ph.D.

Professor, Senior Consultant

- Treasurer of the Europe branch of the International League against Epilepsy (ILAE-Europe)
- Member of the commission on Classification and Terminology of the ILAE
- Co-director of VIREPA basic-EEG course of the ILAE
- Board member of the Danish Epilepsy Society (DES)
- Member of Molecular Therapeutic Board in Neurological Channelopathies Epi-CARE: a
- European Reference Network for rare and complex epilepsies.
- Coordinator the registry database and co-founder of the European STXBP1 consortium (ESCO)
- Member of the steering committee of the Danish Epilepsy Database of RKKP (Regionernes Kliniske Kvalitetsudviklingsprogram)
- Editor of Frontiers in Neurology
- SAB SCN8A Italy Foundation
- SAB: SCN8A Europe Foundation
- SAB CURE-GABAA variants Foundation

Marina Nikanorova MD, Ph.D.

Associate Professor, Senior Consultant

Allan Bayat MD, Ph.D.

Associate Professor, Head of Pediatric Research, Senior Consultant

- Board of the Danish Pediatric Neurology Society
- Senior Honorary Research Fellow University of Bristol.
- Member of the ERN-Ithaca KBG Guideline Steering committee.
- Chairman for the advisory board of rare metabolic disorders at the Danish Medical Council (https://medicinraadet.dk/om-os/fagudvalg/fenylketonuri).

Research Team

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Katrine Johannesen, MD, PhD
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Rebekka Dahl, MSc
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Valentina Rizzo, MD, Child Neurologist
Cristina Cioclu, MD
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Tanya Ramdal Techlo, MSc
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Miriam Luca, Neurologist, Italy
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Anne Juul, MSc
Betul Atak, Masterstudent Pharmacy
Ali Laith Ateya, Masterstudent Pharmacy
Rawan Ismaeil, Bachelorstudent Medicine
Vina Shorash Abdulwahed Amin, Bachelorstudent Pharmacy
Sidsel Armand Larsen, PhD student
William T. Lehn-Schiøler, PhD student
Maria Vlachou MD, PhD student
Tamir Avigdor PhD, postdoctoral fellow
Laura Craciun MD, PhD, postdoctoral fellow
Bernadett Molnár MSc
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Stefan Wüstenhagen, MD
Katarzyna Maria Mieszczanek, MD
Tomasz Stanislaw Mieszczanek, MD
Katarzyna Cebula, MD
Britta Inga Ulfsdotter Hesslow, MD

3. Ph.D. projects

Ongoing

- Sidsel Armand Larsen: Digital Technology in Epilepsy. University of Copenhagen. Main supervisor: Sándor Beniczky.
- William T. Lehn-Schiøler: Trustworthy and Explainable AI for Seizure Detection in EEG. Technical University of Denmark (DTU). Main Supervisor: Sadasivan Puthusserypady Kumaran. Co-supervisor: Sándor Beniczky.
- Laura Kristine Rasmussen: Pathomechanistic insights into GRIA3-related neurodevelopmental disorders using human iPSC-derived glutamatergic neurons with gain- or loss-of-function mutations. University of Southern Denmark. Main supervisor: Morten Meyer. Co-supervisor: Allan Bayat.
- Sebastian Ortiz: Deep phenotyping and functional characterization of GABAA-receptor related disorders:
- Translating genetic diagnostics into personalized treatment. Main supervisor: Rikke Steensbjerre Møller and
- co-supervisors: Elena Gardella and Guido Rubboli.
- Sopio Gverdtsiteli: Genotype-Phenotype Associations in Brain-Expressed Sodium Channelopathies: Commonalities, Differences, and Implications for Prognosis and Therapy. University of Southern Denmark. Main supervisor: Rikke Steensbjerre Møller and co-supervisors: Elena Gardella and Thomas Folkmann
- Valentina Rizzo: Clinical Heterogeneity in Genetic Sleep -related Hypermotor Epilepsy". Main supervisor: Elena Gardella and co-supervisors: Rikke Steensbjerre Møller and Guido Rubboli.
- Cathrine Gjerulfsen: New therapy options and experimental drugs for the treatment of severe intractable
- epilepsy and developmental and epileptic encephalopathies. University of Southern Denmark. Main supervisor: Rikke Steensbjerre Møller and co-supervisor: Guido Rubboli.
- Rebekka Dahl: Decoding GABAA Receptor Variants: Disentangling Clinical Phenotypes Through Computational Approaches. University of Southern Denmark. Main supervisor: Rikke Steensbjerre Møller and co-supervisor: Dennis Lal.
- Cristina Cioclu: Focal cortical dysplasia - pathogenetic mechanisms. University of Modena (Italy). Main supervisor: Stefano Meletti and co-supervisor: Guido Rubboli.

Completed

- Francesca Furia: Deep phenotyping of monogenic epilepsies towards the identification of targeted treatments. University of Southern Denmark. Main supervisor: Elena Gardella. Co-supervisors: Rikke Steensbjerre Møller and Guido Rubboli.
- Frederik Nørby Friis Sørensen: Dissecting neuronal heterogeneity and epileptogenesis in focal cortical dysplasia. University of Copenhagen. Main supervisor: Konstantin Khodosevich. Co-supervisor: Rikke Steensbjerre Møller.
- Tanya Ramdal Techlo: Leverage polygenic approaches to genetically diagnose idiopathic severe epilepsy and hemiplegic migraine. University of Copenhagen. Main supervisor: Thomas Folkmann Hansen. Co-supervisor: Rikke Steensbjerre Møller.
- Marie Amanda Bust Levy: Genetic and Functional Mechanisms in Neurodevelopmental Disorders and Epilepsy. University of Copenhagen. Main supervisor: Zeynep Tümer and co-supervisor: Rikke Steensbjerre Møller.
- Maria Vlachou: Evaluation of electro-clinical findings using standardised feature extraction and machine learning. Aarhus University. Main supervisor: Sándor Beniczky.

Maria Vlachou

Francesca Furia

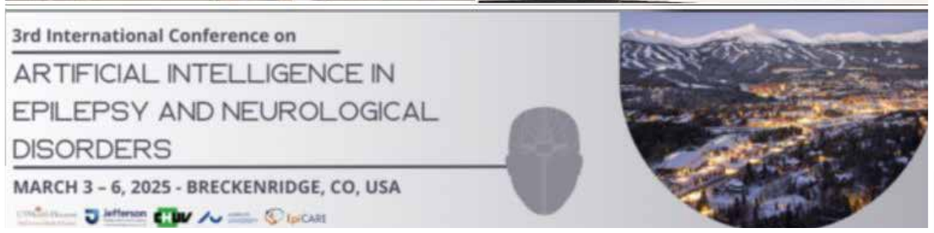
4. Conferences and congresses

3rd International Conference on Artificial Intelligence in Epilepsy and Neurological Disorders

We co-organized the 3rd International Conference on Artificial Intelligence in Epilepsy and Neurological Disorders, held in Breckenridge, Colorado, in March 2025, and co-chaired by Samden Lhatoo, Sándor Beniczky, Philippe Ryvlin, and Michael Sperling.

The scientific program featured state-of-the-art presentations on the application of artificial intelligence across the spectrum of epilepsy and neurological disorders, including EEG analysis, seizure detection and forecasting, neuroimaging, and clinical decision support systems. The meeting comprised a comprehensive program of lectures, oral communications, and poster presentations highlighting cutting-edge methodological advances and their translation into clinical practice.

The conference brought together an international, multidisciplinary audience of clinicians, researchers, and industry partners, fostering active discussion and collaboration in this rapidly evolving field.



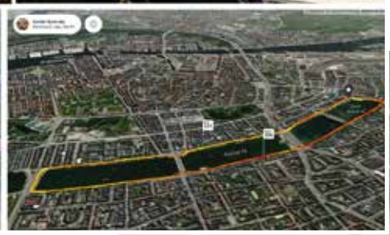
Pictures from the 3rd International Conference on Artificial Intelligence in Epilepsy and Neurological Disorders.

5th International Congress on Mobile Health and Digital Technology in Epilepsy

We co-organized the 5th International Congress on Mobile Health and Digital Technology in Epilepsy, held in Copenhagen, Denmark, in September 2025, and co-chaired by Sándor Beniczky and Philippe Ryvlin.

The scientific program focused on advances in mobile health and digital technologies in epilepsy, including wearable devices for seizure detection and forecasting, mobile applications for patient monitoring and self-management, and the integration of artificial intelligence into clinical decision support systems. The meeting featured a comprehensive program of lectures, oral communications, and poster presentations, highlighting innovative approaches and real-world implementation of digital tools in epilepsy care.

The congress brought together an international, multidisciplinary audience of clinicians, researchers, engineers, and industry partners, fostering collaboration and accelerating the translation of digital innovations into clinical practice.



Pictures from the 5th International Congress on Mobile Health and Digital Technology in Epilepsy in Copenhagen.

Sándor Beniczky has received from the Indian Academy of Neurology the JS Chopra prize and gave the named lecture at the 32nd annual congress of the Academy, in Varanasi, India.

After serving as professor for 8 years at Aarhus University, Sándor Beniczky moved his academic affiliation to the University of Copenhagen. The photo shows the professor inauguration.

Congress/meeting organization – Guido Rubboli

- 7th Annual Nordic Epilepsy Meeting on Excellence in Epilepsy Management Care (in collaboration with Morten Lossius), Oslo, 6 November 2025.
- Expert Perspectives from Denmark and Beyond – Real-World Clinical Experience with Cenobamate (in collaboration with Helle Pfeiffer), Dianalund, 11 December 2025.

Upperpanel: P.Ryvlin, G.Rubboli, R.Kalviainen, M.Lossius; lower panel: the winners of the Young Recipient Awards.

Congress/meeting organization – Elena Gardella

- 3rd SCN2A & SCN8A scientific conference and gathering meeting (in collaboration with Walid Fazeli), Bonn (Germany), May 16th-17th, 2025.

Scientific committee

- Spring DES annual meeting, Middelfart (Denmark), March 6th-7th, 2025.
- 2nd European STXBP1 Summit and Research Roundtable, Heidelberg (Germany), October 8th-10th, 2025.
- Autumn DES annual meeting, Copenhagen (Denmark), November 6th, 2025.
- In Search of Lost Time 6, Rome (Italy), November 12th-14th, 2025.

5. Research projects

Artificial Intelligence: an emerging tool in epilepsy care

Recent literature highlights artificial intelligence (AI) as a rapidly evolving tool with transformative potential across epilepsy care, from diagnosis to prevention. We contributed to the development of a global roadmap, which emphasizes that AI implementation must be ethical, equitable, and clinically effective, calling for robust governance, diverse datasets, and international collaboration to avoid bias and disparities. Complementing this, we outline how AI, digital technologies, and mobile health are already reshaping epilepsy management through automated EEG interpretation, seizure detection, and remote monitoring.

Advances in phenotyping demonstrate AI's ability to integrate multimodal data - clinical, electrophysiological, and imaging - to refine epilepsy classification and support precision medicine. However, evaluation of AI chatbots in EEG-related queries reveals variable accuracy and reliability, underscoring the need for clinical validation before widespread adoption. Importantly, patient-centered perspectives are emerging: studies on AI-driven closed-loop devices for SUDEP prevention highlight both optimism and concerns regarding safety, autonomy, and trust.

Together, these works suggest that while AI holds significant promise to improve outcomes and accessibility in epilepsy care, careful validation, ethical oversight, and user engagement remain essential for responsible integration into clinical practice.

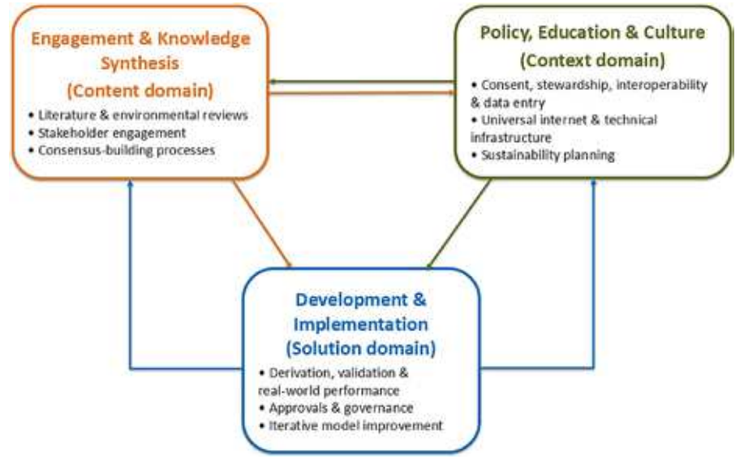


Figure 7. Roadmap for ethical, equitable, and effective artificial intelligence to improve care for all people with epilepsy

Papers

1. Josephson CB, **Beniczky S**, Denaxas S, Ikeda A, Jehi L, Mwesige AK, Jette N, Jones GD, Ryvlin P, Sen A, Triki CC, Waters G, Guekht A, Cross JH. A call for ethical, equitable, and effective artificial intelligence to improve care for all people with epilepsy: A roadmap. A report by the ILAE Global Advocacy Council and Big Data Commission. Epilepsia. 2025 Dec 24. doi: 10.1002/epi.70058. PMID: 41443971.
2. Proença S, Soares JI, Parra J, Maia G, Silva S, Leite J, **Beniczky S**, Jesus-Ribeiro J. Comparative evaluation of artificial intelligence chatbots in answering electroencephalography-related questions. Epileptic Disord. 2025 Dec 16. doi: 10.1002/epd2.70156. PMID: 41399926.
3. Ferreira J, França M, Regalo MC, Rei M, Peixoto R, Aibar JÁ, Robinson T, Matias R, Duprat F, Mantegazza M, Parlak O, Ryvlin P, **Beniczky S**, Lopes L, Perucca E, Claro J, Conde C. Artificial intelligence-driven closed-loop devices in sudden unexpected death in epilepsy prediction and prevention: Insights from persons with epilepsy and caregivers. Epilepsia. 2026 Jan;67(1):175-186. doi: 10.1111/epi.18647. PMID: 40985901; PMCID: PMC12893267.
4. **Beniczky S**, Lhatoo S, Sperling MR, Ryvlin P. Artificial intelligence, digital technology, and mobile health in epilepsy. Epilepsia. 2025 Sep;66 Suppl 3:1-3. doi: 10.1111/epi.18435. PMID: 40286052.
5. Knight A, Gschwind T, Galer P, Worrell GA, Litt B, Soltesz I, **Beniczky S**. Artificial intelligence in epilepsy phenotyping. Epilepsia. 2025 Sep;66 Suppl 3(Suppl 3):39-52. doi: 10.1111/epi.17833. PMID: 37983589; PMCID: PMC11102939.

