FILADELFIA

ANNUAL RESEARCH REPORT 2019

Content

1. Introduction – Filadelfia.

This is Filadelfia`s annual Research report for 2019. – Again a pleasure to introduce the growth within the research projects and papers. The Research Report focuses on projects and papers finalized in 2019 and thereby it does not include ongoing projects and papers.

The Danish Epilepsy Center Filadelfia is a non-profit foundation with an independent Board of Directors, and more than120 years old tradition of medical treatment of patients with epilepsy. It is publically funded and integral part of the Danish Healthcare System.

It is the only specialized hospital in Denmark treating epilepsy and concomitant disorders as well as neurorehabilitation treatment of patients with acquired brain injury.

Besides the epilepsy hospital, Filadelfia comprises specialized social institutions within rehabilitation and the only special school for children and young people with epilepsy in Denmark. 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.

Filadelfia´s expertise is based on many years of specialization in diagnosis and treatment of patients with severe epilepsy.

Long-standing specialized focus has resulted in the attainment of a high level of medical expertise in the fields of diagnostics, treatment, special care, attendance and support of patients diagnosed with epilepsy and/or acquired brain injury.

The hospital has close collaboration with several international centers and our specialists have several positions of trust in national and international scientific societies. We have close collaboration with other Danish research teams as well as Copenhagen, Aarhus and Odense Universities. Several of Filadelfia´s experts have academic affiliations as professors and associated professors at these universities.

Within the broader discipline of epilepsy care there is an ongoing process of also constituting a systematic research approach towards patient care, family perspectives and quality of life. 2019 became the year when systematic and wide steps were taken towards these fields, and we are looking forward to fulfill and strengthen the work in 2020.

2. Core Research Team 2019

Sándor Beniczky
MD and PhD from University of Szeged, Hungary (1997, 2004)
Specialist in Neurology (2002).
Specialist in Clinical Neurophysiology (2006).
European certification as epileptologist (2010).
Fellow of the European Academy of Neurology (2020)
Current position: Professor, consultant, head of department
Editor-in-chief, Epileptic Disorders

Email: sbz@filadelfia.dk

Guido Rubboli
MD and PhD from University of Bologna, Italy
Specialist in Neurology
WHO course in Epileptology and clinical electroencephalography (1985)
Training in experimental and clinical neurophysiology, Chicago USA (1989-91)
Associate Editor: Epileptic Disorders, Behavioral Neurology
Co-Chair: Epilepsy Scientific Panel, European Academy of Neurology
Current position: Professor

Email: guru@filadelfia.dk

Rikke Steensbjerre Møller
M.Sc. Biomedicine from University of Southern Denmark (2003)
PhD in medical genetics from University of Copenhagen (2008)
Specialist in epilepsy genetics
Current Position: Professor, head of department of Epilepsy Genetics and Personalized Medicine

Email: rimo@filadelfia.dk

Elena Gardella
MD and PhD from University of Bologna, Italy (1995, 2003)
Specialist in Neurology (2001). Post doctoral, clinical neurophysiology (2005)
Member of the faculty of 1000 of the ILAE (epilepsy teaching)
Clinical Neurophysiologist 2012-present
Current position: Associate Professor, Senior consultant – Department of Clinical Neurophysiology, Danish Epilepsy Center

Email: elga@filadelfia.dk

Marina Nikanorova
MD and PhD, Moscow Medical University, Russia (1988 and 1999)
Specialist in Neurology (1988)
Neurologist – 1988 - present
Current position: Associate Professor, SDU 2010 – present, senior consultant – Children Department, Danish Epilepsy Center.

Email: mnk@filadelfia.dk

3. Graphic summary - research 2019

4. Positions of Trust

Professor Sàndor Beniczky

- Executive board member and treasurer of ILAE-Europe
- Chair of the IFCN-ILAE EEG taskforce
- Member of the Epilepsy Education Council of the ILAE
- Member of the Diagnostic Methods commission of the ILAE

Professor Guido Ruboli

- Member of the “Sleep and Epilepsy” Task Force of the ILAE (International League Against Epilepsy) ESRS (European Sleep research Society) – EFNS (European Federation of Neurological Societies), 2013-present.
- Member of the ILAE Task Force “Transitions in Child to Adult Care”, 2017-2022.
- Chair of the Epilepsy Scientific Panel, European Academy of Neurology, 2018-present.
- Member of the Scientific Committee “Fuori dall’ombra” - Insieme per l’Epilessia”, 2019 - present

Editorial boards

- Epileptic Disorders - Official Journal of ILAE: Associate Editor since 2017
- Behavioral Neurology: Section Editor since 2015
- Clinical Cases and Reviews in Epilepsy: member of the Editorial Board, since 2014
- Frontiers in Pediatric Neurology, Review Editor since 2018.

Associate Professor Rikke Steensbjerre Møller

- Member of EpiCARE: a European Reference Network for rare and complex epilepsies. Contact person for WP 2: Laboratory Diagnostics, 2019-present
- Member of the leadership team at Department of Regional Health Research, University of Southern Denmark, Odense, Denmark, 2018-present
- Member of the scientific committee of Residras; a European Registry of Dravet Syndrome, Verona, Italy, 2018-present

Associate Professor Elena Gardella

- Executive board member of The Danish Epilepsy Society
- Co-director of the educational program (VIREPA basic-EEG) of the ILAE
- Member of the “Faculty of one thousand for teaching and education” of the ILAE
- Review Editor of Frontiers in Pediatric Neurology, since 2018
- Consultant for Xenon pharma and Neurocrine and adviser for Praxis, all these companies are developing molecules for the selective blockage of SCN8A (Precision medicine of SCN8A-related diseases).

Research Nurse/Coordinator Vibeke Stubbings

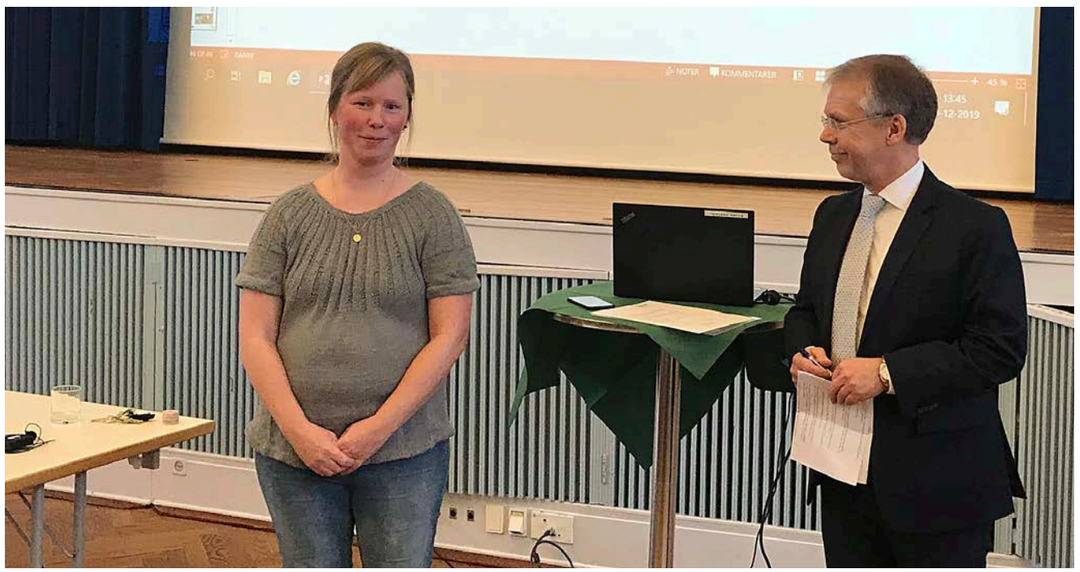
- Board member of the Danish Family Nursing Association, constituted under University Odense.

