FILADELFIA

ANNUAL RESEARCH REPORT 2023

Danish Epilepsy Centre Filadelfia

Content

3 Professors

2 Ass. Professors

2 post docs

12 Ph.D. students

85 Publications *)

105 Oral Presentations

**) 85 papers published in peer reviewed journals including leading journals, such as Lancet Neurology, Nature Genetics, JAMA Neurology, Brain, Annals of Neurology, EBio Medicine, Neurology and Epilepsia.*

1. Introduction – Filadelfia

Filadelfia - The Danish Epilepsy Centre - offers highly specialized healthcare services to epilepsy patients of all ages. 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 are internationally recognized. In 2023, our team, consisting of three professors, two associate professors, two postdocs, and 12 PhD students, published 85 research papers, including in leading journals in the field of clinical neuroscience, and delivered 105 oral presentations at international medical conferences.

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

Filadelfia, The Danish Epilepsy Center, is a non-profit foundation with an independent Board of Directors. In addition to the Epilepsy Hospital, Filadelfia comprises a center 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. The Epilepsy Hospital receives patients from the five Danish Regions, i.e., the public hospitals and practitioners. The specialized social institutions receive clients from the Danish Municipalities.

We are proud to publish this report and hope it will encourage further collaboration for the benefit of persons with epilepsy worldwide.

As the Medical Director, I want to express my gratitude to all the collaborators in the research teams for their efforts and great achievements in 2023, and to Professor Sándor Beniczky for his leading role in producing this research report.

Jens-Otto Skovgaard Jeppesen
Managing Hospital Director
Danish Epilepsy Centre, Filadelfia

Mads Ravnborg
Medical Director, DMSc
Danish Epilepsy Centre, Filadelfia

2. Core Research Team 2022

Sándor Beniczky MD, Ph.D.

Professor, Head of Department

- Editor-in-Chief, Epileptic Disorders
- Founding Co-Chair, ILAE Neurotechnology Section (2022-2023)
- Chair, EEG Task Force, ILAE Commission on Big Data
- Past-chair, Joint Taskforce on EEG of the International Federation of Clinical Neurophysiology (IFCN) and the International League Against Epilepsy (ILAE)
- Steering committee member, European Reference Network EpiCare
- Member, ILAE Commission on Diagnostic Methods
- Member, ILAE Education Council; coordinator of the Virtual Epilepsy Academy (VIREPA)
- Member, ILAE Publication Council
- Member, ILAE Congress Council
- Member, IFCN Guidelines Committee

Rikke Steensbjerre Møller, Ph.D.

Professor, Head of department

- Speaker of a thematic research column within Neuroscience Academy Denmark
- Scientific advisory board member: KCNA2 Foundation
- Scientific advisory board member: KCNT1 Foundation
- Member of EpiCARE: a European Reference Network for rare and complex epilepsies
- Member of the management team at Department of Regional Health Research, University of Southern Denmark, Odense, Denmark
- Member of the scientific committee: Residras; a European Registry of Dravet Syndrome

Guido Rubboli MD, Ph.D.

Professor, Senior Consultant

- Member of the Management Group of the Coordinating Panel on Rare Neurological Diseases of the European Academy of Neurology
- Member, Epilepsy Scientific Panel, European Academy of Neurology
- Member, Task Force “Transition in care from Childhood into Adulthood”, Member International League Against Epilepsy (ILAE)
- Member, scientific committee “Epilessia- Fuori dall’ombra (Epilepsy – Out of the shadow)”
- Director and tutor, VIREPA (Virtual Epilepsy Academy) ILAE, Advanced EEG Course
- Member of the scientific committee KCNT1 Foundation
- Member of the scientific committee KCNA2 Foundation
- Member, Executive Committee. European Reference Network for Rare Disease – EpiCARE
- Member of the Management Group of the Epilepsy Scientific Panel, European Academy of Neurology
- Member, Rare Diseases Scientific Panel on Epilepsy. European Academy of Neurology
- Member, ILAE Task Force “SNOMED-CT”
- Member Joint EAN/ EPNS Task Force on transition from childhood to adulthood
- Associate Editor, Epileptic Disorder
- Section Editor, Behavioral Neurology
- Associate Editor, Frontiers in Neurology
- Editorial board member, Epilepsy and Behavior Reports.

Marina Nikanorova MD, Ph.D.

Associate Professor, Senior Consultant

Elena Gardella MD, Ph.D.

Associate Professor, Senior Consultant

- Member of the commission on Classification and Terminology of the International League against Epilepsy (ILAE)
- Co-director of VIREPA basic-EEG course of the International League against Epilepsy (ILAE)
- Member of the board of the Danish Epilepsy Society (DES)
- Member of Molecular Therapeutic Board in Neurological Channelopathies EpiCARE: a European Reference Network for rare and complex epilepsies.
- Author/revisorof Orphacodes of Orphanet (member of the EpiCare- working group WG2)
- Coordinator the registry database and cofounder of the European STXBP1 consortium (ESCO)
- Member of a thematic research column within Neuroscience Academy Denmark (NAD)
- Member of the BRIDGE team at Department of Regional Health Research, University of Southern Denmark
- Member of the steering committee of the Danish Epilepsy Database of RKKP (Regionernes Kliniske Kvalitetsudviklingsprogram)
- Editor of Frontiers in Neurology
- Grant reviser for COST (European Cooperation in Science and Technology)
- Scientific advisory board member: SCN8A Italy Foundation
- Scientific advisory board member: SCN8A Europe Foundation
- Scientific advisory board member: SCL6A1 Foundation

Allan Bayat MD, Ph.D.

Translational Researcher, Consultant

- Post doctoral BRIDGE Translational researcher at University of Copenhagen in collaboration with Department of Clinical Genetics, Rigshospitalet and the Danish Epilepsy CenteCo-organizer of the Nordic Course on Epilepsy Genetics and Precision Therap
- Member of the ERN-Ithaca KBG Guideline Steering committe
- Member of NorEpiNET, a newly established Nordic educational network dedicated to fostering teaching and collaboration in both clinical and research aspects within the field of epilepsy
- Member of Epicare WG5 on neuropsychology

Research Team

Daniella Terney MD, PhD
Stephan Wüstenhagen, MD, PhD
Pirgit Meritam Larsen, MD, PhD
Jesper Jeppesen, PhD
Maria Vlachou, MD
Levente Hadady, MD
Sidsel Armand Larsen, MSc.
Trine Hammer, MD, PhD
Christina Fenger, PhD
Cathrine Gjerulfsen, MD
Anne Højte Hansen, study nurse
Nazanin Mohammadi, MSc
Francesca Furia, MD
Benedetta Kassabian, MD
Sebastian Ortiz, MD

Leonardo Affronte, MD
Sopio Gverdtsiteli, MD
Matthias De Wacther, MD
Valentina Di Micco, MD
Alessandro Ferretti, MD
Tanya Techlo, MSc
Anne Juul Busch, study nurse
Alessandra Rossi, MD
Alberto Cossu, MD
Roberto Previtali, MD
Caterina Ancora, MD
Anne Vagner Jakobsen, Post doc.
Conny Hjelm, PhD
Trine Arnam-Olsen Moos, PhD

3. Ph.D. projects

Ongoing

- Maria Vlachou: Evaluation of electro-clinical findings using standardised feature extraction and machine learning. Aarhus University. Main supervisor: Sándor Beniczky.
- Sidsel Armand Larsen: Digital Technology in Epilepsy. Aarhus University. Main supervisor: Sándor Beniczky.
- Levente Hadady: Assessment of the clinical impact of electronic applications and wearable devices on the clinical management of patients with epilepsy. University of Szeged, Hungary. Main supervisor: Sándor Beniczky.
- Amir Baroumand: Automated EEG Source imaging. Ghent University, Belgium. Main supervisor: Pieter van Mierlo. Co-supervisor: Sándor Beniczky
- Francesca Furia: Deep phenotyping of monogenic epilepsies towards the identification of targeted treatments. University of Southern Denmark. Main supervisor: Elena Gardella and co-supervisors: Rikke Steensbjerre Møller and Guido Rubboli
- 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
- Frederik Nørby Friis Sørensen: Dissecting neuronal heterogeneity and epileptogenesis in focal cortical dysplasia. University of Copenhagen. Main supervisor: Konstantin Khodosevich and 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
- Tanya Ramdal Techlo: Leverage polygenic approaches to genetically diagnose idiopathic severe epilepsy and hemiplegic migraine. University of Copenhagen. Main supervisor: Thomas Folkmann Hansen and co-supervisor: Rikke Steensbjerre Møller
- 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
- Cristina Cioclu: Focal cortical dysplasia - pathogenetic mechanisms. University of Modena (Italy). Main supervisor: Stefano Meletti and co-supervisor: Guido Rubboli
- Nazanin Azarinejad Mohammadi: “Clinical and functional characterization of GABAA-receptor related disorders: translating genetic diagnostics into personalized treatment”, Syddansk Universitet. Main supervisor: Rikke Møller and co-supervisor: Guido Rubboli.

4. Conferences and congresses

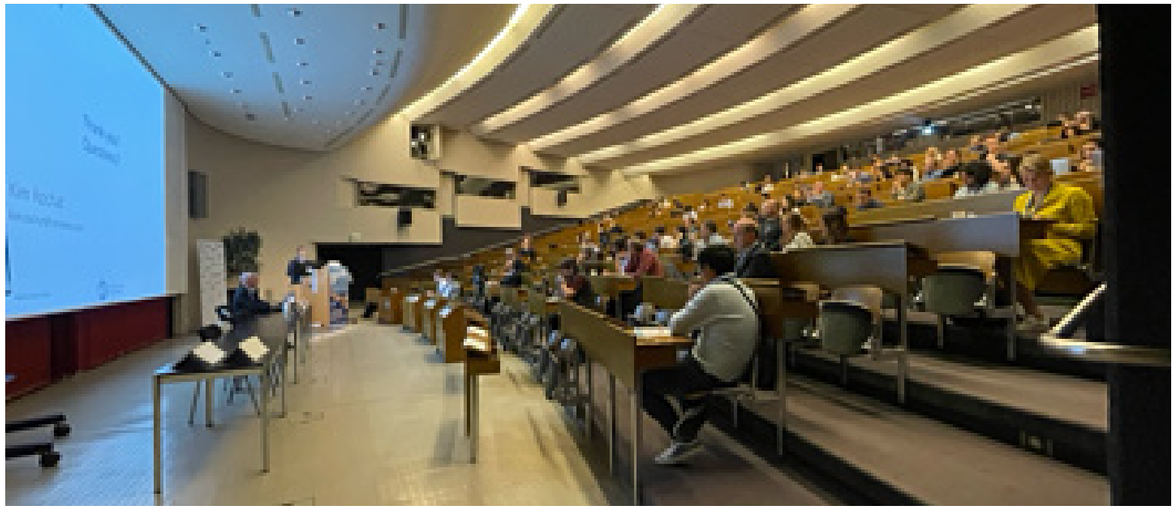
4.1 International Conference on Artificial Intelligence in Epilepsy and Neurological Disorders. March 7-10 2023. Breckenridge, CO, USA.

We have co-organized the first international conference on AI in epilepsy, co-chaired by Sándor Beniczky, Samden Lhatoo, Philippe Ryvlin and Michael Sperling.

The program included 33 lectures and 60 posters on cutting-edge research. The conference attracted over 200 participants, including leading authorities in this field.

4.2 4th International Congress on Mobile Health and Digital Technology in Epilepsy. October 11-13, 2023, Lausanne, Switzerland

We have co-organized the 4th international congress on mobile health and digital technology in epilepsy, co-chaired by Sándor Beniczky and Philippe Ryvlin. The biennial congress alternatingly held in Copenhagen and Lausanne, became a well-established meeting forum for neurotechnology research and development in epilepsy care. The program in 2023 included 25 lectures and 56 posters. The conference attracted 200 participants, including leading authorities in this field, with participants from all over the world.



4.3 Danish-Brazilian Workshop of Wearable Healthcare Technologies for the Central Nervous System. April 1st, 2023, Florianopolis, Brazil.

The objective of the workshop was to provide a dynamic ecosystem of information exchange among Danish and Brazilian health-tech industry, engineers, medical organizations, researchers, and healthcare professionals to develop new strategies for therapeutics and digital health innovations to converge and ultimately deliver better outcomes for patients with epilepsy. The program comprised eight lectures and three panel discussions, and was supported by a grant form the Danish Ministry of Higher Education and Science (International Network Program).

4.4 2nd SCN2A/SCN8A gathering and family meeting, 26-27.05.23, Køge, Denmark.

The objective of the workshop was to provide a dynamic ecosystem of information exchange among Danish and Brazilian health-tech industry, engineers, medical organizations, researchers, and healthcare professionals to develop new strategies for therapeutics and digital health innovations to converge and ultimately deliver better outcomes for patients with epilepsy. The program comprised eight lectures and three panel discussions, and was supported by a grant form the Danish Ministry of Higher Education and Science (International Network Program).

