EpiCARE

European Patient Advocacy Group

Association Posters

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DRAVET ITALIA ONLUS

Vision

We envision a future where children living with Dravet Syndrome and other rare epilepsies can thrive without the limitations of their conditions. By strengthening a global network of families, doctors, and researchers, we aim to accelerate progress toward better treatments and a cure. Through education, advocacy, and collaboration, we seek to raise awareness and ensure that every child receives the care and support they deserve. Our ultimate goal is a world where epilepsy is no longer a barrier to a fulfilling life

Mision

Dravet Italia Onlus is committed to improving the lives of children affected by Dravet Syndrome, a severe and often life-threatening form of epilepsy. Our association focuses on providing support to families, raising awareness, and fostering collaboration with medical professionals and research institutions. While originally founded to address the needs of Dravet Syndrome, we now advocate for all rare and complex epilepsies, working to bridge gaps in care and treatment. By funding research and promoting advancements in the field, we aim to offer hope and real solutions to those facing the daily challenges of these conditions

KEY PROJECTS/ACTIVITIES

- Dravet Registry “RESIDRAS”: A a specialised registry focusing on Dravet Syndrome and other syndromes related to mutations in the SCN1A and PCDH19 genes. Its aim is to establish a comprehensive database by collecting longitudinal, retrospective, and prospective data from both pediatric and adult patients. The registry has since expanded to an international level, where it is known as Platform RESIDRAS for participating centers.
- Dravet Diary: A digital diary to monitor and manage epilepsy and behavioural conditions, very easy to use and to share the data with the physicians
- Dravet Disease Scale for Associated Neuropsychiatric Disorders (D-DAND): focuses on the known comorbidities in DS patients, many of which are shared by other DEEs. The scale is based on interviews with parents, conducted either in person or via videoconference, and evaluates multiple domains.

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- Advocacy for Therapies – The Association has played a key role in securing access to innovative treatments like fenfluramine and cannabidiol for patients in Italy.
- Support Network – The organization has built a strong community, offering educational programs, resources, and emotional support to families affected by Dravet syndrome. It actively engages all relevant stakeholders to promote shared actions aimed at policymakers and social support activities (like the Italian experience that led to the recognition of all rare and complex epilepsies) and fosters collaborations to support epilepsy-related legislation initiatives.
- Research Collaboration – Actively involved in research, the Association supports patient registries and collaborates internationally to improve understanding and treatment of Dravet syndrome

What do we expect from CREA?

Representing the voice of the community to drive meaningful change for all people affected by rare and complex epilepsies — ensuring the best possible quality of life and continuously seeking ways to improve it

What can we offer to CREA?

We offer our dedication, our decade-long experience, and the network of people who have accompanied us throughout this journey. In addition, we bring seven years of active participation in the ePAG EpiCARE group, international collaboration with world-renowned researchers and medical professionals, extensive experience with European research project calls, and a strong track record of publications and articles.

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FUNDACJA PACS2 RESEARCH FOUNDATION

PACS2 RESEARCH FOUNDATION

Vision

Improving the quality of life for patients with PACS2 syndrome through a better understanding of the disease and the development of therapies by 2030. Co-creating a world where patients are engaged in drug research from the very beginning of scientific studies.

Mision

Accelerating drug development for the ultra-rare neurological disorder PACS2 syndrome and improving the quality of life for those affected through the initiation and coordination of international, interdisciplinary scientific research. Advocating for patient partnership in research and the broader involvement of researchers in ultra-rare diseases

KEY PROJECTS/ACTIVITIES

- Drug repurposing leveraging cell painting
- Biomarkers definition for PACS2 syndrome (mouse model phenotyping, multiomics)
- Building community of families engaging in rare disease research at any stage

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- Building a team of 50+ researchers, 10+ research teams (in US, UK, Netherlands, Czech Republik, Canada, Poland) for disease with ca. 150 patients globally
- Advocating successfully for development of innovative therapies including ASO, gene therapy or RNA editing
- Driving agenda of patient engagement in research on national level in Poland - being patient voice in National Rare Disease Council at Ministry of Health and Member of Warsaw Health Innovation Hub

What do we expect from CREA?

Strong voice of the community of families impacted by rare epilepsies for our common needs. Building visibility of the community across Europe.

What can we offer to CREA?