5. PhD Projekts

Defending thesis

MD, PhD. Katrine Marie Johannesen.
Defended her thesis dec. 2019 at Filadelfia on the topic:

Genotype-phenotype correlations in severe epileptic encephalopathies; With special focus on channelopathies.



Present PhD students

Neuropsychologist. Anne Vagner Jacobsen

Defence 2021.
Main supervisor: Professor Ask Elklit.
Co-supervisor: Clinical Associate Professor Marina Nikanorova,and Associate professor Rikke S Møller Filadelfia.
Mapping of experienced stressors and resourcefulness in caregivers of children with severe epilepsy for a better patient centered treatment and care.

Diacon, teacher Conny Hjelm

Defence 2021.
Main supervisor; Anne Austad and Marianne Rodriguez Nygaard. VID Vitenskapelig høgskole, Oslo.
Co supervisor; Mads R.Henriksen.
How does health staff and patients experience the human related conversation in a health care context at Filadelfia.

MD, Allan Bayat

Defence 2022.
Main supervisor: Rikke S Møller. Co supervisors: Guido Rubolli and Elena Gardella. Filadelfia.
Monogenic epilepsies and personalized medicine.

Supervision of PhD students - external

Sándor Beniczky, Professor

- Main supervisor for Mustafa Aykut Kural, at Aarhus University. Thesis: Clinical practice of EEG re-visited: “Improved spike identification, localization and characterization”, Defence in 2021.
- As a guest professor of University of Szeged, Main supervisor of 2 PhD students; Laura Craciun and Bogdan Florea. The students have performed their scientific work in collaboration with the Epilepsy Hospital, and had several fellowship periods of 3-6 month in Dianalund.
- Co -supervisor of PhD student; Cheng-Teng Ip at Copenhagen University.

Guido Rubboli, Professor

- “Endophenotyping patients of genetic generalized epilepsy – a population-based study”(in collaboration with Syddansk Universitet/ Odense). Joanna Gesche, Defence 2020. Co-supervisor

6. Research Projects

Department of Epilepsy Genetics and Personalized Medicine

In department of Epilepsy Genetics and Personalized Medicine pediatricians, neurologists, neurophysiologists, geneticists and basic scientists work together on projects including:

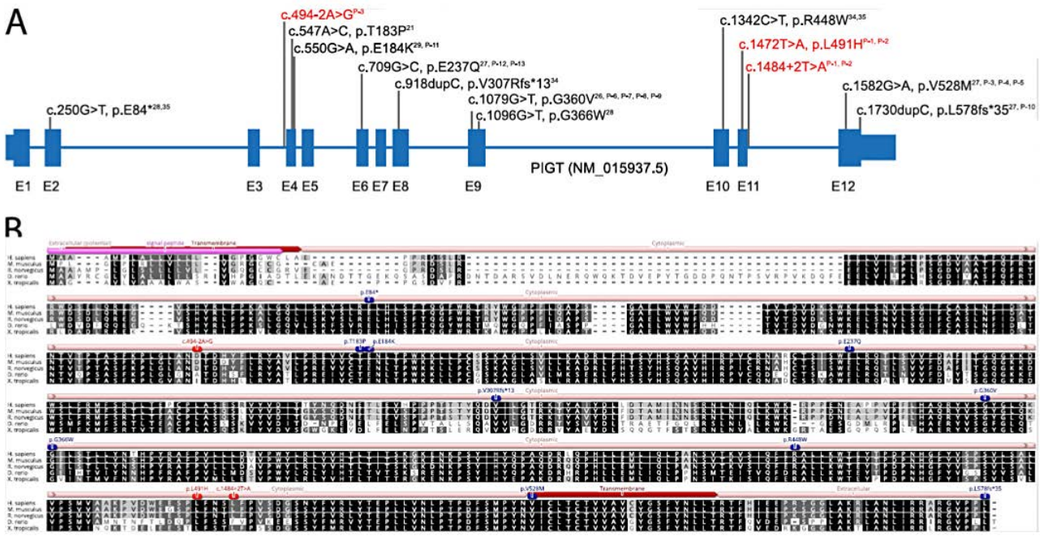
- Gene discovery in neurodevelopmental disorders and epilepsy.
- Genotype-phenotype correlation, including electro-clinical characterization of genetic epilepsy syndromes.
- Functional characterization of genetic defects to understand their pathomechanisms which may lead to improvement of existing or development of new personalized therapies.

Following examples illustrates finalized projects in 2019.

PIGT-CDG, a disorder of the glycosylphosphatidylinositol anchor: description of thirteen novel patients and expansion of the clinical characteristics.

The aim of the study was to provide a detailed electroclinical description and expand the phenotype of PIGT-CDG, and to use computer-assisted facial gestalt analysis to compare the findings with other glycosylphosphatidylinositol (GPI) anchor deficiencies. We evaluated 13 children from eight unrelated families with homozygous or compound heterozygous pathogenic variants in PIGT.

All patients had hypotonia, severe developmental delay, and epilepsy. Epilepsy onset ranged from first day of life to two years of age. Severity of the seizure disorder varied from treatable seizures to severe neonatal onset epileptic encephalopathies. We also showed that computer-assisted facial gestalt analysis accurately assigned PIGT cases to the multiple congenital anomalies-hypotonia-seizures syndrome phenotypic series advocating the additional use of next-generation phenotyping technology.



Paper:

Bayat A, Knaus A, Juul AW, Dukic D, **Gardella E**, Charzewska A, Clement E, Hjalgrim H, Hoffman-Zacharska D, Horn D, Horton R, Hurst JA, Josifova D, Larsen LHG, Lascelles K, Obersztyn E, Pagnamenta A, Pal DK, Pendziwiat M, Ryten M, Taylor J, Vogt J, Weber Y, Krawitz PM, Helbig I, Kini U, **Møller RS**. PIGT-CDG, a disorder of the glycosylphosphatidylinositol anchor: description of thirteen novel patients and expansion of the clinical characteristics. Genetics in Medicine, 2019 Oct;21(10):2216-2223.