4.5 1st European STXBP1 Summit and Research Roundtable Milan on May 16-17-18, 2023

This is the first researchers and families gathering meeting for STXBP1 in Europe.
Including physicians, basic scientists family representatives, and pharma companies from several European countries (UK, Italy, France, Germany, Georgia, Switzerland), Israel, and the USA.
During the meeting were launched the European Consortium on STXBP1, for which we are co-founders, and the European STXBP1 Registry, which will be stored at the Danish Epilepsy Centre (Coordinator: Elena Gardella, Administrator: Francesca Furia) on behalf of the ESCO consortium

4.6 1st Nordic Course on Epilepsy Genetics and Precision Therapy, 20-21.11.23, Stockholm, Sweden

As part of NorEpiNet, Allan Bayat (MD, PhD), Cecilie Johannessen Landmark (Professor MScPharm), Kaja Selmer (MD, PhD), Rikke Steensbjerre Møller (Professor, MSc), Mathilda Modigh (Nurse, PhD student) and Tommy Stödberg (MD, PhD) organized this meeting with 70 physicians and basic scientists from the Nordic countries, UK and USA.

5. Research projects

5.1 Artificial Intelligence in epilepsy

Artificial intelligence (AI) is about to revolutionize healthcare, and this has reached the field of epilepsy. AI holds immense potential to improve efficacy and quality of diagnostic evaluations. Using AI, we developed and validated automated and semi-automated analysis methods of EEG and of video recordings with epileptic seizures.

In collaboration with University of Bergen and Holberg-EEG, we developed an AI-model for automated interpretation of routine, clinical EEGs. The AI-model was developed using an artificial neural network, based on large EEG datasets (over 30 thousand de-identified recordings), systematically labeled using the SCORE software (Standardized Computer-based Organized Reporting of EEG). SCORE-AI was tested on a multicenter test dataset, assessed by international experts, and a large, independent dataset from Oslo University Hospital. SCORE-AI reached performance equivalent to human experts in distinguishing normal from abnormal EEGs, and then classifying the abnormal EEGs into the categories with clinical relevance (focal epileptiform, generalized epileptiform, focal non-epileptiform, diffuse non-epileptiform).

The paper reporting SCORE-AI received much attention internationally. It appeared in numerous editorials, news, posts, tweets and blogs, giving an Altmetric score of 318. It was one of the most viewed articles in JAMA Neurology in 2023 (with close to 30 thousand views in less than 5 months).

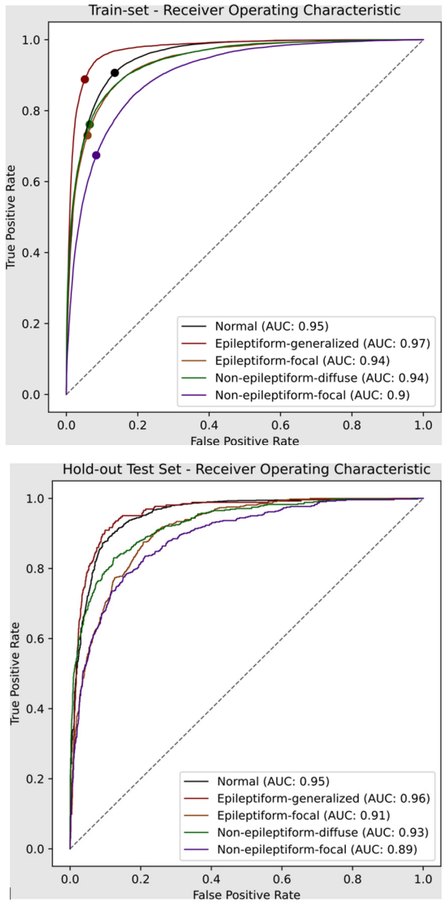


Fig. Receiver Operating Characteristic (ROC) curve of SCORE-AI, in the development and test datasets, demonstrating the high performance and generalizability of the model.

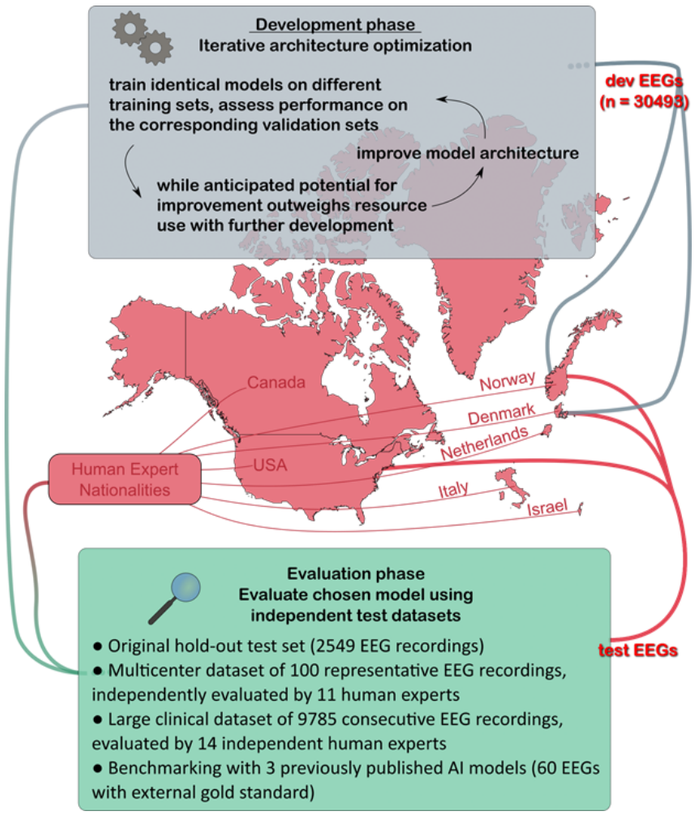


Fig. SCORE-AI: Flowchart of validation.

Papers

Knight A, Gschwind T, Galer P, Worrell GA, Litt B, Soltesz I, **Beniczky S**.
Artificial intelligence in epilepsy phenotyping.
Epilepsia. 2023 Nov 20. doi: 10.1111/epi.17833.

Tveit J, Aurlien H, Plis S, Calhoun VD, Tatum WO, Schomer DL, Arntsen V, Cox F, Fahoum F, Gallentine WB, Gardella E, Hahn CD, Husain AM, Kessler S, Kural MA, Nascimento FA, Tankisi H, Ulvin LB, Wennberg R, **Beniczky S**.
Automated Interpretation of Clinical Electroencephalograms Using Artificial Intelligence.
JAMA Neurol. 2023 Aug 1;80(8):805-812. doi: 10.1001/jamaneurol.2023.1645.

5.2 Big Data

Large structured datasets are essential for medical research. They make possible datamining and addressing clinical questions, based on a data-driven approach.

Since 2013, we have been using the Standardized Computer-based Organized Reporting of EEG (SCORE). The large dataset helped in addressing various clinical questions. In 2023, we published a paper establishing the duration of the epileptic seizure-types, based on real-world data from SCORE. These results are helpful in clinical practice, for identifying and classifying the various seizure types.

Based on a large international dataset with de-identified data from epilepsy surgery programs, we have validated and conducted a head-to-head comparison of tools for predicting outcome after surgery. We developed and validated a method for risk-stratification in this group of patients. We contributed to the analysis of a large Swiss dataset, to evaluate the diagnostic yield of prolonged EEG and MRI after the first seizures.

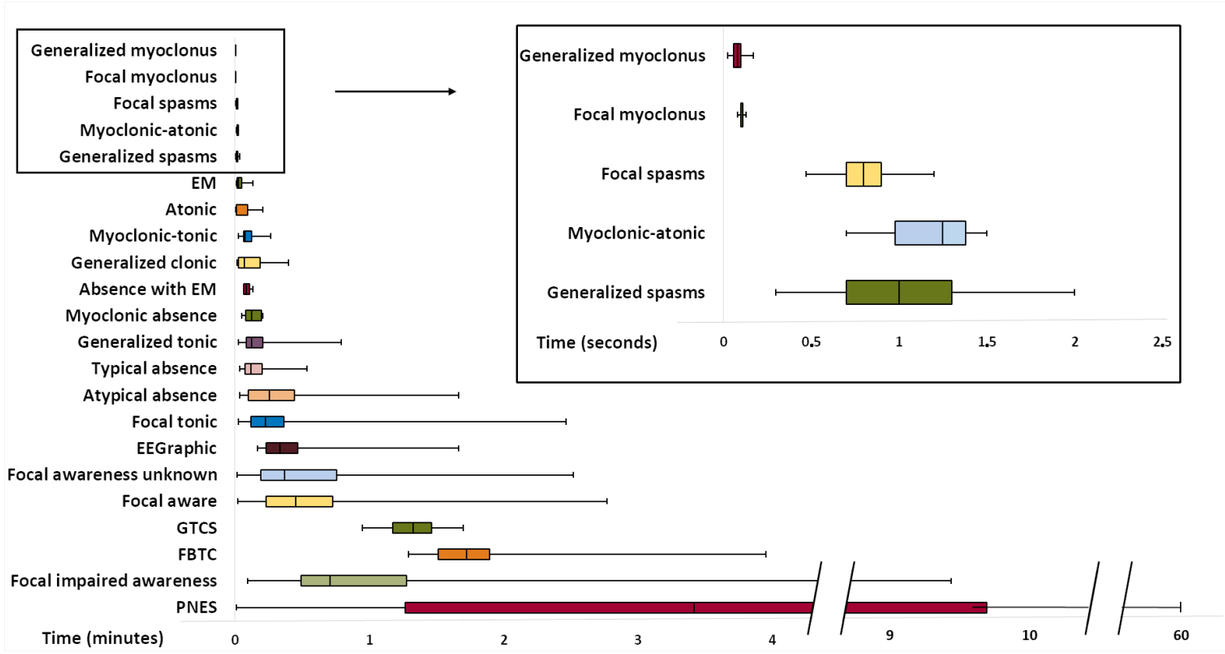


Fig. Duration of epileptic seizures, based on real-world data from SCORE.

Papers

Meritam Larsen P, **Wüstenhagen S**, **Terney D**, Gardella E, Aurlien H, **Beniczky S**.
Duration of epileptic seizure types: A data-driven approach.
Epilepsia. 2023 Feb;64(2):469-478. doi: 10.1111/epi.17492.

Hadady L, Sperling MR, Alcala-Zermeno JL, French JA, Dugan P, Jehi L, Fabó D, Klivényi P, **Rubboli G**, **Beniczky S**.
Prediction tools and risk stratification in epilepsy surgery.
Epilepsia. 2023 Dec 7. doi: 10.1111/epi.17851.

De Stefano P, Ménétré E, Stancu P, Mégevand P, Vargas MI, Kleinschmidt A, Vulliémoz S, Wiest R, **Beniczky S**, Picard F, Seeck M.
Added value of advanced workup after the first seizure: A 7-year cohort study.
Epilepsia. 2023 Dec;64(12):3246-3256. doi: 10.1111/epi.17771.

5.3 Seizure detection & wearables

Automated detection of epileptic seizures using wearable devices is needed for safety indications and for objective quantification of seizure-burden. We continued our decades-long effort to develop and validate seizure detection wearables.

Using machine learning, we optimized our heart-rate variability (HRV) based seizure detection algorithm, which helped us decrease the false alarm rate. We conducted a proof-of-principle study and published a seminal paper on using subcutaneously implanted electrocardiogram (ECG) for automated seizure detection of epileptic seizure. This application has huge potential in clinical practice. It combines a hardware, which is well-established in cardiology, with a novel algorithm for seizure detection.

We have reviewed the state-of-the-art and current clinical practices in using wearables for seizure detection. These results will likely promote further research and development in this important field.

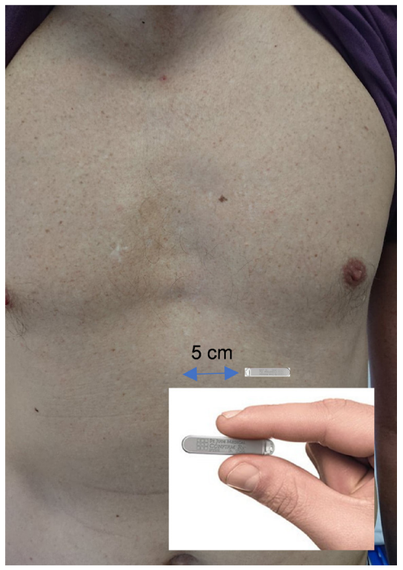


Fig. Automated seizure detection using subcutaneously implanted ECG

Papers

Jeppesen J, Christensen J, Johansen P, **Beniczky S**.
Personalized seizure detection using logistic regression machine learning based on wearable ECG-monitoring device.
Seizure. 2023 Apr;107:155-161. doi:10.1016/j.seizure.2023.04.012.

Jeppesen J, Christensen J, Mølgaard H, **Beniczky S**.
Automated detection of focal seizures using subcutaneously implanted electrocardiographic device: A proof-of-concept study.
Epilepsia. 2023 Dec;64 Suppl 4:S59-S64. doi: 10.1111/epi.17612.