Postgraduate education and research

Postgraduate education in epileptology is undergoing significant transformation, driven by digital innovation, global collaboration, and evolving clinical needs. We are proud to contribute to these important, international endeavors. The ILAE Academy has established a structured, tiered curriculum (Levels 1–3) that standardizes training worldwide while remaining adaptable to local contexts. This framework supports progressive competency development, from foundational knowledge to advanced expertise, and represents a major step toward harmonizing education across regions.

Innovative teaching formats are expanding access and engagement. Experiences from "EEG Talk" demonstrate how interactive e-learning can effectively deliver high-quality EEG education globally, while recent work highlights the growing role of artificial intelligence in augmenting EEG training through automated feedback and decision support. At the same time, disparities persist: studies from sub-Saharan Africa emphasize the need for targeted training of non-specialists, reflecting workforce shortages and resource limitations. Surveys of epilepsy fellowship programs reveal variability in exposure to key areas such as epilepsy surgery, underscoring the need for more standardized curricula.

Complementing these research-focused contributions, several of our 2025 publications provide practical educational material, including step-by-step guides to EEG interpretation and teaching cases on seizure semiology, normal variants, and electromyography patterns.

Together, these efforts illustrate a shift toward more accessible, standardized, and technology-enhanced education, while highlighting ongoing gaps that must be addressed to ensure equitable training worldwide.

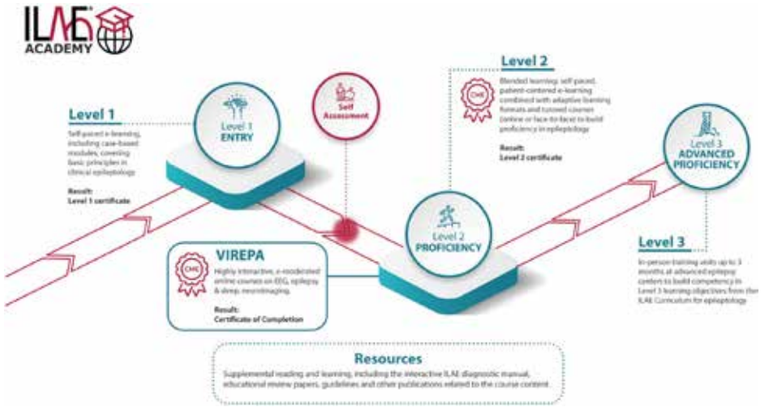


Figure 8. Continuing medical education in epileptology

Papers

1. Nascimento FA, Yuan D, Sheikh IS, Hirsch LJ, **Beniczky S**, Westover MB. The EEG Talk experience: Lessons in e-teaching EEG. Epileptic Disord. 2025 Dec;27(6):1337-1339. doi: 10.1002/epd2.70096PMID: 40904123.
2. Blümcke I, Biesel E, Wiebe S, **Beniczky S**, Wilmshurst JM, Mehndiratta MM, Asadi-Pooya AA, Brandt C, Arzimanoglou A, Singh G; ILAE Academy content development faculty. Continuing medical education in epileptology: The Level 1-2-3 experience of the ILAE academy. Epileptic Disord. 2025 Oct;27(5):707-717. doi: 10.1002/epd2.70045. PMID: 40423759; PMCID: PMC12574487.
3. Butler MR, Marcinski Nascimento KJ, **Beniczky S**, Nascimento FA. How to Read an EEG: A Step-by-Step Guide. Neurol Educ. 2025 Mar 7;4(1):e200208. doi: 10.1212/NE9.0000000000200208. PMID: 40104780; PMCID: PMC11919387.
4. McLaren JR, Yuan D, **Beniczky S**, Westover MB, Nascimento FA. The future of EEG education in the era of artificial intelligence. Epilepsia. 2025 Jun;66(6):1838-1842. doi: 10.1111/epi.18326. PMID: 40035709; PMCID: PMC12169395.
5. Kander V, Valente KD, Carrizosa J, Vidaurre J, Patel AA, Triki CC, Aljandeel G, Singh G, Kato M, Seck L, Kone Z, **Beniczky S**, Asukile MT, Birbeck GL, Jones K, Boylan G, Hardman J, Wilmshurst JM. Expert opinions on pediatric EEG training for non-epilepsy specialists in sub-Saharan Africa. Epileptic Disord. 2025 Jun;27(3):374-388. doi: 10.1002/epd2.70010. PMID: 40035141; PMCID: PMC12203304.
6. Katyal R, Sheikh IS, Gutierrez C, Sinha SR, Day BK, Gavvala JR, Sheth SA, Wirrell E, **Beniczky S**, Nascimento FA. Epilepsy Surgery Education: A Survey of US Epilepsy Fellowship Program Directors. J Clin Neurophysiol. 2026 Jan 1;43(1):1-8. doi: 10.1097/WNP.0000000000001144. Epub 2025 Feb 12. PMID: 39934975.
7. Marcinski Nascimento KJ, King A, **Beniczky S**, Nascimento FA. Teaching Video NeuroImage: Rhythmic Ictal Nonclonic Hand Motions: A Valuable Lateralizing Sign in Focal Epilepsy. Neurology. 2025 Feb 11;104(3):e213346. doi: 10.1212/WNL.0000000000213346. Epub 2025 Jan 15. PMID: 39813631.
8. Marcinski Nascimento KJ, Nascimento FA, **Beniczky S**. Surface electromyography patterns of epileptic seizures. Epileptic Disord. 2025 Feb;27(1):130-134. doi: 10.1002/epd2.20314. Epub 2024 Nov 22. PMID: 39576192.
9. Marcinski Nascimento KJ, **Beniczky S**, Nascimento FA. Slow alpha variant: A normal EEG pattern. Epileptic Disord. 2025 Feb;27(1):127-129. doi: 10.1002/epd2.20313. Epub 2024 Nov 19. PMID: 39560654.

EEG biomarkers I

EEG biomarkers are increasingly central to improving diagnosis, prognosis, and treatment planning in epilepsy. A joint report from the International League Against Epilepsy and the International Federation of Clinical Neurophysiology confirms that interictal epileptiform discharges (IEDs) remain a key prognostic biomarker, significantly increasing the risk of seizure recurrence after a first unprovoked seizure and supporting their role in clinical decision-making.

At a more advanced level, intracranial EEG biomarkers are gaining validation. Multicenter studies demonstrate that fast ripples recorded during stereo-EEG are reliable markers of the epileptogenic zone, while specific visual SEEG features can help predict surgical outcomes. These findings strengthen the role of quantitative and invasive EEG biomarkers in guiding epilepsy surgery.

Standardization and validation frameworks are also evolving. The SzCORE initiative provides an open, community-driven framework for benchmarking automated seizure detection algorithms, promoting transparency and reproducibility in biomarker development.

Together, these studies illustrate a shift from traditional visual EEG interpretation toward quantitative, validated biomarkers across multiple scales. This progression supports more precise risk stratification, improved surgical targeting, and the development of reliable AI-based tools, ultimately advancing personalized epilepsy care.

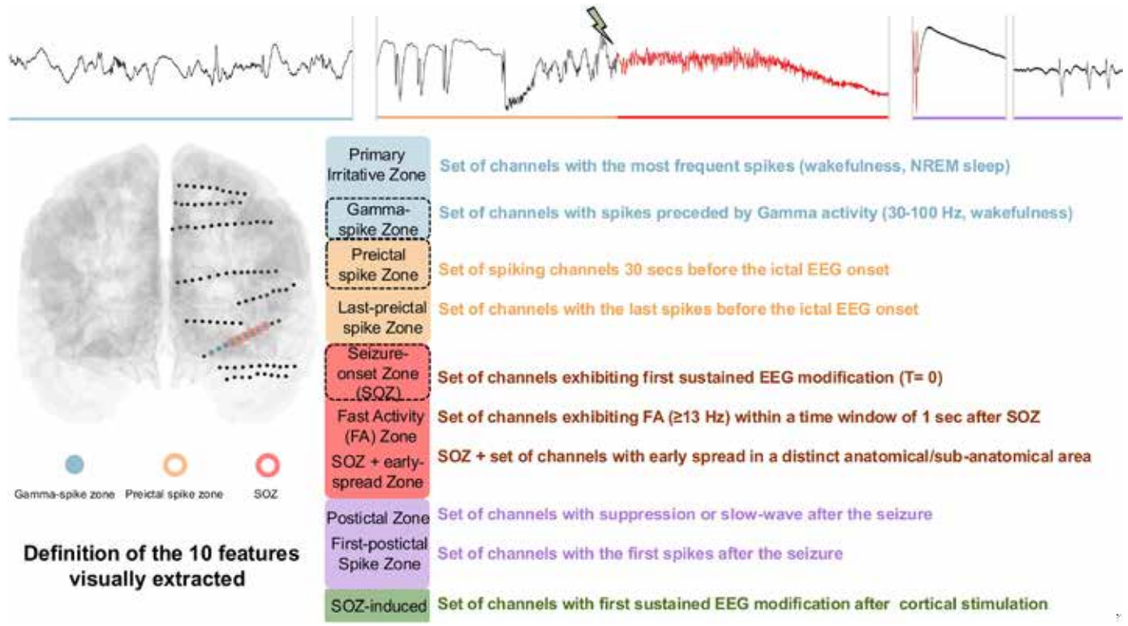


Figure 9. SEEG biomarkers

Papers

1. Baykan B, Dunne J, Wiebe S, Maillard L, **Beniczky S**, Koutroumanidis M, Seeck M. Presence of interictal epileptiform EEG discharges implies increased risk of recurrence after the first unprovoked seizure: Report of the International League Against Epilepsy and International Federation of Clinical Neurophysiology. Epilepsia. 2025 Oct;66(10):3920-3936. doi: 10.1111/epi.18591. PMID: 40875494; PMCID: PMC12605794.
2. Nevalainen P, von Ellenrieder N, Dudley RWR, Balasubramaniam N, **Beniczky S**, Cacic Hribljan M, Fabricius M, Ho A, Jonsson H, Meidahl A, Michaud E, Nikolic M, Rasmussen R, Salli E, Sidaros A, Frauscher B, Gotman J. Fast Ripples Measured From Overnight SEEG Recordings as Markers of the Epileptogenic Zone: A Multicenter Validation Study. Neurology. 2026 Jan 27;106(2):e214511. doi: 10.1212/WNL.0000000000214511. Epub 2025 Dec 26. PMID: 41453125.
3. Abdallah C, Thomas J, Aron O, Avigdor T, Jaber K, Doležalová I, Mansilla D, Nevalainen P, Parikh P, Singh J, **Beniczky S**, Kahane P, Minotti L, Chabardes S, Colnat-Coulbois S, Maillard L, Hall J, Dubeau F, Gotman J, Grova C, Frauscher B. Visual Features in Stereo-Electroencephalography to Predict Surgical Outcome: A Multicenter Study. Ann Neurol. 2025 Sep;98(3):547-560. doi: 10.1002/ana.27278.PMID: 40519108; PMCID: PMC12392059.
4. Dan J, Pale U, Amirshahi A, Cappelletti W, Ingolfsson TM, Wang X, Cossettini A, Bernini A, Benini L, **Beniczky S**, Atienza D, Ryvlin P. SzCORE: Seizure Community Open-Source Research Evaluation framework for the validation of electroencephalography-based automated seizure detection algorithms. Epilepsia. 2025 Sep;66 Suppl 3(Suppl 3):14-24. doi: 10.1111/epi.18113. PMID: 39292446; PMCID: PMC12489712.

Wearable devices

Wearable devices are rapidly advancing as practical tools for continuous, home-based monitoring in epilepsy care. This is one of our major field of research and development. Our recent work highlights how miniaturized neurophysiological technologies, such as wearable EEG, ECG, and motion sensors, enable long-term recording of brain and autonomic signals outside the hospital, supporting more ecologically valid assessment of seizure activity.

Clinical validation studies demonstrate promising performance. Smartphone-connected wearable ECG systems can detect seizures with clinically relevant accuracy, while wrist-worn accelerometers using deep learning show strong potential for identifying generalized convulsive seizures. Simpler systems, including reduced-channel EEG, have also proven capable of detecting clinically important markers such as post-ictal generalized EEG suppression, which may relate to SUDEP risk.

Beyond technical performance, patient perspectives are central to successful implementation. Users generally express strong interest in wearables for safety and seizure tracking, but emphasize the importance of comfort, discretion, reliability, and data privacy. False alarms and device burden remain key concerns that may affect long-term adherence.

Overall, wearable technologies are moving from proof-of-concept toward real-world clinical application. Their integration into routine care could enable continuous monitoring, earlier intervention, and improved patient safety, provided that accuracy, usability, and patient-centered design remain priorities.

