FILADELFIA

ANNUAL RESEARCH REPORT 2022

Danish Epilepsy Centre Filadelfia

Content

*) Papers published in peer-reviewed journals, including leading journals, such as Nature Communications, Brain, Neurology and Epilepsia.

1. Introduction – Filadelfia

Filadelfia - The Danish Epilepsy Centre - offers highly specialized health care services to epilepsy patients of all ages. Being the only tertiary referral center for comprehensive treatment of epilepsy in Denmark, we have a unique access to rare and complex epileptic syndromes on the basis of which our centre is internationally acclaimed. In 2022 Filadelfia's three professors, two associate professors, three and ten Ph.D. students published 111 papers in international and national journals and contributed with 74 oral presentations. Our researchers are affiliated to University of Copenhagen, University of Odense and University of Aarhus. They occupy positions of trust in national and international scientific societies, and participate in a series of international epilepsy research networks.

The Danish Epilepsy Center, Filadelfia is a non-profit foundation with an independent Board of Directors. In 2022 we celebrated Filadelfias 125 years anniversary. Besides 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 publically 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 wish it to encourage further collaboration for the benefit of persons with epilepsy worldwide.

Sándor Beniczky
Professor, Head of Department

Mads Ravnborg
Medical director

2. Core Research Team 2022

Sándor Beniczky MD, Ph.D.
Professor, Head of Department
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- Editor-in-Chief, Epileptic Disorders
- 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)
- 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

Rikke Steensbjerre Møller, Ph.D.
Professor, Head of department
Email: rimo@filadelfia.dk

- 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 leadership team at Department of Regional Health Research, University of Southern Denmark, Odense, Denmark
- Member of the scientific committee of Residras; a European Registry of Dravet Syndrome

Guido Rubboli MD, Ph.D.
Professor, Senior Consultant
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- 2022- present: European Academy of Neurology, member of the Management Group of the Scientific Panel on Epilepsy,
- 2018 - present: ILAE Task Force “Transition in care from Childhood into Adulthood”, Member,
- 2022 - present: ILAE Task Force “SNOMED-CT”, Member,
- 2019 - present: “Epilessia- Fuori dall’ombra”, scientific committee
- 2020 - present: KCNT1 Foundation, member of the scientific committee
- 2021 - present: KCNA2 Foundation, member of the scientific committee
- 2021 - present: VIREPA (Virtual Epilepsy Academy) ILAE, Advanced EEG Course, Director

Elena Gardella MD, Ph.D.
Associate Professor, Senior Consultant
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- Member of the commission on Classification and Terminology of the International League against Epilepsy (ILAE)
- Director of VIREPA basic-EEG course
- 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.
- Member of the BRIDGE team at Department of Regional Health Research, University of Southern Denmark
- Editor of Scientific advisory board member: SCN8A Italy Foundation
- Scientific advisory board member: SCL6A1 Foundation

Marina Nikanorova MD, Ph.D.
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Allan Bayat MD, Ph.D.
Translational Researcher, Consultant
Email: abay@filadelfia.dk

- Post doctoral BRIDGE Translational researcher at University of Copenhagen in collaboration with Department of Clinical Genetics, Rigshospitalet and the Danish Epilepsy Center.

Research Team

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Stephan Wüstenhagen, MD, PhD
Pirgit Meritam Larsen, MD, PhD
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Maria Vlachou, MD
Levente Hadady, MD
Orsolya Györfi, MD, PhD
Pietro Mattioli, MD
Evy Cleeren, PhD
Aykut Kural MD, PhD
Trine Hammer, MD, PhD
Christina Fenger, PhD
Allan Bayat, MD, PhD
Anne Højte Hansen, study nurse
Nazanin Mohammad, MSc

Sabrina Neri, MD
Cristina Cioclu, MD
Margherita Aluffi Valletti (Erasmus+ mobility)
Angelica Pisati (Erasmus+ mobility)
Alessandra Rossi, MD
Roberto Previtali, MD
Alberto Cossu, MD
Morad Kamand, PhD
Francesca Furia, MD
Cathrine Gjerulfsen, MD
Benedetta Kassabian, MD
Catarina Ancora, MD
Katrine M Johannesen, MD, PhD
Carolina Alvarez, MD

3. Ph.D. projects

Completed in 2022

- Allan Bayat: Deep phenotyping, genotype-phenotype correlations and precision medicine in monogenic epilepsies. Faculty of Health Sciences, University of Southern Denmark. Main supervisor: Rikke Steensbjerre Møller, co-supervisors: Guido Rubboli; Elena Gardella.
- Bogdan Florea: Electroencephalography in patients with disturbed level of consciousness. University of Szeged, Hungary. Main supervisor: Sándor Beniczky. Co-supervisor: Péter Klivényi.
- Karin Westin: Extending the clinical applications of magnetoencephalography. Karolinska Institute, Stockholm. Co-supervisor: Sándor Beniczky. Main supervisor: Daniel Lundquist.

Ongoing

- Maria Vlachou: Evaluation of electro-clinical findings using standardised feature extraction and machine learning. 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.
- Nazanin Azarinejad Mohammadi, M.Sc., Clinical and functional characterization of GABAA-receptor related disorders: translating genetic diagnostics into personalized treatment. Faculty of Health Sciences, University of Southern Denmark. Main supervisor: Rikke Steensbjerre Møller, co-supervisor: Philip Ahring.
- Marie Amanda Bust Levy, MSc. Genetic and Functional Mechanisms in Neurodevelopmental Disorders and Epilepsy. Faculty of Health Sciences, University of Copenhagen. Main supervisor: Zeynep Tümer, co-supervisor: Rikke Steensbjerre Møller.
- Tanya Ramdal Techlo, MSc. Leverage polygenic approaches to genetically diagnose idiopathic severe epilepsy and hemiplegic migraine. Faculty of Health Sciences, University of Copenhagen. Main supervisor: Thomas Folkmann Hansen, co-supervisor: Rikke Steensbjerre Møller
- Francesca Furia, MD. Deep phenotyping of monogenic epilepsies towards the identification of targeted treatments. Faculty of Health Sciences, University of Southern Denmark. Main supervisor: Elena Gardella, co-supervisors: Rikke Steensbjerre Møller, Guido Rubboli
- Frederik Nørby Friis Sørensen, MSc. Dissecting neuronal heterogeneity and epileptogenesis in focal cortical dysplasia. Faculty of Health Sciences, University of Copenhagen. Main supervisor: Konstantin Khodosevich, co-supervisor: Rikke Steensbjerre Møller.

DICE 2022

4. Conferences and Summer School

4.1 DICE 2022 - Implementing Epilepsy Genetical In Clinical Practice

Korsør, 7-8 April 2022
Genetic testing has increasingly become an essential part of clinical practice in epilepsy to elucidate the pathogenetic role of genetic variants in different epileptic conditions, and to provide prognostic information and to guide possible targeted therapies.

At the 5th DICE, an international panel of experts have discussed the yields of genetic testing in epilepsy patients and how the use of this diagnostic tool can lead to both better management of the epilepsies and how genetics can contribute to develop novel treatment approaches. In addition, transition from pediatric to adult care, the methodology necessary to perform clinical studies in rare conditions and the current evidences suggesting that genetics can have an impact upon both selection of epilepsy surgery candidates and post-surgical prognoses have been also discussed. Finally combined endeavors of patient associations, international epilepsy networks and registries have been also debated. The Conference was complemented by case-presentations from the audience.

An audience of more than 160 people from all over the world attended the conference, participating to the lively discussions, contributing to the success of this event, and ultimately, emphasizing how the collaboration of epileptologists, geneticists, and patient representatives can be instrumental in further improving epilepsy diagnoses and management.

DICE 2022

4.2 Dianalund Summer School on EEG and Epilepsy

5th edition, July 16-23, 2022
Under the auspices of the International League Against Epilepsy (ILAE), the International Federation of Clinical Neurophysiology and the Danish Epilepsy Society, we held the 5th edition of the Dianalund Summer School on EEG and Epilepsy. This biennial course addresses advanced learning objectives and it is an official core course of the ILAE.

The course was practically-oriented, with many hands-on sessions, and it was highly interactive. The main idea of the course is to bridge the gap between epileptologists and neurophysiologists. Therefore, topics both in the realm of signal analysis and topics related to seizure semiology and clinical significance of EEG findings were covered.

The theoretical presentations (in the morning) were supplemented by EEG-reading sessions (in the afternoon), where the participants were guided through video-EEG samples by the tutors. The last day of the course was dedicated to discussion of difficult cases brought by the participants.

Twenty-seven students from 16 countries attended the course in 2022. They gave a very positive evaluation of the course, using the online evaluation site of the ILAE: 92% of the students considered that the course exceeded their expectations.

Summer School 2022

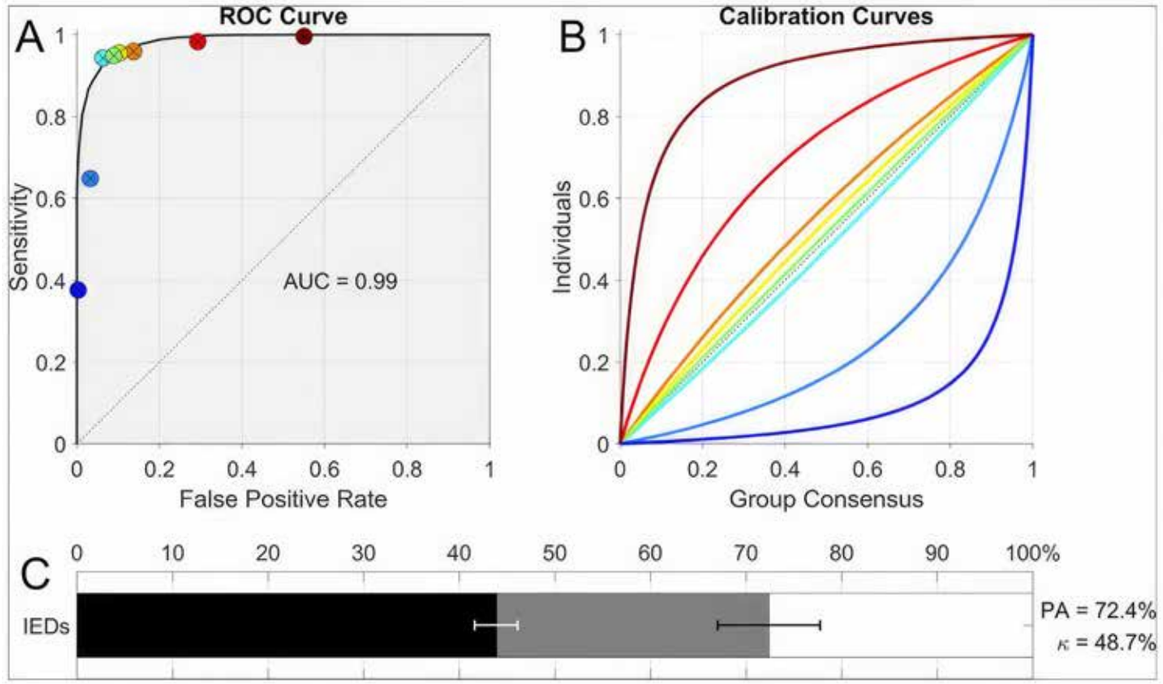
Summer School 2022

5. Research projects

5.1 Standardization and quality assurance in Clinical Neurophysiology

Standardization is essential for improving the quality of care. Clinical Practice Guidelines are important tools to achieve this goal. Guidelines must be developed using a robust methodology, based on systematic review of published evidence. Developing guidelines is resource demanding, yet much deeded for clinical practice. We were happy to contribute to the clinical practice guideline on standards for inpatient long-term video-electroencephalographic monitoring. The guideline has been endorsed both by the International League against Epilepsy and the International Federation of Clinical Neurophysiology
.
A significant technical impediment in clinical EEG is the lack of a universal data format. This makes shared-care and collaborative research difficult. Under the auspices of the International Federation of Clinical Neurophysiology, a working group developed a new, international standard for a universal data format, using DICOM. Our group contributed to this work.

Bias from clinical data (the text of the EEG referral) is detrimental for an objective interpretation of clinical EEG. We showed the discrepancy between what experts consider optimal, and their own clinical practice. We published a manifesto for changing the practice and improve the quality of clinical EEG interpretation.



A: Receiver operating characteristic curve fit to all experts' scores.
B: Parametric calibration curve fit to the binary scores of each expert.
C: Inter-rater reliability (IRR): Kappa (κ) values in relation to percent agreement

Papers

Tatum WO, Mani J, Jin K, Halford JJ, Gloss D, Fahoum F, Maillard L, Mothersill I, Beniczky S. Minimum standards for inpatient long-term video- electroencephalographic monitoring: A clinical practice guideline of the International League Against Epilepsy and International Federation of Clinical Neurophysiology. Epilepsia. 2022 Feb;63(2):290-315. doi: 10.1111/epi.16977.

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. 2022 Dec 28;147:11-13. doi: 10.1016/j.clinph.2022.12.008.

Nascimento FA, Jing J, Beniczky S, Olandoski M, Benbadis SR, Cole AJ, Westover MB. EEG reading with or without clinical information - a real-world practice study. Neurophysiol Clin. 2022 Oct;52(5):394-397. doi: 10.1016/j.neucli.2022.08.002

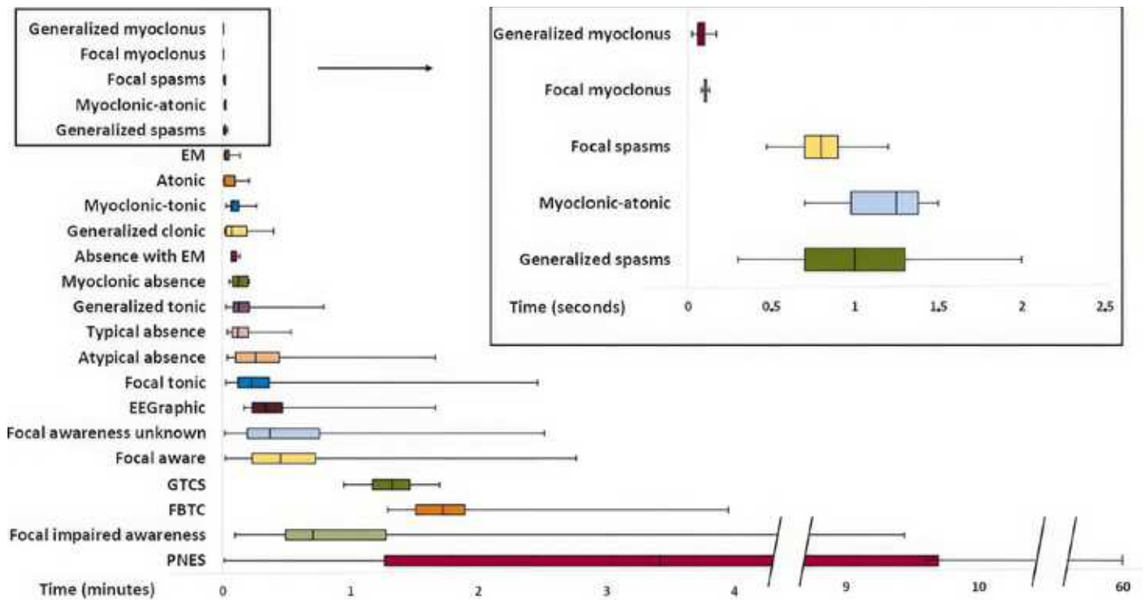
Nascimento FA, Jing J, Beniczky S, Benbadis SR, Gavvala JR, Yacubian EMT, Wiebe S, Rampp S, van Putten MJAM, Tripathi M, Cook MJ, Kaplan PW, Tatum WO, Trinka E, Cole AJ, Westover MB. One EEG, one read - A manifesto towards reducing interrater variability among experts. Clin Neurophysiol. 2022 Jan;133:68-70. doi: 10.1016/j.clinph.2021.10.007.