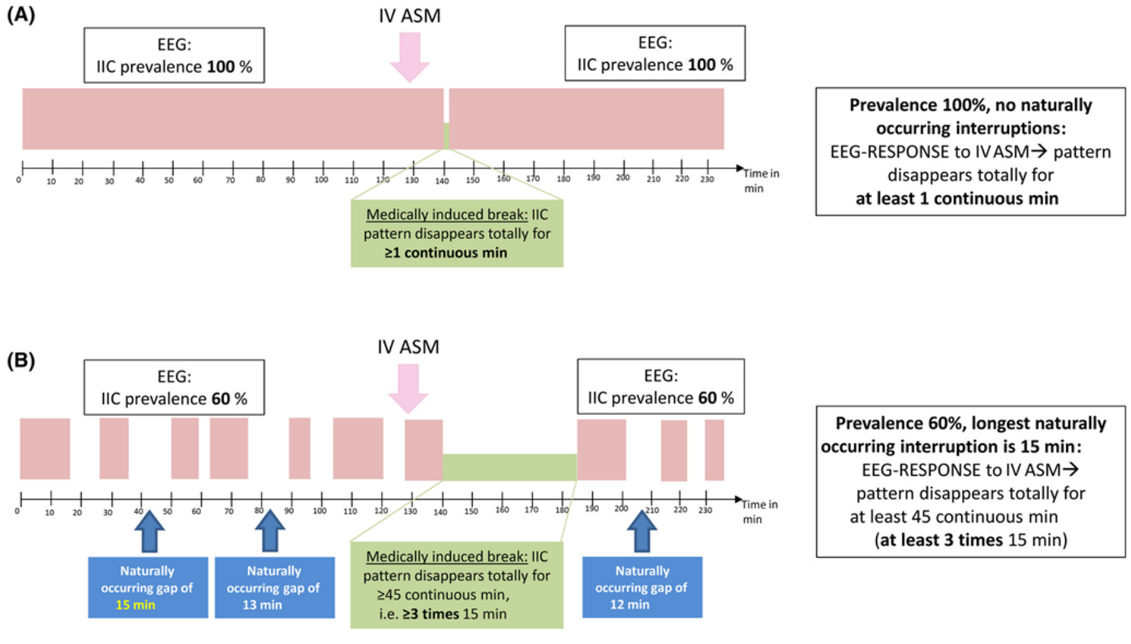
Zelano J, **Beniczky S**, Ryvlin P, Surges R, Tomson T; ILAE SUDEP Task Force.
Report of the ILAE SUDEP Task Force on national recommendations and practices around the world regarding the use of wearable seizure detection devices: A global survey.
Epilepsia Open. 2023 Dec;8(4):1271-1278. doi: 10.1002/epi4.12801.

Beniczky S, Ryvlin P.
Mobile health and digital technology in epilepsy: The dawn of a new era.
Epilepsia. 2023 Dec;64 Suppl 4:S1-S3. doi: 10.1111/epi.17813.

Meritam Larsen P, **Beniczky S**.
Non-electroencephalogram-based seizure detection devices: State of the art and future perspectives.
Epilepsy Behav. 2023 Nov;148:109486. doi: 10.1016/j.yebeh.2023.109486.

5.4 Guidelines & recommendations

Guidelines and recommendations, based on systematic assessment of published evidence, has an upmost importance for high quality clinical care. In 2023, we contributed to development of international guidelines on minimum standards for routine and sleep EEG, endorsed by both the International Federation of Clinical Neurophysiology and the International League Against Epilepsy. We participated in consensus-discussions and published a paper on using intravenous antiseizure medications for diagnosing status epilepticus. We contributed to the work of an international consortium to develop DICOM-format for EEG.



EEG reponse to intravenous anti-seizure medication.

Fig. Diagnosing nonconvulsive status epilepticus: Defining electroencephalographic and clinical response to diagnostic intravenous antiseizure medication trials

Papers

Peltola ME, Leitinger M, Halford JJ, Vinayan KP, Kobayashi K, Pressler RM, Mindruta I, Mayor LC, Lauronen L, **Beniczky S**.
Routine and sleep EEG: Minimum recording standards of the International Federation of Clinical Neurophysiology and the International League Against Epilepsy.
Dual publication in:
Clin Neurophysiol. 2023 Mar;147:108-120. doi: 10.1016/j.clinph.2023.01.002.
Epilepsia. 2023 Mar;64(3):602-618. doi: 10.1111/epi.17448.

Leitinger M, Gaspard N, Hirsch LJ, **Beniczky S**, Kaplan PW, Husari K, Trinka E.
Diagnosing nonconvulsive status epilepticus: Defining electroencephalographic and clinical response to diagnostic intravenous antiseizure medication trials.
Epilepsia. 2023 Sep;64(9):2351-2360. doi: 10.1111/epi.17694.

Halford JJ, Brinkmann BH, Clunie DA, Gotman J, **Beniczky S**, Rampp S, Rémi J, Husain A, Andrew Ehrenberg J, Winkler S.
Continued progress in DICOM neurophysiology standardization.
Clin Neurophysiol. 2023 Mar;147:11-13. doi: 10.1016/j.clinph.2022.12.008.

5.5 Education

Education and research in postgraduate education receive high priority in our hospital. We published educational review papers and multimedia teaching material to spread the new knowledge cumulated in our field. We critically addressed the impact and efficacy of various curricula and teaching methods.

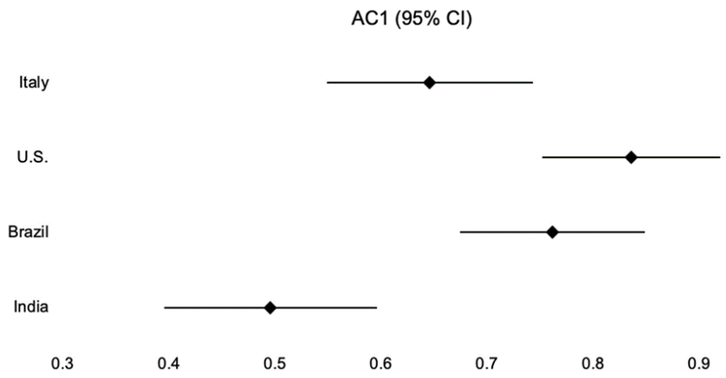


Fig. Expert accuracy and inter rater agreement of “must know” EEG findings for adult and child neurology residents, in different geographic regions.

Papers

Frauscher B, Mansilla D, Abdallah C, Astner-Rohracher A, **Beniczky S**, Brazdil M, Gnatkovsky V, Jacobs J, Kalamangalam G, Perucca P, Ryvlin P, Schuele S, Tao J, Wang Y, Zijlmans GJM, McGonigal A.
Learn how to interpret and use intracranial EEG findings.
Epileptic Disord. 2023 Dec 20. doi: 10.1002/epd2.20190.

Nascimento FA, Katyal R, Olandoski M, Gao H, Yap S, Matthews R, Rampp S, Tatum W, Strowd R, **Beniczky S**.
Expert accuracy and inter-rater agreement of “must-know” EEG findings for adult and child neurology residents.
Epileptic Disord. 2023 Nov 30. doi: 10.1002/epd2.20186.

Greenblatt AS, **Beniczky S**, Nascimento FA.
Pitfalls in scalp EEG: Current obstacles and future directions.
Epilepsy Behav. 2023 Dec;149:109500. doi: 10.1016/j.yebeh.2023.109500.

Nascimento FA, Salazar M, Colonetti J, Schomer D, **Beniczky S**.
How to conduct EEG recordings-A video-based educational resource.
Epileptic Disord. 2023 Dec;25(6):911-913. doi: 10.1002/epd2.20089.

Nascimento FA, **Beniczky S**.
Sawtooth waves: An EEG normal variant.
Epileptic Disord. 2023 Feb;25(1):120-121. doi: 10.1002/epd2.20032.

Nascimento FA, Friedman D, Peters JM, Bensalem-Owen MK, Cendes F, Rampp S,
Wirrell E, Blümcke I, Tatum W, **Beniczky S**.
Focal epilepsies: Update on diagnosis and classification.
Epileptic Disord. 2023 Feb;25(1):1-17. doi: 10.1002/epd2.20045.
Gogou M, Sheikh IS, Tamula ORM 3rd, Katyal R, **Beniczky S**, Nascimento FA.
Competency-based epilepsy training: A comparison between U.S.-based milestones and ILAE curriculum.
Epileptic Disord. 2023 Aug;25(4):586-589. doi: 10.1002/epd2.20037.

Nascimento FA, **Beniczky S**.
Teaching the 6 Criteria of the International Federation of Clinical Neurophysiology for Defining Interictal Epileptiform Discharges on EEG Using a Visual Graphic.
Neurology® Education 2023;2:e200073. doi:10.1212/NE9.0000000000200073

Nascimento FA, Gao H, Katyal R, Matthews R, Yap SV, Rampp S, Tatum WO, Strowd RE, **Beniczky S**.
Education Research: Competency-Based EEG Education. An Online Routine EEG Examination for Adult and Child Neurology Residents.
Neurology® Education 2023;2:e200094. doi:10.1212/NE9.0000000000200094

5.5 GARA-A receptor related epilepsies

Developmental and Epileptic Encephalopathies (DEEs) are devastating early-onset conditions associated with intractable epilepsy, intellectual disability, developmental delay/regression, movement, and autism spectrum disorders. The condition is highly debilitating for both the impacted children and their caretakers. Genetic factors play a major role in the underlying cause of DEEs, and pathogenic variants in subunits of the γ-aminobutyric acid type A receptor (GABAAR), the major inhibitory protein of the brain, are found to be a common cause of DEE. Until recently, these were assumed to lead to loss-of-function (hypoactive) receptors supporting a hypothesis that reduced GABAergic inhibition causes overexcitation in the brain leading to epilepsy.

Recently, we have published exciting new data that contradicts the belief that only loss-of-function GABAARs cause DEE. We discovered that the functional assessment of pathogenic variants from a subset of GABAAR subunits, led to an equal distribution of loss- and gain-of-function receptors. It is currently unknown how increased GABAAR function leads to DEE, however, the paradoxical finding of gain GABAARs has subsequently been confirmed by others. Furthermore, we discovered that patient phenotypes were linked to the functional outcome of the variants and that gain variants are associated with more severe forms of treatment resistant epilepsy.

Papers

Absalom NL, Lin SXN, Liao VWY, Chua HC, **Møller RS**, Chebib M, Ahring PK. GABAA receptors in epilepsy: Elucidating phenotypic divergence through functional analysis of genetic variants. J Neurochem. 2023 Aug 24. doi: 10.1111/jnc.15932. Online ahead of print.

Musto E, Liao VWY, **Johannesen KM**, **Fenger CD**, Lederer D, Kothur K, Fisk K, Bennetts B, Vrielynck P, Delaby D, Ceulemans B, Weckhuysen S, Sparber P, Bouman A, Ardern-Holmes S, Troedson C, Battaglia DI, Goel H, Feyma T, Bakhtiari S, Tjoa L, Boxill M, Demina N, Shchagina O, Dadali E, Kruer M, Cantalupo G, Contaldo I, Polster T, Isidor B, Bova SM, Fazeli W, Wouters L, Miranda MJ, Darra F, Pede E, Le Duc D, Jamra RA, Küry S, Proietti J, McSweeney N, Brokamp E, Andrews PI, Gouray Garcia M, Chebib M, **Møller RS**, Ahring PK, Gardella E. GABRA1-Related Disorders: From Genetic to Functional Pathways. Ann Neurol. 2023 Aug 22. doi: 10.1002/ana.26774. Online ahead of print.

Gjerulfsen CE, **Mieszczanek TS**, **Johannesen KM**, Liao VWY, Chebib M, Nørby HAJ, Gardella E, **Rubboli G**, Ahring P, **Møller RS**. Vinpocetine improved neuropsychiatric and epileptic outcomes in a patient with a GABRA1 loss-of-function variant. Ann Clin Transl Neurol. 2023 Aug;10(8):1493-1498. doi: 10.1002/acn3.51838. Epub 2023 Jul 11.

5.6 Education

Clinical and electrophysiological features of SCN8A variants causing episodic or chronic ataxia

Pathogenic variants in SCN8A are associated with a wide spectrum of epilepsies and neurodevelopmental disorders, including movement disorders and ataxia. Though, the non-epilepsy symptoms of SCN8A developmental and epileptic encephalopathy (DEE) have not been well studied. We set out to investigate disease mechanisms and genotype–phenotype correlations of SCN8A-related ataxia.

Variants associated with chronic progressive ataxia either decreased Na+ current densities and shifted activation curves towards more depolarized potentials. Three variants were associated with episodic ataxia causing loss-of-function by decreasing Na+ current densities or a hyperpolarizing shift of the inactivation curve. Two additional episodic ataxia-associated variants caused mixed gain- and loss-of function effects. Treatment with sodium channel blockers might exacerbate symptoms.

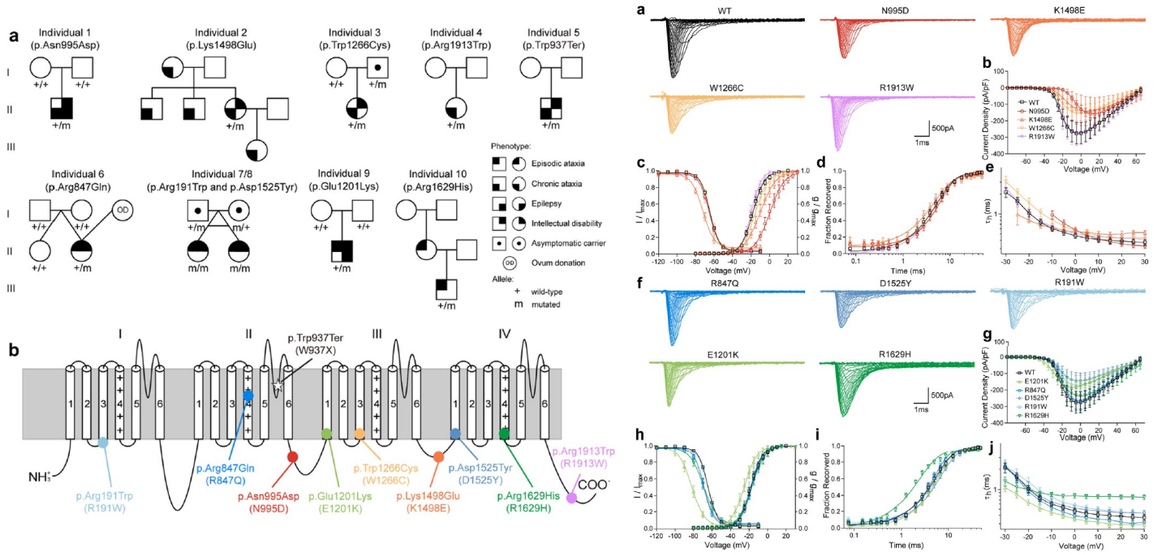


Fig. Right panels: SCN8A variants associated with episodic or chronic ataxia. (a) Pedigrees of study participants. (b) Position of variants in human Nav1.6 channel.