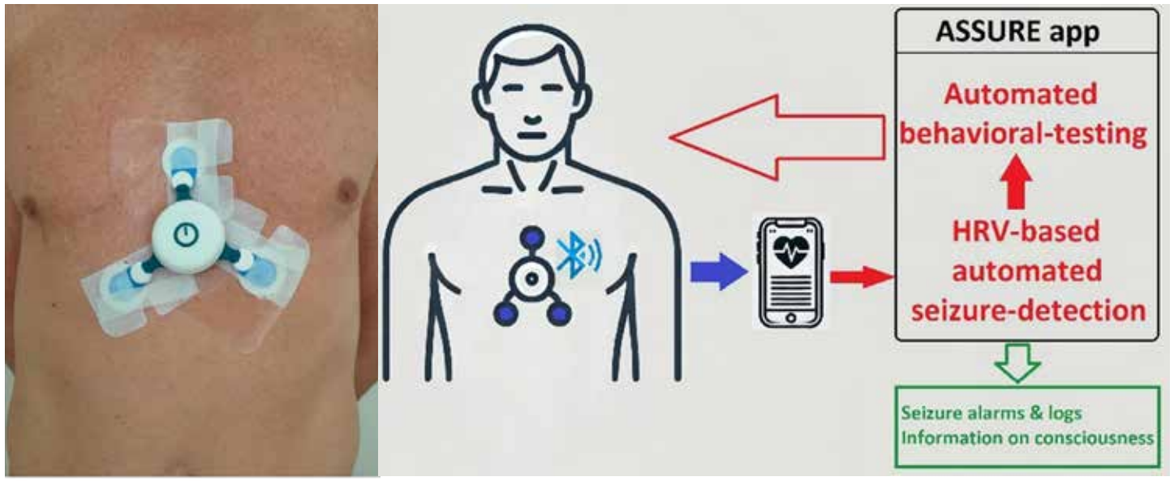


Figure 10. ECG-based seizure detection using wearable device

Papers

1. Musaeus CS, Viana PF, Cook M, Duun-Henriksen J, **Beniczky S**, Kidmose P, Vanrumste B, Filtjens B, Kjaer TW. Home-Based sensing of the nervous system with clinical neurophysiology technologies: IFCN handbook chapter. Clin Neurophysiol Pract. 2025 Sep 21;10:453-463. doi: 10.1016/j.cnp.2025.09.003. PMID: 41142046; PMCID: PMC12547184.
2. Jeppesen J, Christensen J, Ahrenfeldt Petersen O, **Fenger S**, **Armand Larsen S**, **Wüstenhagen S**, Wagner SR, Johansen P, **Beniczky S**. Seizure detection using wearable electrocardiogram connected to a smartphone: a phase 3 clinical validation study. EBioMedicine. 2025 Oct;120:105952. doi: 10.1016/j.ebiom.2025.105952. PMID: 41027311; PMCID: PMC12516532.
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5. Hadady L, Robinson T, Bruno E, Richardson MP, **Beniczky S**. Users' perspectives and preferences on using wearables in epilepsy: A critical review. Epilepsia. 2025 Sep;66 Suppl 3(Suppl 3):4-13. doi: 10.1111/epi.18280. PMID: 39871791; PMCID: PMC12489716.

Seizure classification

We had the honor to lead the international working group that developed the update of the International League Against Epilepsy (ILAE) seizure classification. This represents an important step toward improving clarity, usability, and global applicability. Building on earlier frameworks, the revised classification refines seizure descriptors, emphasizes observable clinical features, and aligns terminology more closely with underlying neurobiology. A key objective is to make classification practical across diverse clinical settings, including regions with limited access to advanced diagnostics.

The accompanying ILAE position paper and practical guide highlight a structured approach based on seizure onset (focal, generalized, or unknown), with expanded descriptors to better capture motor and non-motor features. Greater flexibility is introduced to accommodate incomplete information, supporting real-world clinical decision-making. Importantly, the update also aims to improve communication with patients and caregivers by promoting clearer, more intuitive terminology.

Early evaluations suggest good feasibility in secondary referral centers, with clinicians able to apply the updated system consistently in routine practice. Surveys further indicate broad acceptance among professionals, although they underscore that precise use of terminology remains essential to avoid misclassification.

Overall, the updated classification enhances both clinical utility and global relevance, supporting more accurate diagnosis, research standardization, and communication—key elements for advancing epilepsy care worldwide.

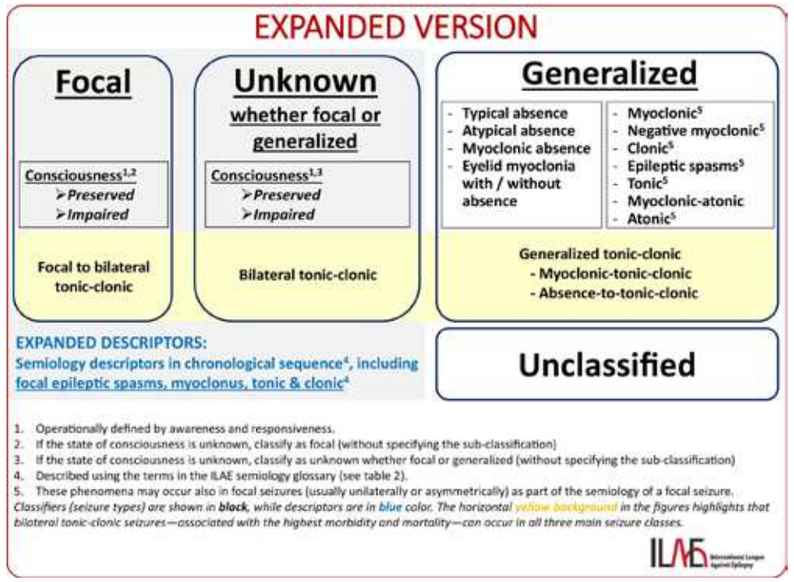

Figure 11. Updated ILAE seizure classification 2025

Papers

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Guidelines, standards and best practices

In 2025 we continued to work on quality assurance, based on the growing need for standardized frameworks to improve the quality, reproducibility, and clinical utility of EEG and epilepsy research. A major advance is the development of structured data annotation systems, such as the Hierarchical Event Descriptor (HED) schema, which enables consistent and machine-readable labeling of EEG events, facilitating data sharing and large-scale analyses.

Efforts to refine clinical standards are equally important. Proposals to include explicit filter settings within ACNS terminology highlight how technical variability can influence EEG interpretation, emphasizing the need for greater transparency in routine practice. Similarly, application of IFCN criteria using “decision hygiene” approaches has been shown to improve inter-rater reliability in identifying interictal epileptiform discharges, addressing a longstanding challenge in EEG interpretation.

In clinical research, the introduction of the GREENBEAN checklist provides structured guidance for reporting studies on EEG-based biomarkers, promoting methodological rigor and reproducibility. Complementing these initiatives, surveys of neurology professionals reveal ongoing variability in perceived standards of care, indicating gaps between recommendations and real-world practice.

Collectively, these contributions reflect a concerted move toward harmonization of terminology, methodology, and reporting. Strengthening guidelines and best practices is essential to enhance reliability, enable multicenter collaboration, and support the integration of advanced technologies into clinical neurophysiology.

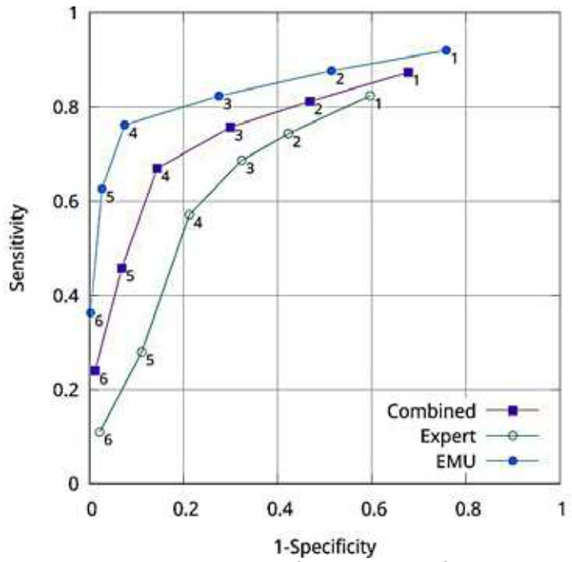

Figure 12. ROC curves condtructed from the number of IFCN criteria.

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1. Hermes D, Pal Attia T, **Beniczky S**, Bosch-Bayard J, Delorme A, Lundstrom BN, Rogers C, Rampp S, Shirazi SY, Truong D, Valdes-Sosa P, Worrell G, Makeig S, Robbins K. Hierarchical Event Descriptor library schema for EEG data annotation. Sci Data. 2025 Aug 19;12(1):1448. doi: 10.1038/s41597-025-05791-2. PMID: 40830348; PMCID: PMC12365201.
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5. Nascimento FA, Katyal R, Kass NR, Yuan D, Sirven JI, Westover MB, **Beniczky S**. Electroencephalography in Clinical Practice: Neurology Professionals’ Views on Optimal Standards of Care. J Clin Neurophysiol. 2025 Nov 1;42(7):639-642. doi: 10.1097/WNP.0000000000001142. PMID: 39820182.

Developmental and Epileptic Encephalopathies: From Gene Discovery to Personalized Therapy

Developmental and epileptic encephalopathies (DEEs) are a group of severe early-onset epilepsies characterized by refractory seizures and significant developmental impairment, where both the underlying etiology and the epileptic activity itself contribute to cognitive and behavioral deterioration. Many DEEs have a monogenic cause, and advances in genomic technologies have substantially improved diagnostic yield. Although individual syndromes are rare, collectively DEEs represent an important cause of childhood epilepsy with considerable morbidity and long-term healthcare impact.

Our research spans the full translational spectrum - from patient registries and natural history studies to mechanistic laboratory investigations and clinical trial readiness. We aim to systematically characterize the phenotypic spectrum of genetic epilepsies and identify demographic, genetic, and clinical variables that influence disease progression and prognosis. By leveraging structured patient registries and longitudinal natural history studies (see section below), we generate high-quality datasets that inform patient care, enable biomarker identification (see section below), and define clinically meaningful outcome measures.

At the same time, we work in close collaboration with basic scientists to integrate functional laboratory data with detailed clinical phenotyping. By linking molecular mechanisms to patient-based observations, we aim to establish actionable genotype–phenotype correlations and identify therapeutic targets.
Through this integrated bench-to-bedside approach, we seek to accelerate the translation of genetic discoveries into precision diagnostics and mechanism-based, personalized therapies for individuals with monogenic epilepsies, ultimately improving outcomes and access to innovative treatments. Multiple papers have been published under this theme, some of which are highlighted below.

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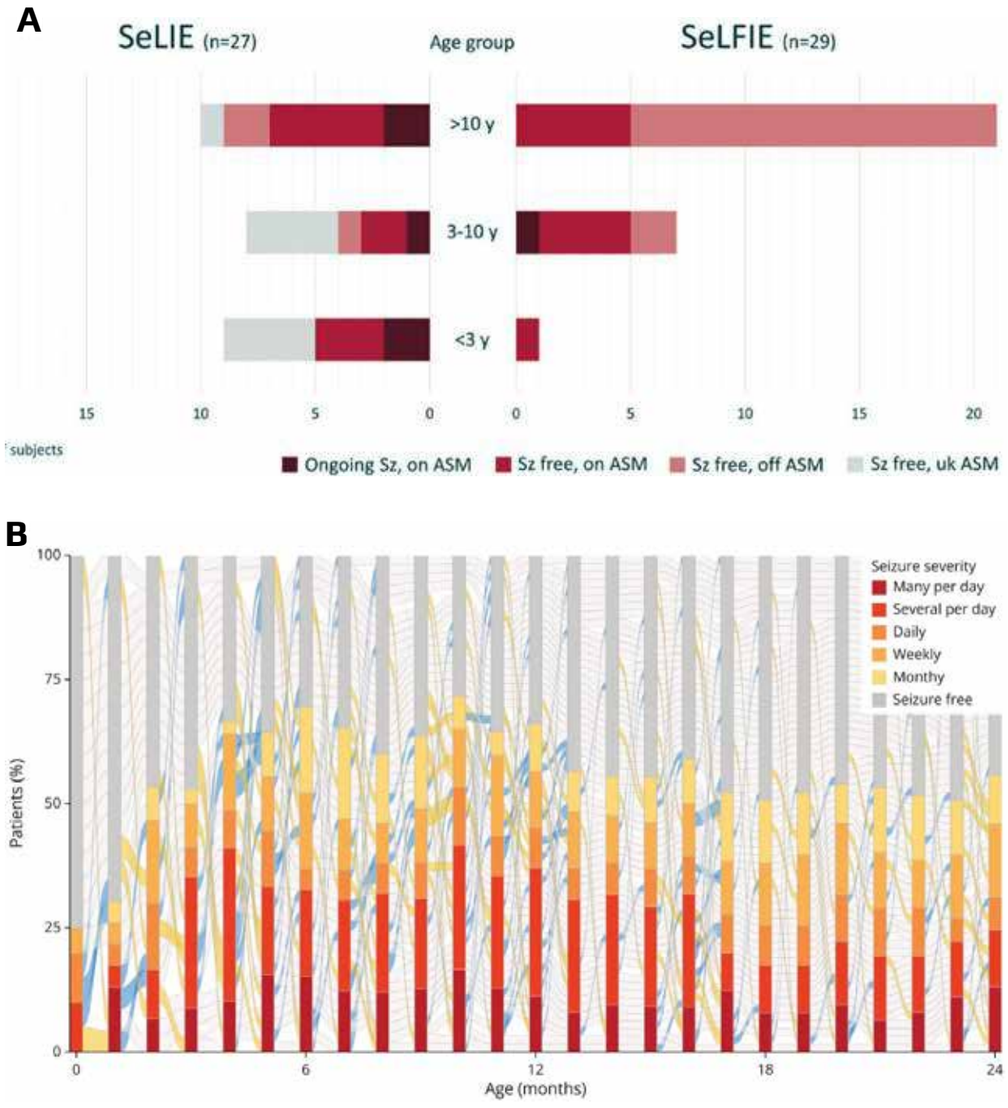
3. **De la Rosa SO**, **Rizzo V**, Jauss RT, Bartolomaeus T, Escolar M, Bernard G, Gavrilova R, Ahrens-Nicklas R, Lemire G, Boycott KM, Mercimek-Andrews S, Prontera P, Costa C, Rakic B, Boerkoel CF, Huynh S, Huh L, Sherr E, Argili E, Ortigoza-Escobar JD, Casas-Alba D, Nunes T, Koolen DA, Platzer K, **Khinchi MS**, **Gardella E**, **Fenger CD**, **Møller RS**, **Bayat A**. MBOAT7 encephalopathy: Characterizing the neurology and epileptology. Epilepsia. 2025 Jul;66(7):2379-2390. doi: 10.1111/epi.18376. Epub 2025 Mar 21.
4. Magielski JH, Cohen S, Kaufman MC, Parthasarathy S, Xian J, Brimble E, Fitter N, **Furia F**, **Gardella E**, **Møller RS**, Helbig I, McKee JL. Deciphering the Natural History of SCN8A-Related Disorders. Neurology. 2025 May 13;104(9):e213533. doi: 10.1212/WNL.0000000000213533. Epub 2025 Apr 14.
5. **De Wachter M**, Millevert C, Nicolai J, Cats E, Kluger G, Milh M, Cloarec R, Syrbe S, Arts K, Jansen K, Krygier M, Smigiel R, Auvin S, **Olofson K**, **Gjerulfsen CE**, Ceulemans B, **Møller RS**, **Bayat A**, Weckhuysen S. Amitriptyline use in individuals with KCNQ2/3 gain-of-function variants: A retrospective cohort study. Epilepsia. 2025 May;66(5):1628-1640. doi: 10.1111/epi.18310. Epub 2025 Feb 17.
6. Lemke JR, Eoli A, Krey I, Popp B, Strehlow V, Wittekind DA, Vuorinen AL, Aldhalaan HM, Baer S, de Saint Martin A, **Hammer TB**, Herman I, Hornemann F, Ingebrigtsen T, Lederer D, Lesca G, Marafie D, Mathot M, Rosenfeld JA, **Møller RS**, Schelhaas HJ, Stillman C, Orsini A, Patel AD, Piard J, Veggiotti P, Vlaskamp DRM, Weckhuysen S, Traynelis SF, Benke TA, Heyne HO, Syrbe S. GRIN2A null variants confer a high risk for early-onset schizophrenia and other mental disorders and potentially enable precision therapy. Mol Psychiatry. 2026 Jan;31(1):374-382. doi: 10.1038/s41380-025-03279-4. Epub 2025 Oct 14.
7. **Bayat A**, Borroto MC, Salian S, Zaki MS, Benkerroum H, Elbendary HM, Nguyen TTM, Sadek AA, Carli D, Brusco A, Ferrero GB, Tartaglia M, Hay E, Krey I, A Jamra R, Bartolomaeus T, Knaus A, Gleeson JG, Houlden H, Dominik N, Jackson A, Douzgou Houge S, Banka S, Mohammadi-Asl J, Hajjari M, Azizimalamiri R, Nourbakhsh P, Neissi M, Scardamaglia A, Li D, Kinoshita T, Maroofian R, Murakami Y, Campeau PM. PIGC-related encephalopathy: Lessons learned from 18 new probands. Eur J Hum Genet. 2025 Dec;33(12):1636-1646. doi: 10.1038/s41431-025-01923-9. Epub 2025 Sep 17.