Enthusiasm, can do attitude, love for work with other passionate people, project management skills, knowledge on drug development process

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GEORGIAN RETT SYNDROME AND OTHER RARE DISEASES ASSOCIATION

Georgian Rett Syndrome and other rare diseases Association

Vision

Promote the creation of an inclusive environment to empower individuals with Rett syndrome and their families, accelerate comprehensive research on the full spectrum of the disease, which is a prerequisite for eliminating Rett syndrome. Establish continuous communication among families of individuals with Rett syndrome, provide information to the public, and raise awareness and education about Rett syndrome

Mision

Unite and empower individuals affected by rare diseases, their families, caregivers, patient organizations, and medical professionals across all regions of Georgia with a special focus on Rett syndrome.The Association advocates for better access to diagnosis, treatment, and support services, promotes policy changes, and fosters collaboration with national and international stakeholders. Through education, awareness, and research support, the organization strives to improve the quality of life for those living with rare diseases with a special focus on Rett syndrome and ensure that no patient is left behind.

KEY PROJECTS/ACTIVITIES

- Georgia Alliance of Rare Diseases: This initiative focuses on strengthening the Georgian Alliance for Rare Diseases as a national umbrella organization that unites patient advocacy groups, individuals, caregivers, and medical professionals. The project aims to enhance collaboration among stakeholders, advocate for improved healthcare policies, expand access to treatments, and ensure the inclusion of rare disease patients across all regions of Georgia.
- National Mental Health Program development for Caregivers: This program focuses on supporting the mental health and well-being of caregivers and care partners of individuals with rare diseases. Recognizing the emotional, physical, and psychological challenges they face, the initiative aims to provide access to mental health services, counseling, peer support groups, and educational resources
- Advocating for access to treatment and AAC high technology tools for well being of Rett patients in health, communication and education at the national level by working with the Ministry of Health to establish government programs for Rett syndrome. One such program is already in place, providing management based on international guidelines, including therapies and specialized medical consultations, covering the entire country

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- National Program for Rett Syndrome Management Successfully advocated for and helped implement a government-funded program for the management of Rett syndrome, ensuring access to therapies and specialized medical care across Georgia.
- The AAC training program- This initiative helped shift perspectives, highlighting the importance of access to communication technologies in education and daily life. By promoting the role of AAC in Rett syndrome, the program ultimately advocated for greater inclusion and support, ensuring that individuals with complex needs are recognized for their cognitive potential and provided with the necessary tools to express themselves.
- Involvement in the Establishment of the Georgian Alliance for Rare Diseases- Played a leading role in the creation of the Georgian Alliance for Rare Diseases, a national umbrella organization that unites patient advocacy groups, caregivers, medical professionals, and policymakers. The Alliance works to strengthen the rare disease community, improve healthcare policies, and expand access to essential treatments and support services.

What do we expect from CREA?

Collaboration and Shared Resources-
Collaboration in creating a unified voice for advocating for better access to treatment, diagnosis, and support services for individuals with rare epilepsies and complex conditions in Europe. Sharing resources, research, and best practices will strengthen our collective impact. Collaboration with Pharma, Biopharma companies in clinical researches to fill the gap of clinical trial and treatment access in Europe.

What can we offer to CREA?

Expertise in Clinical Researches and Advocacy Experience-
The Georgian Rett Syndrome and Rare Diseases Association can offer its clinical research and advocacy experience in navigating the healthcare system, engaging policymakers, and raising awareness about Rare and Complex Epilepsies.
Education and Training Initiatives-With our experience in organizing training programs (such as for AAC devices in Rett syndrome), we can collaborate on training initiatives for healthcare professionals, caregivers, and families of individuals with rare and complex epilepsies, particularly focusing on enhancing communication and care strategies.

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INTERNATIONAL MITO-PATIENTS

imp

international mito patients

Vision

IMP has a global view of mitochondrial diseases with the specificity of rare and drug-resistant epilepsy

Mision

To increase quality of life for people with mitochondrial disease by facilitating cross-border cooperation and collaboration among national patient organisations

KEY PROJECTS/ACTIVITIES

- Collecting information from all our international patient organisation for the quality of life
- Support cross border research
- Promote education and awarness

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- IMP is building a patient registrie that will connect to the clinical registrie
- IMP is and has been part of different research projectto give the overview of patients
- IMP facilitates patient care in different countries

What do we expect from CREA?

CREA could facilitate and perhaps create a European pathway plan for rare epilepsy, there will surely be a strong impact on the fact that our diseases are different

What can we offer to CREA?