Left panels: Functional studies. (a) Representative traces of Na+ current for wild-type (WT) and SCN8A variants associated with chronic ataxia. (b) Peak Na+ currents normalised by cell capacitances were plotted versus voltage. (c) Voltage-dependent steady state activation and inactivation curves. (d) Time course of recovery from fast inactivation at −100 mV. (e) Voltage-dependence of the time constant of fast inactivation. (f) Representative traces of Na+ current for WT and variants associated with episodic ataxia. (g) Peak Na+ currents normalised by cell capacitances were plotted versus voltage. (h) Voltage dependent steady state activation and inactivation curves. (i) Time course of recovery from fast inactivation at −100 mV. (j) Voltage-dependence of the time constant of fast inactivation

Papers

Lyu H, Boßelmann CM, **Johannesen KM**, Koko M, Ortigoza-Escobar JD, Aguilera-Albesa S, Garcia-Navas Núñez D, Linnankivi T, Gaily E, van Ruiten HJA, Richardson R, Betzler C, Horvath G, Brilstra E, Geerdink N, Orsucci D, Tessa A, Gardella E, Fleszar Z, Schöls L, Lerche H, **Møller RS**, Liu Y. Clinical and electrophysiological features of SCN8A variants causing episodic or chronic ataxia. EBioMedicine. 2023 Dec;98:104855. doi: 10.1016/j.ebiom.2023.104855. Epub 2023 Oct 28.

5.7 IRF2BPL as a novel gene for progressive myoclonus epilesy

Progressive myoclonus epilepsies (PMEs) are a heterogeneous group of severe diseases, still genetically unsolved in one third of the cases. IRF2BPL has recently been described as a novel cause of neuro-developmental disorders with multi-systemic regression, epilepsy, cerebellar and pyramidal signs.

The achievements of this study were:

1. to describe a novel IRF2BPL phenotype consistent with PME and
2. to identify clear-cut genotype-phenotype correlations, permitting to predict the evolution of the symptoms based on the type and position of the gene variant.
3. to provide insights into the pathophysiological mechanism leading to the IRF2BPL-PME phenotype. We observed that the skin biopsy revealed massive intracellular glycogen inclusions, resembling Lafora bodies. Notably, both the dysfunctional protein causing Lafora disease (malin) and the IRF2BPL protein act as E3 ubiquitin ligase. This suggests a possible similar pathogenic pathway between IRF2BPL PME, Lafora disease, and other storage disorder PME, opening the pathway for precision treatment approaches of IRF2BPL-PME.

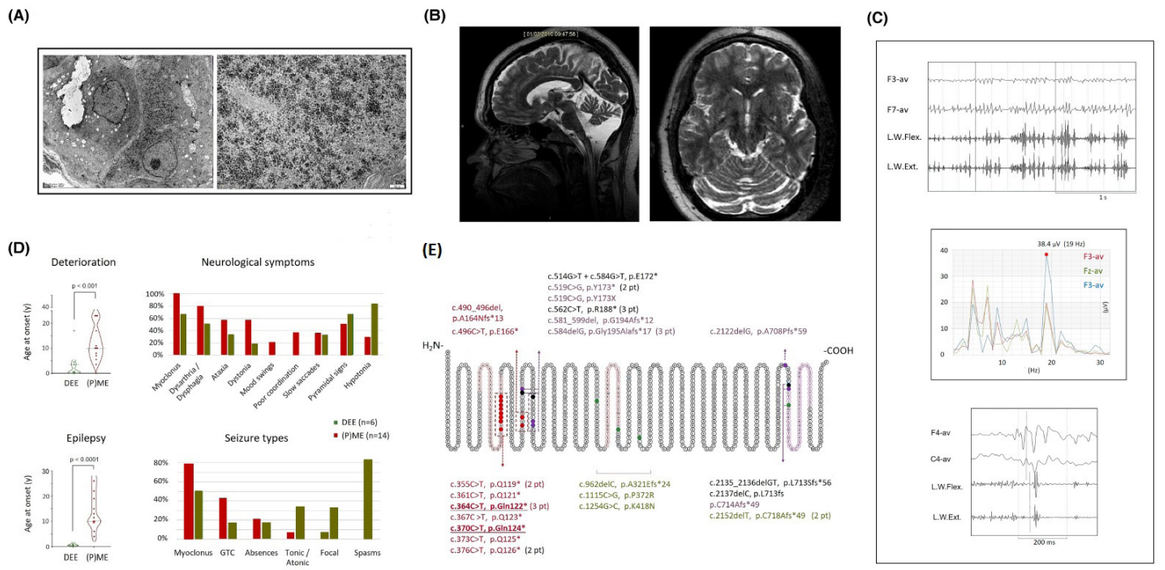


Fig. (A) Axillary skin biopsy. The electronic microscopy shows massive accumulations of glycogen particles free in the cytoplasm of the epithelial cells of a sweat gland. (B) Brain magnetic resonance imaging. CUBE T2-weighted images showing diffuse atrophy of the cerebellum and the bulbo-pontine structures. (C) EEG–EMG correlations. Surface EMG revealed rhythmic myoclonic jerks at a frequency of 11–22 Hz. The spectral EEG analysis revealed a frequency peak in the beta band (18–21 Hz). The EEG activity in the frontal regions (F4/F3) preceded the EMG activity in the contralateral wrist flexor muscles by 12.5–13.5 ms. (D) Clinical differences between subjects with IRF2BPL- developmental and epileptic encephalopathy [DEE] and [progressive] myoclonus epilepsy [(P)ME]). (D) IRF2BPL protein structure and variant position. The IRF2BPL includes two highly conserved domains—the coiled-coil (red circles) and the RING finger (purple circles), present in all members of the E3 ligase family, including malin. The positions of all the pathogenic IRF2BPL variants are indicated in green ([P]ME), red (DEE), purple (no epilepsy), or black (not further classified).

Papers

Gardella E, Michelucci R, Christensen HM, **Fenger CD**, Reale C, Riguzzi P, Pasini E, Albini-Riccioli L, Papa V, Foschini MP, Cenacchi G, Furia F, Marjanovic D, **Hammer TB**, **Møller RS**, **Rubboli G**. IRF2BPL as a novel causative gene for progressive myoclonus epilepsy. Epilepsia. 2023 Aug;64(8):e170-e176. doi: 10.1111/epi.17634. Epub 2023 Jun 8.

5.8 Atypical evolution of PRRT2 benign familial infantile seizures (BFIS) to encephalopathy related to status epilepticus during sleep (ESES)

Heterozygous variants in the PRRT2 gene are mostly associated with benign phenotypes, being the major genetic cause of benign familial infantile seizures (BFIS), as well as in paroxysmal disorders.
We report two children from unrelated families with BFIS that evolved to encephalopathy related to status epilepticus during sleep (ESES).

The mechanism leading to epilepsy and the phenotypic variability of PRRT2 variants remain poorly understood. However, its wide cortical and subcortical expression, in particular in the thalamus, could partially explain both the focal EEG pattern and the evolution to ESES. No variants in the PRRT2 gene have been previously reported in patients with ESES. Due to the rarity of this phenotype, other possible causative cofactors are likely contributing to the more severe course of BFIS in our probands.

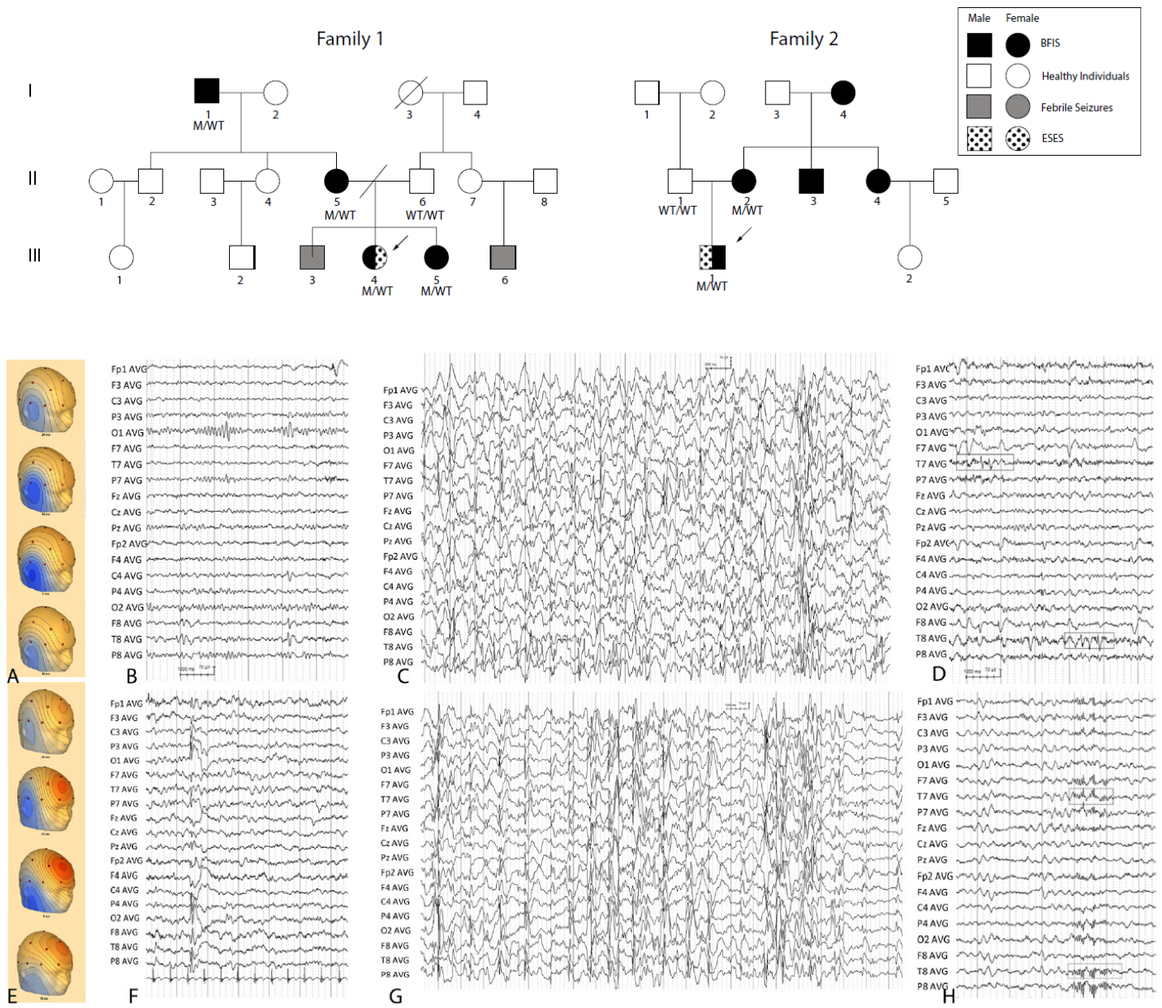


Fig. Pedigree of the Danish family (left) and of the Italian/Slovenian family (right) with benign familial infantile seizures (BFIS), febrile seizures (FS), and one case of encephalopathy related to status epilepticus during sleep (ESES). EEG analysis of P.1 (upper row) and P.2 (lower row): (A, E) sequential amplitude maps (10 ms interval) of IEDs during wakefulness, showing a dipole rotation between onset, peak, and slow wave. IEDs during wakefulness (B,F), NREM sleep (C, G = ESES pattern) and REM sleep (D, H).

Papers

Cossu A, Santos JL, Galati G, **Nikanorova M**, Costa P, Mang Y, Silahtaroglu A, **Rubboli G**, Tommerup N, Dalla Bernardina B, **Møller RS**, Cantalupo G, Gardella E. PRRT2 benign familial infantile seizures (BFIS) wit **Cossu A**, Santos JL, Galati G, **Nikanorova M**, Costa P, Mang Y, Silahtaroglu A, **Rubboli G**, Tommerup N, Dalla Bernardina B, **Møller RS**, Cantalupo G, Gardella E. PRRT2 benign familial infantile seizures (BFIS) with atypical evolution to encephalopathy related to status epilepticus during sleep (ESES). Neurol Sci. 2023 Jun;44(6):2173-2176. doi: 10.1007/s10072-023-06735-7. Epub 2023 Mar 13.h atypical evolution to encephalopathy related to status epilepticus during sleep (ESES). Neurol Sci. 2023 Jun;44(6):2173-2176. doi: 10.1007/s10072-023-06735-7. Epub 2023 Mar 13.