5.2 Standardized Computer-based Organized Reporting of EEG: SCORE

SCORE is a digital tool developed using the international standards we contributed to. The tool helps improving the quality of clinical EEG interpretation and in the same time facilitates research, by building a database. We helped colleagues in an underprivileged area of the world, to implement a free version of the SCORE software. In a prospective study, we showed that this contributed to improving the quality of patient care.

We extracted data form the anonymized clinical dataset, to investigate the prevalence of normal variants. We compiled comprehensive list and characterized the normal variants in our database. To make this useful for postgraduate training we included typical examples of each normal variant. This paper gives a realistic picture about the frequency of EEG normal variants, and it is a valuable resource for trainees.

From our large database, we extracted data on the duration of various seizure-types. Our data-driven approach resulted in accurate characterization of this important feature of the epileptic seizures. Our results are useful for accurate identification of the seizure types in clinical practice, and in setting time-limits for status epilepticus.



Duration of epileptic seizure types, extracted from a large SCORE dataset

Papers

Japaridze G, Kasradze S, Aurlien H, Beniczky S. Implementing the SCORE system improves the quality of clinical EEG reading. Clin Neurophysiol Pract. 2022 Sep 1;7:260-263. doi: 10.1016/j.cnp.2022.07.004

Wüstenhagen S, Terney D, Gardella E, Meritam Larsen P, Rømer C, Aurlien H, Beniczky S. EEG normal variants: A prospective study using the SCORE system. Clin Neurophysiol Pract. 2022 Jun 30;7:183-200. doi: 10.1016/j.cnp.2022.06.001.

Meritam Larsen P, Wüstenhagen S, Terney D, Gardella E, Aurlien H, Beniczky S. Duration of epileptic seizure types: A data-driven approach. Epilepsia. 2022 Dec 15. doi: 10.1111/epi.17492.

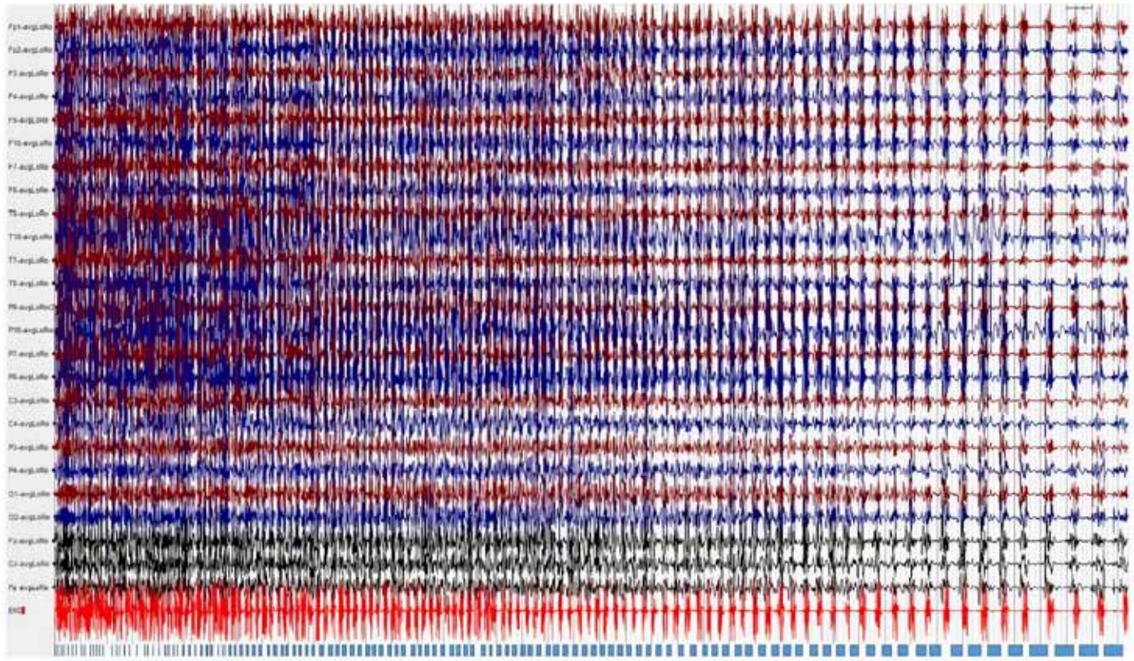
5.3 Electro-clinical phenomena in epileptic seizures

Accurate feature extraction and characterization of the EEG and clinical phenomena (semiology) observed in epileptic seizures is of paramount importance. In 2022 we published our studies on generalized seizures: absence seizures and generalized tonic-clonic seizures.

We found that semiological features, in addition to behavioral arrest and non-responsiveness, were common in typical absence seizures, but they did not predict long-term therapeutic outcome. Using machine learning, we found that the presence of polyspikes had a high positive predictive value for unfavorable therapeutic outcome, and their presence should therefore be included when reporting EEGs in patients with typical absence seizures.

Postictal generalized electroencephalography (EEG) suppression (PGES) is a surrogate marker of sudden unexpected death in epilepsy (SUDEP). In our large dataset, we demonstrated that progressive slowing of clonic phase (PSCP) in generalized tonic-clonic seizures, predicts prolonged PGES, emphasizing the importance of gradually increasing inhibitory phenomena at the end of the seizures. Our findings shed more light on the ictal phenomena leading to increased risk of SUDEP. These phenomena may provide basis for algorithms implemented into wearable devices for identifying GCS with increased risk of SUDEP.

We lead an international working group which, under the auspices of the International League Against Epilepsy revised the semiology glossary. This work was based on systematic search of the published literature. The paper includes video-examples of the various semiology phenomena, and is an important educational resource.



Progressive slowing of the clonic phase in generalized tonic-clonic seizures.

Papers

Vlachou M, Ryvlin P, Arbune AA, Armand Larsen S, Skraep Sidaros A, Cacic Hribljan M, Fabricius M, Beniczky S. Progressive slowing of clonic phase predicts postictal generalized EEG suppression. Epilepsia. 2022 Dec;63(12):3204-3211. doi: 10.1111/epi.17434. Epub 2022 Oct 29.

Vlachou M, Skrimpas GA, Kural MA, Rackauskaite G, Nikanorova N, Christensen J, Nikanorova M, Beniczky S. Electroclinical features and long-term therapeutic response in patients with typical absence seizures. Epileptic Disord. 2022 Apr 1;24(2):315-322. doi: 10.1684/epd.2021.1392.

Beniczky S, Tatum WO, Blumenfeld H, Stefan H, Mani J, Maillard L, Fahoum F, Vinayan KP, Mayor LC, Vlachou M, Margitta S, Ryvlin P, Philippe K. Seizure semiology: ILAE glossary of terms and their significance. Epileptic Disord. 2022 Jun 1;24(3):447-495. doi: 10.1684/epd.2022.1430. PMID: 35770761.

5.4 EEG Source Imaging

Using mathematical algorithms, the source of the EEG signal can be estimated in the brain. EEG Source Imaging (ESI) has been one of the research topics our group has been focusing on for more than a decade. In 2022 we continued this work, using ESI for presurgical evaluation of patients with drug-resistant focal epilepsy.

We investigated the performance of automated and semi-automated spike-detection, in relation to their localization accuracy. We found that these methods show significant agreement with visually detected spikes in the long-term recordings and concordance with the seizure onset zone. In short-term, high-density EEG, semi-automated detection of spikes is concordant with the visually detected ones, and the seizure-onset zone if high spike-counts were detected.

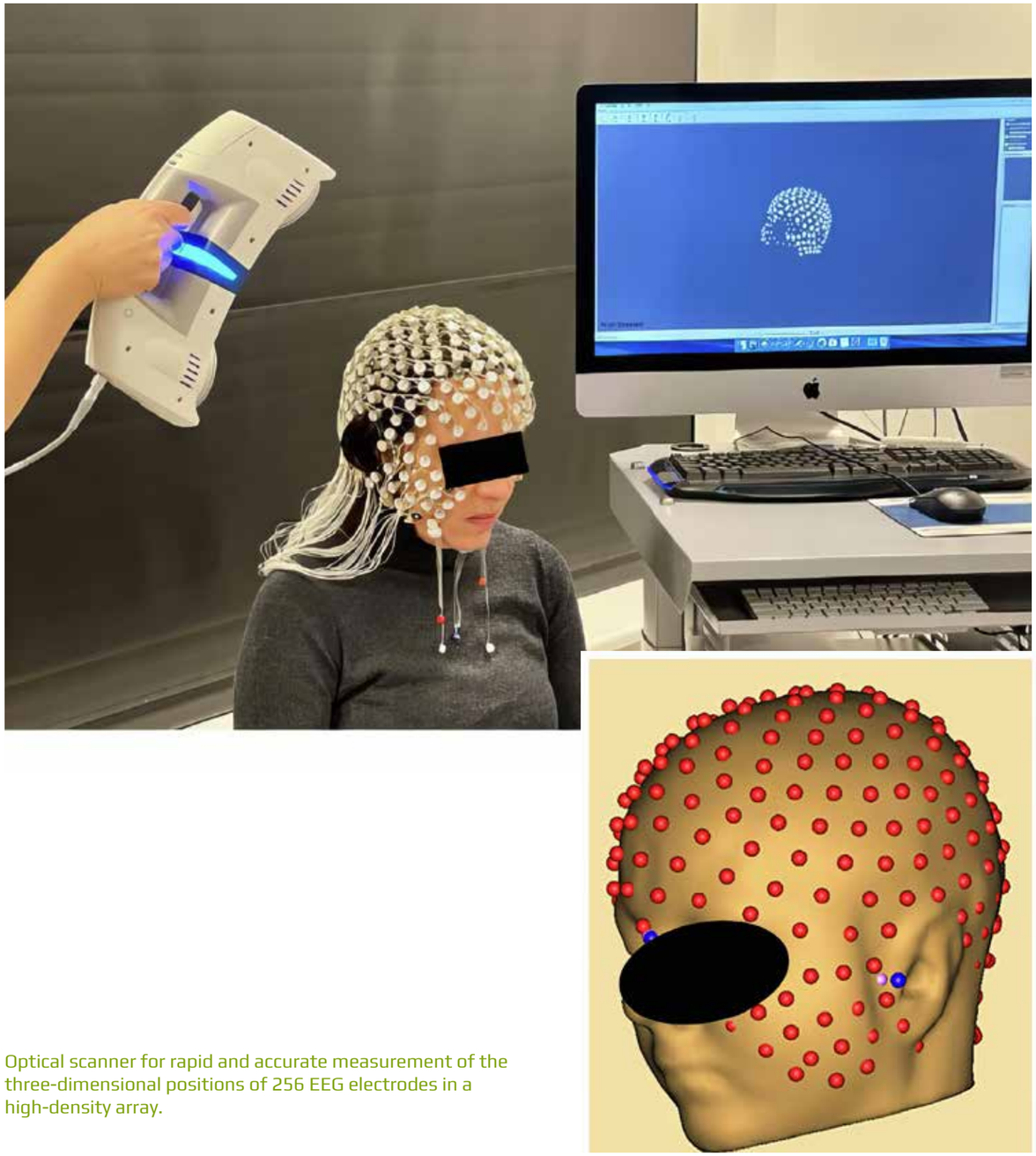
We assessed the accuracy of high-density EEG electrode position measurement, using an optical scanner compared with the classical, photogrammetry method. We found that the handheld optical scanner is more accurate and feasible, compared to the photogrammetry method. This warrants for the clinical implementation of the novel method.

We have validated the accuracy of a novel method of source localization – the relative source power (RSP) imaging of extratemporal interictal epileptiform discharge. A source region with 20 mm radius contained lesioned tissue in all cases. Using localization of the resection site and operation outcome as gold standard, we achieved a sensitivity of 82% and specificity of 50%.

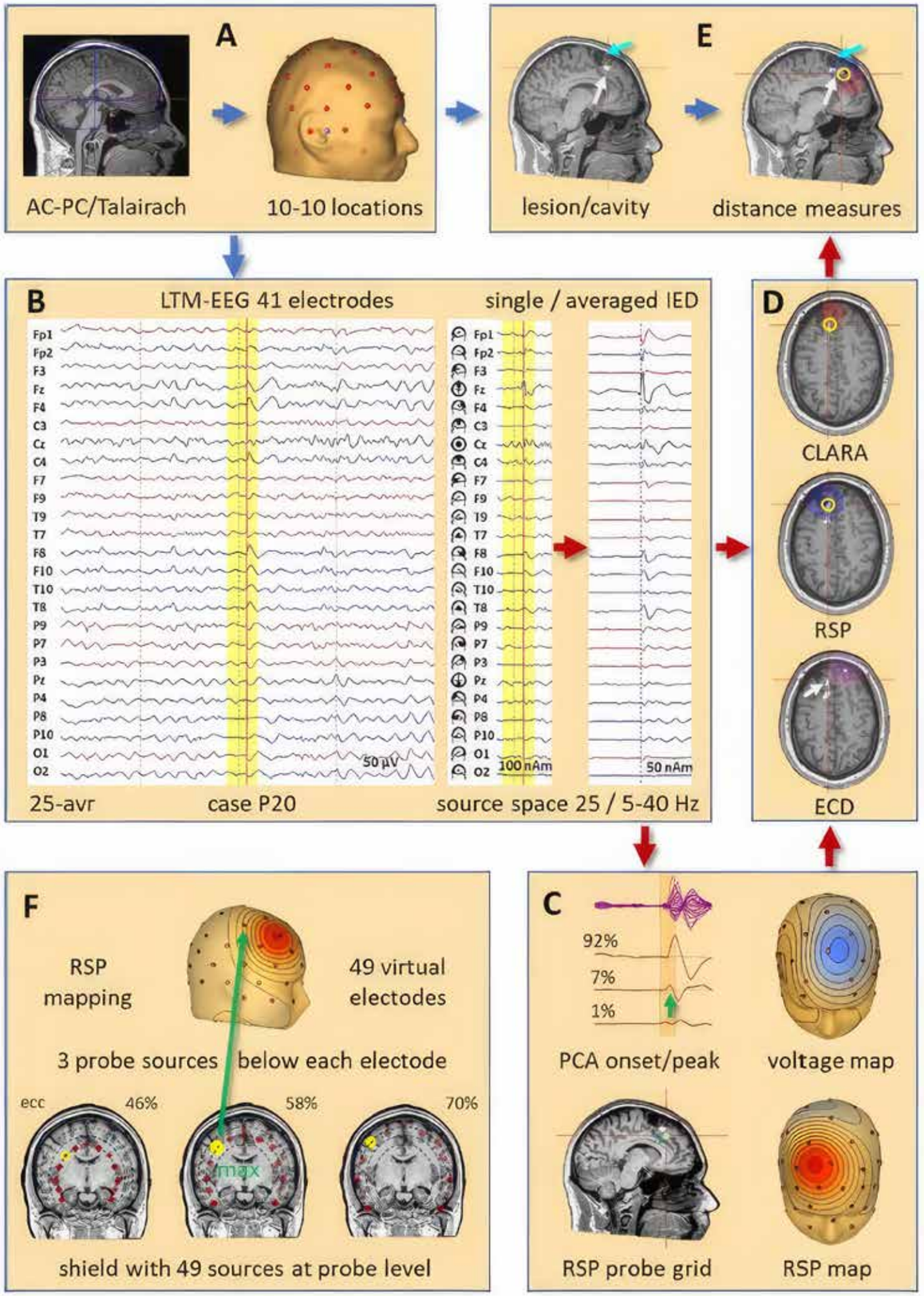
We have investigated the diagnostic utility of ESI in the presurgical evaluation of children with focal cortical dysplasia, and to compared it with other imaging techniques. Highest localization accuracy (80%) was obtained with ESI, followed by PET and ictal SPECT (75%). Our findings demonstrate that ESI using a high-density EEG array allows accurate localization of the epileptogenic zone in children with focal cortical dysplasia.