Natural History Studies_SCN8A-related disorders and beyond

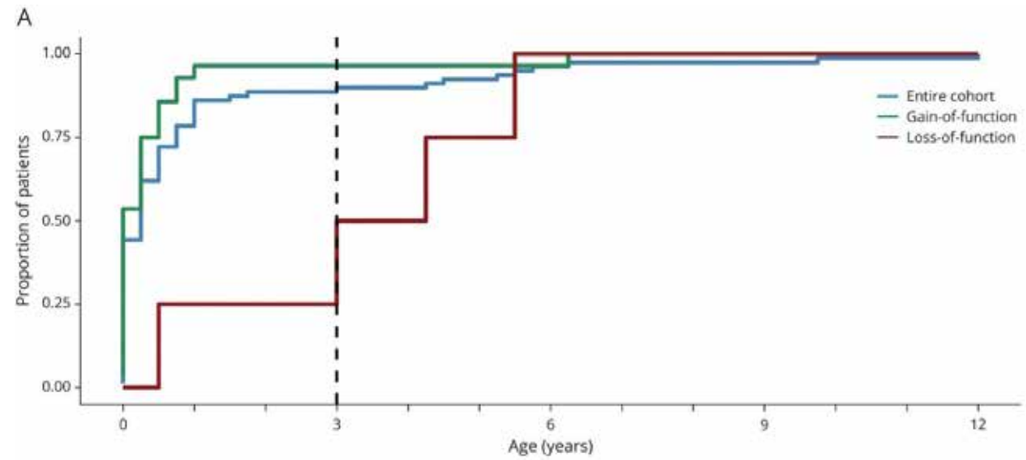
Natural History Studies (NHS) are essential for understanding the progression, clinical features, and underlying mechanisms of rare diseases, including monogenic epilepsies. We are developed several protocols for retrospective and prospective NHS, based on solid disease specific registries and sub-phenotypes stratification (Figure 13 and 14). These studies follow individuals over time, examining not only epilepsy features but also comorbidities and cognitive, behavioral, developmental, and social dimensions. By offering detailed insight into how monogenic epilepsies present and change across lifespan, NHS establish critical reference points for evaluating emerging therapies.

In the setting of clinical trials for monogenic epilepsies, such studies are fundamental in assessing trial readiness. A comprehensive characterization of disease trajectories enables researchers to identify suitable patient populations, select meaningful outcome measures, and design studies tailored to the specific challenges of these disorders.

Our most advanced NHS model is for SCN8A related disorders, on this basis other studies have been developed (STXBP1, CDKL5, SLC6A1, etc).



Example of patients' stratification in SCN8A in self-limited infantile epilepsy (SeLIE) versus self-limited familial infantile epilepsy (SeLFIE), showing that the different populations have different epilepsy outcomes and antiseizure medication (ASM) response. There was a significant association between the group (SeLFIE vs. SeLIE) and epilepsy outcome/ASM dependence (p = .009) in the >10 years age group. (2) Global approach to retrospective NHS for SCN8A related disorders, exploring (A) Seizure onset in individuals with GOF vs LOF subcohort. (B) Seizure history reconstruction over time.



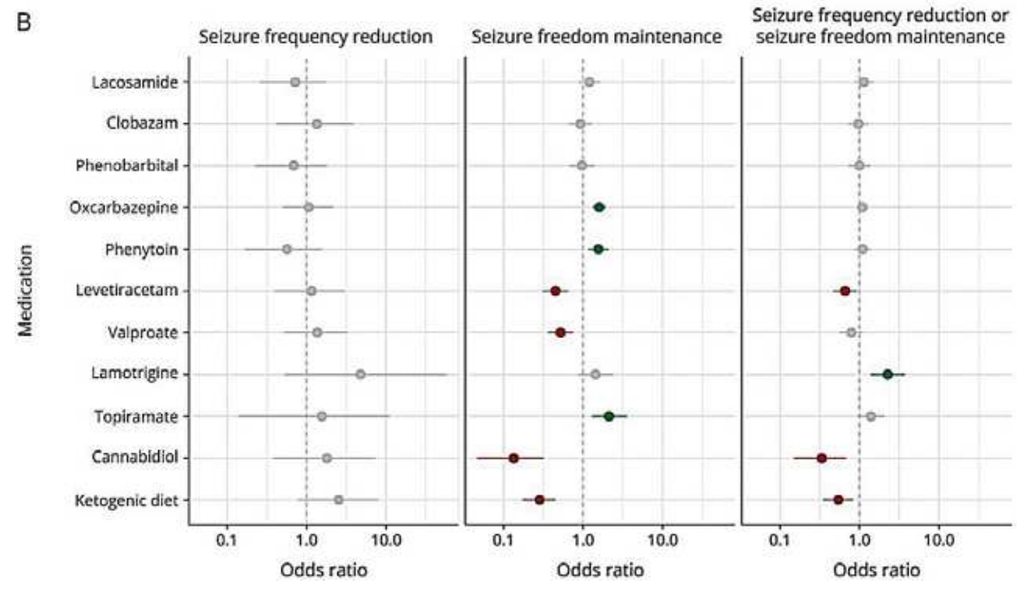


Figure 14. (A) Prescription patterns before and after the SCN8A diagnosis. (B) Medication effectiveness analysis.

Papers

1. **Furia F**, **Gverdtsiteli S**, Janzarik W, Korff C, Lesca G, Mancardi MM, Montomoli M, **Nikanorova M**, Romaniello R, **Rubboli G**, Syrbe S, Vigevano F, **Møller RS**, **Gardella E**. Differential outcomes in familial and sporadic SCN8A self-limited infantile epilepsies: Insights from a large international registry. Epilepsia. 2025 Oct;66(10):4066-4069. doi: 10.1111/epi.18569. Epub 2025 Aug 18.
2. **SCN8A Research Consortium** (**Gardella E**). A research roadmap for SCN8A-related disorders: addressing knowledge gaps and aligning research priorities across stakeholders. Orphanet J Rare Dis. 2025 Aug 19;20(1):444. doi: 10.1186/s13023-025-03672-w.
3. Magielski JH, Cohen S, Kaufman MC, Parthasarathy S, Xian J, Brimble E, Fitter N, **Furia F**, **Gardella E**, **Møller RS**, Helbig I, McKee JL. Deciphering the Natural History of SCN8A-Related Disorders. Neurology. 2025 May 13;104(9):e213533. doi: 10.1212/WNL.0000000000213533. Epub 2025 Apr 14.

Life Beyond Childhood: Experiences from KBG Syndrome

KBG syndrome is a rare genetic condition affecting brain development, often causing learning difficulties, epilepsy, and behavioral challenges (figure 15). While it is well described in children, adult life has remained largely unexplored—until now.

In 2025, we contributed two exciting and complementary studies that shed new light on adulthood in KBG syndrome. Our first study (physician-reported) brought together international clinical expertise to describe the natural history of adults with KBG syndrome. We showed that many adults continue to live with intellectual disability, epilepsy, and psychiatric challenges, but also highlighted the remarkable diversity in abilities, independence, education, and work (figure 16). Our second study (patient- and family-reported) captured something even more powerful—the real-life experiences of 91 individuals aged 16–86 years. Here, all data came directly from patients and families. We found that development continues into adulthood, with many gaining independence over time. At the same time, ongoing challenges such as seizures, mental health issues, and physical health problems still significantly affect daily life—especially for those with active epilepsy.

Together, these studies provide a unique, 360° view of KBG syndrome in adulthood. They underline that this is a lifelong condition—but also one where growth, adaptation, and individuality truly matter. Importantly, they highlight the value of combining clinical insight with the voices of patients and families to better shape future care.

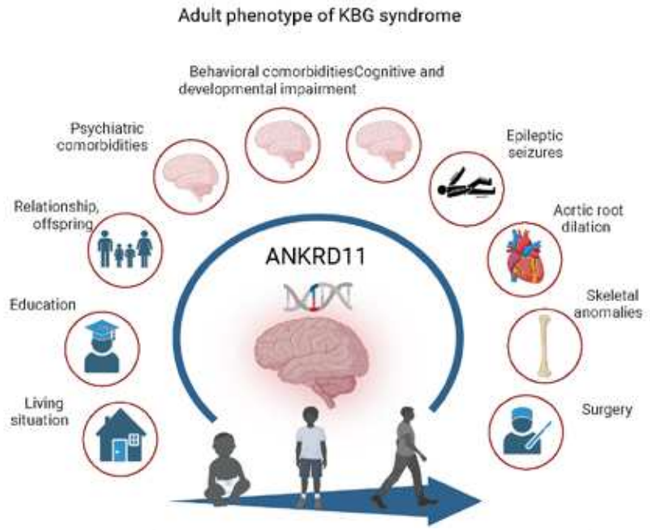
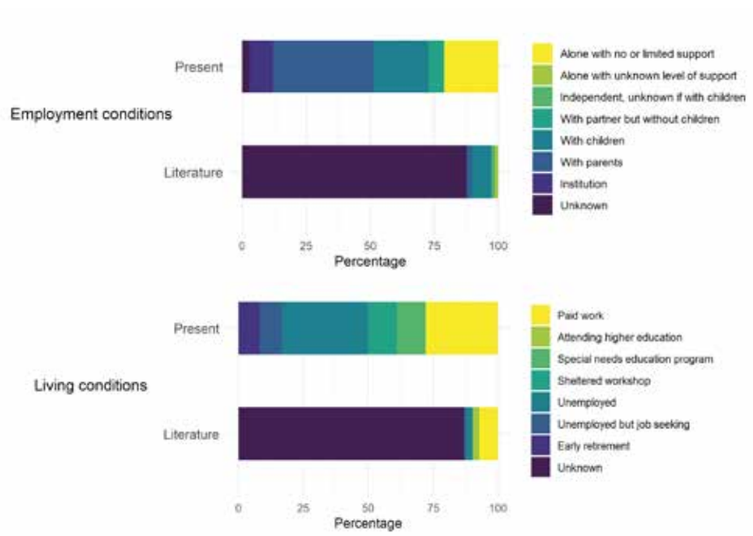


Figure 15. Summary of health issues associated with KBG syndrome across a lifetime.



Summary of employment and living conditions in our KBG syndrome cohort compared with the literature.

Papers

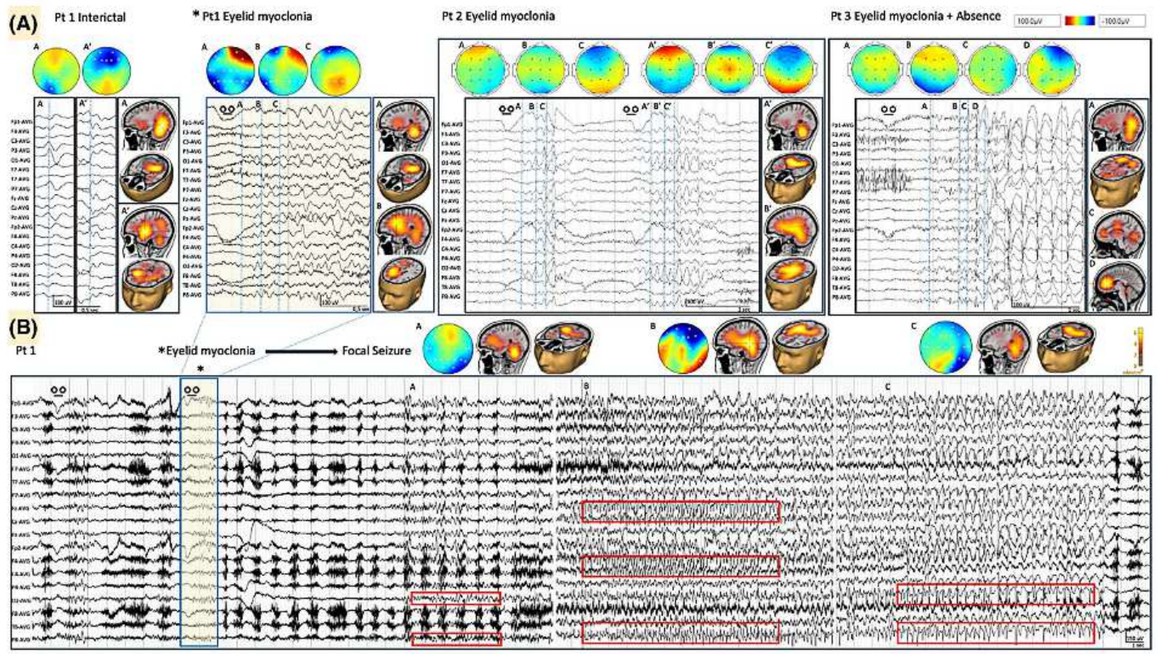
1. **Bayat A**, Grimes H, de Boer E, Herlin MK, **Dahl R**S, Lund ICB, Bayat M, Bolund ACS, **Gjerulfsen CE**, Gregersen PA, **Zilmer M**, **Juhl S**, **Cebula K**, Rahikkala E, Maystadt I, Peron A, Vignoli A, Alfano RM, Stanzial F, Benedicenti F, Currò A, Luk HM, Jouret G, Zurita E, Heuft L, Schnabel F, Busche A, Veenstra-Knol HE, Tkemaladze T, Vrielynck P, Lederer D, Platzer K, Ockeloen CW, Goel H, Low KJ. Natural history of adults with KBG syndrome: A physician-reported experience. Genet Med. 2025; doi: 10.1016/j.gim.2024.101170.
2. Low KJ, Walker M, Treneman-Evans G, Bramswig NC, Herlin MK, Lesca G, Scarano E, Ockeloen CW, **Bayat A**. Life beyond childhood: Insight into the lived experience of adults with KBG syndrome through an online patient/caregiver-reported co-produced questionnaire. Brain Behav. 2025;doi: 10.1002/brb3.70553.