The phenotypic spectrum of SCN8A-related disorders

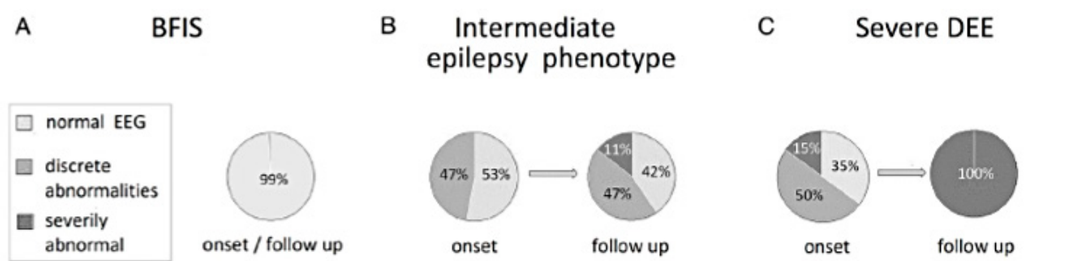
Pathogenic variants in *SCN8A* have originally been described in patients with developmental and epileptic encephalopathy (DEE). However, recent studies have shown that *SCN8A* variants can be associated with a broader phenotypic spectrum, including the following: (1) Patients with early onset, severe DEE, developing severe cognitive and motor regression, pyramidal/extrapyramidal signs, and cortical blindness. Severe *SCN8A*-DEE is characterized by intractable seizures beginning in the first months of life. The seizures are often prolonged focal hypomotor and occur in clusters, with prominent vegetative symptoms (apnea, cyanosis, mydriasis), evolving to clonic or bilateral tonic-clonic manifestations. Spasm-like episodes, cortical myoclonus, and recurrent episodes of status epilepticus are also common. Electroencephalograms (EEGs) show progressive background deterioration and multifocal abnormalities, predominant in the posterior regions. (2) Sporadic and familial patients with mild-to-moderate intellectual disability, discrete neurological signs, and treatable epilepsy. EEG is abnormal in half of the cases, showing multifocal or diffuse epileptiform abnormalities. (3) Familial cases with benign infantile seizures, sometimes associated with paroxysmal dyskinesia later in life, with no other neurological deficits, normal cognition, and usually normal interictal EEG. (4) Patients without epilepsy but with cognitive and/or behavioral disturbances, or with movement disorders. Extrapyramidal features, such as dyskinesia, ataxia, and choreoathetosis are common in all groups. Early death has been reported in about 5% of the patients, most often in the subgroup of severe DEE. Premature death occurs during early childhood and often for causes other than sudden unexpected death in epilepsy. All epilepsy subgroups exhibit better seizure control with sodium channel blockers, usually at supratherapeutic doses in the severe cases. In severe SCN8A-DEE, ketogenic diet often has a good effect, whereas levetiracetam has a negative effect, if any. The familial SCN8A-related epilepsies show an autosomal dominant pattern of inheritance, whereas the vast majority of *SCN8A*-DEEs occur de novo.

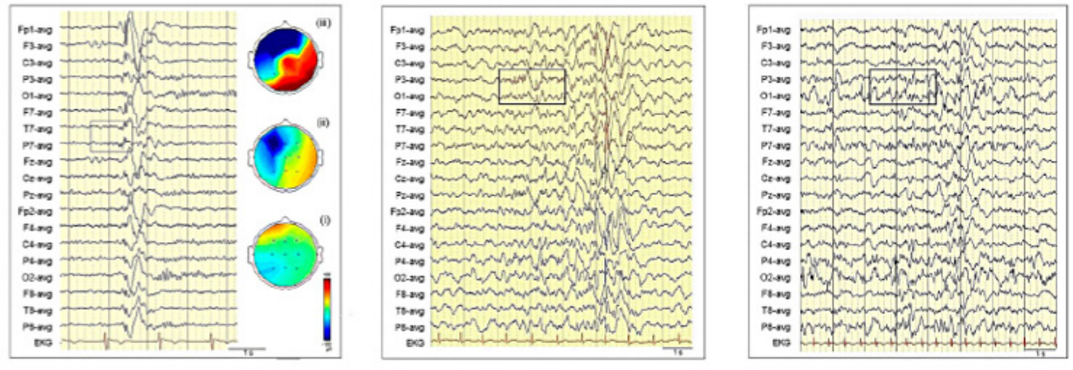
Paper:

Johannesen KM, Gardella E, Encinas AC, Lehesjoki AE, Linnankivi T, Petersen MB, Lund ICB, Blichfeldt S, Miranda MJ, Pal DK, Lascelles K, Procopis P, Orsini A, Bonuccelli A, Giacomini T, Helbig I, Fenger CD, Sisodiya SM, Hernandez-Hernandez L, Krithika S, Rumple M, Masnada S, Valente M, Cereda C, Giordano L, Accorsi P, Bürki SE, Mancardi M, Korff C, Guerrini R, von Spiczak S, Hoffman-Zacharska D, Mazurczak T, Coppola A, Buono S, Vecchi M, Hammer MF, Varesio C, Veggiotti P, Lal D, Brünger T, Zara F, Striano P, **Rubboli G, Møller RS**. The spectrum of intermediate SCN8A-related epilepsy. Epilepsia, 2019 60(5):830-844.

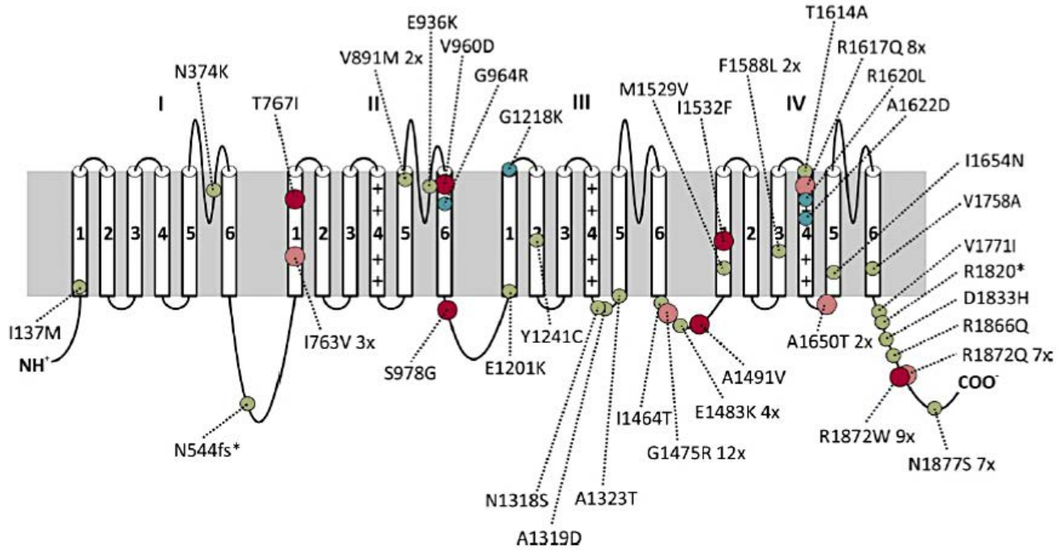
Gardella E, Møller RS, Phenotypic and genetic spectrum of SCN8A related disorders, treatment options and outcomes. Epilepsia. 2019 Dec;60 Suppl 3:S77-S85.

Musto E, Gardella E, Møller RS. Recent advances in treatment of epilepsy-related sodium channelopathies. Eur J Paediatr Neurol. 2020 Jan;24:123-128. Epub 2019 Dec 18.





	BFIS	Intermediate	Severe DEE
Age at onset	4-13 mo	14 mo (1.5 mo - 7 y)	4 mo (1 d - 3 y)
Sz Type	F (apnea) -> bilat TCS	TCS, M, F, Abs, T, AT, Sp	F (apnea) -> bilat TCS, T, Sp, M
Cognition / behavior	19 normal / 1 mild ID	Mild/moderate ID +/- autistic features	Severe / profound ID (+/- regression)
Outcome	All sz free	Ca. 50% sz free	Rare / short periods sz freedom
Neurological symptoms	Paroxysmal Kinesigenic Dykinesia	Ataxia, hypotonia, fine motor imp, speech delay, autistic features	No speech / eye contact, hypotonia tetraparesis, dyskinesia



Expanding the clinical and EEG spectrum of CNKSR2-related encephalopathy with status epilepticus during slow sleep (ESES)

Objective: To investigate the clinical and EEG features of Encephalopathy with Status Epilepticus during slow Sleep (ESES) related to CNKSR2 pathogenic variants.

Methods: Detailed clinical history, repeated wakefulness/overnight sleep EEGs, brain MRI were collected in five patients, including one female, with CNKSR2-related ESES.