In spite of a standardized analysis pipeline, several aspects tailored to the individual patient involve subjective decisions of the expert performing the analysis. We investigated the inter-analyzer agreement of ESI in presurgical evaluations of epilepsy, using the same software and analysis pipeline. The overall agreement among experts for the ESI methods was substantial, and there was no significant difference between the methods. Our results suggest that using a standardized analysis pipeline, newly trained experts reach similar ESI solutions, calling for more standardization in this emerging clinical application in neuroimaging.

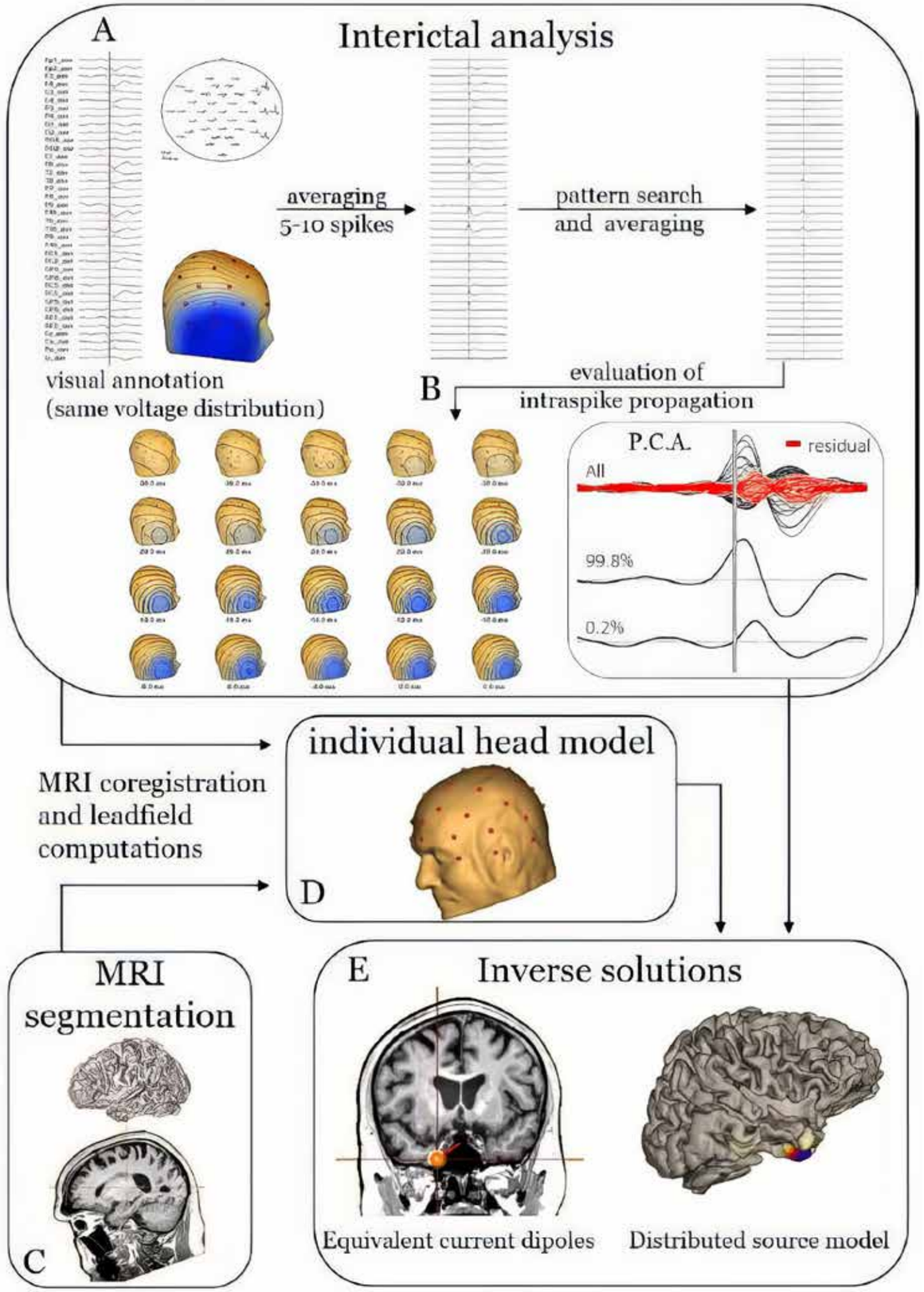
ESI requires special expertise and it is underutilized. To circumvent this, automated analysis pipelines have been previously developed and validated for the interictal discharges. In a new study published in 2022, we presented the clinical validation of an automated ESI for ictal EEG signals. The accuracy of the automated ESI was 74%. Automating the ESI of the ictal EEG signals will facilitate implementation of this tool in the presurgical evaluation.



Optical scanner for rapid and accurate measurement of the three-dimensional positions of 256 EEG electrodes in a high-density array.



Analysis pipeline of a novel ESI method: Relative Source Power.



Standardized analysis pipeline of the interictal EEG source imaging.

Publication

Heers M, Böttcher S, Kalina A, Katletz S, Altenmüller DM, Baroumand AG, Strobbe G, van Mierlo P, von Oertzen TJ, Marusic P, Schulze-Bonhage A, Beniczky S, Dümpelmann M. Detection of interictal epileptiform discharges in an extended scalp EEG array and high-density EEG-A prospective multicenter study. Epilepsia. 2022 Jul;63(7):1619-1629. doi: 10.1111/epi.17246.

Györfi O, Ip CT, Justesen AB, Gam-Jensen ML, Rømer C, Fabricius M, Pinborg LH, Beniczky S. Accuracy of high-density EEG electrode position measurement using an optical scanner compared with the photogrammetry method. Clin Neurophysiol Pract. 2022 May 2;7:135-138. doi: 10.1016/j.cnp.2022.04.002.

Scherg M, Schulz R, Berg P, Cho JH, Bornfleth H, Kural MA, Woermann FG, Bien CG, Beniczky S. Relative Source Power: A novel method for localizing epileptiform EEG discharges. Clin Neurophysiol. 2022 Jan;133:9-19. doi: 10.1016/j.clinph.2021.10.005.

Wanders A, Garibotto V, Spinelli L, Beniczky S, Vulliémoz S, Daniel RT, Schaller K, Bartoli A, Korff C, Seeck M. High density electric source imaging in childhood-onset epilepsy due to focal cortical dysplasia. Clin Neurophysiol Pract. 2022 Jul 26;7:245-251. doi: 10.1016/j.cnp.2022.07.002.

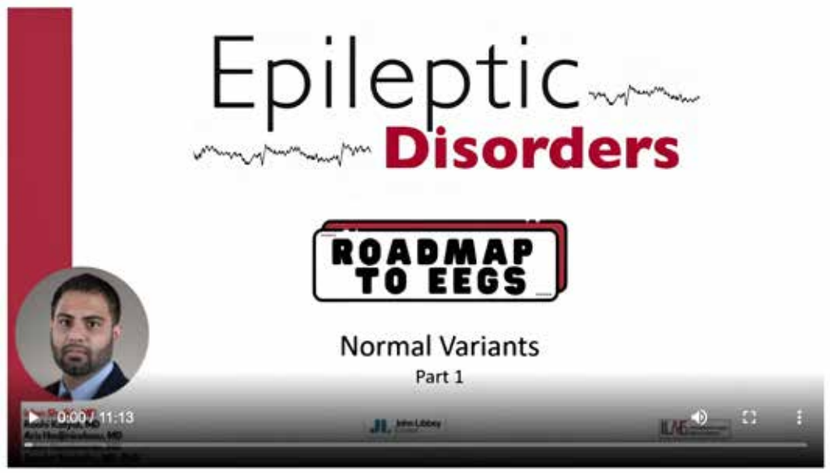
Mattioli P, Cleeren E, Hadady L, Cossu A, Cloppenborg T, Arnaldi D, Beniczky S. Electric Source Imaging in Presurgical Evaluation of Epilepsy: An Inter-Analyser Agreement Study. Diagnostics (Basel). 2022 Sep 24;12(10):2303. doi: 10.3390/diagnostics12102303.

Baroumand AG, Arbune AA, Strobbe G, Keereman V, Pinborg LH, Fabricius M, Rubboli G, Gøbel Madsen C, Jespersen B, Brennum J, Mølby Henriksen O, Mierlo PV, Beniczky S. Automated ictal EEG source imaging: A retrospective, blinded clinical validation study. Clin Neurophysiol. 2022 Sep;141:119-125. doi: 10.1016/j.clinph.2021.03.040.

5.5 Research in postgraduate education

Postgraduate education utilizes important new knowledge in the field of adult learning. We have contributed to the development of interactive, online, self-paced learning elements, addressing the learning objectives of the ILAE curriculum in epileptology, and we have summarized these educational offerings and their performance in a paper published in 2022.

Based on a consensus discussion of a broad, international expert panel, we compiled a curriculum for the EEG patterns which neurology residents must learn during their training. We have developed an internship program at Epileptic Disorders, the official educational journal of the ILAE. One of the main objectives were to promote the educational activities and improve the outreach. With the interns, we developed a series of educational videos, addressing important learning objectives in EEG. These videos are available now on our YouTube channel too. We published a comprehensive, educational review paper on how to read voltage maps in EEG. We have evaluated and reported the results of the student evaluation of the educational courses which we organized at 34th International Epilepsy Congress. This content was available online too. We showed that an online interactive teaching session about the operational criteria of epileptiform discharges, significantly improved the accuracy of young trainees in interpreting EEG.



Screenshot of the online educational material on EEG normal variants.

Papers

Blümcke I, Biesel E, Bedenlier S, Händel M, Wilmshurst J, Mehndiratta MM, Yacubian EM, Cendes F, Arzimanoglou A, Beniczky S, Wolf P, Giavasi C, Baxendale S, Shisler P, Wiebe S. A structured, blended learning program towards proficiency in epileptology: the launch of the ILAE Academy Level 2 Program. Epileptic Disord. 2022 Oct 1;24(5):737-750. doi: 10.1684/epd.2022.1462.

Nascimento FA, Jing J, Strowd R, Sheikh IS, Weber D, Gavvala JR, Maheshwari A, Tanner A, Ng M, Vinayan KP, Sinha SR, Yacubian EM, Rao VR, Perry MS, Fountain NB, Karakis I, Wirrell E, Yuan F, Friedman D, Tankisi H, Rampp S, Fasano R, Wilmshurst JM, O'Donovan C, Schomer D, Kaplan PW, Sperling MR, Benbadis S, Westover MB, Beniczky S. Competency-based EEG education: a list of "must-know" EEG findings for adult and child neurology residents. Epileptic Disord. 2022 Oct 1;24(5):979-982. doi: 10.1684/epd.2022.1476.

Nascimento FA, Gavvala JR, Tankisi H, Beniczky S. Neurology resident EEG training in Europe. Clin Neurophysiol Pract. 2022 Aug 24;7:252-259. doi: 10.1016/j.cnp.2022.08.001.

Sheikh IS, Katyal R, Hadjinicolaou A, Beniczky S, Nascimento FA. Introducing the Epileptic Disorders Internship Program. Epileptic Disord. 2022 Dec 1;24(6):1139-1140. doi: 10.1684/epd.2022.1485.

Sheikh IS, Katyal R, Hadjinicolaou A, Nascimento FA, Beniczky S. Roadmap to EEGs: video-based e-learning modules addressing clinical EEG reading. Epileptic Disord. 2022 Dec 1;24(6):1132-1138. doi: 10.1684/epd.2022.1495.

Kural MA, Aydemir ST, Levent HC, Ölmez B, Özer IS, Vlachou M, Witt AH, Yilmaz AY, Beniczky S. The operational definition of epileptiform discharges significantly improves diagnostic accuracy and inter-rater agreement of trainees in EEG reading. Epileptic Disord. 2022 Apr 1;24(2):353-358. doi: 10.1684/epd.2021.1395.

Foged MT, Scherg M, Fabricius M, Beniczky S. Learn to interpret voltage maps: an atlas of topographies. Epileptic Disord. 2022 Apr 1;24(2):229-248. doi: 10.1684/epd.2021.1396.

Nascimento FA, Kural MA, Beniczky S. Learning about e-learning – the 34th International Epilepsy Congress experience. Epileptic Disord. 2022 Jun 1;24(3):623-625. doi: 10.1684/epd.2022.1412.

5.6 Artificial intelligence in epilepsy diagnosis and monitoring

Medical applications of artificial intelligence (AI) are likely to be a game-changer in our field too. We contributed to training and validation of AI models for automated and semi-automated analyses of videos and EEGs.

Automated video analyses was sensitive for detecting major motor seizures, such as generalized tonic-clonic seizures. However, for assessment and classification of the other seizure-types, a semi-automated ("hybrid") approach was necessary, where human experts inspected the video-epochs highlighted by the AI model. The hybrid approach significantly reduced the work-load of the human experts.

We found similar outcome for AI models detecting interictal epileptiform discharges. The fully automated application of the AI model gave high specificity, but the sensitivity was too low for clinical implementation. However, the hybrid system in which human experts inspected the automated detections, have high sensitivity and specificity, and significantly decreased the work-load.

We contributed to training and validation of an AI model which predicts impaired consciousness in absence epilepsy, based on analysis of the EEG signals.

Graphical user interface view of the video-analysis system using artificial intelligence

Papers

Armand Larsen S, Terney D, Østerkjerhuus T, Vinding Merinder T, Annala K, Knight A, Beniczky S. Automated detection of nocturnal motor seizures using an audio-video system. Brain Behav. 2022 Sep;12(9):e2737. doi: 10.1002/brb3.2737.

Peltola J, Basnyat P, Armand Larsen S, Østerkjaerhuus T, Vinding Merinder T, Terney D, Beniczky S. Semiautomated classification of nocturnal seizures using video recordings. Epilepsia. 2022 Feb 23. doi: 10.1111/epi.17207.

Kural MA, Jing J, Fürbass F, Perko H, Qerama E, Johnsen B, Fuchs S, Westover MB, Beniczky S. Accurate identification of EEG recordings with interictal epileptiform discharges using a hybrid approach: Artificial intelligence supervised by human experts. Epilepsia. 2022 May;63(5):1064-1073. doi: 10.1111/epi.17206.

Springer M, Khalaf A, Vincent P, Ryu JH, Abukhadra Y, Beniczky S, Glauser T, Krestel H, Blumenfeld H. A machine-learning approach for predicting impaired consciousness in absence epilepsy. Ann Clin Transl Neurol. 2022 Oct;9(10):1538-1550. doi: 10.1002/acn3.51647.