EEG biomarkers II

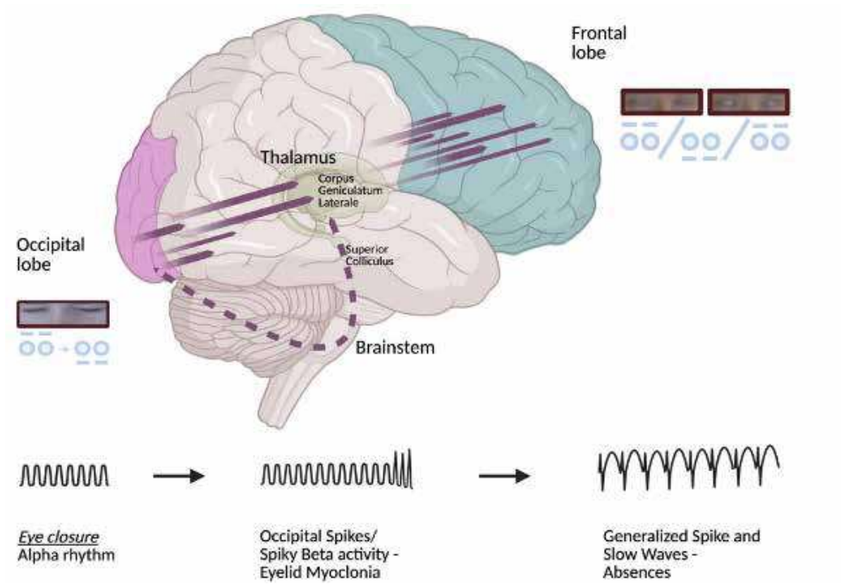
EEG biomarkers are increasingly recognized as valuable tools in the natural history studies of monogenic epilepsies, that typically present with distinct seizure types and varying degrees of impairment.

EEG can capture these patterns, revealing information on the evolution of the disease over time and response to treatment. Therefore, EEG biomarkers play an essential role in tracking disease progression in NHS. In addition, EEG abnormalities often correlate with specific genetic variants, or specific syndromic condition (e.g. epilepsy with eyelid myoclonia (EEM) Figures 17-18) helping to refine diagnostic criteria and quantify potential therapeutic effects.

Namely, EEG biomarkers are instrumental in evaluating the efficacy of treatments in clinical trials, providing an objective, quantifiable measure of seizure control and neurophysiological changes in response to therapeutic interventions.



Integrated quantitative electroencephalography (EEG) analysis. (a) Patient 1 - interictal EEG: Interictal abnormalities are observed in both the occipital (A) and frontal (A′) regions. Eyelid Myoclonia (EM): A posterior-to-anterior progression of the epileptiform discharges is observed after eye closure (A->B->C) as shown in the EEG traces, amplitude maps, and source analysis. Patient 2 - EM: Two episodes show a posterior-to-anterior spread of epileptic discharges after eye closure (A->B->C; A′->B ->C), based on EEG traces, amplitude maps, and source analysis. Patient 3 - EM-Absence: A similar posterior to-anterior progression is observed (A->B->C->D) after eye closure. (b) Patient 1 - EM (detailed above) followed by a right occipital focal onset seizure: The seizure begins with a rhythmic activity in the right occipital region (A), then spreads anteriorly (B), and ends with a rhythmic sequence of high amplitude spikes in the right occipital region (C). Amplitude and source analysis maps confirm right occipital onset of the focal seizure.



Schematic representation of the extended pathophysiological network in epilepsy with eyelid myoclonia (EEM).

Papers

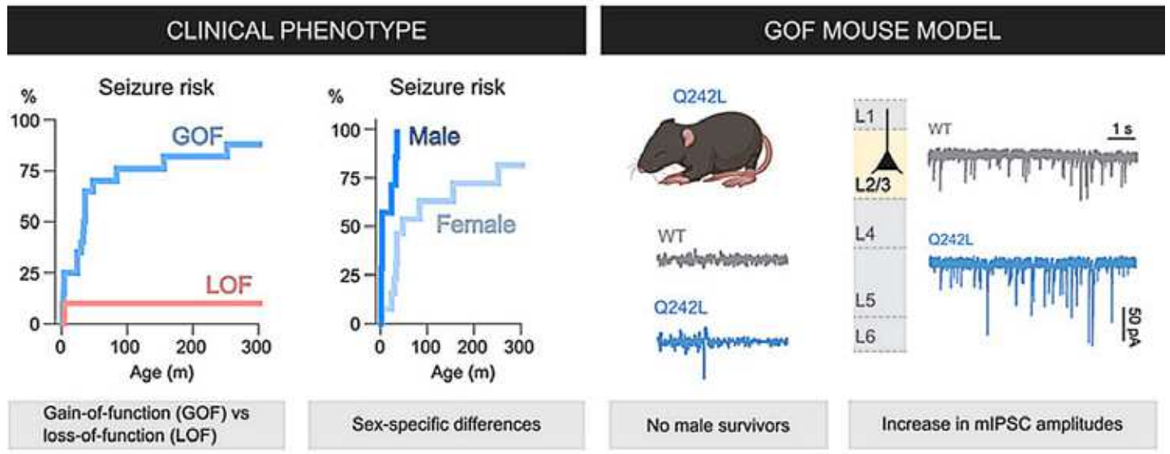
1. **Ricci E, Mieszczanek TS, Zilmer M, Cebula K, Juhl S, Nikanorova M, Mieszczanek KM, Khinchi MS, Hesslow BIU, Thygesen KS, Pedersen CR, Olofsson K, Wüstenhagen S, Møller R, Rubboli G, Beniczky S, Gardella E**. Unmasking the role of the occipital lobe in epilepsy with eyelid myoclonia. Epilepsia. 2025 Jun 13;66(8):e194-201. doi: 10.1111/epi.18477.

From Disease-Causing Variants to Targeted Therapy in GABAA Receptor-Related Epilepsies

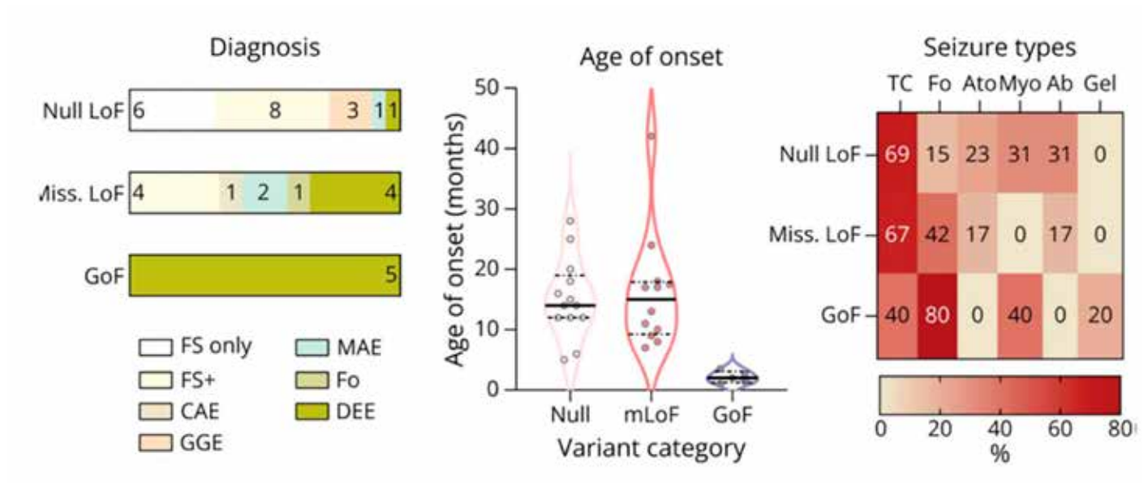
Developmental and Epileptic Encephalopathies (DEEs) are severe early-onset neurodevelopmental disorders characterized by refractory epilepsy, intellectual disability, developmental delay or regression, and high mortality. Pathogenic variants in subunits of the γ-aminobutyric acid type A GABAA receptor, the principal inhibitory receptor in the brain, represent a significant and recurrent genetic cause.

Contrary to the long-held assumption that disease results exclusively from loss-of-function (LOF) variants, we demonstrated that gain-of-function (GOF) variants constitute a distinct and often more severe pathogenic mechanism. Clinical severity correlates strongly with variant function: approximately half of affected individuals harbor GOF variants, which are associated with early-onset DEE, profound intellectual and motor impairment, movement disorders, feeding difficulties, and high early mortality. In contrast, LOF variants typically produce milder phenotypes. These genotype–phenotype relationships were validated in patient-variant knock-in mouse models that recapitulate spontaneous seizures, motor deficits, and increased mortality.

This framework is exemplified by disorders of GABRA3, the only epilepsy-associated GABAA receptor subunit gene located on the X chromosome. Through deep phenotyping and functional characterization of 43 individuals with 19 GABRA3 variants, we identified both GOF and LOF effects linked to distinct clinical profiles (Figure 19). GOF variants cause severe, treatment-resistant epilepsy with profound intellectual disability, predominantly affecting males. LOF variants result in milder phenotypes, with epilepsy uncommon and heterozygous females typically unaffected carriers. A GOF knock-in mouse model (Gabra3Q242L/+) reproduced sex-specific seizure susceptibility, cortical hyperexcitability, and early mortality.



Functional consequence of pathogenic GABRA3 variants determines whether X-linked inheritance is dominant or recessive. Johannesen et al., JCI, 2025.



Epilepsy features for Individuals With Null LoF, Missense LOF and GoF GABRG2 variants. Rossi et al, Neurology, 2025.

Together, these findings demonstrate that the functional consequence of a variant, rather than its presence alone, determines disease severity and inheritance pattern, providing a mechanistic foundation for precision diagnostics and targeted therapy in GABAA receptor epilepsies.

In 2025 we have published four papers on GABAA receptor related disorders:

Papers

1. **Ortiz S**, Bagliani C, Lin SXN, Kusay AS, Silvennoinen K, Kälviäinen R, Jutila L, Saarela A, Dahl RS, Stödberg T, Marini C, Cesaroni E, Bisulli F, Licchetta L, Fallica E, Barco TL, Torta F, Rizzo V, Castro-Villablanca F, Yabumoto M, Mirzaa G, Rossi A, Laugaard-Jacobsen C, Nobili L, Liin SI, **Gardella E**, Ahring PK, **Møller RS**, **Rubboli G**. Early neurological symptoms and epilepsy outcomes in individuals with the recurrent GABRG2 p.(Ala106Thr) gain-of-function variant: Structural and phenotypic insights. Epilepsia. 2025 Dec 6. doi: 10.1111/epi.70045. Online ahead of print.
2. **Johannesen KM**, Aung KP, Liao VW, Absalom N, Chua HC, Gan XN, Mao M, McKenzie CE, Lee HM, **Ortiz S**, Spillmann RC, Shashi V, Radtke RA, Mirzaa GM, Weisner PA, Flores Daboub J, Hagedorn C, Bayrak-Toydemir P, DeMille D, Zhao J, Bajaj N, Capri Y, Keren B, Schmidts M, van de Laar IM, van Slegtenhorst MA, Ploski R, Bogotko M, Bourque DK, Alkhunaizi E, Chad L, Quercia N, Elloumi H, Wentzensen IM, Kruer MC, Bisarad P, Galaz-Montoya CI, Rusu V, Braun D, Angione K, Win JC, Espinosa-Jovel C, Zacher P, Platzer K, Berkovic SF, Scheffer IE, Chebib M, **Rubboli G**, **Møller RS**, Reid CA, Ahring PK. Functional consequence of pathogenic GABRA3 variants determines whether X-linked inheritance is dominant or recessive. J Clin Invest. 2025 Nov 25;136(2):e189830. doi: 10.1172/JCI189830. eCollection 2026 Jan 16.

3. Rossi A, Lin SXN, Absalom NL, **Ortiz-De la Rosa S**, Liao VWY, Mohammadi NA, Viswanathan S, Stödberg T, Danieli A, Bonanni P, Aeby A, Orsini A, Bonuccelli A, Rüegger A, Giraldez BG, Isidor B, Stüve B, Marini C, Cesaroni E, **Fenger CD**, Philippe C, Meunier C, Lederer D, Moortgat S, Spinelli E, Fallica E, Zeiner F, Bauman M, Licchetta L, Bisulli F, Operto FF, Benkel-Herrenbrueck I, Gorman KM, **Johannesen KM**, Platzer K, Schnabel F, Lagae L, Laufs M, Zordania R, Malone S, Messana T, Werckx W, Jonsson C, Afawi Z, Foiadelli T, Halleb Y, Stoeva R, Jennesson-Lyver M, Lesca G, Guerrini R, Berkovic SF, Scheffer IE, Chebib M, **Gardella E**, **Møller RS**, **Rubboli G**, Ahring PK. Phenotypic Spectrum in Individuals With Pathogenic GABRG2 Loss- and Gain-of-Function Variants. Neurology. 2025 Jul 22;105(2):e213644. doi: 10.1212/WNL.0000000000213644. Epub 2025 Jun 26.
4. Millevert C, Kan ASH, Hanke M, Koko M, Omidvar ME, Hedrich UBS, Wuttke TV, Barišić N, Lagae L, Aledo-Serrano Á, Niehoff EM, Platzer K, Zacher P, Polster T, Dilena R, Monfrini E, Geneviève D, Roubertie A, Bruel AL, Mau-Them FT, Dasouki M, Cohen S, Helbig I, Harrison AG, Ellis C, Dubbs HA, Marsh ED, Lebon S, He N, Meng H, Chebib M, **Møller RS**, Marini C, Ahring PK, Lerche H, Weckhuysen S. The genetic and phenotypic spectrum of GABRB1-related disorders. Brain. 2026 Feb 7;149(2):534-547. doi: 10.1093/brain/awaf213.