5.9 Biology of Juvenile Myoclonic Epilepsy – BIOJUME Consortium" and "Variation in prognosis and treatment outcome in juvenile myoclonic epilepsy".

This research showed that by applying a simplified set of criteria to define phenotypic variations of juvenile myoclonic epilepsy, evidence-based definition and prognostic stratification of juvenile myoclonic epilepsy can be implemented. A simplified set of diagnostic criteria for juvenile myoclonic epilepsy includes the following: (i) myoclonic jerks as mandatory seizure type; (ii) a circadian timing for myoclonia not mandatory for the diagnosis of juvenile myoclonic epilepsy; (iii) age of onset ranging from 6 to 40 years; (iv) generalized EEG abnormalities; and (v) intelligence conforming to population distribution.

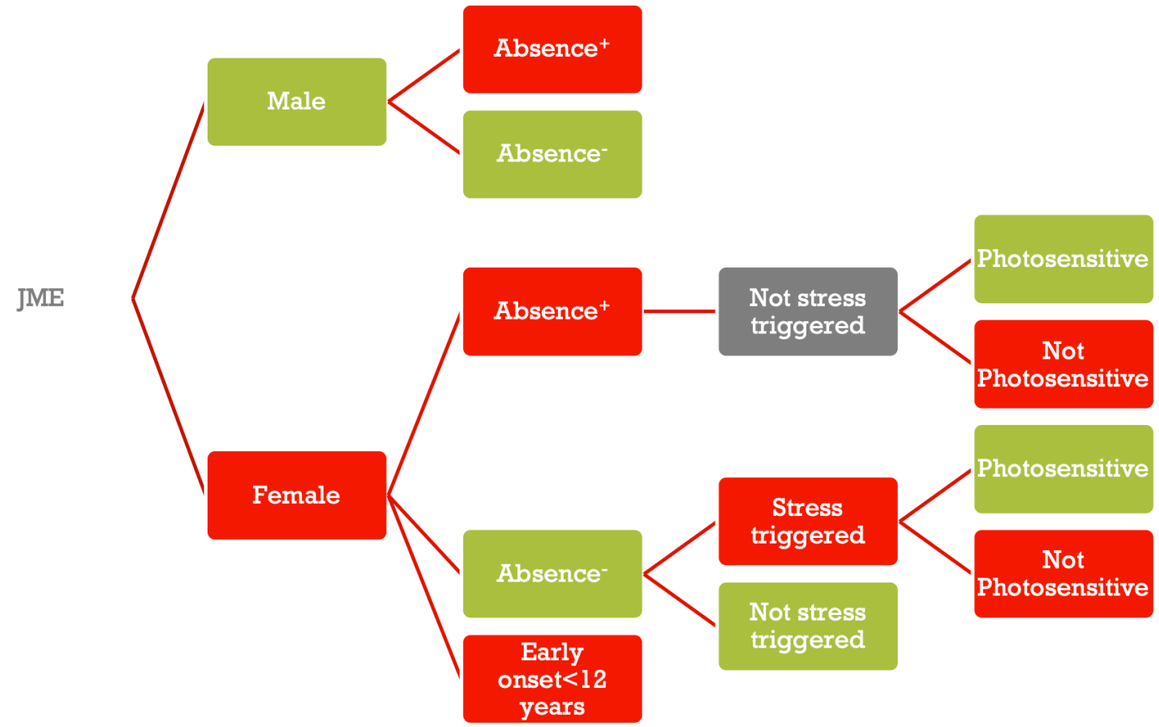


Fig. Proposed predictive model of ASM resistance in JME. At each stratum, blue denotes better prognosis, orange worse prognosis, and grey denotes neutral effect on outcome.

Papers

Rubboli G, Beier CP, Selmer KK, Syvertsen M, Shakeshaft A, Collingwood A, Hall A, Andrade DM, Fong CY, Gesche J, Greenberg DA, Hamandi K, Lim KS, Ng CC, Orsini A; **BIOJUME Consortium**; Striano P, Thomas RH, Zarubova J, Richardson MP, Strug LJ, Pal DK. Variation in prognosis and treatment outcome in juvenile myoclonic epilepsy: a Biology of Juvenile Myoclonic Epilepsy Consortium proposal for a practical definition and stratified medicine classifications. Brain Commun. 2023 Jun 9;5(3):fcad182. doi: 10.1093/braincomms/fcad182. eCollection 2023.

5.10 KCNT2-Related Disorders: Phenotypes, Functional, and Pharmacological Properties

This study outlined the phenotypic and genotypic spectrum of KCNT2-related disorders, highlighting novel genotype-phenotype associations. Pathogenic KCNT2 variants cause GoF or LoF in vitro phenotypes, and each shows a unique pharmacological profile.

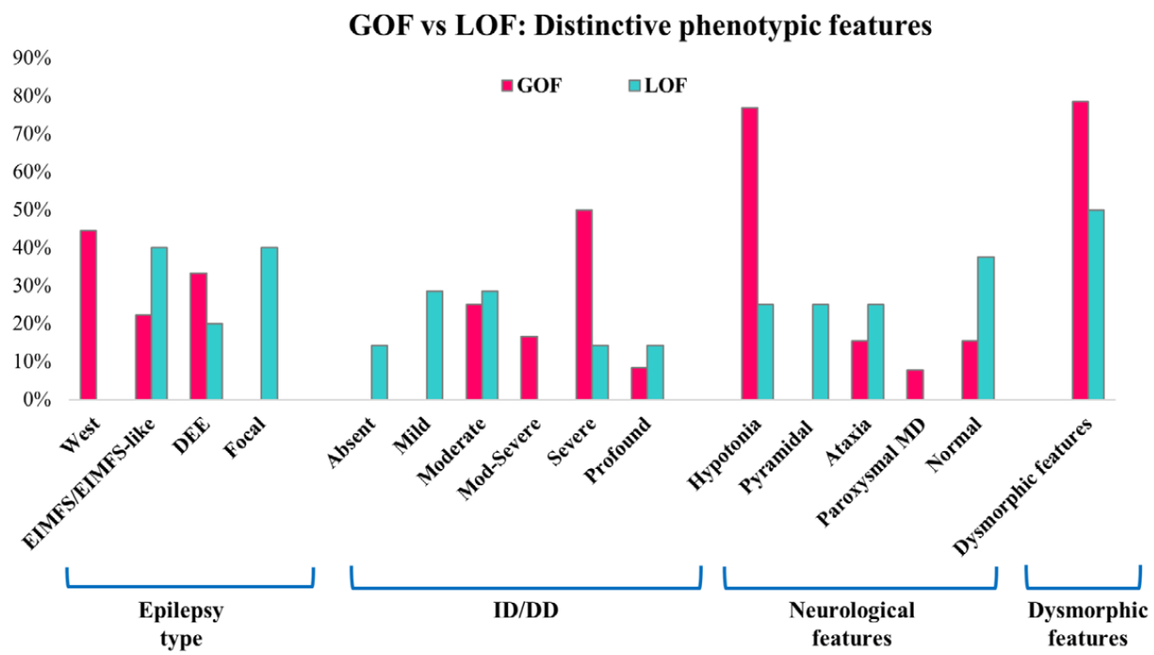


Fig. Representation of the main phenotypic features in patients carrying GoF (red bars) versus LoF variants (green bars); values are expressed as the percentage of patients presenting the trait out of the number of patients

Papers

Cioclu MC, Mosca I, Ambrosino P, Puzo D, **Bayat A**, Wortmann SB, Koch J, Strehlow V, Shirai K, Matsumoto N, Sanders SJ, Michaud V, Legendre M, Riva A, Striano P, Muhle H, Pendziwiat M, Lesca G, Mangano GD, Nardello R; KCNT2-study group; Lemke JR, **Møller RS**, Soldovieri MV, **Rubboli G**, Taglialatela M. KCNT2-Related Disorders: Phenotypes, Functional, and Pharmacological Properties. Ann Neurol. 2023 Aug;94(2):332-349. doi: 10.1002/ana.26662. Epub 2023 May 22.

5.11 SLC6A1: the past, present and future

A series of articles illustrated the current knowledge and the recent advances on SLC6A1-related neurodevelopmental disorders (SLC6A1-NDDs). However, there are still many aspects of this disorder that needs further elucidating. Moving closer to targeted treatment in SLC6A1-NDDs, further studies and research are warranted for a clear understanding of the disorder including the phenotypic range and natural history.

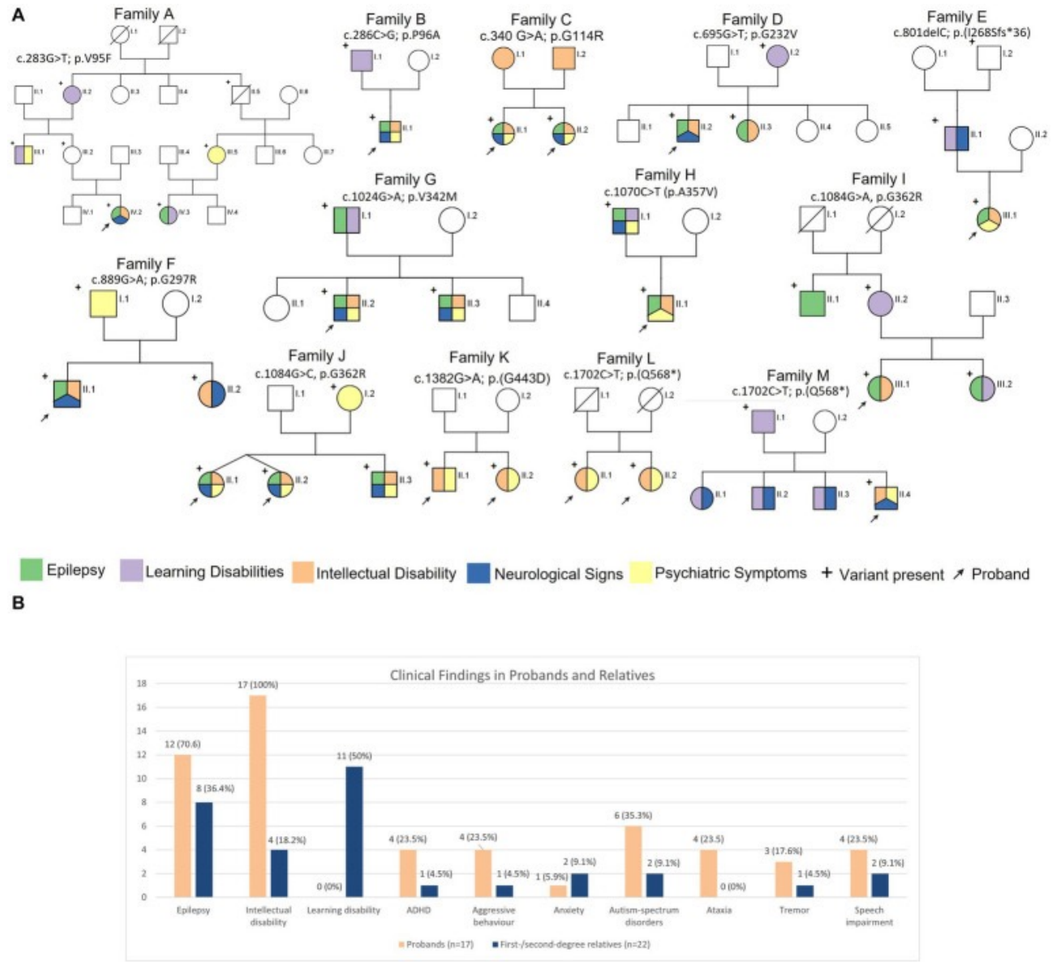


Fig. (A) Pedigrees of the 12 families featuring SLC6A1-relalted DEE and main clinical features in the reported families. (B) Bar graph illustrating the prevalence of the neurological features in the probands and first/second degree relatives. (Kassabian et al., Front Neurol 2023).