5.7 Wearable devices in epilepsy monitoring

One of the main research topics of our group is development and validation of wearable devices for automated seizure detection and characterization. In 2022 we continued working on numerous projects in this area.

We have validated an artificial intelligence-based algorithm for automated detection of absence seizures, using a wearable non-invasive EEG device. In a proof of principle study, we demonstrated the feasibility and clinical utility of automated behavioral testing triggered by automated seizure detection. We contributed to a multicenter study on detection of temporal lobe seizures using ultra-long term monitoring with a subcutaneously implanted EEG device. Our findings demonstrated that semi-automated seizure detection/review process can be performed with high sensitivity and clinically applicable specificity. In a large, international survey study on the ultra-long-term experience of patients with using wearable devices, we showed the clinical utility of these devices, and we found that the devices formally validated in phase-3 studies performed better than the other devices.

Graphical user interface view of the video-analysis system using artificial intelligence

Papers

Japaridze G, Loeckx D, Buckinx T, Armand Larsen S, Proost R, Jansen K, MacMullin P, Paiva N, Kasradze S, Rotenberg A, Lagae L, Beniczky S. Automated detection of absence seizures using a wearable electroencephalographic device: a phase 3 validation study and feasibility of automated behavioral testing. Epilepsia. 2022 Feb 17. doi: 10.1111/epi.17200.

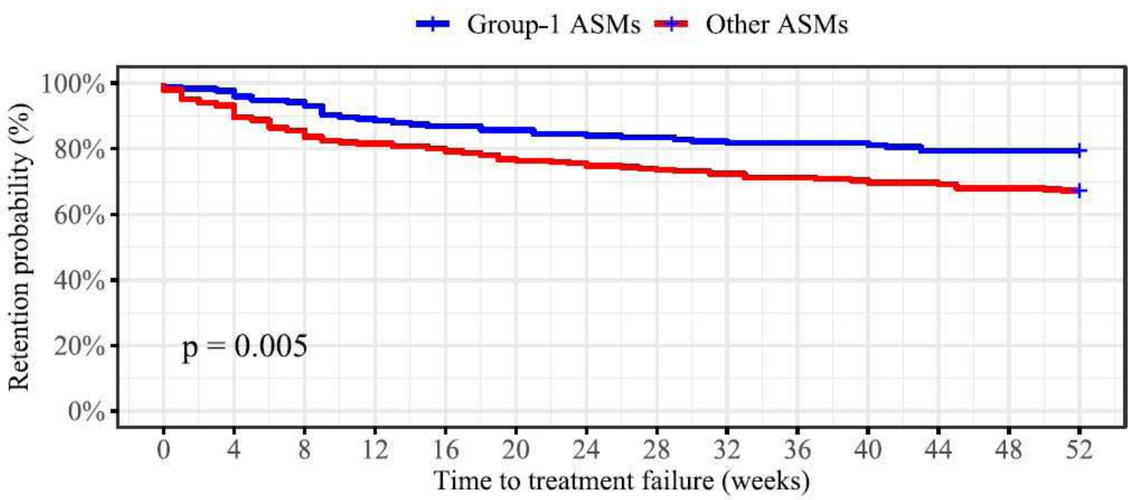
Remvig LS, Duun-Henriksen J, Fürbass F, Hartmann M, Viana PF, Kappel Overby AM, Weisdorf S, Richardson MP, Beniczky S, Kjaer TW. Detecting temporal lobe seizures in ultra long-term subcutaneous EEG using algorithm-based data reduction. Clin Neurophysiol. 2022 Oct;142:86-93. doi: 10.1016/j.clinph.2022.07.504.

Hadady L, Klivényi P, Fabó D, Beniczky S. Real-world user experience with seizure detection wearable devices in the home environment. Epilepsia. 2022 Feb 23. doi: 10.1111/epi.17189.

5.8 Epipick: a web-based decision support system for optimal, patient-tailored choice of antiseizure medication

We developed the Epipick - a freely accessible web-based application (https://epipick.org) to help health care professionals select the most appropriate, patient-tailored antiseizure medication (ASM) in patients with epilepsy (seizure onset at 10 years of age or older). EpiPick considers seizure types and patient-specific variables to provide treatment recommendations, ranking ASMs in order of appropriateness based on the available scientific evidence and expert judgement. The app also provides a summary of prescribing information for each of the ASMs being suggested.

After the original publication of the application in 2021, we continued monitoring the published evidence and we updated the application accordingly in 2022. We completed a large-scale validation study on 425 consecutive patients with newly diagnosed epilepsy, who were followed for at least 1 year after starting medication. ASMs classified by the algorithm as best options had a significantly higher retention rate, higher seizure freedom rate and lower rate of discontinuation due to adverse effects than ASMs ranked as less desirable by the algorithm. We concluded that the use of the freely available decision support system is associated with improved outcomes. This drug selection application can provide valuable assistance to health care professionals prescribing medication for individuals with epilepsy.



Kaplan–Meier analysis of the time to treatment failure. Patients treated with best option antiseizure medications (ASMs) recommended by the algorithm (Group 1) versus patients treated with other drugs

Papers

Hadady L, Klivényi P, Perucca E, Rampp S, Fabó D, Bereczki C, Rubboli G, Asadi-Pooya AA, Sperling MR, Beniczky S. Web-based decision support system for patient-tailored selection of antiseizure medication in adolescents and adults: An external validation study. Eur J Neurol. 2022 Feb;29(2):382-389. doi: 10.1111/ene.15168.

Asadi-Pooya AA, Beniczky S, Rubboli G, Sperling MR, Rampp S, Perucca E. The EpiPick algorithm to select appropriate antiseizure medications in patients with epilepsy: Validation studies and updates. Epilepsia. 2022 Jan;63(1):254-255. doi: 10.1111/epi.17129.

5.9 GABA-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, Liao VWY, Johannesen KMH, Gardella E, Jacobs J, Lesca G, Gokce-Samar Z, Arzimanoglou A, Zeidler S, Striano P, Meyer P, Benkel-Herrenbrueck I, Mero IL, Rummel J, Chebib M, Møller RS, Ahring PK. Gain-of-function and loss-of-function GABRB3 variants lead to distinct clinical phenotypes in patients with developmental and epileptic encephalopathies. Nat Commun. 2022 Apr 5;13(1):1822.

Ahring PK, Liao VWY, Lin S, Absalom NL, Chebib M, Møller RS. The de novo GABRA4 p.Thr300Ile variant found in a patient with early-onset intractable epilepsy and neurodevelopmental abnormalities displays gain-of-function traits. Epilepsia. 2022 Sep;63(9):2439-2441.

5.10 GRIA-related disorders

Developmental and Epileptic Encephalopathies (DEEs) are rare and severe neurological conditions often associated with intellectual disability, developmental delay, autism spectrum disorders and movement disorders. Seizures often begin in early infancy, and patients are often resistant to antiepileptic treatment.

Genetic factors play a major role in the underlying cause of many DEEs, and the identification of the causative genes have disclosed unique information on the different pathomechanisms and opened novel therapeutic perspectives. Recently, human variants in the α-amino-3-hydroxy-5methyl-4-isoxazole propionic acid receptor (AMPAR) have been reported to cause of DEE. This receptor is important as it helps to maintain normal brain activity and variants in GRIA genes that encode the AMPAR will cause changes to normal brain function.

The overall aim of this proposal is to establish specific correlations between phenotype, genotype, functional effects and therapeutic response to translate genetic diagnostics into therapy. Knowing the functional effect of a genetic variant can assist clinicians to avoid ineffective or even disease-aggravating treatments. Our findings will help change the current paradigm of treating patients with DEE currently uses a trial-and-error approach to one that utilizes precision medicine based on a patient's genetic, functional and clinical diagnoses (Figure 1).

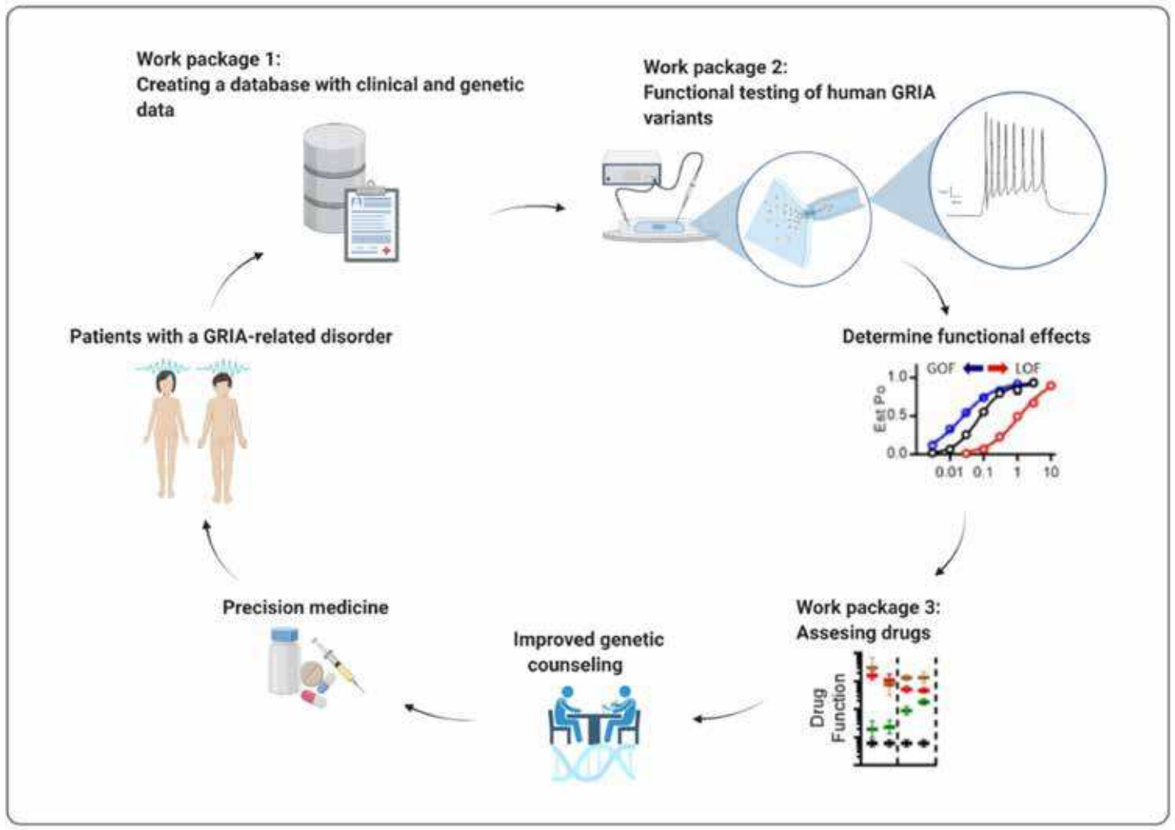


Illustration of the research strategy and how the three work packages bridge the translational gab an ultimately enable precision therapy.

5.11 Ultra-long sub-cutaneous EEG monitoring: reliability,, safety and impact on clinical management in uncontrolled epilepsies

In epileptology, seizure classification and quantification are among the main parameters that contributes to define the degree of severity of an epileptic condition, and they are crucial to guide individual treatments. However, patient self-evaluation compared with objective evaluation by videoEEG monitoring or long-term ambulatory EEG revealed that patients self-assessment is highly imprecise, documenting fewer than 50% of their seizures, on average. Recently, a novel EEG recording device consisting of an EEG electrode designed for subcutaneous implantation (subcutaneous EEG, 24/7 EEG™ SubQ) connected to an external device has been shown to be able to record EEG for periods of time up to 3 months. We have initiated a study in drug-resistant epilepsy patient using sqEEG with the purposes of:

- assess the sensitivity, reliability and safety of the 24/7 EEG™ SubQ device for recording seizures over longer periods (over 1 month) in people suffering from either focal or generalized uncontrolled epilepsy
- assess sensitivity and reliability of automated seizure recognition software in the 24/7 EEG™ SubQ equipment
- evaluate how and whether data collected with the 24/7 EEG™ SubQ equipment can influence the clinical treatment of the participating subjects

In 2021-2022, 3 patients have been implanted, showing that sqEEG can be a promising technique fro long-term seizure recording and possibly for drug effects assessment.

5.12 The Human Related Conversation and the Professional Tension Between Diakonia and Nursing

The article explores how nurses experience tension in a professional collaboration between nursing and work with a diaconal profile in Filadelfia, Denmark. Filadelfia was founded in 1897 as a diaconal institution with a hospital and deaconess house. Today, Filadelfia appears primarily as a highly specialized Epilepsy Hospital unrelated to diakonia. The collaboration this article investigates is called The Human Related Conversation (hereafter THRC). It is a practice developed within a diaconal unit of the institution, Filadelfia Uddannelse, and performed by nurses at Filadelfia Epilepsy Hospital. The article is based on a discourse analysis of five interviews conducted using THRC. The analysis finds that the nurses' experience of THRC generates different discourses, including a nursing discourse and a legitimacy discourse, and encourages them to work in a more value-conscious way.

Papers

Hjelm, Conny (2021 [2022]): The Human Related Conversation and the Professional Tension Between Diakonia and Nursing. Diaconia, vol. 12, p. 137–160. doi:10.13109/diac.2021.12.2.137

Conny Hjelm
Deacon and Ph.d. stud.