Understanding GRIA related disorder and translating knowledge into precision therapy for AMPA-receptor related epilepsies.

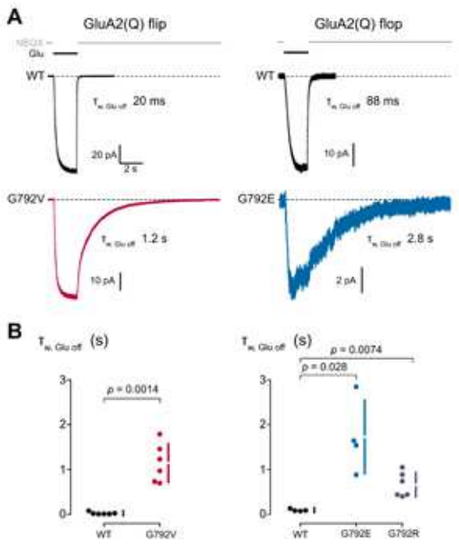
GRIA syndrome is a rare genetic condition caused by changes in the GRIA1–4 genes, which affect AMPA receptors—the brain's main system for fast communication between nerve cells. When this system is disrupted, it can lead to developmental delay, epilepsy, and movement or behavioral difficulties. At the Danish Epilepsy Center, we are proud to be among the world leaders in this field, with a unique global database of more than 200 GRIA patients helping us understand these complex disorders.

A key challenge in GRIA research is determining whether a genetic variant causes a gain-of-function (GoF) or loss-of-function (LoF) effect. This is crucial, because treatment decisions depend on it—but laboratory testing is often slow, expensive, and not always available. Therefore, new ways to predict these effects are urgently needed.
In 2025, our research made important advances.

In our first study, we described a child with a severe GRIA3-related disorder, including epilepsy and parkinsonism - a rare and striking feature. We showed that the genetic variant had a mixed effect, causing both gain- and loss-of-function at the same time. This was associated with a particularly severe disease course, suggesting that mixed effects may be more harmful than previously thought. This finding highlights how complex GRIA disorders can be and why precise functional understanding is essential.

In our second study, we demonstrated that paralog positions - similar locations across related GRIA genes - can help predict whether a variant leads to gain- or loss-of-function. By studying patients with variants at matching positions in GRIA2 and GRIA3, we showed that these shared positions consistently resulted in gain-of-function effects and similar clinical features, including developmental delay and epilepsy. This is highly important, as it provides a faster and more accessible way to interpret genetic findings without always relying on complex laboratory experiments.

Together, these studies strengthen our translational approach: combining genetics, clinical data, and functional insights to guide precision medicine. They show that GRIA syndrome is not only complex but also predictable in new ways—bringing us closer to faster diagnoses and more targeted treatments for affected individuals.



GRIA2 variants slow glutamate unbinding from homomeric GluA2 AMPARs, suggesting increased affinity and gain of function. (A) Representative currents from wild type (WT) flip and flop GluA2(Q)/γ2 receptors jumped into 10 mM glutamate (black bars) from NBQX at −60 mV (in the presence of CTZ and PEPA, respectively, to block desensitization). In each case, the weighted time constant of current decay (τw,Glu off) is shown. Lower traces show corresponding records from the flip variant G792V and the flop variant G792E. The dashed lines indicate the original baseline current. (B) Swarmplots showing pooled values τw,Glu off for flip receptors (left) and flop receptors (right). Summary data (mean ± standard deviation) for each group are shown as gapped lines to the right of the raw data. Indicated p values indicate differences from respective WT controls.

Papers

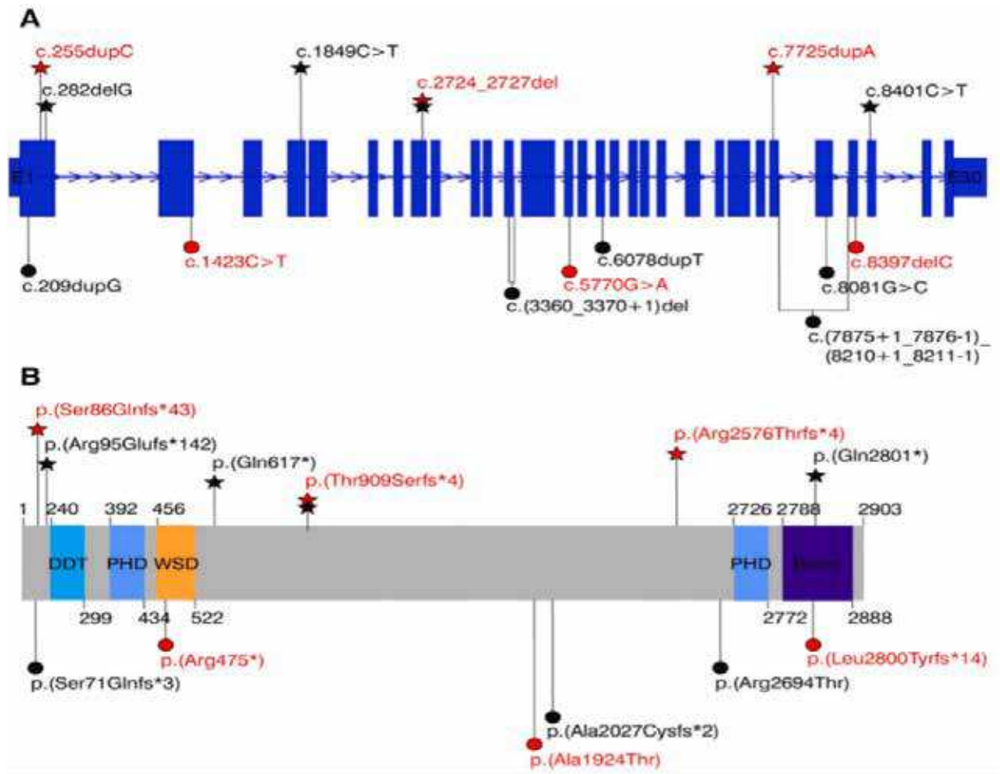
1. Fons C, Ge YH, **Rasmussen LK**, Shi YS, **Bayat A**. Mixed functional consequences of the N651D GRIA3 variant: a case of early-onset developmental and epileptic encephalopathy with parkinsonism. J Med Genet. 2025 Nov 21;62(12):808-811. doi: 10.1136/jmg-2025-110855.
2. **Sjøstrøm E**, Studniarczyk D, Dou X, Dahl RS, Cruz V, Wang H, Mercier S, Deb W, Besnard T, Friedman J, Essid M, Karoui S, Ben Jemaa L, Benyounes T, Lesca G, Tonduti D, Iascone M, Orcesi S, Fradin M, Dubourg C, Napuri S, Cull-Candy SG, Coombs ID, Farrant M, **Bayat A**. Clinical and neurodevelopmental characteristics of paralogous gain-of-function variants at GRIA2 p.Gly792 and GRIA3 p.Gly803. Clin Genet. 2025;108(5):553-565. doi: 10.1111/cge.14770.

Deep phenotyping of monogenic epilepsies: new insights

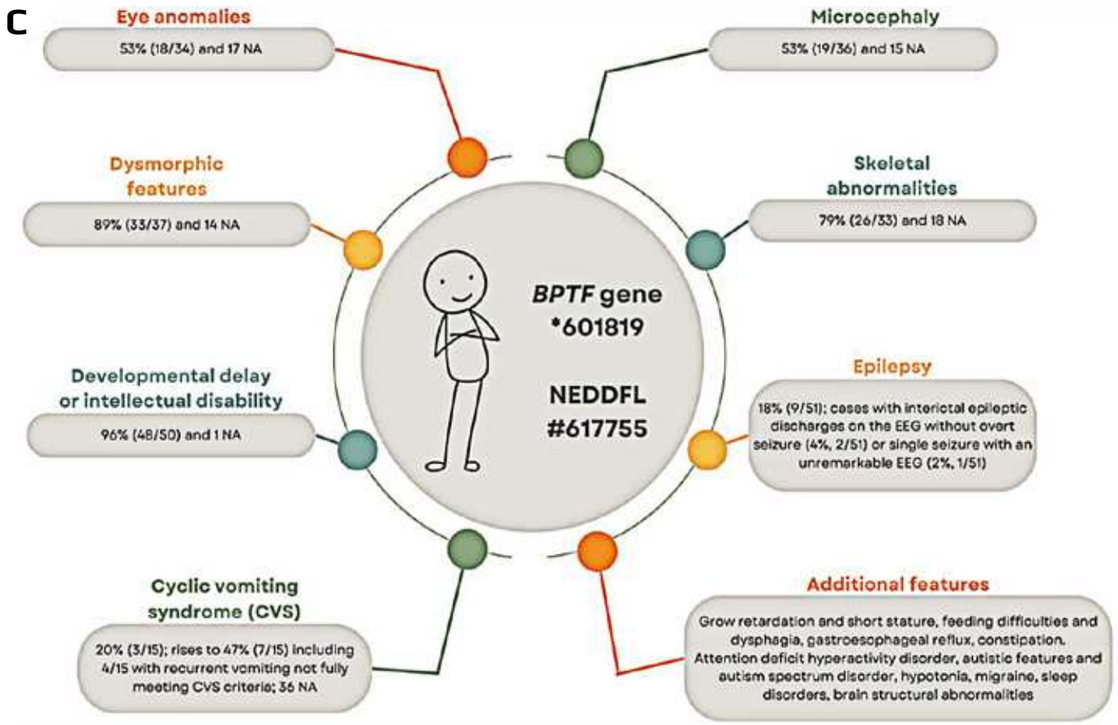
Deep phenotyping in monogenic epilepsies is essential to define the full clinical spectrum beyond seizures. In BPTF-related disorders, it uncovers key features such as cyclical vomiting syndrome, refining diagnosis and management. It strengthens genotype–phenotype correlations and provides a critical foundation for precision therapies and future clinical trials.

Families with BPTF -related neurodevelopmental disorders, with or without gastrointestinal symptoms, were recruited through an international collaboration, focusing on a novel, previously unrecognized clinical manifestation, namely cyclical vomiting syndrome (CVS). CVS was diagnosed using criteria from the International Classification of Headache Disorders, 3rd edition (ICHD-3). Genetic variants were analyzed for pathogenicity, and effectiveness of therapies was assessed.

This study broadens the syndromic phenotype associated with BPTF haploinsufficiency, highlighting CVS as a core feature. The findings raise clinician awareness, guide management, and enhance understanding of this rare condition.



Schematic of the BPTF gene, protein, and variant positions reported in this study. (A) Schematic of the BPTF gene and (B) the corresponding protein structure. The stars indicate the variants of individuals affected by cyclical vomiting syndrome (CVS). (C) Primary and secondary characteristics of BPTF haploinsufficiency, as reported in the 51 individuals described so far.



Schematic of the BPTF gene, protein, and variant positions reported in this study. (A) Schematic of the BPTF gene and (B) the corresponding protein structure. The stars indicate the variants of individuals affected by cyclical vomiting syndrome (CVS). (C) Primary and secondary characteristics of BPTF haploinsufficiency, as reported in the 51 individuals described so far.

Papers

1. Ferretti A, Furlan M, Glinton KE, **Fenger CD**, Boschann F, Zeidler S, Stoltenburg C, Barakat TS, Martinez-Agosto JA, Devinsky O, **Furia F**, **Rubboli G**, Di Napoli A, Bellone G, Furio S, Piccirillo M, Mennini M, Di Nardo G, Parisi P, **Møller RS**, Yang Y, Stankiewicz P, **Gardella E**. Cyclical Vomiting Syndrome in Individuals With BPTF Haploinsufficiency. Pediatr Neurol. 2025 Sep;170:58-65. doi: 10.1016/j.pediatrneurol.2025.06.010. Epub 2025 Jun 13.

Cenobamate – a promising drug in highly refractory focal epilepsies and developmental epileptic encephalopathies

Thirty-forty per cent of patients with epilepsy do not achieve adequate seizure control despite the availability of a large selection of anti-seizure medications (ASM) and the introduction of several new drugs in the last few years.

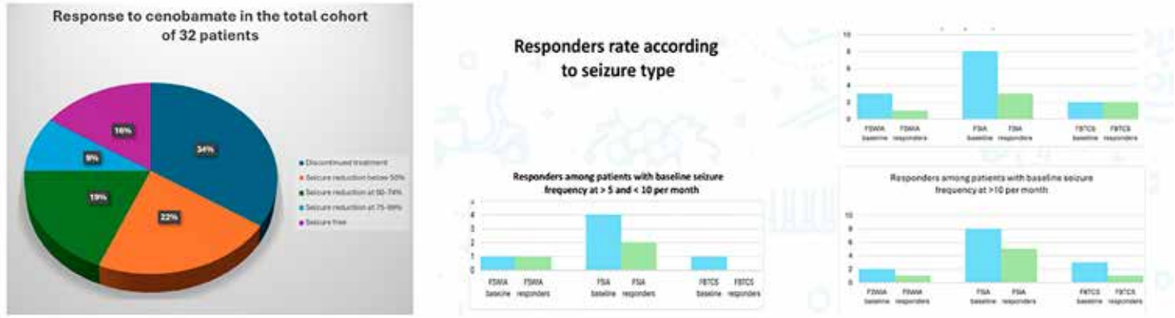
Cenobamate was developed as a new aryl-carbamate with a dual-action mechanism. Cenobamate positively modulates γ-aminobutyric acid A (GABAA) receptors and blocks persistent sodium currents. A recent systematic review evaluated cenobamate as the most efficacious third-generation ASM. In Europe, it is approved as an adjunctive drug for the treatment of focal-onset seizures in adults with epilepsy.

Only few studies have reported the effect of cenobamate in pediatric patients with drug-resistant epilepsy and there are even more limited reports of treatment with cenobamate in genetic epilepsies.