Papers

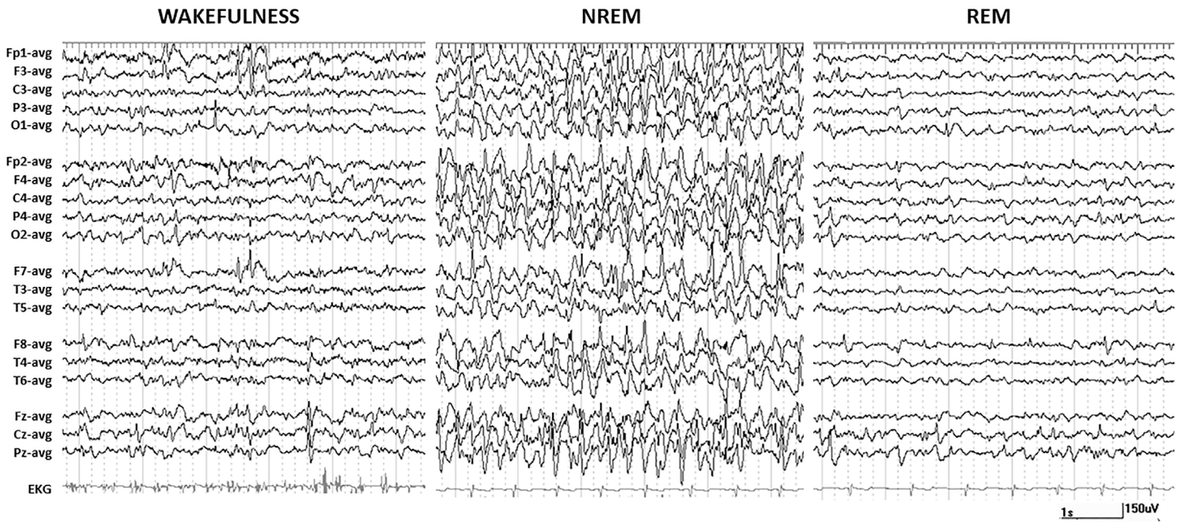
Johannesen, K. M., Nielsen, J., Sabers, A., Isidor, B., Kattentidt-Mouravieva, A. A., Zieglgänsberger, D., Heidlebaugh, A. R., Oetjens, K. F., Vidal, A. A., Christensen, J., Tiller, J., Freed, A. N., Møller, R. S., & Rubboli, G. (2023). The phenotypic presentation of adult individuals with SLC6A1-related neurodevelopmental disorders. Frontiers in neuroscience, 17, 1216653. https://doi.org/10.3389/fnins.2023.1216653

Johannesen, K. M., Pérez-Palma, E., & Rubboli, G. (2023). Editorial: SLC6A1: the past, present and future. Frontiers in neuroscience, 17, 1289821. https://doi.org/10.3389/fnins.2023.1289821

Kassabian, B., Fenger, C. D., Willems, M., Aledo-Serrano, A., Linnankivi, T., McDonnell, P. P., Lusk, L., Jepsen, B. S., Bayat, M., Kattentidt, A., Vidal, A. A., Valero-Lopez, G., Alarcon-Martinez, H., Goodspeed, K., van Slegtenhorst, M., Barakat, T. S., Møller, R. S., Johannesen, K. M., & Rubboli, G. (2023). Intrafamilial variability in SLC6A1-related neurodevelopmental disorders. Frontiers in neuroscience, 17, 1219262. https://doi.org/10.3389/fnins.2023.1219262

5.12 Developmental epileptic encephalopathy in DLG4-related synaptopathy

This study, in collaboration with Z. Tümer (University of Copenhagen) defined the phenotype of DLG4-related synaptopathy, demonstrating that a subgroup of individuals with DLG4-related synaptopathy has DEE, and around one fourth of them have ESES/DEE-SWAS, confirming DEE as part of the DLG4-related phenotypic spectrum.



Kassabian B, Levy AM, Gardella E, Aledo-Serrano A, Ananth AL, Brea-Fernández AJ, Caumes R, Chatron N, Dainelli A, De Wachter M, Denommé-Pichon AS, Dye TJ, Fazzi E, Felt R, Fernández-Jaén A, Fernández-Prieto M, Gantz E, Gasperowicz P, Gil-Nagel A, Gómez-Andrés D, Greiner HM, Guerrini R, Haanpää MK, Helin M, Hoyer J, Hurst ACE, Kallish S, Karkare SN, Khan A, Kleinendorst L, Koch J, Kothare SV, Koudijs SV, Lagae L, Lakeman P, Leppig KA, Lesca G, Lopergolo D, Lusk L, Mackenzie A, Mei D, **Møller RS**, Pereira EM, Platzer K, Quelin C, Revah-Politi A, Rheims S, Rodríguez-Palmero A, **Rossi A**, Santorelli F, Seinfeld S, Sell E, Stephenson D, Szczaluba K, Trinka E, Umair M, Van Esch H, van Haelst MM, Veenma DCM, Weber S, Weckhuysen S, Zacher P, Tümer Z, **Rubboli G**. Developmental epileptic encephalopathy in DLG4-related synaptopathy. Epilepsia. 2023 Dec 22. doi: 10.1111/epi.17876.

5.13 Gain-of-function and loss-of-function variants in GRIA3 lead to distinct neurodevelopmental phenotypes

In an exciting advancement, we explored the impact of rare variants in AMPA receptors on neurodevelopmental disorders (NDD). Specifically, we investigated 43 missense and one frameshift variant in the GRIA3 gene, which encodes AMPA receptor subunits, unique on the X chromosome. Electrophysiological on oocytes and HEK cell assays revealed that 31 variants significantly altered receptor function, leading to loss-of-function (LoF) or gain-of-function (GoF) properties. By merging clinical data with functional results, we identified clinical biomarkers that could preduct if a GRIA3 variant could cause LoF or GoF.

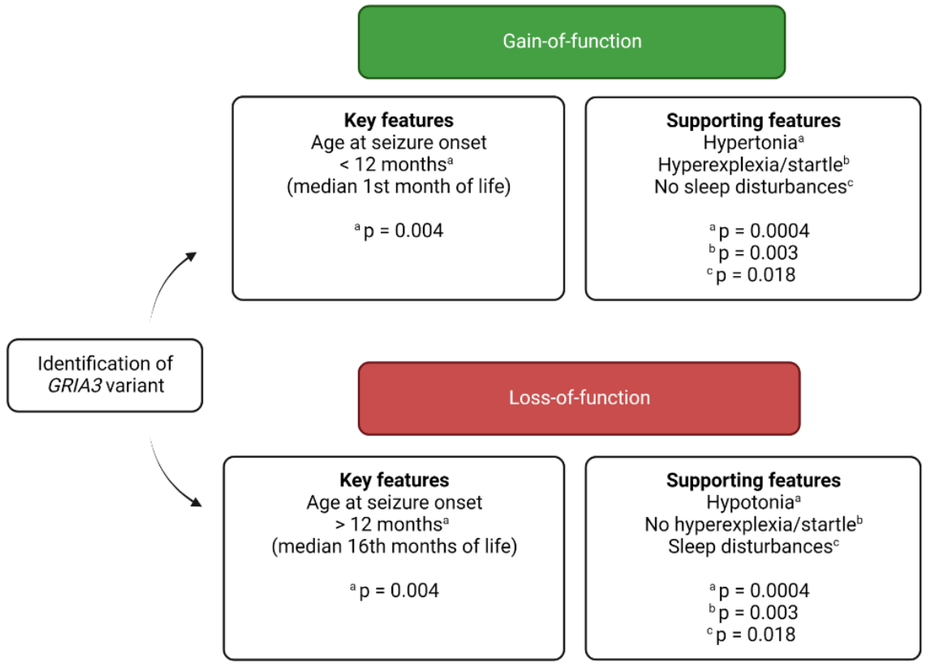


Fig. GRIA3 missense variants with gain-of-function and loss-of-function effects on AMPA receptors function lead to distinct clinical phenotype

Papers

Rinaldi B, **Bayat A**, Zachariassen LG, Sun JH, Ge YH, Zhao D, Bonde K, Madsen LH, Awad IAA, Bagiran D, Sbeih A, Shah SM, El-Sayed S, Lyngby SM, Pedersen MG, Stenum-Berg C, Walker LC, Krey I, Delahaye-Duriez A, Emrick LT, Sully K, Murali CN, Burrage LC, Plaud Gonzalez JA, Parnes M, Friedman J, Isidor B, Lefranc J, Redon S, Heron D, Mignot C, Keren B, Fradin M, Dubourg C, Mercier S, Besnard T, Cogne B, Deb W, Rivier C, Milani D, Bedeschi MF, Di Napoli C, Grilli F, Marchisio P, Koudijs S, Veenma D, Argilli E, Lynch SA, Au PYB, Ayala Valenzuela FE, Brown C, Masser-Frye D, Jones M, Patron Romero L, Li WL, Thorpe E, Hecher L, Johannsen J, Denecke J, McNiven V, Szuto A, Wakeling E, Cruz V, Sency V, Wang H, Piard J, Kortüm F, Herget T, Bierhals T, Condell A, Zeev BB, Kaur S, Christodoulou J, Piton A, Zweier C, Kraus C, Micalizzi A, Trivisano M, Specchio N, Lesca G, **Møller RS**, Tümer Z, Musgaard M, Gerard B, Lemke JR, Shi YS, Kristensen AS. Gain-of-function and loss-of-function variants in GRIA3 lead to distinct neurodevelopmental phenotypes. Brain. 2023 Dec 1:awad403. doi: 10.1093/brain/awad403.

5.14 Phenotypic and functional assessment of two novel KCNQ2 gain-of-function variants Y141N and G239S and effects of amitriptyline treatment

This study investigates the clinical features of patients with de novo KCNQ2 Y141N or G239S variants, revealing mild global developmental delay with language deficits but absence of neonatal seizures. Functional testing indicated a gain-of-function effect, supported by increased current densities and hyperpolarizing shifts in activation gating. Treatment with the Kv7 channel blocker amitriptyline showed promising results in improving motor, verbal, social, sensory, and adaptive behavior skills in one patient, suggesting it as a potential targeted treatment for KCNQ2 gain-of-function variants.

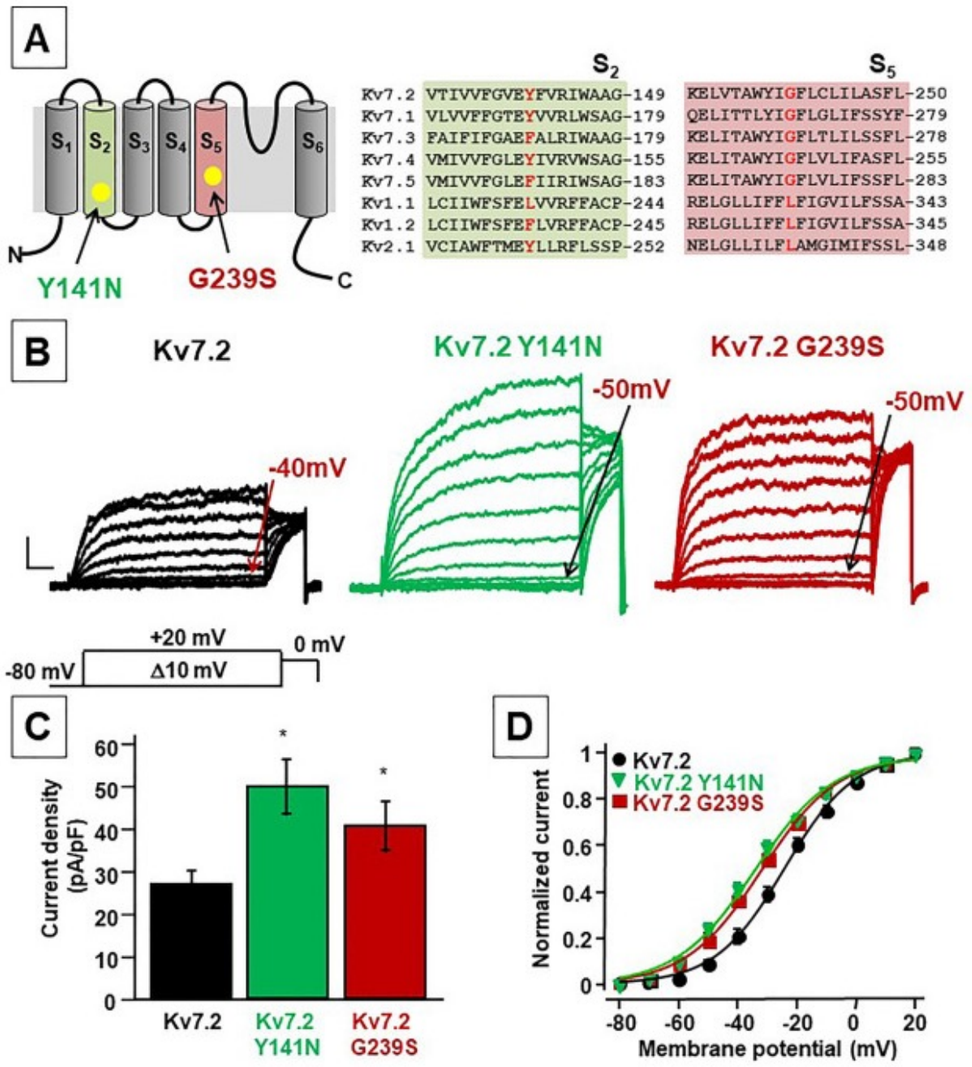


Fig. Schematic representation of a single Kv7.2 subunit, alignment and functional analysis of mutations at position Y141 and G239.