6. Publication list in 2022

1. 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. 2022 Dec 28;147:11-13. doi: 10.1016/j.clinph.2022.12.008.
2. **Meritam Larsen P**, **Wüstenhagen S**, **Terney D**, **Gardella E**, Aurlien H, **Beniczky S**. Duration of epileptic seizure types: A data-driven approach. Epilepsia. 2022 Dec 15. doi: 10.1111/epi.17492.
3. Stevelink R, Al-Toma D, Jansen FE, Lamberink HJ, Asadi-Pooya AA, Farazdaghi M, Cação G, Jayalakshmi S, Patil A, Özkara Ç, Aydın Ş, Gesche J, Beier CP, Stephen LJ, Brodie MJ, Unnithan G, Radhakrishnan A, Höfler J, Trinka E, Krause R; EpiPGX Consortium; Irelli EC, Di Bonaventura C, Szaflarski JP, Hernández- Vanegas LE, Moya-Alfaro ML, Zhang Y, Zhou D, Pietrafusa N, Specchio N, Japaridze G, **Beniczky S**, Janmohamed M, Kwan P, Syvertsen M, Selmer KK, Vorderwülbecke BJ, Holtkamp M, Viswanathan LG, Sinha S, Baykan B, Altindag E, von Podewils F, Schulz J, Seneviratne U, Viloria-Alebesque A, Karakis I, D'Souza WJ, Sander JW, Koeleman BPC, Otte WM, Braun KPJ. Individualised prediction of drug resistance and seizure recurrence after medication withdrawal in people with juvenile myoclonic epilepsy: A systematic review and individual participant data meta- analysis. EClinicalMedicine. 2022 Nov 11;53:101732. doi: 10.1016/j.eclinm.2022.101732.
4. **Mattioli P, Cleeren E**, **Hadady L**, **Cossu A**, Cloppenborg T, Arnaldi D, **Beniczky S**. Electric Source Imaging in Presurgical Evaluation of Epilepsy: An Inter- Analyser Agreement Study. Diagnostics (Basel). 2022 Sep 24;12(10):2303. doi: 10.3390/diagnostics12102303.
5. Sheikh IS, Katyal R, Hadjinicolaou A, Nascimento FA, **Beniczky S**. Roadmap to EEGs: video-based e-learning modules addressing clinical EEG reading. Epileptic Disord. 2022 Dec 1;24(6):1132-1138. doi: 10.1684/epd.2022.1495.
6. **Vlachou M**, Ryvlin P, Arbune AA, **Armand Larsen S**, Skraep Sidaros A, Cacic Hribljan M, Fabricius M, **Beniczky S**. Progressive slowing of clonic phase predicts postictal generalized EEG suppression. Epilepsia. 2022 Dec;63(12):3204-3211. doi: 10.1111/epi.17434.
7. Japaridze G, Kasradze S, Aurlien H, **Beniczky S**. Implementing the SCORE system improves the quality of clinical EEG reading. Clin Neurophysiol Pract. 2022 Sep 1;7:260-263. doi: 10.1016/j.cnp.2022.07.004.
8. Nascimento FA, Gavvala JR, Tankisi H, **Beniczky S**. Neurology resident EEG training in Europe. Clin Neurophysiol Pract. 2022 Aug 24;7:252-259. doi: 10.1016/j.cnp.2022.08.001.
9. Nascimento FA, Jing J, **Beniczky S**, Olandoski M, Benbadis SR, Cole AJ, Westover MB. EEG reading with or without clinical information - a real-world practice study. Neurophysiol Clin. 2022 Oct;52(5):394-397. doi: 10.1016/j.neucli.2022.08.002.
10. Sheikh IS, Katyal R, Hadjinicolaou A, **Beniczky S**, Nascimento FA. Introducing the Epileptic Disorders Internship Program. Epileptic Disord. 2022 Dec 1;24(6):1139-1140. doi: 10.1684/epd.2022.1485.
11. Springer M, Khalaf A, Vincent P, Ryu JH, Abukhadra Y, **Beniczky S**, Glauser T, Krestel H, Blumenfeld H. A machine-learning approach for predicting impaired consciousness in absence epilepsy. Ann Clin Transl Neurol. 2022 Oct;9(10):1538-1550. doi: 10.1002/acn3.51647.
12. Wanders A, Garibotto V, Spinelli L, **Beniczky S**, Vulliémoz S, Daniel RT, Schaller K, Bartoli A, Korff C, Seeck M. High density electric source imaging in childhood-onset epilepsy due to focal cortical dysplasia. Clin Neurophysiol Pract. 2022 Jul 26;7:245-251. doi: 10.1016/j.cnp.2022.07.002. PMID: 36062078;
13. Remvig LS, Duun-Henriksen J, Fürbass F, Hartmann M, Viana PF, **Kappel Overby AM**, Weisdorf S, Richardson MP, **Beniczky S**, Kjaer TW. Detecting temporal lobe seizures in ultra long-term subcutaneous EEG using algorithm-based data reduction. Clin Neurophysiol. 2022 Oct;142:86-93. doi: 10.1016/j.clinph.2022.07.504.
14. **Armand Larsen S**, **Terney D**, Østerkjerhuus T, Vinding Merinder T, Annala K, Knight A, **Beniczky S**. Automated detection of nocturnal motor seizures using an audio-video system. Brain Behav. 2022 Sep;12(9):e2737. doi: 10.1002/brb3.2737.
15. Nascimento FA, Jing J, Strowd R, Sheikh IS, Weber D, Gavvala JR, Maheshwari A, Tanner A, Ng M, Vinayan KP, Sinha SR, Yacubian EM, Rao VR, Perry MS, Fountain NB, Karakis I, Wirrell E, Yuan F, Friedman D, Tankisi H, Rampp S, Fasano R, Wilmshurst JM, O'Donovan C, Schomer D, Kaplan PW, Sperling MR, Benbadis S, Westover MB, **Beniczky S**. Competency-based EEG education: a list of "must-know" EEG findings for adult and child neurology residents. Epileptic Disord. 2022 Oct 1;24(5):979-982. doi: 10.1684/epd.2022.1476.
16. **Wüstenhagen S**, **Terney D**, **Gardella E**, **Meritam Larsen P**, **Rømer C**, Aurlien H, **Beniczky S**. EEG normal variants: A prospective study using the SCORE system. Clin Neurophysiol Pract. 2022 Jun 30;7:183-200. doi: 10.1016/j.cnp.2022.06.001.
17. Blümcke I, Biesel E, Bedenlier S, Händel M, Wilmshurst J, Mehndiratta MM, Yacubian EM, Cendes F, Arzimanoglou A, **Beniczky S**, Wolf P, Giavasi C, Baxendale S, Shisler P, Wiebe S. A structured, blended learning program towards proficiency in epileptology: the launch of the ILAE Academy Level 2 Program. Epileptic Disord. 2022 Oct 1;24(5):737-750. doi: 10.1684/epd.2022.1462.
18. **Beniczky S**, Jeppesen J, Kjær TW, Fabricius M. Wearable devices for automated seizure detection. Ugeskr Laeger. 2022 Jun 27;184(26):V10210770.
19. **Beniczky S**, Tatum WO, Blumenfeld H, Stefan H, Mani J, Maillard L, Fahoum F, Vinayan KP, Mayor LC, **Vlachou M**, Margitta S, Ryvlin P, Philippe K. Seizure semiology: ILAE glossary of terms and their significance. Epileptic Disord. 2022 Jun 1;24(3):447-495. doi: 10.1684/epd.2022.1430. PMID: 35770761.
20. Bonardi CM, **Bayat A**, Madsen CG, **Hammer TB**, Reale C, **Gardella E**, **Marjanovic D**, **Beniczky S**, **Møller RS**, **Rubboli G**. Trisomy 20p/monosomy 18p associated with congenital bilateral perisylvian syndrome. Epileptic Disord. 2022 Jun 1;24(3):577-582. doi: 10.1684/epd.2022.1423.
21. Nascimento FA, **Kural MA**, **Beniczky S**. Learning about e-learning – the 34th International Epilepsy Congress experience. Epileptic Disord. 2022 Jun 1;24(3):623-625. doi: 10.1684/epd.2022.1412.
22. **Györfi O**, Ip CT, **Justesen AB**, **Gam-Jensen ML**, **Rømer C**, Fabricius M, Pinborg LH, **Beniczky S**. Accuracy of high-density EEG electrode position measurement using an optical scanner compared with the photogrammetry method. Clin Neurophysiol Pract. 2022 May 2;7:135-138. doi: 10.1016/j.cnp.2022.04.002.
23. Westin K, **Beniczky S**, Lundqvist D. Reply to "Slow oscillations anticipate interictal epileptic discharges". Clin Neurophysiol. 2022 Jul;139:130-131. doi: 10.1016/j.clinph.2022.04.014.
24. Roberg LE, Monsson O, Kristensen SB, Dahl SM, Ulvin LB, Heuser K, Taubøll E, Strzelczyk A, Knake S, Bechert L, Rosenow F, Beier D, **Beniczky S**, Krøigård T, Beier CP. Prediction of Long-term Survival After Status Epilepticus Using the ACD Score. JAMA Neurol. 2022 Jun 1;79(6):604-613. doi: 10.1001/jamaneurol.2022.0609.
25. Heers M, Böttcher S, Kalina A, Katletz S, Altenmüller DM, Baroumand AG, Strobbe G, van Mierlo P, von Oertzen TJ, Marusic P, Schulze-Bonhage A, **Beniczky S**, Dümpelmann M. Detection of interictal epileptiform discharges in an extended scalp EEG array and high-density EEG-A prospective multicenter study. Epilepsia.2022 Jul;63(7):1619-1629. doi: 10.1111/epi.17246.
26. Westin K, Cooray G, **Beniczky S**, Lundqvist D. Interictal epileptiform discharges in focal epilepsy are preceded by increase in low-frequency oscillations. Clin Neurophysiol. 2022 Apr;136:191-205. doi: 10.1016/j.clinph.2022.02.003.
27. **Hadady L**, Klivényi P, Fabó D, **Beniczky S**. Real-world user experience with seizure detection wearable devices in the home environment. Epilepsia. 2022 Feb 23. doi: 10.1111/epi.17189.
28. Peltola J, Basnyat P, **Armand Larsen S**, Østerkjaerhuus T, Vinding Merinder T, **Terney D**, **Beniczky S**. Semiautomated classification of nocturnal seizures using video recordings. Epilepsia. 2022 Feb 23. doi: 10.1111/epi.17207.
29. **Kural MA**, Jing J, Fürbass F, Perko H, Qerama E, Johnsen B, Fuchs S, Westover MB, **Beniczky S**. Accurate identification of EEG recordings with interictal epileptiform discharges using a hybrid approach: Artificial intelligence supervised by human experts. Epilepsia. 2022 May;63(5):1064-1073. doi: 10.1111/epi.17206.
30. Japaridze G, Loeckx D, Buckinx T, **Armand Larsen S**, Proost R, Jansen K, MacMullin P, Paiva N, Kasradze S, Rotenberg A, Lagae L, **Beniczky S**. Automated detection of absence seizures using a wearable electroencephalographic device: a phase 3 validation study and feasibility of automated behavioral testing. Epilepsia. 2022 Feb 17. doi: 10.1111/epi.17200.
31. Nielsen TØ, Herlin MK, Linnet KM, **Beniczky S**, Sommerlund M, Granild-Jensen JB, Gregersen PA. Autosomal dominant sleep-related hypermotor epilepsy caused by a previously unreported CHRNA4 variant. Eur J Med Genet. 2022 Mar;65(3):104444. doi: 10.1016/j.ejmg.2022.104444.
32. Krøigård T, Andersen KV, Tankisi H, **Beniczky S**, Kristensen AG. Reply to "Conduction studies on the sural nerve". Clin Neurophysiol Pract. 2021 Dec 13;7:25-26. doi: 10.1016/j.cnp.2021.11.003.
33. Dalsgaard FF, Moeslund N, Zhang ZL, Pedersen M, Qerama E, **Beniczky S**, Ryhammer P, Ilkjær LB, Erasmus M, Eiskjær H. Clamping of the Aortic Arch Vessels During Normothermic Regional Perfusion After Circulatory Death Prevents the Return of Brain Activity in a Porcine Model. Transplantation. 2022 Sep 1;106(9):1763-1769. doi: 10.1097/TP.0000000000004047.
34. Engedal TS, Johnsen B, Sidaros A, Fabricius M, Christensen J, **Beniczky S**. EEG diagnosticsof non-convulsive status epilepticus in critically ill patients. Ugeskr Laeger. 2022 Jan 17;184(3):V07210570.
35. Foged MT, Scherg M, Fabricius M, **Beniczky S**. Learn to interpret voltage maps: an atlas of topographies. Epileptic Disord. 2022 Apr 1;24(2):229-248. doi: 10.1684/epd.2021.1396.
36. Tatum WO, Mani J, Jin K, Halford JJ, Gloss D, Fahoum F, Maillard L, Mothersill I, **Beniczky S**. Minimum standards for inpatient long-term video-EEG monitoring: A clinical practice guideline of the international league against epilepsy and international federation of clinical neurophysiology. Clin Neurophysiol. 2022 Feb;134:111-128. doi: 10.1016/j.clinph.2021.07.016.
37. **Kural MA**, Aydemir ST, Levent HC, Ölmez B, Özer IS, **Vlachou M**, Witt AH, Yilmaz AY, **Beniczky S**. The operational definition of epileptiform discharges significantly improves diagnostic accuracy and inter-rater agreement of trainees in EEG reading. Epileptic Disord. 2022 Apr 1;24(2):353-358. doi: 10.1684/epd.2021.1395.
38. Tatum WO, Mani J, Jin K, Halford JJ, Gloss D, Fahoum F, Maillard L, Mothersill I, **Beniczky S**. Minimum standards for inpatient long-term video-electroencephalographic monitoring: A clinical practice guideline of the International League Against Epilepsy and International Federation of Clinical Neurophysiology. Epilepsia. 2022 Feb;63(2):290-315. doi: 10.1111/epi.16977.
39. **Vlachou M**, Skrimpas GA, **Kural MA**, Rackauskaite G, Nikanorova N, Christensen J, **Nikanorova M**, **Beniczky S**. Electroclinical features and long-term therapeutic response in patients with typical absence seizures. Epileptic Disord. 2022 Apr 1;24(2):315-322. doi: 10.1684/epd.2021.1392.
40. Nascimento FA, Jing J, **Beniczky S**, Benbadis SR, Gavvala JR, Yacubian EMT, Wiebe S, Rampp S, van Putten MJAM, Tripathi M, Cook MJ, Kaplan PW, Tatum WO, Trinka E, Cole AJ, Westover MB. One EEG, one read - A manifesto towards reducing interrater variability among experts. Clin Neurophysiol. 2022 Jan;133:68-70.doi: 10.1016/j.clinph.2021.10.007.
41. Asadi-Pooya AA, **Beniczky S**, **Rubboli G**, Sperling MR, Rampp S, Perucca E. The EpiPick algorithm to select appropriate antiseizure medications in patients with epilepsy: Validation studies and updates. Epilepsia. 2022 Jan;63(1):254-255. doi: 10.1111/epi.17129.