Results: Neurodevelopment in infancy was normal in two patients, delayed in three. Epilepsy onset (agerange: 2–6 years) was associated with appearance or aggravation of cognitive impairment, language regression and/or behavioral disorders. Worsening of epilepsy and of cognitive/behavioral disturbances paralleled by enhancement of non-rapid eye movement (NREM) sleep-related, frontally predominant, EEG epileptic discharges [spike-wave-index (SWI): range 60–96%] was consistent with ESES. In three patients, episodes of absence status epilepticus or aggravation of atypical absences occurred, in this latter case associated with striking increment of awake SWI. Speech/oro-motor dyspraxia was diagnosed in four patients. In two patients, long-term follow-up showed epilepsy remission and persistence of mild/-moderate cognitive disorders and behavioral disturbances into adulthood.

Conclusions: Novel findings of our study are occurrence also in females, normal neurodevelopment before epilepsy onset, epilepsy aggravation associated with enhanced awake SWI, mild/moderate evolution in adulthood and language disorder due to speech/oro-motor dyspraxia.

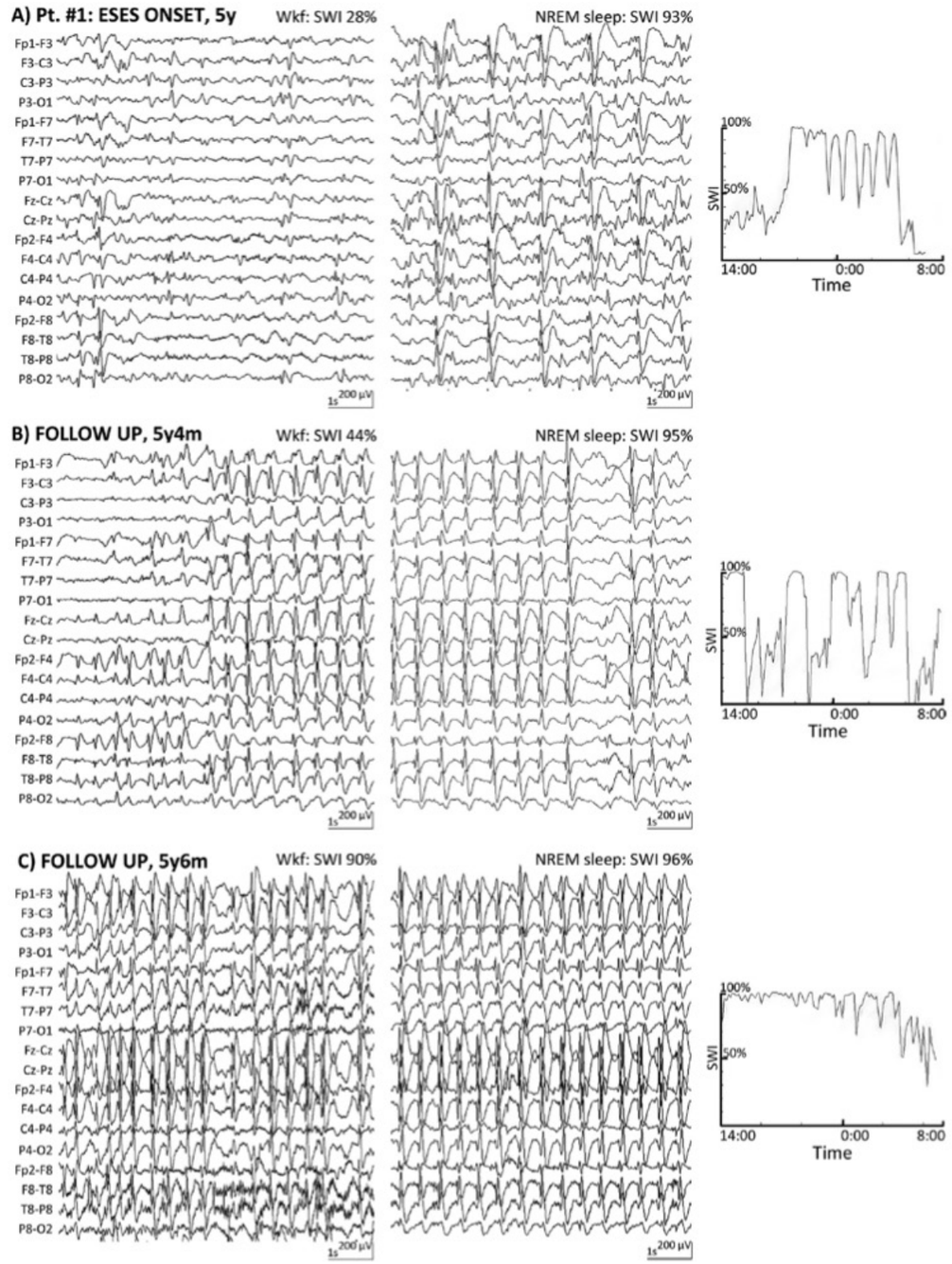
Significance: Our findings expand the phenotypic spectrum of CNKSR2-related ESES.

Paper

CM Bonardi, C Mignot, JM Serratosa, BG Giraldez, R Moretti, G Rudolf, C Reale, PM Gellert, KM Johannesen, G Lesca, CA Tassinari, **E Gardella, RS Møller**, G Rubboli.

Expanding the clinical and EEG spectrum of CNKSR2-related encephalopathy with status epilepticus during slow sleep (ESES).

Clinical Neurophysiology 131 (2020) 1030–1039

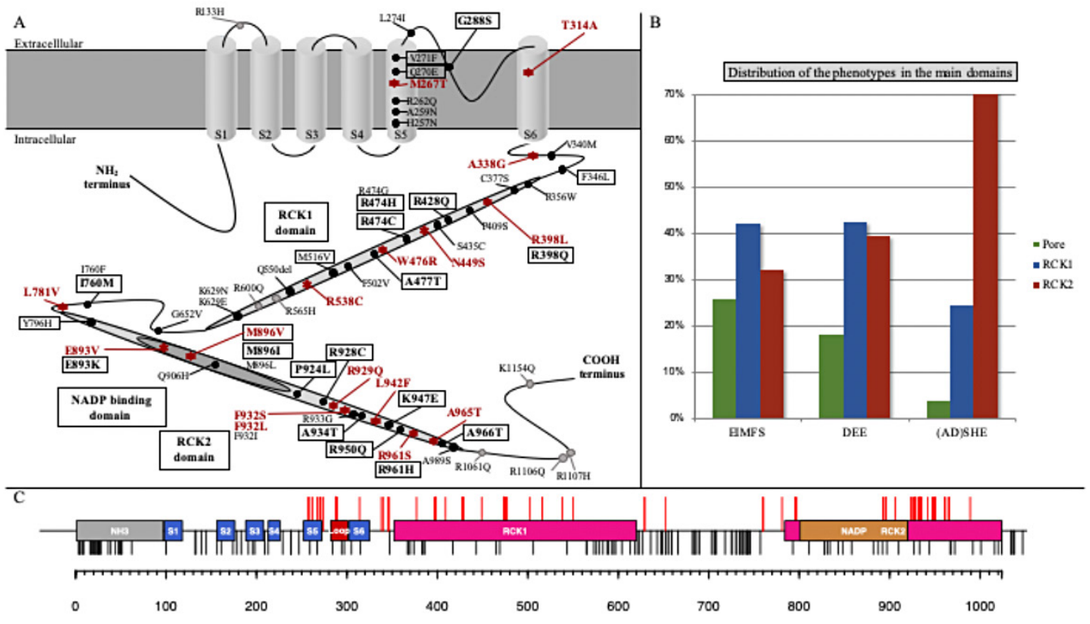


The phenotypic and mutational spectrum of KCNT1-related epilepsies and epileptic encephalopathies