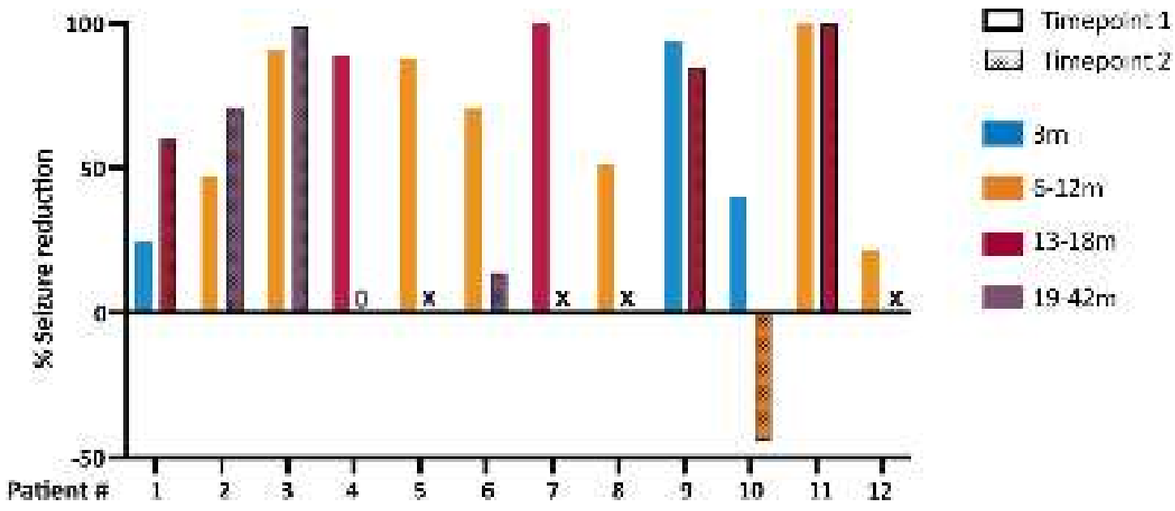
In a population of adult patients suffering from ultra-refractory focal epilepsies (defined as focal epilepsies in which≥6 treatments, including ASM trials, epilepsy surgery, and vagus nerve stimulator, failed to achieve seizure control) add-on cenobamate in a real-life setting achieved a 44% responder rate (≥50% reduction in seizure frequency), including five patients who became seizure-free (15.6% of the total cohort) (Figure 23). Discontinuation due to adverse events or lack of efficacy occurred in 34% of patients. AEs most frequently reported were tiredness and dizziness and in two-thirds they resolved by dose-reduction of ASMs or cenobamate. Follow-up extended up to 32 months.

In a cohort of 12 patients, including children (median age 8 years) suffering from developmental and epileptic encephalopathies caused by GOF pathogenic variants in SCN8A, the treatment with cenobamate as add-on achieved a meaningful reduction of countable motor seizures in 83% (6 patients experienced a seizure reduction above 70%, including two who achieved seizure freedom). An increase in seizure-free days per patient was also reported. Furthermore, 80% of patients reported non–seizure-related improvements (included increased alertness, better sleep, and improved muscle tone). Adverse effects were reported by 50% of patients and half resolved by reducing concomitant antiseizure medications.

These findings underlines the usefulness of cenobamate for the treatment of patients with ultra-refractory epilepsy and indicates its long-term effectiveness in real-world clinical practice. In addition, cenobamate appears to be a promising and safe treatment for SCN8A-DEE, even during early childhood, either by reducing the seizure burden and by improving non–seizure-related symptoms.



Left: responder rate in the total cohort of patients (left) and according to seizure type (right). Gjerulfsen et at, Neurol Sci, 2025.



Percentage of seizure reduction for each patient with SCN8A-related DEE with follow-up at the two timepoints for data collection. X, missing data at time point; 0, no reduction of seizure frequency (Gjerulfsen et al., Epilepsia, 2025).

Papers

1. **Gjerulfsen CE, Juhl S, Mieszczanek KM, Spanilá L, Thygesen KS, Pavbro A, Møller RS, Rubboli G**. Cenobamate as add-on treatment in ultra-refractory focal epilepsy: Real-world results from The Danish Epilepsy Centre, Dianalund, Denmark. Neurol Sci. 2025 Aug;46(8):3875-3884.
2. **Gjerulfsen CE**, Oudin MJ, **Furia F**, Gverdtsiteli S, Landmark CJ, Trivisano M, Balestrini S, Guerrini R, Aledo-Serrano A, Morcos R, Previtali R, Veggiotti P, Ricci E, **Rubboli G**, **Gardella E**, **Møller RS**. Cenobamate as add-on treatment for SCN8A developmental and epileptic encephalopathy Epilepsia. 2025 Apr;66(4):1119-1128

6. Lectures - oral presentations in 2025

Sándor Beniczky:

1. My EEG "Evergreen Paper" and beyond. Valentinssymposium, February 22, online.
2. Guidelines for Reporting EEG & Neurophysiology Biomarker Evaluation for Application to Neurology: GREENBEAN. 3rd International Conference on Artificial Intelligence in Epilepsy and Neurological Disorders, March 3-6, Breckenridge, USA.
3. Seizure detection devices and wearables. Canadian Epilepsy Teaching Network – Grand Rounds, March 21st, online.
4. The role of EEG in acute and structural epilepsies. International Congress on Structural Epilepsy & Symptomatic Seizures (STESS), April 2, Gothenburg, Sweden.
5. EEG: Focal, Diffuse, and Epileptiform Abnormalities in Adults. American Academy of Neurology's 77th Annual Meeting, scheduled for April 5-9, 2025, in San Diego, USA.
6. Seizure semiology. April 15, Faculty of Medicine of Sfax – webinar.
7. Automated seizure detection. April 23, Diagnostics & Tracking Research Roundtable for Epilepsy, Washington DC, USA.
8. The role of artificial intelligence for diagnostication and prognostication of status epilepticus. 3rd Nordic Status Epilepticus Meeting, 22-23 May, Oslo, Norway.
9. Automated EEG interpretation. International Conference for Technology and Analysis of Seizures (ICTALS), June 2-6, Montréal, Canada.
10. From vision to practice: Ensuring research becomes clinical reality. International Conference for Technology and Analysis of Seizures (ICTALS), June 2-6, Montréal, Canada.
11. Artificial Intelligence in epilepsy. 8th Congress of the Society of Neurologists of the Republic of Moldova, June 13-14, Chisinau, Moldova.
12. Artificial intelligence (AI) and EEG: not the graveyard of the human EEG reader. 11th Congress of the European Academy of Neurology, June 21-24, Helsinki, Finland.
13. How to localize the source of the EEG signals. Webinar of the Argentinian Epilepsy Society, Juni 25, online.
14. Dipoles and source estimation. XVIII Congreso de la Liga Colombiana Contra la Epilepsia, July 24-27, Cali, Colombia.
15. Artificial intelligence in EEG. XVIII Congreso de la Liga Colombiana Contra la Epilepsia, July 24-27, Cali, Colombia.
16. EEG post-processing. XVIII Congreso de la Liga Colombiana Contra la Epilepsia, July 24-27, Cali, Colombia.
17. Seizure classification. XVIII Congreso de la Liga Colombiana Contra la Epilepsia, July 24-27, Cali, Colombia.
18. Let's read EEGs together! International Epilepsy Congress. August 30-September 3, Lisbon, Portugal.
19. How to test a patient during seizures. International Epilepsy Congress. August 30-September 3, Lisbon, Portugal.
20. EEG source imaging. International Epilepsy Congress. August 30-September 3, Lisbon, Portugal.
21. SCORE-AI. International Epilepsy Congress. August 30-September 3, Lisbon, Portugal.
22. Magnetoencephalography-based source imaging. International Epilepsy Congress. August 30-September 3, Lisbon, Portugal.
23. A tale of 365 nights: a yearlong electroencephalography investigation into human sleep in natural conditions. 5th International Congress on Mobile Health and Digital Technology in Epilepsy, September 4-6, Copenhagen, Denmark.
24. Artificial Intelligence in epilepsy: The low-hanging fruits - SCORE-AI. 5th International Congress on Mobile Health and Digital Technology in Epilepsy, September 4-6, Copenhagen, Denmark.
25. Challenges in epilepsy diagnosis. European Congress of Clinical Neurophysiology, September 9-12, London, UK.
26. Artificial intelligence for EEG. European Congress of Clinical Neurophysiology, September 9-12, London, UK.
27. Epileptic seizure classification: Electroclinical perspectives. European Congress of Clinical Neurophysiology, September 9-12, London, UK.
28. Artificial intelligence: How to turn promises into results. European Congress of Clinical Neurophysiology, September 9-12, London, UK.
29. Exploring a new frontier in brain research and neural codes. European Congress of Clinical Neurophysiology, September 9-12, London, UK.
30. AI in EEG interpretation. 27th World Congress of Neurology (WCN), October 12-15, Seul, South Korea.
31. EEG in status epilepticus. 27th World Congress of Neurology (WCN), October 12-15, Seul, South Korea.
32. Automated seizure detection using wearable devices. 32nd annual congress of the Indian Academy of Neurology, September 28-November 1, Varanasi, India.
33. Identifying Interictal Epileptiform Discharges. 32nd annual congress of the Indian Academy of Neurology, September 28-November 1, Varanasi, India.
34. Automatisk Anfaldsdetektion. November 21-22. Dansk Neuropædiatrisk Selskab, Kolding, Denmark.
35. EEG source imaging in epilepsy—practicalities and pitfalls. Brazilian Congress of Clinical Neurophysiology, November 26-29, Belo Horizonte, Brazil.
36. Multimodal EEG: towards an integrated view on brain disease. Brazilian Congress of Clinical Neurophysiology, November 26-29, Belo Horizonte, Brazil.

Rikke Steensbjerre Møller:

1. Utility of Genetic Testing for Therapeutic Decision-Making in Individuals with Epilepsy, Copenhagen University, December, 2025.
2. State of the art of genetic investigations in epilepsy in 2025, NorEpiNet Genetics Course, Oslo, November, 2025.
3. Emerging therapeutic insights into GABA-A receptor modulation: ASMs, vinpocetine, and novel agents, GABAA receptor meeting, Sydney, October 2025.
4. Bridging clinical and basic GABA-A receptor research, GABAA receptor family session Sydney, October 2025.
5. Utility of Genetic Testing for Therapeutic Decision-Making in Individuals with Epilepsy, Flagship Seminar, USYD, Sydney, October 2025.
6. Genetik – Hvad ved vi, hvor langt er vi? Psykolog dag, Epilepsihospitalet, oktober 2025.
7. Hvordan kan gentest bane vejen for mere præcis og personlig behandling af epilepsi? Epilepsiforeningen, Århus, oktober 2025.
8. Hvordan kan gentest bane vejen for mere præcis og personlig behandling af epilepsi? Epilepsiforeningen, Ålborg, oktober 2025.
9. Hvordan kan gentest bane vejen for mere præcis og personlig behandling af epilepsi? Epilepsiforeningen, Odense, oktober 2025.
10. Hvordan kan gentest bane vejen for mere præcis og personlig behandling af epilepsi? Epilepsiforeningen, Kreds Øst, Herlev, oktober 2025.
11. From disease-causing variants to targeted therapy in GABA-A receptor related disorders, 3rd Channelopathy Meeting, Tübingen, September 2025.
12. Future perspectives in treatment - Precision medicine, Nordic Epilepsy Masterclass, Copenhagen, September 2025.
13. The role of genetic testing in the treatment of DRE, Nordic Epilepsy Masterclass, Copenhagen, September 2025.
14. Hvordan kan genetikken fremme individualiseret behandling af epilepsi? National netværksgruppe for sundhedsfaglige på bosteder under Epilepsiforeningen, Odense, September 2025.
15. Precision medicine in genetic epilepsies, the Chilean Society of Epileptology, September 2025 (online).
16. Precision Medicine: The changing face of epilepsy diagnosis and management through genetics, the 36th International Epilepsy Congress, Lisbon, September, 2025.
17. Neurological Clues: Identifying Symptoms of GABAA-Receptor Related Disorders, European SLC6A1 and Cure GABAA Conference, Lisbon, August 2025.
18. Precision medicine in genetic epilepsies, Genetic Epilepsy Course, Buenos Aires, Argentina, August 2025 (online).
19. How to interpret genetic data in epilepsies: Interpretation of variants in GABAA-receptor genes, YES webinar, June 2025 .
20. State of the art of genetic investigations in epilepsy in 2025, Annual EpiCARE meeting, Salzburg, June 2025.
21. From disease-causing variants to targeted therapy in GABA-A receptor related epilepsies, Annual EpiCARE meeting, Salzburg, June 2025.
22. Genetic mechanisms of sodium-channelopaties, SCN2A/SCN8A scientific meeting and family gathering, Bonn, May 2025.
23. Ongoing and upcoming clinical trials in Denmark, NorEpiNet webinar, April, 2025.
24. From disease-causing variants to targeted therapy in GABA-A receptor related epilepsies; Genetics of Neurodevelopmental Disorders, Troina, Italy, April 2025.
25. Clinical neuroscience – Epilepsy, NAD, Copenhagen University, April, 2025.
26. The Danish Epilepsy Centre, Filadelfia: Multidisciplinary treatment of epilepsy on a nation-wide level; 1st Latin American Congress on Epilepsy Genetics, Santiago, Chile, January, 2025.
27. Precision medicine in genetic epilepsies; 1st Latin American Congress on Epilepsy Genetics, Santiago, Chile, January, 2025.

Guido Rubboli:

1. DEE in adults: Beyond seizures. Epilepsy Academy, 28 January 2025, webinar.
2. ESES/DEE-SWAS. State of the art and challenges, SWAS Expert Meeting, Barcelona (Spain), 1-2 March, 2025.
3. The complex relationship between epilepsy and sleep in DEEs. 4th Rome Debate on Developmental and Epileptic Encephalopathies, Rome (Italy), 11 March 2025.
4. Polygraphic recordings in the study of epilepsy. School of Child Neuropsychiatry, University of Verona, Verona (Italy), 11 April 2025.