Papers

Bayat, A., Iavarone, S., Miceli, F., Jakobsen, A. V., Johannesen, K. M., **Nikanorova, M.**, Ploski, R., Szymanska, K., Flamini, R., Cooper, E. C., Weckhuysen, S., Taglialatela, M., & Møller, R. S. (2024). Phenotypic and functional assessment of two novel KCNQ2 gain-of-function variants Y141N and G239S and effects of amitriptyline treatment. Neurotherapeutics : the journal of the American Society for Experimental NeuroTherapeutics, 21(1), e00296. https://doi.org/10.1016/j.neurot.2023.10.006

6. Lectures - oral presentations in 2023

Sándor Beniczky:

- Automated interictal and ictal EEG source imaging.
  Winter conference on brain research. January 20-25 2023, Snowbird, Utah, USA.
- Non-convulsive status epilepticus.
  10th NATIONAL EEG WORKSHOP UNDER THE AGEIS OF INDIAN EPILEPSY SOCIETY 2023. Online. February 22, 2023.
- Automated seizure detection using wearables: are we there yet?
  Automated interpretation of clinical EEG using AI.
  International Conference on Artificial Intelligence in Epilepsy and Neurological Disorders. March 7-10 2023. Breckenridge, CO, USA.
- New advances in Clinical Neurophysiology.
  Presidential symposium at the Annual Meeting of the Epilepsy Societies of Austria, Germany and Switzerland. March 16, Berlin, Germany.
- Science of EEG Source Localization- Understanding the Value of Electrical Dipoles.
  Annual Meeting of the American Clinical Neurophysiology Society (ACNS). March 25-29, 2023, Austin, Texas, USA•
- Automated seizure detection using wearables: state-of-the-art.
  First Danish-Brazilian workshop on automated seizure detection. April 1 2023, Florianopolis, Santa Catarina, Brazil.
- Epileptiform and Non-epileptiform Abnormalities on Routine EEG.
  The 75th Annual Meeting of the American Academy of Neurology. April 22-27, 2023, Boston, Massachusetts, USA.
- Paroxysmal non-epileptic events.
  EEG and semiology.
  Interictal EEG abnormalities.
  Annual workshop of the Irish Society of Clinical Neurophysiology. April 28 2023, Dublin, Ireland.
- How to find lesion in non-lesional focal epilepsies, using EEG source imaging
  European Congress of Clinical Neurophysiology. May 9-12, Marseille, France.
- What is a Spike and does it harm the Brain?
  Colloquium Update on Rare Neurological Diseases. May 12, 2023, Salzburg, Austria.
- Machine learning and future of EEG.
  Annual meeting of the Turkish Epilepsy Society. May 26-28, 2023 Sapanca, Turkey.
- Behavioral testing during and after seizures.
  EpiCare: Nurses & EEG technicians workshop on best practices in Epilepsy Monitoring Units. May 31, 2023. Utrecht, the Netherlands.
- When is a spike a spike and a seizure a seizure?
  Online Epilepsy Seminars of the George Washington University, Washington DC, USA. June 13, 2023.
- Seizure detection using wearable devices.
  Online Grand Rounds of the New Your University, NY, USA. June 14, 2023.
- Using digital solutions to transform the delivery of clinical trials and research.
  The 9th European Academy of Neurology congress. July 1-4, Budapest, Hungary.
- Seizure detection using wearable devices.
  Systematic EEG reading.
  EEG source imaging.
  35th International Epilepsy Congress, September 2-6, 2023, Dublin, Ireland.
- Challenges to unify seizure classification and semiology terminology.
  15th International Epilepsy Colloquium. September 19-22. Cleveland, Ohio, USA.
- Artificial Intelligence and Epilepsy.
  McMaster Pediatric Neurology Rounds. September 22, 2023, online.
- Focal and generalized seizures: electroclinical correlations.
  Epilepsy Symposium at Karolinska University. September 29, Stockholm, Sweden.
- Digital technology in epilepsy trials.
  EpiCare webinar. October 5, 2023, online.
- Automated and semi-automated interpretation of EEG.
  4th International Congress on Mobile Health and Digital Technology in Epilepsy. October 11-13, 2023, Lausanne, Switzerland.
- Wearables and seizure counting: are we there yet?
  XXVI World Congress of Neurology (WCN 2023). October 15-19, 2023. Montreal, Canada.
- Wearable and seizure counting: Are we there yet?
  Lundbeck Science Forum Neurology. November 16-17, 2023. London, UK.
- Nonconvulsive status epilepticus in critically ill patients should not be treated aggressively.
  Annual Meeting of the American Epilepsy Society. December 1-5, 2023. Orlando, Florida, USA.

Sándor Beniczky:

- ILAE Residential Comprehensive Epilepsy Surgery Course: 11th EPODES (online, January 25, 2022)
  Scalp video-EEG monitoring in adults
- American Clinical Neurophysiology Society Annual meeting (Orlando, January 30 2022).
  Quantitative Analysis of Surface EMG in Functional Seizures
- Annual Meeting of the European Reference Network for epilepsy: EpiCare (Lyon, February 17, 2022)
  Update on the Clinical Neurophysiology work-package
- 16th World Congress on Controversies in Neurology (CONY) (online, March 26, 2022)
  Do seizure detection devices have a significant role in managing people with epilepsy?
- The Triennial Course of the British Society of Clinical Neurophysiology (Oxford, March 28, 2022)
  Identifying interictal epileptiform EEG discharges
- Clinical and Scientific Meeting of the Irish Society of Clinical Neurophysiology (Dublin, April 1, 2022)
  EEG in Critically Ill Patients
- Fourth International Taiwanese Congress of Neurology (Taiwan, April 15, 2022)
  EEG source image in clinical practice
- 63rd Annual Meeting of the Japanese Society of Neurology (Tokyo, May 18, 22022)
  Automated seizure detection using wearable device
- Electroencephalography Teaching Course (Santander, May 26-27, 2022)
  EEG in status epilepticus
  EEG in critically ill patients
- BrainDrugs Annual Meeting (Snekkersten, June 10, 2022)
  Epipick: a web-based algorithm for optimal, patient-tailored choice of antiseizure medication
- ICTALS 2022 – The tipping point (Bern, July 8, 2022)
  Devices for epilepsy

Rikke Steensbjerre Møller:

- Genetics of early onset epilepsies, Rome workshop (EpiCare, In search of lost time, Rome, Italy, December, 2024
- GABAA-receptor related disorders, ERN-ITHACA meeting, Dublin, Ireland, December, 2024
- Genotyping/phenotyping clinical interpretation, NorEpiNet course: from genes to treatment, Stockholm, November, 2023
- Epilepsy genetics, NorEpiNet course: from genes to treatment, Stockholm, November, 2023
- Drug repurposing and targeted treatments, 5th Annual Meeting, Danish Epilepsy Center & Norwegian Epilepsy Center, Oslo, October, 2023
- Personlig medicin, neuropædiatermøde, Sorø, Oktober, 2023
- Treatment responsiveness in LOF and GOF GABA-A receptor related epilepsies, 2nd Channelopathy Meeting, Tübingen, September, 2023
- Pathogenic GABRA3 variants lead to dominant and recessive X-linked disorders depending on functional outcome, 2nd Channelopathy Meeting, Tübingen, September, 2023
- How to streamline up-to-date diagnostic processes in epilepsy when suspecting a rare or complex epilepsy: the ERN EpiCARE approach, 35th International Epilepsy Congress, Dublin, September, 2023
- Utility of Genetic Testing on Therapeutic Decision-making in Individuals with Epilepsy, EPNS2023, Prague, June, 2023
- The role of genetic testing in rare epilepsy disorders, Orion satellite symposium, EPNS2023, Prague, June, 2023
- Impact of Genetic Testing on Therapeutic Decision-making in Individuals with Epilepsy, Bonner Epilepsie Seminar, Bonn, June, 2023
- Utility of Genetic Testing on Therapeutic Decision-making in Individuals with Epilepsy, Nordic Epilepsy Academy (webinar), June, 2023
- SCN2A/SCN8A Mixed Functions: Recent Discoveries, Underlying Mechanisms, Trends in Therapy, SCN2A/SCN8A Conference and Family Gathering, Køge, May, 2023
- Garden variety genetic epileptic encephalopathies: treatment snapshots, EpiPed course, Girona, April, 2023
- Utility of Genetic Testing for Therapeutic Decision-making in Individuals with Epilepsy, BRIDGE, SDU, April, 2023
- Utility of Genetic Testing for Therapeutic Decision-making in Individuals with Epilepsy, Personalized Medicine Research Cluster - Match Making Day, Copenhagen University, April, 2023
- Utility of Genetic Testing for Therapeutic Decision-making in Individuals with Epilepsy, McMaster/Western Combined Pediatric Neurology Rounds (webinar), March, 2023
- Genetisk udredning ved epilepsi – fra genetisk testning til personlig medicin?, Yngre genetikeres fyraftensmøde, Klinisk Genetisk Afdeling, Rigshospitalet, March, 2023
- Genetisk udredning af børn og voksne med epilepsi – hvem, hvornår og hvorfor? Epilepsi fra svært til lært, Middelfart, February, 2023
- Current practice in diagnostic genetic testing of the epilepsies, Ology Neurodiem: Expert Talks presentation, February 2023

Guido Rubboli:

- "Transition of care per le persone con epilessia - Il punto di vista dell'epilettologo dell'adulto" at the meeting Update in Epilessia, Padova (Italy), Febr 10th, 2023.
- "Encephalopathy related to status epilepticus during slow sleep, Italian League against Epilepsy, Feb 14th, 2023, (webinar)
- Lecture on "Future treatment trends within epilepsy: the path towards precision medicine" Epilepsi Symposium 2023, Copenhagen (Denmark), March 8th, 2023
- "Epilepsy genetics and the path toward precision medicine" at the Grand Rounds, Neurology Department, Northwestern University, Chicago (USA), March 16th, 2023 (webinar)
- "Ion-channel genes in epilepsy: implications in heart electrophysiology" at the 1st Danish- Brazilian Workshop on wearable healthcare technologies for the central nervous system Florianopolis (Brazil) April 1st, 2023.
- "Epilepsy in neuro-degenerative disease", UCB webinar, May 23rd, 2023
- "Genetics in epilepsy. When and who to test?" at the Norsk Epilepsiselskap Årskonferanse, Oslo (Norway), 8th June, 2023.
- Chairman at the Symposium on "Personalized medicine for the diagnosis and care of patients with complex epilepsy syndromes", EAN Budapest (Hungary), July 4th, 2023
- "Personalized treatment in rare epilepsies: What we have and what is the horizon? at the Symposium "Personalized medicine for the diagnosis and care of patients with complex epilepsy syndromes", EAN Budapest (Hungary) July 4th, 2023
- Chair at the Half-Day Teaching Course on "Genetic testing: whom, when and what to test", IEC ILAE, Sept 1st, Dublin (Ireland).
- "Yields of genetic testing in clinical practice – a path toward precision medicine" at the Half-Day Teaching Course on "Genetic testing: whom, when and what to test", IEC ILAE, Dublin (Ireland), Sep 1st, 2023
- "Generalized seizures –adults", Videosession at the IEC ILAE, Dublin (Ireland), Sept 5th, 2023
- "Le Crisi non epilettiche, at the XIII Corso di aggiornamento- non solo Parkinson. I disturbi neurologici funzionali. La terapia del Parkinson oggi e domani" Reggio Emilia (Italy), September 22nd, 2023
- "Chirurgia dell'epilessia in Regione Emilia Romagna – l'esperienza di un centro inegrato Dianalund-Copenhagen", Bologna (Italy), september 22nd, 2023
- "The EEG background interpretation in the pediatric epilepsy: variations from the norm and significance", EPIPED EEG Course on EEG interpretation in pediatric epilepsies, Bologna (Italy), October 3rd, 2023 (webinar)
- "Advances in rare epilepsies – Highlights of the EAN Congress 2023", EAN Webinar October 4th, 2023
- Chairman and organizer of the 5th Annual Meeting Danish Epilepsy Center & Norwegian Epilepsy Center "Advances in management of complex epilepsies", Oslo (Noorway), October 12th 2023
- "Ultra-long EEG monitoring. Yield and challenges" at the 5th Annual Meeting Danish Epilepsy Center & Norwegian Epilepsy Center, Oslo (Norway), October the 12th, 2023.
- "Monitoraggio neurofisiologico: quando e come? Epilessia" 53° Congress of the Italian Society of Neurology, Napoli (Italy), October 23rd, 2023
- Lecture on "Cenobamate – the Dianalund experience " Webinar, Angelini, October 25th 2023
- Chairman at the EpiCARE meeting "In search of lost time – 4. Epilepsy Care Challenges from Neonates to Adults. Rome (Italy), December 14th, 2023

Elena Gardella:

- Speaker/panel, STXBP1 summit- Lundbeck Foudation project, annual meeting, Copenhagen, Denmark, 20.01.23
- "Natural history studies - why harmonizing registry variables are key task for NorEpiNet", NorEpiNet meeting, annual meeting, Goteborg, Sweden, 03.02.23
- "Illustrative cases of movement disorder in complex epilepsy syndromes", ERN-EpiCare - Pediatric Movement disorders, webinar, 08.02.23
- "One gene with various epilepsy and epilepsy syndromes phenotypes: the example of SCN8A related epilepsy", Séminaire Pierre Royer, Paris, France, 16.03.23
- "Garden variety genetic epileptic encephalopathies: treatment snapshots", 3rd Girona EpiPed course (speaker/trainer), Girona, Spain, 26-29.04.23
- "From genetic testing to the phenotype", 10th International Residential Course on Drug Resistant Epilepsies, course (speaker/trainer), Tagliacozzo, Italy, 01-03.05.23
- "The contribute of EEG analysis to deep phenotyping of STXBP1-DEE", STXBP1 summit & family gathering, Milan, Italy, 17-18.05.23
- "SCN8A phenotypes and long term evolution", SCN8A/SCN2A Conference and Family Gathering,, Køge, Denmark, 26-27.05.23
- "Clinical characteristics of the main types of DEEs", 1st Advanced Course on the Pharmacological Treatment of Drug Resistant Epilepsies, Mallorca, Spain, 30-31.05.23
- "Genotyping / phenotyping clinical interpretation", Master University of Genoa, Italy, webinar 08.07.23