IMP being an international association we can help to have patient feedback at the global level, determine together the common problems of rare epilepsy and participate in the different working groups

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ASSOCIATION CONTRE LES MALADIES MITOCHONDRIALES (AMMI)

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Vision

All Mitochodrial desease and rare epilepsie in France and cross boarder. Help families to have access to diagnostic, treatment and have a better quality of life

Mision

The association creates a family network, helps families psychologically and helps with the purchase of adapted equipment, informs families of the progress of research, helps and participates in various research projects and participates in projects to improve the quality of life of patients. AMMi is in contact with health institutions and public authorities

KEY PROJECTS/ACTIVITIES

- The association actively participates in the national diagnostic and health protocols, and reference centers
- The association is one of the founding members of the Brain House, Rare Diseases Alliance and IMP. We are actif in these organisations
- Unified protocol for mitochondrial disease and epilepsie for france

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- Involved in therapeutic education
- We are actively involved in research in france and cross boarder
- Awarness for mitochondrial disease and rare épilepsie

What do we expect from CREA?

Creat a strong community and creat common projects

What can we offer to CREA?

Being part of working groups and helping organising events in france

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UNIONE ITALIANA NEXMIF

Unione Italiana NeXmif

Vision

Our Vision is that all clinical aspects related to the Nexmif mutation can be cured.

Mision

Our Mission is to promote awareness of Nexmif encephalopathy signs and symptoms, raising funds for scientific research

KEY PROJECTS/ACTIVITIES

- Raise awareness of our organization among clinicians
- Involve existing patients in our activities including potential first case register
- Fundraising to allow follow up to our first Telethon Seed Grant launched in Fall 2024

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- Research collaboration: in parallel we funded the Telethon Fall 2024 Seed Grant and We are trying to Explore gene therapy with Prof Lignani at UCL
- Creation of a website with comprehensive information and clear picture of us as an organization
- We are part of Alleanza Epilessie Rare e Complesse so we can benefit of all the achievments Isabella is driving

What do we expect from CREA?

I would like Crea to help us creating awareness about Nexmif mutation, eventually help in liasing with other EU family groups and potentially create a good network among researches to ensure growth to our first steps done with Telethon (Dr Cancedda) and with Prof Lignani.

What can we offer to CREA?

Our full availability for what we know

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European Patient Advocacy Group

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GRIN EUROPE Care 4 GRIN

GRIN EUROPE

Vision

GRIN Europe is dedicated to improving the lives of individuals affected by GRIN-related disorders by driving research, fostering collaboration, and advocating for better medical care. Our vision is to ensure that all individuals with GRIN disorders receive early diagnosis, personalized treatments, and comprehensive support. We work to bridge the gap between families, clinicians, and researchers, using collaborative platforms and digital solutions to advance understanding and care for these disorders

Mision

GRIN Europe represents a growing alliance of GRIN patient organizations across Europe, supporting families affected by GRIN-related neurodevelopmental disorders. These ultra-rare conditions often involve epilepsy, movement disorders, cognitive disabilities, and autonomic dysfunctions. Our mission is to connect patient groups, facilitate research, and advocate for improved clinical care, ensuring equal access to treatments and specialized support. We collaborate with leading researchers, clinicians, and industry partners to push forward clinical trials, cross-border collaborations, and AI-assisted research initiatives. By joining CREA, we strengthen our voice in rare epilepsy advocacy, integrate GRIN-related disorders into broader neurological research, and promote data-sharing initiatives to improve patient outcomes.

KEY PROJECTS/ACTIVITIES

- Uniting GRIN patient organizations into an active European alliance- Making sure no family is underrepresented
- Organizing and hosting the European GRIN conferences- bringing together families, researchers, and clinicians from Europe and the rest of the world to address the needs of the patients.
- Engaging in clinical trials and participating in grant applications to advance research

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- Network and support - by building a collaboration between the different associations and family groups to ensure a more unified approach to poatient care across Europe
- Supporting Eduation and Rersearch- by hosting the annual, patient-focused GRIN conferences to drive translational medicine and research collaborations among the experts.
- Engaged in research and drug development- Actively participating in clinical trials and participating in multiple Rare Disease Networks, to enhance research and access to treatments accross Europe.

What do we expect from CREA?

Enhance collaboration between the different associations to ensure a more unified approach to complex and rare epilepsy care. Supporting and securing funding for clinical research, patient advocacy, and digital health initiatives across Europe and across disorders. To have a platform to strengthen our voice in policy discussions, ensuring better care and access for all DEE patients including the GRIN families.

What can we offer to CREA?