Variants in KCNT1, a gene encoding a sodium-gated potassium channel (subfamily T member 1), have been associated with a spectrum of epilepsies and neurodevelopmental disorders ranging from familial autosomal dominant or sporadic sleep-related hypermotor epilepsy ((AD)SHE) to epilepsy of infancy with migrating focal seizures (EIMFS) and to a wide group of heterogeneous developmental and epileptic encephalopathies (DEE). This study aims to provide a comprehensive overview of the phenotypic and genotypic spectrum of KCNT1-related epileptic disorders in 207 individuals, the largest cohort reported so far. The described cohort includes 66 unpreviously published and 141 published cases. Four phenotypes emerged from our analysis : i) EIMFS (120 patients, of whom 33 were novel); ii) DEE other than EIMFS (non-EIMFS DEE) (31 patients, 17 novel); iii) (AD)SHE (52 patients, 14 novel); iv) other phenotypes (4 patients, 2 novel). In our cohort of new cases, the main phenotypic features were: a) in EIMFS, great heterogeneity of seizure types, including epileptic spasms, epilepsy improvement over time (seizure freedom was achieved in about 10% of patients), no death; b) in non-EIMFS DEE, more frequent occurrence than previously reported, high seizure type variability, possible evolution to DEE with SHE features; c) in (AD)SHE, high prevalence of drug-resistance, although seizure frequency could improve over the years, appearance of cognitive regression after seizure onset in all patients, no severe psychiatric disorders although behavioural/psychiatric comorbidities were reported in about 50% of the patients, SUDEP in one patient; d) in the other phenotype group, temporal lobe epilepsy, and epilepsy with tonic-clonic seizures and cognitive regression. Genotypic analysis of the whole cohort showed only missense variants and one inframe deletion, all of them with a presumed gain-of-function effect. Although the variants were distributed among the different domains, genotype-phenotype considerations showed (AD)SHE-associated variants mainly clustered in the RCK2 domain in the C-terminus distal to the NADP domain, non-EIMFS DEE-associated variants clustered primarily around RCK1 and RCK2.

Paper

Bonardi CM, Heyne HO, Fiannacca M, Fitzgerald MP, **Gardella E**, Gunning B, Olofsson K, Lesca G, Verbeek N, Stamberger H, Striano P, Zara F, Mancardi MM, Nava C, Syrbe S, Buono S, Dibbens LM, Baulac S, Coppola A, Weckhuysen S, Ceulemans B A-S S, Sarret C, Baumgartner T, Muhle H, Portes V des, Toulouse J, Nougues M-C, Rossi M, Demarquay G, Ville D, Hirsch Ed, Maurey H, Willems M, Bellescize J de, Altuzarra C Desmettre, Villeneuve N, Bartolomei F, Picard F, Hornemann F, Koolen DA, Kroes HY, Tan W-H, Reale C, **Fenger CD**, Bearden DR, **Møller RS, Rubboli G.** The phenotypic and mutational spectrum of KCNT1-related epilepsies and epileptic encephalopathies
Brain (submitted)



Clinical Trials

Collaboration between Pediatric department and Neurological department; Adults with epilepsy

Fenfluramin Dravet syndrom:

- **An open-label** extension trial to assess the long-term safety of ZX008 (Fenfluramine hydrochloride) oral solution as an adjunctive therapy in children and young adults with Dravet syndrome.
  First patient started in August 2016. Now carried on to open –label study 1900.
  Principal investigator: **Marina Nikanorova**
  Study coordinator: **Klaus Ehrenreich**

Fenfluramin Lennox-Gastaut syndrome

- A two-part study of ZX008 in children and adults with Lennox-Gastaut syndrome (LGS);
  Part I: A randomized, double-blind, placebo controlled trial of two fixed doses of ZX008 (Fenfluramine hydrochloride) oral solution as adjunctive therapy for seizures in children and adults with LGS.
  Part II: An open-label extension trial to assess long-term safety of ZX008 in children and adults with LGS
  First patient started January 2019.
  Principal investigator: **Dragan Marjanovic**
  Co-investigator: **Marina Nikanorova**
  Study coordinators: **Klaus Ehrenreich and Lone Olsen.**

Department of Clinical Neurophysiology

The department of clinical neurophysiology is mainly serving as a diagnostic facility for patients managed at the Epilepsy Hospital. However, outpatients are referred to clinical neurophysiology investigations form other hospitals too. The department performs a wide variety of neurophysiological investigations, including EEG, evoked potentials, electromyography and nerve conduction studies. The staff consists of five physicians, 12 neurophysiology technician and a nurse. The main research interest of the department is clinical application of EEG in epilepsy.

Standardization and quality assurance in clinical neurophysiology

Most of the neurophysiological methods in clinical neurophysiology are based on local traditions, expert-opinions and on old studies lacking proper reference standard and study design. To improve patient care it is important to revisit the old techniques and to validate new methods, using robustly designed diagnostic studies.
Our group contributed to this in several areas of clinical neurophysiology. We demonstrated that recording duration is more important for the diagnostic yield of EEGs than increasing spatial sampling beyond the standard IFCN electrode array and we proved the diagnostic added value of sleep EEG recordings. We contributed to the IFCN recommendation for examination and diagnostic strategies of polyneuropathy electrodiagnosis.

Papers:

Bach Justesen A, Foged MT, Fabricius M, **Skaarup C**, Hamrouni N, Martens T, Paulson OB, Pinborg LH, **Beniczky S.** Diagnostic yield of high-density versus low-density EEG: The effect of spatial sampling, timing and duration of recording. Clin Neurophysiol. 2019 Nov;130(11):2060-2064. doi: 10.1016/j.clinph.2019.08.007.

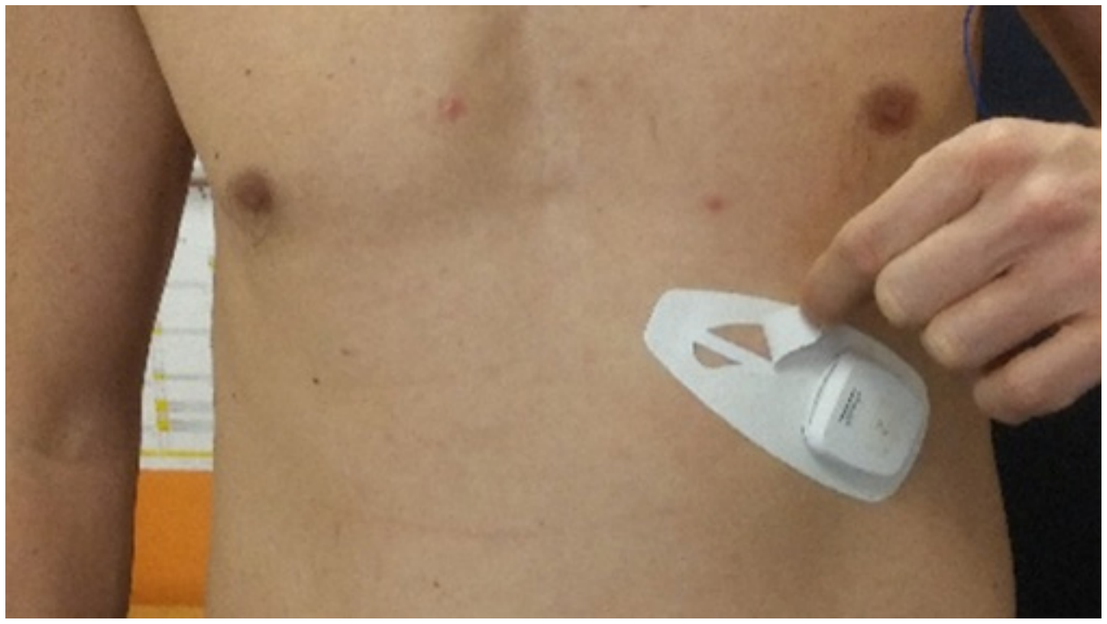
Tankisi H, Pugdahl K, **Beniczky S**, Andersen H, Fuglsang-Frederiksen A. Evidence-based recommendations for examination and diagnostic strategies of polyneuropathy electrodiagnosis. Clin. Neurophysiology Pract. 2019 Nov 18;4:214-222. doi: 10.1016/j.cnp.2019.10.005