42. Scherg M, Schulz R, Berg P, Cho JH, Bornfleth H, **Kural MA**, Woermann FG, Bien CG, **Beniczky S**. Relative Source Power: A novel method for localizing epileptiform EEG discharges. Clin Neurophysiol. 2022 Jan;133:9-19. doi: 10.1016/j.clinph.2021.10.005.
43. **Hadady L**, Klivényi P, Perucca E, Rampp S, Fabó D, Bereczki C, **Rubboli G**, Asadi-Pooya AA, Sperling MR, **Beniczky S**. Web-based decision support system for patient-tailored selection of antiseizure medication in adolescents and adults: An external validation study. Eur J Neurol. 2022 Feb;29(2):382-389. doi: 10.1111/ene.15168.
44. Baroumand AG, Arbune AA, Strobbe G, Keereman V, Pinborg LH, Fabricius M, **Rubboli G**, Gøbel Madsen C, Jespersen B, Brennum J, Mølby Henriksen O, Mierlo PV, **Beniczky S**. Automated ictal EEG source imaging: A retrospective, blinded clinical validation study. Clin Neurophysiol. 2022 Sep;141:119-125. doi: 10.1016/j.clinph.2021.03.040.
45. Macnee M, Pérez-Palma E, López-Rivera JA, Ivaniuk A, May P, **Møller RS**, Lal D. Data-driven historical characterization of epilepsy-associated genes. Eur J Paediatr Neurol. 2022 Dec 14;42:82-87.
46. **Johannesen KM**, **Bayat A**. [Christmas article: Genetic analysis in syndromic patients - d'oh!]. Ugeskr Laeger. 2022 Dec 12;184(50):V80109.
47. Alvarez C, Grimmel M, Ebrahimi-Fakhari D, Paul VG, Deininger N, Riess A, Haack T, **Gardella E**, **Møller RS**, **Bayat A**. Expansion of the phenotypic and molecular spectrum of CWF19L1-related disorder. Clin Genet. 2022 Dec 1. doi: 10.1111/cge.14275.
48. Rychkov GY, Shaukat Z, Lim CX, Hussain R, Roberts BJ, Bonardi CM, **Rubboli G**, Meaney BF, Whitney R, **Møller RS**, Ricos MG, Dibbens LM. Functional Effects of Epilepsy Associated KCNT1 Mutations Suggest Pathogenesis via Aberrant Inhibitory Neuronal Activity. Int J Mol Sci. 2022 Dec 1;23(23):15133.
49. Amadori E, Pellino G, Bansal L, Mazzone S, **Møller RS**, **Rubboli G**, Striano P, Russo A. Answer to: Genetic paroxysmal neurological disorders featuring episodic ataxia and epilepsy (Amadori E et al., 2022). EJMG-D-22-00384. Eur J Med Genet. 2022 Dec;65(12):104634.
50. Amadori E, Pellino G, Bansal L, Mazzone S, **Møller RS**, **Rubboli G**, Striano P, Russo A. Eur J Med Genet. 2022 Dec;65(12):104634. doi: 10.1016/j.ejmg.2022.104634. Genetic paroxysmal neurological disorders featuring episodic ataxia and epilepsy (Amadori E et al., 2022). EJMG-D-22-00384. Epub 2022 Oct 2.
51. Happ HC, Sadleir LG, Zemel M, de Valles-Ibáñez G, Hildebrand MS, McConkie-Rosell A, McDonald M, May H, Sands T, Aggarwal V, Elder C, Feyma T, **Bayat A**, **Møller RS**, **Fenger CD**, Klint Nielsen JE, Datta AN, Gorman KM, King MD, Linhares ND, Burton BK, Paras A, Ellard S, Rankin J, Shukla A, Majethia P, Olson RJ, Muthusamy K, Schimmenti LA, Starnes K, Sedláčková L, Štěrbová K, Vlčková M, Laššuthová P, Jahodová A, Porter BE, Couque N, Colin E, Prouteau C, Collet C, Smol T, Caumes R, Vansenne F, Bisulli F, Licchetta L, Person R, Torti E, McWalter K, Webster R, Gerard EE, Lesca G, Szepetowski P, Scheffer IE, Mefford HC, Carvill GL. Neurodevelopmental and Epilepsy Phenotypes in Individuals With Missense Variants in the Voltage-Sensing and Pore Domains of KCNH5. Neurology. 2023 Feb 7;100(6):e603-e615.
52. Neri S, Maia N, Fortuna AM, Damasio J, Coale E, Willis M, Jorge P, Højte AF, **Fenger CD**, **Møller RS**, **Bayat A**. Expanding the pre- and postnatal phenotype of WASHC5 and CCDC22 -related Ritscher-Schinzel syndromes. Eur J Med Genet. 2022 Nov;65(11):104624
53. Shakeshaft A, Laiou P, Abela E, Stavropoulos I, Richardson MP, Pal DK; **BIOJUME Consortium**. Heterogeneity of resting-state EEG features in juvenile myoclonic epilepsy and controls. Brain Commun. 2022 Jul 8;4(4):fcac180. doi: 10.1093/braincomms/fcac180. eCollection 2022.
54. **Møller RS**, Zhao L, Shoaff JR, Duno M, Andersen BN, Nguyen V, Fang TC, Kupelian V, Thorén R. Incidence of Aicardi-Goutières syndrome and KCNT1-related epilepsy in Denmark. Mol Genet Metab Rep. 2022 Oct 13;33:100924.
55. Mattison KA, Tossing G, Mulroe F, Simmons C, Butler KM, Schreiber A, Alsadah A, Neilson DE, Naess K, Wedell A, Wredenberg A, Sorlin A, McCann E, Burghel GJ, Menendez B, Hoganson GE, Botto LD, Filloux FM, Aledo-Serrano Á, Gil-Nagel A, Tatton-Brown K, Verbeek NE, van Hirtum-Das M, Breckpot J, **Hammer TB**, **Møller RS**, Whitney A, Douglas AGL, Kharbanda M, Brunetti-Pierri N, Morleo M, Nigro V, May HJ, Tao JX, Argili E, Sherr EH, Dobyns WB, Consortium GER, Baines RA, Warwicker J, Parker JA, Banka S, Campeau PM, Escayg A. ATP6V0C variants impair vacuolar V-ATPase causing a neurodevelopmental disorder often associated with epilepsy. Brain. 2022 Sep 8:awac330.
56. Jensen JM, Nielsen US, **Bayat A**, Rasmussen MB, **Møller RS**, Bisgaard AM, **Hammer TB**. [Genetic testing in autism spectrum disorder]. Ugeskr Laeger. 2022 Aug 22;184(34):V04220253.
57. Brünger T, Pérez-Palma E, Montanucci L, Nothnagel M, **Møller RS**, Schorge S, Zuberi S, Symonds J, Lemke JR, Brunklaus A, Traynelis SF, May P, Lal D. Conserved patterns across ion channels correlate with variant pathogenicity and clinical phenotypes. Brain. 2022 Aug 29:awac305.
58. **Johannesen KM**, **Gardella E**, Ahring PK, **Møller RS**. De novo SCN3A missense variant associated with self-limiting generalized epilepsy with fever sensitivity. Eur J Med Genet. 2022 Oct;65(10):104577.
59. **Bayat A**, **Fenger CD**, Techlo TR, Højte AF, Nørgaard I, Hansen TF, **Rubboli G**, **Møller RS**, Group DCCRS. Impact of Genetic Testing on Therapeutic Decision-Making in Childhood-Onset Epilepsies-a Study in a Tertiary Epilepsy Center. Neurotherapeutics. 2022 Jul;19(4):1353-1367.
60. Döring JH, Saffari A, Bast T, Brockmann K, Ehrhardt L, Fazeli W, Janzarik WG, Klabunde-Cherwon A, Kluger G, Muhle H, Pendziwiat M, **Møller RS**, Platzer K, Santos JL, Schröter J, Hoffmann GF, Kölker S, Syrbe S. Efficacy, Tolerability, and Retention of Antiseizure Medications in PRRT2-Associated Infantile Epilepsy. Neurol Genet. 2022 Sep 28;8(5):e200020
61. Stamberger H, Crosiers D, Balagura G, Bonardi CM, Basu A, Cantalupo G, Chiesa V, Christensen J, Dalla Bernardina B, Ellis CA, Furia F, Gardiner F, Giron C, Guerrini R, Klein KM, Korff C, Krijtova H, Leffner M, Lerche H, Lesca G, Lewis-Smith D, Marini C, **Marjanovic D**, Mazzola L, McKeown Ruggiero S, Mochel F, Ramond F, Reif PS, Richard-Mornas A, Rosenow F, Schropp C, Thomas RH, Vignoli A, Weber Y, Palmer E, Helbig I, Scheffer IE, Striano P, **Møller RS**, **Gardella E**, Weckhuysen S. Natural History Study of STXBP1-Developmental and Epileptic Encephalopathy Into Adulthood. Neurology. 2022 Jul 19;99(3):e221-e233.
62. Krey I, Platzer K, Esterhuizen A, Berkovic SF, Helbig I, Hildebrand MS, Lerche H, Lowenstein D, **Møller RS**, Poduri A, Sadleir L, Sisodiya SM, Weckhuysen S, Wilmshurst JM, Weber Y, Lemke JR, Berkovic SF, Cross JH, Helbig I, Lerche H, Lowenstein D, Mefford HC, Perucca P, Tan NC, Caglayan H, Helbig K, Singh G, Weber Y, Weckhuysen S. Current practice in diagnostic genetic testing of the epilepsies. Epileptic Disord. 2022 Oct 1;24(5):765-786. doi: 10.1684/epd.2022.1448.
63. **Johannesen KM**, **Bayat A**, **Hammer TB**, **Møller RS**. [Genetic factors provide individualised targeted treatment of epilepsy]. Ugeskr Laeger. 2022 Jun 27;184(26):V02220122.
64. Ahring PK, Liao VWY, Lin S, Absalom NL, Chebib M, **Møller RS**. The de novo GABRA4 p.Thr300Ile variant found in a patient with early-onset intractable epilepsy and neurodevelopmental abnormalities displays gain-of-function traits. Epilepsia. 2022 Sep;63(9):2439-2441.
65. Miceli F, Millevert C, Soldovieri MV, Mosca I, Ambrosino P, Carotenuto L, Schrader D, Lee HK, Riviello J, Hong W, Risen S, Emrick L, Amin H, Ville D, Edery P, de Bellescize J, Michaud V, Van-Gils J, Goizet C, Willemsen MH, Kleefstra T, **Møller RS**, **Bayat A**, Devinsky O, Sands T, Korenke GC, Kluger G, Mefford HC, Brilstra E, Lesca G, Milh M, Cooper EC, Taglialatela M, Weckhuysen S. KCNQ2 R144 variants cause neurodevelopmental disability with language impairment and autistic features without neonatal seizures through a gain-of-function mechanism. EBioMedicine. 2022 Jul;81:104130.
66. Brunklaus A, Brünger T, Feng T, Fons C, Lehikoinen A, Panagiotakaki E, Vintan MA, Symonds J, Andrew J, Arzimanoglou A, Delima S, Gallois J, Hanrahan D, Lesca G, MacLeod S, **Marjanovic D**, McTague A, Nuñez-Enamorado N, Perez-Palma E, Scott Perry M, Pysden K, Russ-Hall SJ, Scheffer IE, Sully K, Syrbe S, Vaher U, Velayutham M, Vogt J, Weiss S, Wirrell E, Zuberi SM, Lal D, **Møller RS**, Mantegazza M, Cestèle S. The gain of function SCN1A disorder spectrum: novel epilepsy phenotypes and therapeutic implications. Brain. 2022 Nov 21;145(11):3816-3831.
67. Guerrini R, Mei D, Kerti-Szigeti K, Pepe S, Koenig MK, Von Allmen G, Cho MT, McDonald K, Baker J, Bhambhani V, Powis Z, Rodan L, Nabbout R, Barcia G, Rosenfeld JA, Bacino CA, Mignot C, Power LH, Harris CJ, **Marjanovic D**, **Møller RS**, **Hammer TB**; DDD Study; Keski Filppula R, Vieira P, Hildebrandt C, Sacharow S; Undiagnosed Diseases Network; Maragliano L, Benfenati F, Lachlan K, Benneche A, Petit F, de Sainte Agathe JM, Hallinan B, Si Y, Wentzensen IM, Zou F, Narayanan V, Matsumoto N, Boncristiano A, la Marca G, Kato M, Anderson K, Barba C, Sturiale L, Garozzo D, Bei R; ATP6V1A collaborators; Masuelli L, Conti V, Novarino G, Fassio A. Phenotypic and genetic spectrum of ATP6V1A encephalopathy: a disorder of lysosomal homeostasis. Brain. 2022 Aug 27;145(8):2687-2703.
68. Christensen MB, Levy AM, Mohammadi NA, Niceta M, Kaiyrzhanov R, Dentici ML, Al Alam C, Alesi V, Benoit V, Bhatia KP, Bierhals T, Boßelmann CM, Buratti J, Callewaert B, Ceulemans B, Charles P, De Wachter M, Dehghani M, D'haenens E, Doco-Fenzy M, Geßner M, Gobert C, Guliyeva U, Haack TB, **Hammer TB**, Heinrich T, Hempel M, Herget T, Hoffmann U, Horvath J, Houlden H, Keren B, Kresge C, Kumps C, Lederer D, Lermine A, Magrinelli F, Maroofian R, Vahidi Mehrjardi MY, Moudi M, Müller AJ, Oostra AJ, Pletcher BA, Ros-Pardo D, Samarasekera S, Tartaglia M, Van Schil K, Vogt J, Wassmer E, Winkelmann J, Zaki MS, Zech M, Lerche H, Radio FC, Gomez-Puertas P, **Møller RS**, Tümer Z. Biallelic variants in ZNF142 lead to a syndromic neurodevelopmental disorder. Clin Genet. 2022 Aug;102(2):98-109.
69. Neri S, Mastroianni G, **Gardella E**, Aguglia U, **Rubboli G**. Epilepsy in neurodegenerative diseases. Epileptic Disord. 2022 Apr 1;24(2):249-273.
70. Gesche J, Cornwall CD, Delcomyn L, **Rubboli G**, Beier CP. Pseudoresistance in idiopathic/genetic generalized epilepsies - Definitions, risk factors, and outcome. Epilepsy Behav. 2022 May;130:108633. doi: 10.1016/j.yebeh.2022.108633.
71. Shakeshaft A, Panjwani N, Collingwood A, Crudgington H, Hall A, Andrade DM, Beier CP, Fong CY, **Gardella E**, Gesche J, Greenberg DA, Hamandi K, Koht J, Lim KS, **Møller RS**, Ng CC, Orsini A, Rees MI, **Rubboli G**, Selmer KK, Striano P, Syvertsen M, Thomas RH, Zarubova J, Richardson MP, Strug LJ, Pal DK. Sex-specific disease modifiers in juvenile myoclonic epilepsy. Sci Rep. 2022 Feb 21;12(1):2785. doi: 10.1038/s41598-022-06324-2.
72. **Rubboli G**, **Johannesen KM**. Expanding the phenotype of PURA-related developmental epileptic encephalopathy. Epileptic Disord. 2022 Apr 1;24(2):445-446.
73. Absalom NL, Liao VWY, **Johannesen KM**H, **Gardella E**, Jacobs J, Lesca G, Gokce-Samar Z, Arzimanoglou A, Zeidler S, Striano P, Meyer P, Benkel-Herrenbrueck I, Mero IL, Rummel J, Chebib M, **Møller RS**, Ahring PK. Gain-of-function and loss-of-function GABRB3 variants lead to distinct clinical phenotypes in patients with developmental and epileptic encephalopathies. Nat Commun. 2022 Apr 5;13(1):1822.
74. Beltrán-Corbellini Á, Aledo-Serrano Á, **Møller RS**, Pérez-Palma E, García-Morales I, Toledano R, Gil-Nagel A. Epilepsy Genetics and Precision Medicine in Adults: A New Landscape for Developmental and Epileptic Encephalopathies. Front Neurol. 2022 Feb 17;13:777115.
75. **Johannesen KM**, Liu Y, **Gardella E**, Lerche H, **Møller RS**. Reply: Genotype-phenotype correlations in SCN8A-related epilepsy: a cohort study of Chinese children in southern China. Brain. 2022 May 24;145(4):e28-e30.
76. Xian J, Parthasarathy S, Ruggiero SM, Balagura G, Fitch E, Helbig K, Gan J, Ganesan S, Kaufman MC, Ellis CA, Lewis-Smith D, Galer P, Cunningham K, O'Brien M, Cosico M, Baker K, Darling A, Veiga de Goes F, El Achkar CM, Doering JH, Furia F, García-Cazorla Á, **Gardella E**, Geertjens L, Klein C, Kolesnik-Taylor A, Lammertse H, Lee J, Mackie A, Misra-Isrie M, Olson H, Sexton E, Sheidley B, Smith L, Sotero L, Stamberger H, Syrbe S, Thalwitzer KM, van Berkel A, van

Haelst M, Yuskaitis C, Weckhuysen S, Prosser B, Son Rigby C, Demarest S, Pierce S, Zhang Y, **Møller RS**, Bruining H, Poduri A, Zara F, Verhage M, Striano P, Helbig I. Assessing the landscape of STXBP1-related disorders in 534 individuals. Brain. 2022 Jun 3;145(5):1668-1683.