Expertise in GRIN-related disorders, focusing on epilepsy, movement disorders, neurodevelopmental and behavioral challenges. A network of GRIN patient associations, researchers, and clinicians to contribute to broader collaborations. Active participation in clinical trials, grant applications, and ERN discussions, ensuring that patient needs drive research and policy decisions.

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CROATIAN ASSOCIATION FOR EPILEPSY

HUE

Vision

È inoltre prevista la possibilità di accedere a iniziative solidali finanziate da enti quali GAL, PNRR e diverse Fondazioni. Parallelamente, si potrà fare ricorso a mutui a tasso agevolato destinati a progetti sociali, come quelli proposti da Banca Etica

Mision

Since 1997, the Croatian Association for Epilepsy (HUE) has been dedicated to improving the quality of life for people living with epilepsy and their families. Through education, advocacy, and support, HUE works to raise public awareness, reduce stigma, and promote greater understanding of epilepsy and its impact on daily life. Our mission is to empower individuals with epilepsy by providing accessible information, creating supportive communities, and advocating for equal rights in healthcare, education, employment, and social participation. We organize awareness campaigns, educational events, support groups, and innovative projects such as recreational-educational camps and online counseling.

As a proud member of the International Bureau for Epilepsy, HUE collaborates globally to break down prejudice and promote dignity for every person with epilepsy. Guided by the motto "Spreading knowledge about epilepsy to break down prejudice," we remain committed to building a more inclusive, informed, and compassionate society.

KEY PROJECTS/ACTIVITIES

- Educational Camps: Eleven recreational and educational camps for children and adolescents with epilepsy have been organized, providing a safe environment for participants to build confidence, learn coping skills, and interact with peers facing similar challenges.
- Febrile Seizures Project: This project involved educating caregivers, educators, and health professionals on managing febrile seizures, fostering teamwork among all stakeholders in pediatric epilepsy care.
- Online Resources: Projects such as the "Online Counseling for Epilepsy" and the "Educational Internet Platform on Psychogenic Non-Epileptic Seizures" cater to patients, healthcare professionals, and the general public, enhancing understanding and support.

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- Educating the Public and Professionals.
- Community Support and Awareness: The organization hosts in-person and virtual meetings to provide support groups and thematic lectures by medical and other professionals. These sessions foster mutual support and learning, helping individuals better navigate the challenges of living with epilepsy.
- Special Events and Campaigns: HUE annually commemorates key epilepsy awareness days: International Epilepsy Day (the second Monday in February), National Epilepsy Day (February 14), and Purple Day (March 26), with the hallmark event, "Cycle for Epilepsy!" These events raise awareness and support for the epilepsy community.

What do we expect from CREA?

From the Complex and Rare Epilepsies Alliance (C.R.E.A), we expect collaboration through knowledge exchange, participation in international initiatives, support in raising awareness of rare and complex epilepsies, and strengthened advocacy for improved diagnosis, treatment, and quality of life for those affected.

What can we offer to CREA?

We can offer our long-standing experience in public education, advocacy, and patient support, as well as active participation in awareness campaigns and research initiatives. Through our established national network, educational resources, and strong collaboration with healthcare professionals, we can contribute to shared goals by promoting understanding of complex and rare epilepsies, supporting affected individuals and families, and raising the visibility of these conditions across Europe and beyond.

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CDKL5 SLOVAKIA

Vision

The CDKL5 Slovakia was established primarily to help families affected by the birth of a child with a rare genetic disease called the CDKL5 deficiency disorder.
We aim to spread and increase awareness and information about the disease among experts and the public, to better understand the challenges of the lives of families and improve the inclusion of disabled people. To raise funds supporting the disease research.
To facilitate the life path for the families by sharing the useful information about the clinical symptoms of the disease, methods of treating the symptoms and advances in research.

Mision

To help and support the health of people with special needs, i.e. people with any physical or mental disability, or people with any genetic, oncological or other serious illness: Provide social assistance, assistance in obtaining alternative sources of financing, or direct co-financing of the treatment, rehabilitation, methodological, didactic or aids according to the financial capabilities of the association; / Supporting the education, rehabilitation and comprehensive development of disabled people / Cooperation with the Slovak and foreign experts / Supporting and facilitating the education of therapists, educators, assistants and family members / Organizing of trainings, conferences, courses, seminars and other forms of education / Spreading the awareness of the rare diseases and challenges related of caring disabled people / Establishing and operating a day center / Providing various types of stimulation (physiotherapy, swimming, special pedagogical stimulation, etc.) / Raising and obtaining the funds /Events organizing.