5. Linking epilepsy, sleep disruption and cognitive impairment in Encephalopathy related to Status Epilepticus during Slow Sleep, School of Child Neuropsychiatry, University of Verona, Verona (Italy), 17 April 2025.
6. Epilepsy and sleep disorder and how does it influence cognition and prognosis in refractory epilepsy»? EpiCARE WG 18, Meeting Beyond seizures: Advancing Holistic Care for rare and complex epilepsies, Ålesund (Norway), 26-27 May, 2025.
7. Ultra-long subcutaneous EEG monitoring in drug-resistant epilepsies. Clinical yields and challenges. 5th Mobile Health and Digital Technology in Epilepsy Congress, Copenhagen 4-6 September 2025.
8. Myoclonus and myoclonic seizures. Videosession, International Epillepsy Congress, Lisbon, 31 august-5 september 2025.
9. Genetic testing in epilepsy – which patients, which tests, and which yields in the clinics?, Teaching Course, International Epilepsy Congress, Lisbon, 31 august-5 september 2025.
10. Dissecting the KCNA2-LOF phenotype, Legolas meeting, Dianalund 5-6 June 2025.
11. Juvenile and Adult Forms of Progressive Myoclonus Epilepsies, European Congress of Clinical Neurophysiolgy, London (UK), 9.9.2025.
12. ESES/DEE-SWAS: Definición, actualización y desafíos futuros. Simposio Internacional de Encefalopatía Epiléptica del Desarrollo/ Encefalopatía Epiléptica con punta onda continua durante el sueño (EED/EE con POCSL), Salta (Argentina), 27 september 2025, webinar.
13. Genetica in epilessia: chi testare e per quale motivo. Dalla clinica alla ricerca. San Giovanni Rotonso (Italy), 1-2 October 2025.
14. Ultra-long subcutaneous EEG monitoring in drug-resistant epilepsies. Filadelfia, Dianalund, October 2025.
15. Treatment of epilepsy in patients with intellectual disability. Update on epilepsy surgery. Meeting of Danish Epilepsy Society, Copenhagen, 7 november 2025.
16. Treatment of drug resistant IGEs, Advanced Course on the Pharmacological Treatment of Drug Resistant Epilepsies, Palma di Maiorca (Spain), 8-9 November 2025.
17. Ethics in AI - where can (or shouldn't) we go from here? EpiCARE AI experts to interview the speakers with epilepsy-specific questions, EpiCARE 6th Rome Workshop, 12-14 November 2025.
18. Cenobamate as add-on in ultra-refractory focal epilepsies: The Dianalund experience. Meeting: Expert perspectives from Denmark and beyod, Dianalaund, 11 December 2025.

Elena Gardella:

1. Emerging genetic insights in DEEs and associated movement disorders, International Symposium on Neurodevelopmental Syndromes and Movement Disorders, Barcelona (Spain), February 6th-7th, 2025.
2. Genetic form of ESES / (D)EE-SWAS, VIII Curso de manejo práctico de la epilepsia en la edad pediátrica, Barcelona (Spain), February 28th-March 1st, 2025.
3. Quantitative EEG in ESES / (D)EE-SWAS: clinical applicability, VIII Curso de manejo práctico de la epilepsia en la edad pediátrica, Barcelona (Spain), February 28th-March 1st, 2025.
4. The role and impact of abnormal sleep in Developmental and Epileptic Encephalopathies, 4th Rome debate on DEEs, Rome (Italy), March 15th, 2025.
5. From genetic testing to the phenotype, 12th International Residential Course on Drug Resistant Epilepsies, Tagliacozzo (Italy), May 4th-10th, 2025.
6. SCN8A clinical spectrum, 3rd SCN2A & SCN8A scientific conference and gathering meeting, Bonn (Germany), May 16th-17th, 2025.
7. SCN8A anti-seizure medication, 3rd SCN2A & SCN8A scientific conference and gathering meeting, Bonn (Germany), May 16th-17th, 2025.
8. SCN8A: summary of day1 for families, 3rd SCN2A & SCN8A scientific conference and gathering meeting, Bonn (Germany), May 16th-17th, 2025.
9. Natural History Studies in Epilepsy, NorEpiNet meeting (Scandinavic), Virtual, June 2, 2025
10. Natural History Studies in Epilepsy and Drug Trial Readiness, 10th Legolas meeting, Dianalund (Denmark), June 05th-06th, 2025.
11. Precision medicine in Genetic epilepsies hype or hope, Webinar – Second-Level Master's Degree in Epileptology – Genoa University (Italy), June 7th, 2025.
12. SCN8A Phenotypes and long-term evolution, Webinar YES-ILAE, June 12, 2025
13. From genes to waves: EEG Biomarkers in GABAA Related Disorders, GABA family meeting; Lisbon (Portugal), September 29th, 2025.
14. The VIREPA BEEG Course, International Epilepsy Congress, Lisbon (Portugal), August 30-September 3rd, 2025.
15. Simposio Internacional de Encefalopatía Epiléptica del Desarrollo/Encefalopatía Epiléptica con punta onda continua durante el sueño (EED/EE con POCSL), Genética EED/EE con POCSL, Salta (Argentina), September 26th, 2025.
16. Lo Spettro delle DEEs monogeniche, Dalla clinica lla ricercar neurologica nel sollievo della sofferenza, San Giovanni Rotondo (Italy), October 1st-2nd, 2025.
17. The ESCO natural history study & registry, 2nd European STXBP1 Summit and Research Roundtable, Heidelberg (Germany), October 8th-10th, 2025.
18. Genetic Epilepsies – a clinic and neurophysiological angle, Annual meeting of the Danish Society of Clinical Neurophysiology, Vejle (Denmark), October 31st-November 1st, 2025.
19. Treatment of Dravet disease; Lennox Gastaut syndrome, and other DEEs, 3rd Advanced Course on the Pharmacological Treatment of Drug-Resistant Epilepsies (DRE), Palma de Mallorca (Spain), November 7th-9th, 2025.
20. Clinical characteristics of the main types of DEEs, 3rd Advanced Course on the Pharmacological Treatment of Drug-Resistant Epilepsies (DRE), Palma de Mallorca (Spain), November 7th-9th, 2025.
21. Clinical and Genetic Perspectives: Genotype–phenotype correlations, biomarkers, pharmacogenomics, and clinical translation, Dravet research gathering 1st European Meeting on Preclinical Research in Dravet Syndrome, virtual, November 19th, 2025.
22. A genotype-phenotype clinical interpretation approach, From genes to treatment: Understanding epilepsy genetics and precision medicine, Oslo, November 24th-25th, 2025.

Marina Nikanorova:

1. 2nd Master class "Div´in Dravet syndrome" – Paris, 28-29th March 2025.
2. 18th Baltic Sea summer school on epilepsy – 3 sessions, September 2025.

Allan Bayat:

1. "Drug-repurposing". The Dianalund Experience. January 2025, Chile.
2. "GRIA1 and GRIA3 related disorders". January 2025, Chile.
3. Kan Medicinrådets principper bane vejen for et mere robust og bæredygtigt sundhedsvæsen? - Medicinske Tidsskrifter. February 2025, Online.
4. "Recent advances in treating DEEs – lessons from Dravet & Lennox Gestaut". 2nd European STXBP1 Summit, Heidelberg, October 2025.
5. "Utility of genetic testing for personalized treatment". Danish Epilepsy Society Summit, November 2025.
6. "Future treatments for genetic epilepsies". Annual conference at the Danish Society for Pediatric Neurology, November 2025.

Francesca Furia:

1. Invited lecture "Mild cases of STXBP1-related disorders" at the second European STXBP1 summit in Heidelberg in October 2025.
2. Ph.d. defence "Deep phenotyping of monogenic epilepsies towards the identification of targeted treatments" in May 2025.
3. Invited lecture "Sleep disturbances in SCN8A-related disorders" at the 10th LEGOLAS meeting in Dianalund in June 2025.
4. Invited lecture "Sleep disturbances in SCN8A-related disorders" at the third SCN2A & SCN8A Scientific Conference & Family Gathering in Bonn in May 2025.
5. Invited lecture "Deep phenotyping of monogenic epilepsies towards the identification of targeted treatments" at the annual spring meeting and general assembly of the Danish Epilepsy Society in Middelfart in March 2025.

Sopio Gverdtsiteli:

1. 3rd SCN2A-SCN8A Scientific Conference and Family Gathering, Bonn, Germany – "Episodic ataxia in SCN2A", "NDEEMA across sodium channelopathies".
2. 36th International Epilepsy Cogress, Lisbon, Portugal – "Clinical and genetic insights from a large cohort of individuals with SCN2A-related disorders".
3. 7th Annual Nordic EpiXcel Meeting, Oslo, Norway – "Clinical and genetic insights of SCN2A-related disorders".

Rebekka Staal Dahl:

1. CURE GABA-A 2025 Family & Scientific Conference – "The GABAA-Receptor Portal".
2. 2025 Rare Reception – "The GABAA-Receptor Portal".
3. IEEE Women in Engineering 'Precision by Design' Webinar – "Genetic Glitches: When the Code Misfires".

7. Publication list in 2025

1. Nevalainen P, von Ellenrieder N, Dudley RWR, Balasubramaniam N, **Beniczky S**, Cacic Hribljan M, Fabricius M, Ho A, Jonsson H, Meidahl A, Michaud E, Nikolic M, Rasmussen R, Salli E, Sidaros A, Frauscher B, Gotman J. Fast Ripples Measured From Overnight SEEG Recordings as Markers of the Epileptogenic Zone: A Multicenter Validation Study. Neurology. 2026 Jan 27;106(2):e214511. doi: 10.1212/WNL.0000000000214511. Epub 2025 Dec 26. PMID: 41453125.
2. McKee HR, Ovalle AB, Giambarberi L, Lekoubou A, Rossetti AO, Block C, Habela CW, Alexopoulos AV, Tripathi M, **Beniczky S**, Van Haerents S, Wright SK, Muh CR, King-Stephens D, Richardson RM, Ali A, Karakis I, Ruiz AR, Bui E. Annual Course / From Then to Now: The Evolving Spectrum of Epilepsy Care. Epilepsy Curr. 2025 Dec 22:15357597251406694. doi: 10.1177/15357597251406694. Epub ahead of print. PMID: 41446561; PMCID: PMC12722146.
3. Josephson CB, **Beniczky S**, Denaxas S, Ikeda A, Jehi L, Mwesige AK, Jette N, Jones GD, Ryvlin P, Sen A, Triki CC, Waters G, Guekht A, Cross JH. A call for ethical, equitable, and effective artificial intelligence to improve care for all people with epilepsy: A roadmap. A report by the ILAE Global Advocacy Council and Big Data Commission. Epilepsia. 2025 Dec 24. doi: 10.1002/epi.70058. Epub ahead of print. PMID: 41443971.
4. Proença S, Soares JI, Parra J, Maia G, Silva S, Leite J, **Beniczky S**, Jesus-Ribeiro J. Comparative evaluation of artificial intelligence chatbots in answering electroencephalography-related questions. Epileptic Disord. 2025 Dec 16. doi: 10.1002/epd2.70156. Epub ahead of print. PMID: 41399926.
5. Seeck M, Nascimento FA, Benbadis S, Tatum WO, Wirrell E, Ramantani G, **Beniczky S**. Comment on "Incidental interictal epileptiform discharges in infants with nonepileptic events" by Montenegro et al. Epilepsia. 2026 Feb;67(2):1007-1009. doi: 10.1111/epi.18686. Epub 2025 Dec 12. PMID: 41388233; PMCID: PMC12927699.
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7. Marcinski Nascimento KJ, Yuan D, Guerriero RM, Hirsch LJ, **Beniczky S**, Nascimento FA. How to Read an EEG: Epileptiform Abnormalities. Neurol Educ. 2025 Oct 29;4(4):e200265. doi: 10.1212/NE9.0000000000200265. PMID: 41180596; PMCID: PMC12576837.
8. Musaeus CS, Viana PF, Cook M, Duun-Henriksen J, **Beniczky S**, Kidmose P, Vanrumste B, Filtjens B, Kjaer TW. Home-Based sensing of the nervous system with clinical neurophysiology technologies: IFCN handbook chapter. Clin Neurophysiol Pract. 2025 Sep 21;10:453-463. doi: 10.1016/j.cnp.2025.09.003. PMID: 41142046; PMCID: PMC12547184.
9. **Beniczky S**, Trinka E, Wirrell E, Singh MB, Blumenfeld H, Bogacz Fressola A, Cendes F, Craiu D, Frauscher B, Jansen FE, Kahane P, Kander V, Kishk N, Khoo CS, Lizcano A, De Palma L, Ryvlin P, Specchio N, Sperling MR, Tatum W, Yacubian EM, Wilmshurst J, Cross JH. A practical guide to the updated seizure classification 2025. Epileptic Disord. 2025 Dec;27(6):1087-1104. doi: 10.1002/epd2.70110. Epub 2025 Oct 13. PMID: 41081650; PMCID: PMC12747708.
10. **Beniczky S**, Trinka E, Caraballo R, Carreno M, Cendes F, Bosque-Varela P, Lerda-Casaccia T, Wiebe S, Yacubian EM. Acceptance and perceptions on the 2025 update of the ILAE classification of epileptic seizures: details are important. Epilepsy Behav. 2025 Nov;172:110738. doi: 10.1016/j.yebeh.2025.110738. Epub 2025 Oct 3. PMID: 41045783.
11. Jeppesen J, Christensen J, Ahrenfeldt Petersen O, **Fenger S**, **Armand Larsen S**, **Wüstenhagen S**, **Wagner SR**, **Johansen P**, **Beniczky S**. Seizure detection using wearable electrocardiogram connected to a smartphone: a phase 3 clinical validation study. EBioMedicine. 2025 Oct;120:105952. doi: 10.1016/j.ebiom.2025.105952. Epub 2025 Sep 29. PMID: 41027311; PMCID: PMC12516532.
12. Baykan B, Dunne J, Wiebe S, Maillard L, **Beniczky S**, Koutroumanidis M, Seeck M. Presence of interictal epileptiform EEG discharges implies increased risk of recurrence after the first unprovoked seizure: Report of the International League Against Epilepsy and International Federation of Clinical Neurophysiology. Clin Neurophysiol Pract. 2025 Aug 28;10:380-391. doi: 10.1016/j.cnp.2025.07.007. PMID: 41018165; PMCID: PMC12464601.
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14. Nascimento FA, Yuan D, Sheikh IS, Hirsch LJ, **Beniczky S**, Westover MB. The EEG Talk experience: Lessons in e-teaching EEG. Epileptic Disord. 2025 Dec;27(6):1337-1339. doi: 10.1002/epd2.70096. Epub 2025 Sep 4. PMID: 40904123.
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16. Baykan B, Dunne J, Wiebe S, Maillard L, **Beniczky S**, Koutroumanidis M, Seeck M. Presence of interictal epileptiform EEG discharges implies increased risk of recurrence after the first unprovoked seizure: Report of the International League Against Epilepsy and International Federation of Clinical Neurophysiology. Epilepsia. 2025 Oct;66(10):3920-3936. doi: 10.1111/epi.18591. Epub 2025 Aug 28. PMID: 40875494; PMCID: PMC12605794.
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