77. **Bayat A**, de Valles-Ibáñez G, Pendziwiat M, Knaus A, Alt K, Biamino E, Bley A, Calvert S, Carney P, Caro-Llopis A, Ceulemans B, Cousin J, Davis S, des Portes V, Edery P, England E, Ferreira C, Freeman J, Gener B, Gorce M, Heron D, Hildebrand MS, Jezela-Stanek A, Jouk PS, Keren B, Kloth K, Kluger G, Kuhn M, Lemke JR, Li H, Martinez F, Maxton C, Mefford HC, Merla G, Mierzewska H, Muir A, Monfort S, Nicolai J, Norman J, O'Grady G, Oleksy B, Orellana C, Orec LE, Peinhardt C, Pronicka E, Rosello M, Santos-Simarro F, Schwaibold EMC, Stegmann APA, Stumpel CT, Szczepanik E, Terczyńska I, Thevenon J, Tzschach A, Van Bogaert P, Vittorini R, Walsh S, Weckhuysen S, Weissman B, Wolfe L, Reymond A, De Nittis P, Poduri A, Olson H, Striano P, Lesca G, Scheffer IE, **Møller RS**, Sadleir LG. PIGN encephalopathy: Characterizing the epileptology. Epilepsia. 2022 Apr;63(4):974-991.

78. **Bayat A**, Aledo-Serrano A, Gil-Nagel A, Korff CM, Thomas A, Boßelmann C, Weber Y, **Gardella E**, Lund AM, de Sain-van der Velden MGM, **Møller RS**. Pyridoxine or pyridoxal-5-phosphate treatment for seizures in glycosylphosphatidylinositol deficiency: A cohort study. Dev Med Child Neurol. 2022 Jun;64(6):789-798.

79. Brunklaus A, Pérez-Palma E, Ghanty I, Xinge J, Brilstra E, Ceulemans B, Chemaly N, de Lange I, Depienne C, Guerrini R, Mei D, **Møller RS**, Nabbout R, Regan BM, Schneider AL, Scheffer IE, Schoonjans AS, Symonds JD, Weckhuysen S, Kattan MW, Zuberi SM, Lal D. Development and Validation of a Prediction Model for Early Diagnosis of SCN1A-Related Epilepsies. Neurology. 2022 Mar 15;98(11):e1163-e1174.

80. Koko M, Motelow JE, Stanley KE, Bobbili DR, Dhindsa RS, May P; Canadian Epilepsy Network; Epi4K Consortium; Epilepsy Phenome/Genome Project; EpiPGX Consortium; **EuroEPINOMICS-CoGIE Consortium**. Association of ultra-rare coding variants with genetic generalized epilepsy: A case-control whole exome sequencing study. Epilepsia. 2022 Mar;63(3):723-735.

81. Krey I, von Spiczak S, **Johannesen KM**, Hikel C, Kurlemann G, Muhle H, Beysen D, Dietel T, **Møller RS**, Lemke JR, Syrbe S. L-Serine Treatment is Associated with Improvements in Behavior, EEG, and Seizure Frequency in Individuals with GRIN-Related Disorders Due to Null Variants. Neurotherapeutics. 2022 Jan;19(1):334-341.

82. Manivannan SN, Roovers J, Smal N, Myers CT, Turkdogan D, Roelens F, Kanca O, Chung HL, Scholz T, Hermann K, Bierhals T, Caglayan HS, Stamberger H; MAE Working Group of EuroEPINOMICS RES Consortium; Mefford H, de Jonghe P, Yamamoto S, Weckhuysen S, Bellen HJ. De novo FZR1 loss-of-function variants cause developmental and epileptic encephalopathies. Brain. 2022 Jun 3;145(5):1684-1697

83. Aledo-Serrano Á, Cabal-Paz B, **Gardella E**, Gómez-Porro P, Martínez-Múgica O, Beltrán-Corbellini A, Toledano R, García-Morales I, Gil-Nagel A. Effect of fenfluramine on seizures and comorbidities in SCN8A-developmental and epileptic encephalopathy: A case series. Epilepsia Open. 2022 Sep;7(3):525-531.

84. Liang D, **Gardella E**, Kragholm K, Polcwiartek C, Sessa M. The Relationship Between Valproate and Lamotrigine/Levetiracetam Use and Prognosis in Patients With Epilepsy and Heart Failure: A Danish Register-Based Study. J Card Fail. 2022 Apr;28(4):630-638.

85. Parenti I, Leitão E, Kuechler A, Villard L, Goizet C, Courdier C, **Bayat A**, Rossi A, Julia S, Bruel AL, Tran Mau-Them F, Nambot S, Lehalle D, Willems M, Lespinasse J, Ghoumid J, Caumes R, Smol T, El Chehadeh S, Schaefer E, Abi-Warde MT, Keren B, Afenjar A, Tabet AC, Levy J, Maruani A, Aledo-Serrano Á, Garming W, Milleret-Pignot C, Chassevent A, Koopmans M, Verbeek NE, Person R, Belles R, Bellus G, Salbert BA, Kaiser FJ, Mazzola L, Convers P, Perrin L, Piton A, Wiegand G, Accogli A, Brancati F, Benfenati F, Chatron N, Lewis-Smith D, Thomas RH, Zara F, Striano P, Lesca G, Depienne C. The different clinical facets of SYN1-related neurodevelopmental disorders. Front Cell Dev Biol. 2022 Dec 8;10:1019715.

86. Kolvenbach CM, Felger T, Schierbaum L, Thiffault I, Pastinen T, Szczepańska M, Zaniew M, Adamczyk P, **Bayat A**, Yilmaz Ö, Lindenberg TT, Thiele H, Hildebrandt F, Hinderhofer K, Moog U, Hilger AC, Sullivan B, Bartik L, Gnyś P, Grote P, Odermatt B, Reutter HM, Dworschak GC. X-linked variations in SHROOM4 are implicated in congenital anomalies of the urinary tract and the anorectal, cardiovascular and central nervous systems. J Med Genet. 2022 Nov 15:jmedgenet-2022-108738.

87. Loong L, Tardivo A, Knaus A, Hashim M, Pagnamenta AT, Alt K, Böhrer-Rabel H, Caro-Llopis A, Cole T, Distelmaier F, Edery P, Ferreira CR, Jezela-Stanek A, Kerr B, Kluger G, Krawitz PM, Kuhn M, Lemke JR, Lesca G, Lynch SA, Martinez F, Maxton C, Mierzewska H, Monfort S, Nicolai J, Orellana C, Pal DK, Płoski R, Quarrell OW, Rosello M, Rydzanicz M, Sabir A, Śmigiel R, Stegmann APA, Stewart H, Stumpel C, Szczepanik E, Tzschach A, Wolfe L, Taylor JC, Murakami Y, Kinoshita T, **Bayat A**, Kini U. Biallelic variants in PIGN cause Fryns syndrome, multiple congenital anomalies-hypotonia-seizures syndrome, and neurologic phenotypes: A genotype-phenotype correlation study. Genet Med. 2023 Jan;25(1):37-48.

88. Vitobello A, Mazel B, Lelianova VG, Zangrandi A, Petitto E, Suckling J, Salpietro V, Meyer R, Elbracht M, Kurth I, Eggermann T, Benlaouer O, Lall G, Tonevitsky AG, Scott DA, Chan KM, Rosenfeld JA, Nambot S, Safraou H, Bruel AL, Denommé-Pichon AS, Tran Mau-Them F, Philippe C, Duffourd Y, Guo H, Petersen AK, Granger L, Crunk A, **Bayat A**, Striano P, Zara F, Scala M, Thomas Q, Delahaye A, de Sainte Agathe JM, Buratti J, Kozlov SV, Faivre L, Thauvin-Robinet C, Ushkaryov Y. ADGRL1 haploinsufficiency causes a variable spectrum of neurodevelopmental disorders in humans and alters synaptic activity and behavior in a mouse model. Am J Hum Genet. 2022 Aug 4;109(8):1436-1457.

89. El Chehadeh S, Han KA, Kim D, Jang G, Bakhtiari S, Lim D, Kim HY, Kim J, Kim H, Wynn J, Chung WK, Vitiello G, Cutcutache I, Page M, Gecz J, Harper K, Han AR, Kim HM, Wessels M, **Bayat A**, Jaén AF, Selicorni A, Maitz S, de Brouwer APM, Silfhout AV, Armstrong M, Symonds J, Küry S, Isidor B, Cogné B, Nizon M, Feger C, Muller J, Torti E, Grange DK, Willems M, Kruer MC, Ko J, Piton A, Um JW. SLITRK2 variants associated with neurodevelopmental disorders impair excitatory synaptic function and cognition in mice. Nat Commun. 2022 Jul 15;13(1):4112.

90. Bayat M, **Bayat A**, Blauenfeldt RA. Atypical painful stroke presentations: A review. Acta Neurol Scand. 2022 Nov;146(5):465-474.

91. Ruaud L, Drunat S, Elmaleh-Bergès M, Ernault A, Guilmin Crepon S; **MCPH Consortium**; El Ghouzzi V, Auvin S, Verloes A, Passemard S. Neurological outcome in WDR62 primary microcephaly. Dev Med Child Neurol. 2022 Apr;64(4):509-517.

92. Aledo-Serrano A, Hariramani R, Gonzalez-Martinez A, Álvarez-Troncoso J, Toledano R, **Bayat A**, Garcia-Morales I, Becerra JL, Villegas-Martínez I, Beltran-Corbellini A, Gil-Nagel A. Anakinra and tocilizumab in the chronic phase of febrile infection-related epilepsy syndrome (FIRES): Effectiveness and safety from a case-series. Seizure. 2022 Aug;100:51-55

93. Peng SX, Pei J, Rinaldi B, Chen J, Ge YH, Jia M, Wang J, Delahaye-Duriez A, Sun JH, Zang YY, Shi YY, Zhang N, Gao X, Milani D, Xu X, Sheng N, Gerard B, Zhang C, **Bayat A**, Liu N, Yang JJ, Shi YS. Dysfunction of AMPA receptor GluA3 is associated with aggressive behavior in human. Mol Psychiatry. 2022 Oct;27(10):4092-4102.

94. Ismail V, Zachariassen LG, Godwin A, Sahakian M, Ellard S, Stals KL, Baple E, Brown KT, Foulds N, Wheway G, Parker MO, Lyngby SM, Pedersen MG, Desir J, **Bayat A**, Musgaard M, Guille M, Kristensen AS, Baralle D. Identification and functional evaluation of GRIA1 missense and truncation variants in individuals with ID: An emerging neurodevelopmental syndrome. Am J Hum Genet. 2022 Jul 7;109(7):1217-1241.

95. **Bayat A**, Krett B, Dunø M, Torring PM, Vissing J. Novel truncating variants in FGD1 detected in two Danish families with Aarskog-Scott syndrome and myopathic features. Am J Med Genet A. 2022 Jul;188(7):2251-2257.

96. Schwarz N, Seiffert S, Pendziwiat M, Rademacher AV, Brünger T, Hedrich UBS, Augustijn PB, Baier H, **Bayat A**, Bisulli F, Buono RJ, Bruria BZ, Doyle MG, Guerrini R, Heimer G, Iacomino M, Kearney H, Klein KM, Kousiappa I, Kunz WS, Lerche H, Licchetta L, Lohmann E, Minardi R, McDonald M, Montgomery S, Mulahasanovic L, Oegema R, Ortal B, Papacostas SS, Ragona F, Granata T, Reif PS, Rosenow F, Rothschild A, Scudieri P, Striano P, Tinuper P, Tanteles GA, Vetro A, Zahnert F, Goldberg EM, Zara F, Lal D, May P, Muhle H, Helbig I, Weber Y. Spectrum of Phenotypic, Genetic, and Functional Characteristics in Patients With Epilepsy With KCNC2 Pathogenic Variants. Neurology. 2022 May 17;98(20):e2046-e2059.

97. Lines MA, Goldenberg P, Wong A, Srivastava S, **Bayat A**, Hove H, Karstensen HG, Anyane-Yeboa K, Liao J, Jiang N, May A, Guzman E, Morleo M, D'Arrigo S, Ciaccio C, Pantaleoni C, Castello R; TUDP Study Group; McKee S, Ong J, Zibdeh-Lough H, Tran-Mau-Them F, Gerasimenko A, Heron D, Keren B, Margot H, de Sainte Agathe JM, Burglen L, Voets T, Vriens J, Innes AM, Dyment DA. Phenotypic spectrum of the recurrent TRPM3 p.(Val837Met) substitution in seven individuals with global developmental delay and hypotonia. Am J Med Genet A. 2022 Jun;188(6):1667-1675

98. Bayat M, **Bayat A**. Neurological manifestation of 22q11.2 deletion syndrome. Neurol Sci. 2022 Mar;43(3):1695-1700

99. Hamanaka K, Miyoshi K, Sun JH, Hamada K, Komatsubara T, Saida K, Tsuchida N, Uchiyama Y, Fujita A, Mizuguchi T, Gerard B, **Bayat A**, Rinaldi B, Kato M, Tohyama J, Ogata K, Shi YS, Saito K, Miyatake S, Matsumoto N. Amelioration of a neurodevelopmental disorder by carbamazepine in a case having a gain-of-function GRIA3 variant. Hum Genet. 2022 Feb;141(2):283-293

100. Cherik F, Reilly J, Kerkhof J, Levy M, McConkey H, Barat-Houari M, Butler KM, Coubes C, Lee JA, Le Guyader G, Louie RJ, Patterson WG, Tedder ML, Bak M, **Hammer TB**, Craigen W, Démurger F, Dubourg C, Fradin M, Franciskovich R, Frengen E, Friedman J, Palares NR, Iascone M, Misceo D, Monin P, Odent S, Philippe C, Rouxel F, Saletti V, Strømme P, Thulin PC, Sadikovic B, Genevieve D. DNA methylation episignature in Gabriele-de Vries syndrome. Genet Med. 2022 Apr;24(4):905-914.

101. Ramond F, Dalgliesh C, Grimmel M, Wechsberg O, Vetro A, Guerrini R, FitzPatrick D, Poole RL, Lebrun M, **Bayat A**, Grasshoff U, Bertrand M, Witt D, Turnpenny PD, Faundes V, Santa María L, Mendoza Fuentes C, Mabe P, Hussain SA, Mullegama SV, Torti E, Oehl-Jaschkowitz B, Salmon LB, Orenstein N, Shahar NR, Hagari O, Bazak L, Hoffjan S, Prada CE, Haack T, Elliott DJ. Clustered variants in the 5′ coding region of TRA2B cause a distinctive neurodevelopmental syndrome. Genet Med. 2022 Dec 20;25(4):100003

102. Houtman SJ, Lammertse HCA, van Berkel AA, Balagura G, **Gardella E**, Ramautar JR, Reale C, **Møller RS**, Zara F, Striano P, Misra-Isrie M, van Haelst MM, Engelen M, van Zuijen TL, Mansvelder HD, Verhage M, Bruining H, Linkenkaer-Hansen K. STXBP1 Syndrome Is Characterized by Inhibition-Dominated Dynamics of Resting-State EEG. Front Physiol. 2021 Dec 23;12:775172. doi: 10.3389/fphys.2021.775172. PMID: 35002760.

103. **Johannesen KM**, Iqbal S, Guazzi M, Mohammadi NA, Pérez-Palma E, Schaefer E, De Saint Martin A, Abiwarde MT, McTague A, Pons R, Piton A, Kurian MA, Ambegaonkar G, Firth H, Sanchis-Juan A, Deprez M, Jansen K, De Waele L, Briltra EH, Verbeek NE, van Kempen M, Fazeli W, Striano P, Zara F, Visser G, Braakman HMH, Haeusler M, Elbracht M, Vaher U, Smol T, Lemke JR, Platzer K, Kennedy J, Klein KM, Au PYB, Smyth K, Kaplan J, Thomas M, Dewenter MK, Dinopoulos A, Campbell AJ, Lal D, Lederer D, Liao VWY, Ahring PK, **Møller RS**, **Gardella E**. Structural mapping of GABRB3 variants reveals genotype-phenotype correlations. Genet Med. 2022;24(3):681-693. doi: 10.1016/j.gim.2021.11.004.PMID: 34906499.