Automated seizure detection using wearable devices

There is need for automated seizure detection using mobile or wearable devices, for objective seizure documentation and decreasing morbidity and mortality associated with seizures. Due to technological development, a high number of articles have addressed non-electroencephalography (EEG)-based seizure detection. However, the quality of study-design and reporting is extremely heterogeneous. Fifteen studies of phase-2 or above, demonstrated that non-EEG-based devices detected generalized tonic–clonic seizures (GTCS) with high sensitivity (≥90%) and low false alarm rate (FAR) (down to 0.2/day). There has been limited published evidence for detection of motor seizures other than GTCS, mostly from subgroups in larger studies, targeting GTCS. There has been little evidence for non-EEG-based detection of non-motor seizures: sensitivity was low (19–74%) with extremely high FAR (50–216/day).

To assess the feasibility and accuracy of seizure detection based on heart rate variability (HRV) using a wearable electrocardiography (ECG) device. In this phase 2 study, we prospectively recruited patients admitted to long-term video–EEG monitoring (LTM). ECG was recorded using a dedicated wearable device. Seizures were automatically detected using HRV parameters computed off-line, blinded to all other data. We compared the performance of 26 automated algorithms with the seizure time-points marked by experts who reviewed the LTM recording. Patients were classified as responders if >66% of their seizures were detected. We recruited 100 consecutive patients and analyzed 126 seizures (108 non-convulsive and 18 convulsive) from 43 patients who had seizures during monitoring. The best performing HRV algorithm combined a measure of sympathetic activity with a measure of how quickly HR changes occurred. The algorithm identified 53.5% of the patients with seizures as responders. Among responders, detection sensitivity was 93.1% (95% CI: 86.6%-99.6%) for all seizures and 90.5% (95% CI: 77.4%-97.3%) for non-convulsive seizures. FAR was 1.0/24 h (0.11/night). Median seizure detection latency was 30 s. Typically, patients with prominent autonomic nervous system changes were responders: An ictal change of >50 heartbeats per minute predicted who would be responder with a positive predictive value of 87% and a negative predictive value of 90%.

The automated HRV algorithm, using ECG recorded with a wearable device, has high sensitivity for detecting seizures, including the non-convulsive ones. FAR was low during the night. This approach is feasible in patients with prominent ictal autonomic changes.



Papers:

Jeppesen J, Fuglsang-Frederiksen A, Johansen P, Christensen J, Wüstenhagen S, Tankisi H, Qerama E, Hess A, **Beniczky S**. Seizure detection based on heart rate variability using a wearable electrocardiography device. Epilepsia. 2019 Oct;60(10):2105-2113. doi: 10.1111/epi.16343.

Beniczky S, Jeppesen J. Non-electroencephalography-based seizure detection. Curr Opin Neurol. 2019 Apr;32(2):198-204. doi: 10.1097/WCO.0000000000000658.

Electromagnetic Source Imaging

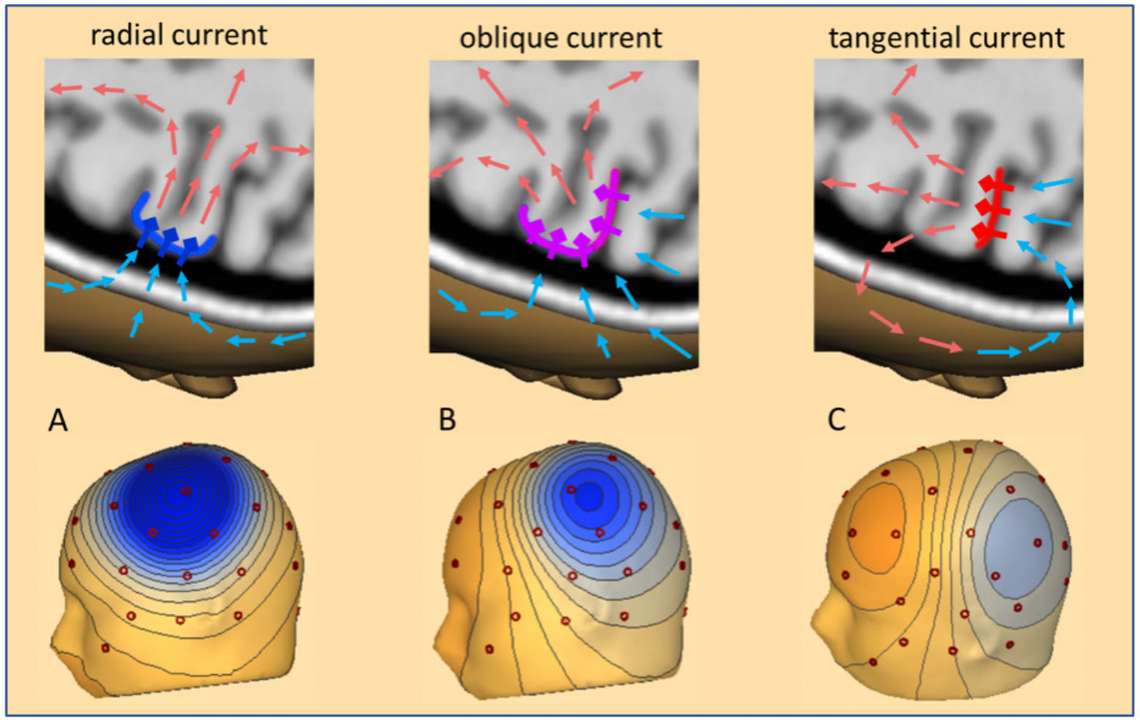
Electric and magnetic source imaging methods (ESI, MSI) estimate the location in the brain of the sources generating the interictal epileptiform discharges (II-ESI, II-MSI) and the ictal activity (IC-ESI, IC-MSI). These methods provide potentially valuable clinical information in the presurgical evaluation of patients with drug-resistant focal epilepsy, evaluated for surgical therapy. However, they are still underutilized in most epilepsy centers performing a presurgical evaluation, due to lack of robust clinical validation studies. We have conducted several prospective validation studies on ESI and MSI.

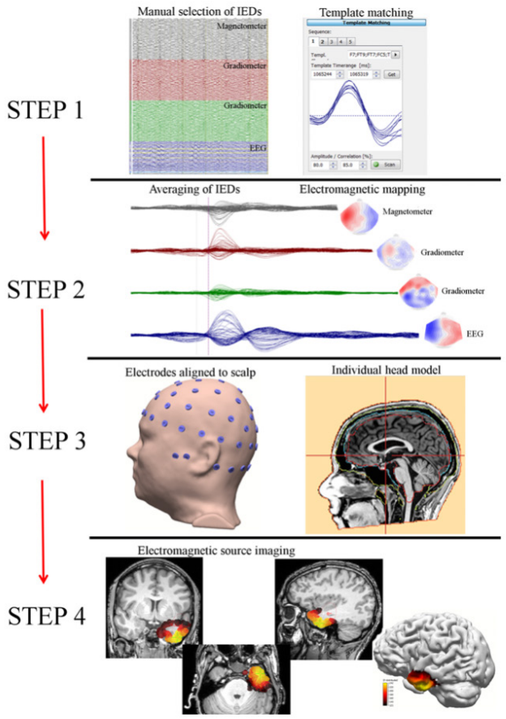
We described a novel method for transforming EEG signals from sensor space into source space. Multiple discrete sources have the power to transform the EEG back into the brain by defining new EEG traces in source space. Using standard source space 25, these can provide for improved clinical interpretation of EEG.