- “Basic EEG course”, 35th International Epilepsy Congress, Dublin, Ireland, 02-06.09.23
- “SCN8A: Clinical complexity and natural history study”, 2nd Channelopathy Meeting, Tubingen, Germany, 04-06.10.23
- “Genotyping / phenotyping clinical interpretation”, NorEpinet from genes to treatment, course (speaker/trainer), Stockholm, Sweden, 20-21.11.23
- “From the electro-clinical presentation to genetic diagnosis of monogenic epilepsies”, Romanian Epilepsy Society, annual meeting, Bucharest, Romania, 23-25.11.23
- “The genetics of focal cortical dysplasia and other malformations of cortical development”, Romanian Epilepsy Society, annual meeting, Bucharest, Romania, 23-25.11.23
- Chairman, EpiCare meeting- In search of lost time 4, Rome, Italy, 13-15.12.23

Marina Nikanorova:

- Rational approach to children with drug-resistant epilepsy – Fra svært til lært, Middelfart, Denmark, 3rd February 2023
- Neonatal seizures – webinar with Danish neuropædiatricians, 13th March, 2023
- Modern treatment of Dravet syndrome - Finnish Neuropediatric Society meeting, Turku, Finland, 23-24 March 2023
- Danish experience with Fenfluramine in treatment of Dravet syndrome – UCB meeting, Brussels, 25 April 2023
- How to extract the epilepsy syndrome diagnosis from seizure semiology –webinar with Danish, Norwegian and Finnish neuropediatricians, 23 May 2023
- 16th Baltic Sea summer school on epilepsy – 3 sessions, October 2023
- Non-epileptic paroxysmal conditions of childhood – Fra svært til lært, Middelfart, 27th October 2023
- Epileptic encephalopathies in childhood – Fra svært til lært, Middelfart, 27th October 2023
- Danish experience with Fenfluramin in treatment of Dravet syndrome – Danish-Norwegian meeting, Oslo, November 2023

Allan Bayat:

- “Gain-of-function and loss-of-function GRIA3 variants lead to distinct neurodevelopmental phenotypes”. Neuroscience academy of Denmark, NAD symposium, Panum. University of Copenhagen, Denmark.
- “Epilepsy genetics and personalized medicine”. Clinical Genetic Dept., Aarhus University Hospital, Denmark
- “GRIA3 related disorders”. ILAE Conference, Dublin, Ireland.
- “Epilepsy genetics and personalized medicine”. Neurology conference 2023, Hvidovre University Hospital, Denmark
- “GRIA3 related disorders”. GRI Conference, Online webinar.
- “Dravet syndrome and future perspectives”. Exploring connections, 2023. Online webinar https://www.exploring-connections.com/static/Video-hub.html
- “GRIA3 related disorders”. 2023, Leipzig, Germany.
- “Impact of genetic testing on therapeutic decision making in childhood-onset epilepsies”. Danish Epilepsy Society Conference, 2023, Denmark

Francesca Furia:

- “STXBP1 deep phenotyping registry” STXBP1 summit- Lundbeck Foundation project, annual meeting, Copenhagen, Denmark, 20.01.23
- “The phenotype and genotype spectrum of individuals with ANK3 variants”, 9th LEGOLAS Meeting, Leipzig, Germany, 20-21.04.2023
- “ Launch of the European registry”, STXBP1 summit & family gathering, Milan, Italy, 17-18.05.23
- “ Sleep disorders in sodium channelopathies”, SCN8A/SCN2A Conference and Family Gathering, Køge, Denmark, 26-27.05.23
- “From large registries to natural history studies”, Advances in management of complex epilepsies - 5th Annual Meeting - Danish Epilepsy Center & Norwegian Epilepsy Center, Oslo, Norway, 12.10.23
- “ The Danish experience of a precision medicine approach to STXBP1 disorders”, NorEpinet from genes to treatment, course (speaker/trainer), Stockholm, Sweden, 20-21.11.23
- “Sleep disturbances in patients with SCN8A related disorders”, XXXIII national congress AIMS, Milan, Italy, 24-26 November 2023

7. Publication list in 2023

1. **Meritam Larsen P**, **Wüstenhagen S**, **Terney D**, **Gardella E**, Aurlien H, **Beniczky S**. Duration of epileptic seizure types: A data-driven approach. Epilepsia. 2023 Feb;64(2):469-478. doi: 10.1111/epi.17492.
2. Bayat M, **Beniczky S**, Thomsen JLS. Very late onset methylmalonic academia (cblB type) as a cause of status epilepticus, leukoencephalopathy and myelopathy. Neurol Sci. 2023 Dec doi: 10.1007/s10072-023-07270-1.
3. Hadady L, Sperling MR, Alcala-Zermeno JL, French JA, Dugan P, Jehi L, Fabó D, Klivényi P, **Rubboli G**, **Beniczky S**. Prediction tools and risk stratification in epilepsy surgery. Epilepsia. 2023 Dec 7. doi: 10.1111/epi.17851.
4. Frauscher B, Mansilla D, Abdallah C, Astner-Rohracher A, **Beniczky S**, Brazdil M, Gnatkovsky V, Jacobs J, Kalamangalam G, Perucca P, Ryvlin P, Schuele S, Tao J, Wang Y, Zijlmans GJM, McGonigal A. Learn how to interpret and use intracranial EEG findings. Epileptic Disord. 2023 Dec 20. doi: 10.1002/epd2.20190.
5. Nascimento FA, Katyal R, Olandoski M, Gao H, Yap S, Matthews R, Rampp S, Tatum W, Strowd R, **Beniczky S**. Expert accuracy and inter-rater agreement of ”must-know” EEG findings for adult and child neurology residents. Epileptic Disord. 2023 Nov 30. doi: 10.1002/epd2.20186.
6. Knight A, Gschwind T, Galer P, Worrell GA, Litt B, Soltesz I, **Beniczky S**. Artificial intelligence in epilepsy phenotyping. Epilepsia. 2023 Nov 20. doi: 10.1111/epi.17833.
7. Westin K, **Beniczky S**, Pfeiffer C, Hämäläinen M, Lundqvist D. On the clinical utility of on-scalp MEG: A modeling study of epileptic activity source estimation. Clin Neurophysiol. 2023 Dec;156:143-155. doi: 10.1016/j.clinph.2023.10.006.
8. Greenblatt AS, **Beniczky S**, Nascimento FA. Pitfalls in scalp EEG: Current obstacles and future directions. Epilepsy Behav. 2023 Dec;149:109500. doi: 10.1016/j.yebeh.2023.109500.
9. **Beniczky S**, Ryvlin P. Mobile health and digital technology in epilepsy: The dawn of a new era. Epilepsia. 2023 Dec;64 Suppl 4:S1-S3. doi: 10.1111/epi.17813.
10. **Meritam Larsen P**, **Beniczky S**. Non-electroencephalogram-based seizure detection devices: State of the art and future perspectives. Epilepsy Behav. 2023 Nov;148:109486. doi: 10.1016/j.yebeh.2023.109486.
11. De Stefano P, Ménétré E, Stancu P, Mégevand P, Vargas MI, Kleinschmidt A, Vulliémoz S, Wiest R, **Beniczky S**, Picard F, Seeck M. Added value of advanced workup after the first seizure: A 7-year cohort study. Epilepsia. 2023 Dec;64(12):3246-3256. doi: 10.1111/epi.17771.
12. Zelano J, **Beniczky S**, Ryvlin P, Surges R, Tomson T; ILAE SUDEP Task Force. Report of the ILAE SUDEP Task Force on national recommendations and practices around the world regarding the use of wearable seizure detection devices: A global survey. Epilepsia Open. 2023 Dec;8(4):1271-1278. doi: 10.1002/epi4.12801.
13. **Beniczky S**. An interview with Mustafa Aykut Kural, the 2023 Epileptic Disorders prize winner. Epileptic Disord. 2023 Oct;25(5):797-798. doi: 10.1002/epd2.20094.
14. Nascimento FA, Salazar M, Colonetti J, Schomer D, **Beniczky S**. How to conduct EEG recordings-A video-based educational resource. Epileptic Disord. 2023 Dec;25(6):911-913. doi: 10.1002/epd2.20089.
15. Leitinger M, Gaspard N, Hirsch LJ, **Beniczky S**, Kaplan PW, Husari K, Trinka E. Diagnosing nonconvulsive status epilepticus: Defining electroencephalographic and clinical response to diagnostic intravenous antiseizure medication trials. Epilepsia. 2023 Sep;64(9):2351-2360. doi: 10.1111/epi.17694.
16. Tveit J, Aurlien H, Plis S, Calhoun VD, Tatum WO, Schomer DL, Arntsen V, Cox F, Fahoum F, Gallentine WB, **Gardella E**, Hahn CD, Husain AM, Kessler S, Kural MA, Nascimento FA, Tankisi H, Ulvin LB, Wennberg R, **Beniczky S**. Automated Interpretation of Clinical Electroencephalograms Using Artificial Intelligence. JAMA Neurol. 2023 Aug 1;80(8):805-812. doi: 10.1001/jamaneurol.2023.1645.
17. Aanestad E, Gilhus NE, Olberg HK, Kural MA, **Beniczky S**, Brogger J. Spike count and morphology in the classification of epileptiform discharges. Front Neurol. 2023 May 23;14:1165592. doi: 10.3389/fneur.2023.1165592.
18. 18. **Wüstenhagen S**, Juhl S, **Beniczky S**. Eyelid myoclonia versus eyelid fluttering. Epileptic Disord. 2023 Jun;25(3):441-444. doi: 10.1002/epd2.20067.
19. Jeppesen J, Christensen J, Johansen P, **Beniczky S**. Personalized seizure detection using logistic regression machine learning based on wearable ECG-monitoring device. Seizure. 2023 Apr;107:155-161. doi:10.1016/j.seizure.2023.04.012.
20. Jeppesen J, Christensen J, Mølgaard H, **Beniczky S**. Automated detection of focal seizures using subcutaneously implanted electrocardiographic device: A proof-of-concept study. Epilepsia. 2023 Dec;64 Suppl 4:S59-S64. doi: 10.1111/epi.17612.
21. Tamula G, Katyal R, **Beniczky S**, Nascimento FA. Focal cortical dysplasias: New advances for curing epilepsy. Epileptic Disord. 2023 Apr;25(2):284. doi: 10.1002/epd2.20013.
22. Nascimento FA, **Beniczky S**. Sawtooth waves: An EEG normal variant. Epileptic Disord. 2023 Feb;25(1):120-121. doi: 10.1002/epd2.20032.
23. Nascimento FA, Friedman D, Peters JM, Bensalem-Owen MK, Cendes F, Rampp S, Wirrell E, Blümcke I, Tatum W, **Beniczky S**. Focal epilepsies: Update on diagnosis and classification. Epileptic Disord. 2023 Feb;25(1):1-17. doi: 10.1002/epd2.20045.
24. Gogou M, Sheikh IS, Tamula ORM 3rd, Katyal R, **Beniczky S**, Nascimento FA. Competency-based epilepsy training: A comparison between U.S.-based milestones and ILAE curriculum. Epileptic Disord. 2023 Aug;25(4):586-589. doi: 10.1002/epd2.20037.
25. Peltola ME, Leitinger M, Halford JJ, Vinayan KP, Kobayashi K, Pressler RM, Mindruta I, Mayor LC, Lauronen L, **Beniczky S**. Routine and sleep EEG: Minimum recording standards of the International Federation of Clinical Neurophysiology and the International League Against Epilepsy. Dual publication in: Clin Neurophysiol. 2023 Mar;147:108-120. doi: 10.1016/j.clinph.2023.01.002. Epilepsia. 2023 Mar;64(3):602-618. doi: 10.1111/epi.17448.
26. Halford JJ, Brinkmann BH, Clunie DA, Gotman J, **Beniczky S**, Rampp S, Rémi J, Husain A, Andrew Ehrenberg J, Winkler S. Continued progress in DICOM neurophysiology standardization. Clin Neurophysiol. 2023 Mar;147:11-13. doi: 10.1016/j.clinph.2022.12.008.
27. Nascimento FA, **Beniczky S**. Teaching the 6 Criteria of the International Federation of Clinical Neurophysiology for Defining Interictal Epileptiform Discharges on EEG Using a Visual Graphic. Neurology® Education 2023;2:e200073. doi:10.1212/NE9.0000000000200073
28. Nascimento FA, Gao H, Katyal R, Matthews R, Yap SV, Rampp S, Tatum WO, Strowd RE, **Beniczky S**. Education Research: Competency-Based EEG Education. An Online Routine EEG Examination for Adult and Child Neurology Residents. Neurology® Education 2023;2:e200094. doi:10.1212/NE9.0000000000200094

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31. Nascimento FA, Katyal R, Matthews R, Yap SV, Rampp S, Tatum WO, Strowd RE, **Beniczky S**. Education Research: Competency-Based EEG Education. An Online Routine EEG Examination for Adult and Child Neurology Residents. Neurology® Education. 2023; 2(4). https://doi.org/10.1212/NE9.0000000000200094
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33. Rong M, Benke T, Zulfiqar Ali Q, Aledo-Serrano Á, **Bayat A**, **Rossi A**, Devinsky O, Qaiser F, Ali AS, Fasano A, Bassett AS, Andrade DM. Adult Phenotype of SYNGAP1-DEE. Neurol Genet. 2023 Nov 17;9(6):e200105. doi: 10.1212/NXG.0000000000200105. eCollection 2023 Dec.
34. **Johannesen KM**, Tümer Z, Weckhuysen S, Barakat TS, **Bayat A**. Solving the unsolved genetic epilepsies: Current and future perspectives. Epilepsia. 2023 Dec;64(12):3143-3154. doi: 10.1111/epi.17780. Epub 2023 Oct 17.
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