104. **Johannesen KM**, **Gardella E**, Gjerulfsen CE, **Bayat A**, Rouhl RPW, Reijnders M, Whalen S, Keren B, Buratti J, Courtin T, Wierenga KJ, Isidor B, Piton A, Faivre L, Garde A, Moutton S, Tran-Mau-Them F, Denommé-Pichon AS, Coubes C, Larson A, Esser MJ, Appendino JP, Al-Hertani W, Gamboni B, Mampel A, Mayorga L, Orsini A, Bonuccelli A, Suppiej A, Van-Gils J, Vogt J, Damioli S, Giordano L, Moortgat S, Wirrell E, Hicks S, Kini U, Noble N, Stewart H, Asakar S, Cohen JS, Naidu SR, Collier A, Brilstra EH, Li MH, Brew C, Bigoni S, Ognibene D, Ballardini E, Ruivenkamp C, Faggioli R, Afenjar A, Rodriguez D, Bick D, Segal D, Coman D, Gunning B, Devinsky O, Demmer LA, Grebe T, Pruna D, Cursio I, Greenhalgh L, Graziano C, Singh RR, Cantalupo G, Willems M, Yoganathan S, Góes F, Leventer RJ, Colavito D, Olivotto S, Scelsa B, Andrade AV, Ratke K, Tokarz F, Khan AS, Ormieres C, Benko W, Keough K, Keros S, Hussain S, Franques A, Varsalone F, Grønborg S, Mignot C, Heron D, Nava C, Isapof A, Borlot F, Whitney R, Ronan A, Foulds N, Somorai M,

Brandsema J, Helbig KL, Helbig I, Ortiz-González XR, Dubbs H, Vitobello A, Anderson M, Spadafore D, Hunt D, **Møller RS**, **Rubboli G**; PURA study group. PURA-Related Developmental and Epileptic Encephalopathy: Phenotypic and Genotypic Spectrum. Neurol Genet. 2021;7(6):e613. doi: 10.1212/NXG.0000000000000613. PMID: 34790866.

105. Proietti J, Amadori E, Striano P, Ricci E, Cordelli DM, Bana C, Dilena R, **Gardella E**, Klint Nielsen JE, Pisani F, Lo Barco T, Fiorini E, Fontana E, Darra F, Dalla Bernardina B, Cantalupo G. Epilepsy features in ARID1B-related Coffin-Siris syndrome. Epileptic Disord. 2021 Dec 1;23(6):865-874. doi: 10.1684/epd.2021.1356. PMID: 34730517.

106. Ahring PK, Liao VWY, **Gardella E**, **Johannesen KM**, Krey I, Selmer KK, Stadheim BF, Davis H, Peinhardt C, Koko M, Coorg RK, Syrbe S, Bertsche A, Santiago-Sim T, Diemer T, **Fenger CD**, Platzer K, Eichler EE, Lerche H, Lemke JR, Chebib M, **Møller RS**. Gain-of-function variants in GABRD reveal a novel pathway for neurodevelopmental disorders and epilepsy. Brain. 2022;145(4):1299-1309. doi: 10.1093/brain/awab391. PMID: 34633442.

107. **Johannesen KM**, Liu Y, Koko M, Gjerulfsen CE, Sonnenberg L, Schubert J, **Fenger CD**, Eltokhi A, Rannap M, Koch NA, Lauxmann S, Krüger J, Kegele J, Canafoglia L, Franceschetti S, Mayer T, Rebstock J, Zacher P, Ruf S, Alber M, Sterbova K, Lassuthová P, Vlckova M, Lemke JR, Platzer K, Krey I, Heine C, Wieczorek D, Kroell-Seger J, Lund C, Klein KM, Au PYB, Rho JM, Ho AW, Masnada S, Veggiotti P, Giordano L, Accorsi P, Hoei-Hansen CE, Striano P, Zara F, Verhelst H, Verhoeven JS, Braakman HMH, van der Zwaag B, Harder AVE, Brilstra E, Pendziwiat M, Lebon S, Vaccarezza M, Le NM, Christensen J, Grønborg S, Scherer SW, Howe J, Fazeli W, Howell KB, Leventer R, Stutterd C, Walsh S, Gerard M, Gerard B, Matricardi S, Bonardi CM, Sartori S, Berger A, Hoffman-Zacharska D, Mastrangelo M, Darra F, Vøllo A, Motazacker MM, Lakeman P, Nizon M, Betzler C, Altuzarra C, Caume R, Roubertie A, Gélisse P, Marini C, Guerrini R, Bilan F, Tibussek D, Koch-Hogrebe M, Perry MS, Ichikawa S, Dadali E, Sharkov A, Mishina I, Abramov M, Kanivets I, Korostelev S, Kutsev S, Wain KE, Eisenhauer N, Wagner M, Savatt JM, Müller-Schlüter K, Bassan H, Borovikov A, Nassogne MC, Destrée A, Schoonjans AS, Meuwissen M, Buzatu M, Jansen A, Scalais E, Srivastava S, Tan WH, Olson HE, Loddenkemper T, Poduri A, Helbig KL, Helbig I, Fitzgerald MP, Goldberg EM, Roser T, Borggraefe I, Brünger T, May P, Lal D, Lederer D, **Rubboli G**, Heyne HO, Lesca G, Hedrich UBS, Benda J, **Gardella E**, Lerche H, **Møller RS**. Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. Brain. 2022;145(9):2991-3009. doi: 10.1093/brain/awab321. PMID: 34431999.

108. **Moos**, **T. A. O.** & Rydahl-Hansen, S., 'Everything is as before, but nothing is as it was' - A phenomenological-hermeneutic study of meaningfulness in adult patients with refractory epilepsy after interdisciplinary epilepsy rehabilitation 2021 Sept, Epilepsy and Behav.2021;122:108168. DOI:https://doi.org/10.1016/j.yebeh.2021.108168

109. **Larsen JB**, Jørgensen S. Simple and Robust Detection of CYP2D6 Gene Deletions and Duplications Using CYP-2D8P as Reference. Pharmaceuticals (Basel). 2022 Jan 28;15(2):166. doi: 10.3390/ph15020166. PMID: 35215279; PMCID: PMC8880347.

110. **Larsen JB**, Populationsbaserede terapeutiske referenceintervaller ud fra retrospektive laboratoriedata DSKB-Nyt: Nr 1 – Febuar 2022, s.24- s.27

111. **Hjelm, C** (2021 [2022]): The Human Related Conversation and The Professional Tension Between Diakonia and Nursing. Diaconia, vol. 12, p. 137–160. doi:10.13109/diac.2021.12.2.137

7. Lectures - oral presentations in 2022

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
- ILAE - European Epilepsy Congress (Geneva, July 9-12, 2022)
  Interactive, half-day EEG teaching course
  EEG source imaging: basic principles and clinical applications
- Dianalund Summer School on EEG and Epilepsy (5th edition, July 16-23, 2022)
  Systematic approach to clinical EEG reading
  Standardized Computer-based Organized Reporting of EEG
  Electromagnetic Source Imaging
- ILAE – Epilepsy Summer School (Slanic Prahova, September 2, 2022)
  Seizure semiology: ILAE glossary of terms and their significance
- International Congress of Clinical Neurophysiology (Geneva, September 3-8, 2022)
- How to Use Voltage Maps to Estimate the Source
  Interictal Epileptiform Discharges: What's New for This Old Stuff?
  Automated and Semi-Automated Source Imaging
- Epilepsy colloquium (Lausanne, September 14-16, 2022)
  Pitfalls in ictal scalp-EEG interpretation
  Wearable devices for automated seizure detection
- The 55th Annual Congress of the Japan Epilepsy Society (Sendai, September 21, 2022)
  A pragmatic algorithm to classify seizures without EEG
- ILAE – Latin-American Epilepsy Congress (Medellin, October 4, 2022)
  Electric Source Imaging of scalp recordings: impact on patient management.
- Annual meeting of the ILAE British Chapter (Cardiff, October 13, 2022)
  The Salzburg consensus criteria for non-convulsive status epilepticus
- 4th International video-EEG course in pediatric epilepsies (Madrid, October 29, 2022)
  The role of source analysis in epilepsy surgery
- Annual meeting of the Danish Epilepsy Society (Copenhagen, November 4, 2022).
  Is this epilepsy? Or not? An interactive video-EEG session.
- ILAE - Asian & Oceanian Epilepsy Congress (online, November 18, 2022)
  Can seizure detection devices reduce mortality?
- Forum of Excellence in Epilepsy (London, November 19, 2022)
  AI interpretation of EEG in epilepsy
- Epilepsy symposium (Szeged, November 24-25, 2022)
  EEG interpretation in clinical practice
  Seizure semiology and classification
- Annual meeting of the American Epilepsy Society (Nashville, December 6, 2022)
  What is an epileptiform discharge?
- Conference of the Egyptian Epilepsy Society (online, December 23, 2022).
  Accurate identification of interictal epileptiform discharges.

Rikke Steensbjerre Møller:

- Personalized medicine in epilepsy – upcoming new treatments, Annual Meeting of the Danish Society of Epileptology, Copenhagen, Denmark
- Importance of epilepsy genetics for diagnosis, treatment, and prognosis. 4th Annual Meeting between the Danish and the Norwegian Epilepsy Centre, Copenhagen, Denmark
- Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. Childhood Epilepsy and Movement Disorders Workshop, Barcelona, Spain
- Experience with precision medicine for epilepsy in a Danish tertiary reference center. Impact and hurdles. SEED conference 2022, Tunis, Tunisia
- Future perspectives of genetics in epilepsy. Educational course. The Chilean Society of Neurology and Psychiatry of Childhood and Adolescence (SOPNIA) (Virtual)
- Precision Medicine in Genetic Epilepsies: Challenges and Opportunities. 60th Annual Meeting of the German Society of Epileptology, Leipzig, Germany
- Utility of genetic testing for therapeutic decision-making in individuals with epilepsy. University of Washington, Saint Louis (Virtual)
- Sodium channelopathies (SCN2A-SCN8A). Genetic Epilepsies and Precision Medicine, EPIGENS (Virtual)
- Developmental and epileptic encephalopathies: genetic diagnosis in adolescents and adults. Curso de Invierno de Epilepsia, La Granja, Spain

Guido Rubboli:

- "Group Discussion on Topics: Surgery", Speaker at the Nordic Experience Sharing Meeting, Korsør, 6 April 2022.
- "ESES: an update", speaker, virtual meeting, University of Padova, 31 May 2022.
- "Genetics in epilepsy - children and adults. The clinician's perspective." National Anniversary symposium Oslo 9-10 June, 2022
- "Half-day Teaching Course: Genetic testing: whom, when and what to test", European Epilepsy Congress, Geneva 9 July 2022
- "Ultra-long sub-cutaneous EEG monitoring: reliability, safety and impact on clinical management in uncon-

trolled epilepsies. Our experience with 24/7 EEG SubQ24: preliminary results. Speaker in collaboration with S. Beniczky. Meeting sponsored by UNEEG at the European Epilepsy Congress, Geneva, 11 July 2022
- "Teaching Session: Video session – adult", European Epilepsy Congress, , Geneva 13 July 2022
- "Polygraphic recordings" at the 5th ed. Dianalund Summer School on EEG and Epilepsy, Dianalund, 16-23 July 2022, in collaboration with E. Gardella.
- "Erfaringer med PER som tidlig tillæg i klinisk praksis" webinar meeting, Speaker 6 september 2022
- "Understanding pathophysiology and improving outcomes in Electrical Status epilepticus during Sleep (ESES)", 8th London-Innsbruck Colloquium on status epilepticus and acute seizures, Salzburg, 17-20 September 2022
- "EPI_PED course. Genetic and EEG comparison in pediatric epilepsy", speaker and tutor, Bologna, 9-13 October 2022.
- "Photosensitivity in epileptic syndromes" and "From bench-to-bedside II. Interactive presentation of cases. Cases with absences, tonic, atonic and myoclonic seizures", Video-EEG in pediatric epilepsies, "From seizures to syndromes", Madrid, 27-29 October 2022.
- "La classificazione neurofisiologica del mioclono - circuiti sottesi", Diagnostic working group of the Italian League Against Epilepsy, Rome, 17-18 November 2022.
- "Introduction" at the 4th Annual Meeting Danish Epilepsy Center and Norwegian Epilepsy Center " Epilepsy syndromes 2022", Copenhagen , 10 November 2022

Elena Gardella:

- Fuori dallómbra, Padova, Italy
- 5th Dianalund International Conference on Epilepsy (DICE), Køge, Denmark
- LEGOLAS annual meeting, Amsterdam, The Nederland
- 14th European Epilepsy Congress (EEG); Geneva, Switzerland
- STXBP1 Summit and family meeting
- 4th Annual Meeting DEC and NEC, Copenhagen, Denmark
- Nordic Epilepsy Sharing meeting on personalized medicine (part VI)- Natural History Studies and drug trials readiness, Copenhagen, Denmark
- Annual Meetig of the CUTE syndrome foundation / American Epilepsy Society meeting, Nashville, USA
- 5th Dianalund Summer School on EEG and Epilepsy (DSSEES), Dianalund, Denmark
- 9th International Residential Course on Drug Resistant Epilepsies, Tagliacozzo, Italy
- EPIPED-EEG course "Genetic and EEG comparison in pediatric epilepsy", Bologna, Italy
- EpiCARE Workshop: Childhood Epilepsy and Movement Disorders Translational Medicine and Novel Therapeutic Approaches; Barcelona, Spain
- Video-EEG in pediatric epilepsies Madrid

Marina Nikanorova:

- How do we choose ASMs for refractory epilepsies in childhood – online meeting with Finnish neuropediatricians, 08.03.22
- Clinical experiences with Fenfluramin in Denmark – Advisory Board, 28.06.22
- Early adjustment of Perampanel in refractory epilepsy in children and adults – Advisory Board, 06.09.22
- ILAE Epiwebtutorial – 20.09.22 (Case-oriented learning)
- 15th Baltic Sea Summer School on Epilepsy – Case-oriented learning, 3 online sessions, September 2022
- Epileptic encephalopathies of childhood: outcome in adults – meeting of the Lithuanian ILAE Chapter, 14.10.22
- Diagnosing, treating and managing patients with Lennox-Gastaut syndrome – online Nordic meeting for neuropediatricians and neurologists (Danmark, Finland, Norway, Sweden), 25.10.22

Allan Bayat:

- GRIA3 related disorders. ILAE-Europe Epilepsy workshop. Rome.
- Two electrode voltage clamping – TEVC. ERN-ITHACA Research workgroup. Budapest.
- Understanding GPI anchoring disorders, precision medicine and unwinding the cause of a high childhood mortality. LEGOLAS-meeting, Amsterdam.
- Impact of genetic testing on therapeutic decision making in childhood-onset epilepsies. European conference on epilepsy, ILAE, Geneve, Switzerland.
- Pyridoxine or pyridoxal-5-phosphate treatment for seizures in glycosylphosphatidylinositol deficiency: A cohort study. European conference on epilepsy, ILAE, Geneve, Switzerland.
- Phenotypical and functional assessment of four novel KCNQ2 gain-of-function variants and effects of amitriptyline treatment. The 5th Dianalund International Conference on Epilepsy.

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