We investigated the clinical utility of ESI in presurgical evaluation. We found that ESI had diagnostic added value in 34% of the patients. In most cases (85.7%), these changes were related to planning of the invasive recordings. In nine out of 13 patients, invasive recordings confirmed the localization. Out of eight patients in whom the ESI source was resected, six became seizure-free.

In a large prospective study we recorded magnetoencephalography (MEG) simultaneously with EEG and performed EMSI, comprising electric source imaging, magnetic source imaging, and analysis of combined MEG-EEG datasets, using 2 different software packages. As reference standard for irritative zone (IZ) and seizure onset zone (SOZ), we used intracranial recordings and for localization accuracy, outcome 1 year after operation. We included 141 consecutive patients. EMSI showed localized epileptiform discharges in 94 patients (67%). Most of the epileptiform discharge clusters (72%) were identified by both modalities, 15% only by EEG, and 14% only by MEG. Agreement was substantial between inverse solutions and moderate between software packages. EMSI provided new information that changed the management plan in 34% of the patients, and these changes were useful in 80%. Depending on the method, EMSI had a concordance of 53% to 89% with IZ and 35% to 73% with SOZ. Localization accuracy of EMSI was between 44% and 57%, which was not significantly different from MRI (49%-76%) and PET (54%-85%). Combined EMSI achieved significantly higher odds ratio compared to electric source imaging and magnetic source imaging.

We performed a systematic review and meta-analysis of ESI and MSI in presurgical evaluation. We found evidence for the accuracy of source imaging in presurgical evaluation of patients with drug-resistant focal epilepsy. These methods have high sensitivity (up to 90%) and diagnostic odds ratio (up to 7.9), but the specificity is lower (up to 54%). ESI and MSI should be included in the multimodal presurgical evaluation.





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Filadelfia Research Department

The department engages in coordination, support and facilitation of clinical research, and has multidisciplinary clinical research as an area of special interest.

The department works by motivation of employees of all professions to engage in research and evidence based thinking in close cooperation with the individual clinical units, the residential social setting included.

Currently, the department is engaged in the implementation of Family Focused Care as a pathway to derive empiric clinical research.

The overall strategy is closely connected to the well-described "Vision & Strategy" of the hospital, as well as the Research Strategy of Filadelfia.

Altogether, this emphasizes the importance of connecting the clinical as well as the residential social setting in a shared research portfolio.

7. Nursing Research Perspectives

Annual Dianalund Nursing Conference

Dianalund Nursing Conference has been held for 8 years. It covers widely in topics and themes but always aiming to focus on clinical aspects within epilepsy nursing Care. 2019 focused on ***"Family Focused Care"*** from different perspectives.

Family & Caregiver Days – With focus on rare genetic epilepsies

Syndromes like **DRAVET** and ***STXPB1*** represents complex and life changing diagnosis. Filadelfia was in 2019 head organizer for two individual Family / Caregiver days with focus on the disease, everyday living and future research perspectives. All presented by a wide range of health professionals from Filadelfia.

The days have gained in success. Family's needs of having professional and specific information as well as having the opportunity to meet other families in a similar situation have proven successful.

8. Publications in Peer Review 2019

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Book & book chapters

Book

- Encephalopathy related to Status Epilepticus during slow Sleep: linking epilepsy, sleep disruption and cognitive impairment. Edited by **Guido Rubboli**, Carlo Alberto Tassinari. John Libbey Eurotext, Montrouge, France, 2019

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- Alving J. Sabers A, Uldall P and others. **Møller RS, Stubbings V**.; Epilepsi – en basisbog. 2edt. FADLs Forlag 2019.pp 72 & 107-111.

- **Sándor Beniczky** and **Praveen Sharma**. Electromagnetic source imaging, high-density EEG and MEG.In: Oriano Mecarelli (editor): Clinical Electroencephalography. Pages: 329-344. Springer Nature Switzerland, 2019; Cham. Switzerland.

9. Speaker presentations

Paediatric Department

MD, Head of Paediatric department Kern Olofsson

- Annual meeting at the Danish Neuropediatric Society 22 nov 19, Update / status of new anti-epileptic treatments

Ass. Professor Marina Nikanorova

- **Neuropediatric meeting, Viborg**, 5 March 2019: Myoclonic seizure. Myoclonic epilepsies
- **Meeting of the Finnish Neuropediatric Society and Finnish Chapter of ILAE, Kuopio, Finland**, 21-22 March 2019: Treatment of childhood onset intractable epileptic syndromes
- **International Expert group meeting, Salerno, Italy**, 3 – 4 May 2019: Pediatric experience with Perampanel use in children with drug-refractory epilepsy.
- **Baltic Child Neurology Association meeting, Kaunas, Lithuania**, 15 – 17 May 2019: Genetic epileptic encephalopathies: recognition of clinical phenotypes
- **Baltic Sea Summer School on Epilepsy, Rostock, Germany**, 19 - 24 August, 2019: Case-oriented learning Tutorial – Non-epileptic paroxysmal conditions in children Tutorial – interactive seizure semiology Lecture – Epileptic encephalopathies: outcomes in adults

Department of Epilepsy Genetics and Personalized Medicine

Ass. Professor Rikke S. Møller

- **Epilepsy Seminar. Dalum Landbrugsskole, Odense**, Genetic testing of children and adults with epilepsy – whom, when and why.
- **2nd annual joined Denmark/Norway epilepsy specialist meeting, Copenhagen, Denmark**, Personalized treatment approaches in genetic epilepsies
- **STXBP1 Investigator and Family Meeting, CHOP, Philadelphia, USA**, Incorporating Epilepsy Genetics into Clinical Practice,
- **Nordic Forum on Precision Medicine in Genetic Epilepsies, Copenhagen, Denmark**, PCDH19 – girls clustering epilepsy.
- **Danish Epilepsy Society, annual meeting, Copenhagen, Denmark**, Epileptic channelopathies: from gene discovery to personalized treatment.

MD PhD Katrine M Johannesen

- **GEE Symposium, Kings College, London, UK**. Speaker session on SLC6A1. June 2019
- **Nordic Expertise Panel, Copenhagen**. Speaker session on SCN2A. October 2019
- **SLC6A1 connect annual symposium, Baltimore, USA**. Panel discussion. December 2019
- **SCN8A Researcher and Family get together, Baltimore, USA**. Speaker session on SCN8A December 2019.

Professor Guido Rubolli

- **National Congress of the Iranian Epilepsy Association, Teheran (Iran)** 23-25/1/19. 3 talks:
  "Antiepileptic drug treatment after epilepsy surgery"
  "Encephalopathy related to Status Epilepticus during Sleep (ESES)"
  "Levetiracetam efficacy and safety as monotherapy and add on therapy with the optimal dosage in adult
  Patients. Case presentations.
- **Spring Meeting of the Danish Epilepsy Society, Copenhagen**, 9-10/5-2019. "Can genetics be relevant in presurgical evaluation?"
- **International Epilepsy Congress, Bangkok, (Thailand)**, 22-26/6/19 "Generalized seizures": Videosession"
- **Epilepsy Meeting, Barcelona, Spain**. Sept 2019 "The prognosis of ESES"

Adult Neurological Department

Research developmental Nurse Trine Arnam-Olsen Moos

- **Danish Nurse Association, Conference, Copenhagen** Dec. 2019 : Rehabilitation in Nursing, Denmark: Session on Patient Perspective.

Department of Clinical Neurophysiology

Professor Sàndor Beniczky

- **Epilepsy Symposium, Dresden Tyskland**. April 2019. Seizure Detection using surface EMG.
- **The 7th London-Innsbruck Colloquium on Status Epilepticus and acute seizures**. April 2019. EEG in Status Epilepticus and on the intensive Care Unit.
- **Danish Epilepsy Society, Copenhagen** May 2019. Seizure Semiology
- **17th European Congress of Clinical Neurophysiology, Warsaw Poland**. June 2019.
  - Seizure Types; In interactive video-EEG session.
  - Controversies in Clinical Neurophysiology; Common average montage.
  - Diagnostic accuracy and clinical utility of EMG and MEG source localization
  - EEG in the intensive care unit
- **Russian Neurology Congress, Skt. Petersburg, Russia**. June 2019. Clinical utility of EEG in diagnosis and monitoring of patients with epilepsy.
- **33rd International Epilepsy Congress, Bangkok, Thailand**. June 2019.
  - Seizure detection based on movement and accelerometry
  - Seizure types; an inter active video-EEG session
  - Seizure detection using wearable devices; a modern possibility?
  - Basic EEG technology.
- **Latin American Congress of clinical Neurophysiology, Sao Paulo, Brazil.** Aug 2019.
  - Standardized Computer-based organized reporting of EEG.
  - Automated seizure detection using wearable devices.
  - Electromagnetic source imaging.
- **5th SuSIE – Summer School on Imaging in Epilepsy**. Aug 2019. How computered EEG & MEG analysis changed the concept of epilepsy & presurgical evaluation.
- **13th Baltic Sea Summer School on Epilepsy, Rostock, Germany** Aug 2019.
  - Epilepsy – differential diagnosis.
  - Seizure classification.
  - Seizure Semiology

- **2nd International Congress on Mobile Health and Seizure Detection in Epilepsy, Lausanne Switzerland**. Sept 2019 Quantifying severity and generalized tonic-clonic seizures.

- **Neurology Forum, Lisbon Portugal** Oct 2019. Can we detect seizures in epilepsy?

- **6th Hong Kong Neurological Congress**. Annual Scientific Meeting Nov. 2019.
  - EEG in critical setting
  - Wearable devices for detection of epileptic seizures

- **Annual Meeting of the American Epilepsy Society, Baltimore**. Dec 2019.
  - Seizure detection using wearable devices: Potential implications for prevention of SUDEP
  - Ictal Patterns of Surface Electromyography

- **Epilepsy symposium "Anfaldsrelatede risker och hjaelpmedel for detection av anfall" Gothenburg, Sweden** Dec.2019; New devices for seizure detection and risk reduction: what is in the pipeline?

- **Annual meeting of the Israeli Neurological Society, Tel Aviv** Dec, 2019. Seizure Detection Using Wearable Devices

- **Head organizer of the 2nd International Congress on Mobile Health and Seizure Detection in Epilepsy, Lausanne Switzerland**. Sept 2019.

Clinical Neurophysiology Assistants Anders Justesen & Christian Skaarup

- **ILAE Congress Bangkok** June 2019. Presurgical evaluation – from a technician's point of view.

Ass. Professor Elena Gardella:

- **Spring meeting of the Danish Epilepsy Society Copenhagen, DK,** May 9, 2019 Clinical approach to epileptic channelopathies

- **"Hårde Nødder" Odense, Denmark**, May 2019. Common motor generators for epileptic seizures and other paroxysmal motor events.

- **First STXBP1 Investigator's Meeting (SIM) Philadelphia, USA**, June.2019. Electroclinical phenotype of STXBP1 disorders.

- **Summer school of clinical neurophysiology of the Romanian Society of Clinical Neurophysiology. Sulcevita, Romania** July 2019. Video-EEG monitoring in Genetic Epilepsies.

10. Posters

Katrine M Johannesen

- ESHG, Göteborg, June 2019, **Johannesen KMH**,Relationship of clinical phenotype and functional effect in SCN8A-related disorders

Anne Vagner Jacobsen

- ESTSS 2019 - 16th ESTSS conference in Rotterdam – Holland, June 2019: **Jacobsen AV**. Title: The impact of severe pediatric epilepsy on experienced stressors, depression and PTSD in caregivers.

- 4th international epilepsy symposium, Bielefeld Germany, sept 2019: **Jacobsen AV**. Epilepsy and Psychology. Seizures, Cognition and Behavior. Title: Assessing methodological issues evaluating cognitive outcome of pediatric epilepsy surgery: A monocentric 2-year follow-up study

Trine Arnam-Olsen Moos

- ILAE Congress Bangkok June 2019: **Moos T**. "Everything is as before but nothing is as it was" A scientific and qualitative research study exploring the experience of life amongst adult patients with refractory epilepsy after 8 weeks of multidisciplinary rehabilitation interventions.

Lærke Duncan

- ILAE Congress Bangkok June 2019: Duncan L, **Skaarup NC, Cho JH, Scherg M, Beniczky S**: Forty EEG electrode array for long term video - EEG monitoring.

Cecillie Bøttiger Itenov

- ILAE Congress Bangkok June 2019. **Itenov C, Nielsen K, Beniczky S** : Diagnostic added value of recording surface electromyography

Jane Eriksen

- ILAE Congress Bangkok, June 2019. **Eriksen J, Duncan L, Sand L, Terney D, Nikanorova M, Khinchi M, Stubbings V, Olsen T, Olofsson K, Beniczky S, Gardella E**. The Danish protocol for the diagnostic assessment of ESES/CSWS

Anders Justesen

- ILAE Congress Bangkok,: June 2019 **Justesen AB**, Foged MT, Fabricius M, Skaarup NC, Hamrouni N, Martens T, Paulson OB, Pinborg L, **Beniczky S**; Diagnostic yield of high-density versus low-density EEG: The effect of spatial sampling, timing and duration of recording.

Elena Gardella

- Riunione policentrica Lega Italiana contro lÉpilessia Roma, Italy January 24-25, 2019: M Guazzi, **Reveles R. Møller RS, Gardella E**. Lieve ritardo mentale epilessia correlata a mutazione GABRB3.

- Annual congress of the Italian Society of Neurology Bologna (Italy). 12-15, October 2019.: Real C, **Møller RS, Rubboli G**. Bonardi C, Trivisano MA, Cantalupo G, Spagnoli C, Rokkjær M, Lo Barco T, La Selva L, Allen NM, Neu A, Be Gérard, M Pendziwiat, Helbig I, **Gellert PM, Nikanorova M, Reveles R**, Høi-Hansen CE, Specchio N, Fontana E, Fusco C, dalla Bernardina B, Magaudda A, Toscano A, **Gardella E**. Deep electro-clinical phenotyping of KCNB1 developmental and epileptic encephalopathy.

Jan Borg Rasmussen

- 14th Danish Congress in Clinical Biochemistry Aarhus, Denmark. 18.-20. June. **Larsen JB, Jensen PB, Rasmussen JB**. Fighting adverse drug reactions (ADR): a cyp450 genetic test panel with reported allele frequencies of Cyp2C9, 2C19 and 2D6 in a Danish population.

Filadelfia - juni 2020

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