KEY PROJECTS/ACTIVITIES

- Establishing and operating a day care center for disabled people
- Supporting families (financial, psychological, emotional, inclusion)
- Spreading awareness, educating professionals

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- Establishing of day care center providing the complex therapies for disabled people.
- Financial help to families and raising/obtaining the funds
- Educating the physicians, therapeutists and professionals dealing with rare diseases

What do we expect from CREA?

Improvement of collaboration across countries and various organisations, spreading and sharing the information / contacts, having more powerful voices on European level, improvement of engaging "smaller" countries to be a part of clinical trials / research.

What can we offer to CREA?

Project management, Clinical Trials expertise, Coordinating activities

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MATTHEW'S FRIENDS KETOGENIC THERAPIES CHARITY

Ketogenic Dietary Therapies
Information • Training • Research • Support

Vision

Matthew's Friends are a UK registered charity (since 2004) specialising exclusively in medical Ketogenic Dietary Therapies (KDT) for refractory epilepsy, GLUT1DS and PDH, for children and adults. The charity provides evidence-based information and support for families, with inspiring recipes and practical tips. Visit the charity website at www.matthewsfriends.org KetoCollege offers HCP education in Ketogenic Therapies for epilepsy; eLearning modules, free webinars and in-person annual KetoCollege Advance. Established in 2016, with expert national and international Mentors, Advance provides practical insights into the professional experience of using KDT, aiming to improve confidence and ability to manage KDT in challenging circumstances.

Mision

Matthew's Friends mission is to publicise Ketogenic Dietary Therapies and make them more available to all those who should need it, be it child or adult.
To support parents and patients to make an informed choice about the Ketogenic Diet and to source and provide the latest information available.
To support families and patients through their dietary therapy.
To support professionals in their administration of dietary therapies by way of literature, ketogenic starter packs and files, educational conferences and training meetings and provide funding, where possible.
To support research projects into the efficacy of these dietary treatments for use in epilepsy and other conditions, working with medical professionals to ensure our information is safe, relevant and current

KEY PROJECTS/ACTIVITIES

- Introduction to Ketogenic Diets for Adults with epilepsy - A guide to support an informed choice and preparation
- Founding Patrons of the International Neurological Ketogenic Society - supporting the society for health professionals around the globe
- Funding Ketogenic services in the NHS - providing a new Ketogenic Diet service in London for children with epilepsy

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- Advocacy for Ketogenic Therapies - our charity has played a key role in advocating globally for Ketogenic Therapies, over two decades.
- Hosting Global Symposia - our charity has hosted Global Symposiums on Ketogenic Therapies, bringing together international experts to support education and further confidence and understanding of Ketogenic Therapies
- Tools to achieve success - our charity has produced many resources for families and developed recipes, tips and advice to enable successful management of Ketogenic Therapy to maximise the potential of this treatment.

What do we expect from CREA?

We look forward to collaborating with other patient organisations to help achieve better outcomes for people with epilepsy. We appreciate the opportunity to make a difference together, with a unified focus on improving patient outcomes.

What can we offer to CREA?

Our experience of more than two decades in patient support and health professional networking we hope will bring value to the conversation. Our main focus is Ketogenic Therapies for the treatment of refractory epilepsy and we can offer insight into these therapies, including evidence-based research and personal experiences.

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LIFE- ASSOCIATION FOR RARE DISEASES IN CHILDREN

LIFE • ASSOCIATION FOR RARE DISEASES IN CHILDREN •

Vision

The vision of the Association "LIFE" is a society where every person affected by a rare disease has equal access to timely diagnosis, effective treatment, and comprehensive support. "Život" envisions a future where rare disease patients and their families are fully included, empowered, and supported—medically, socially, and emotionally. It aims to eliminate barriers in healthcare and ensure that no one faces the challenges of a rare disease alone. Through collaboration with healthcare systems, policymakers, and communities, the association strives to build a compassionate, informed society that values and protects the rights and well-being of this families.

Mision

The mission of the Association "LIFE" is to improve the lives of individuals and families affected by rare diseases through support, advocacy, and systemic change. The association is dedicated to ensuring timely and accurate diagnosis, access to appropriate treatment, and the availability of necessary medical equipment and therapies for every person living with a rare disease. It works in close collaboration with healthcare professionals, institutions, and decision-makers to influence healthcare policies and promote patient-centered care. "LIFE" also provides emotional and practical support to families, helping them navigate daily challenges and offering a strong sense of community. Through awareness campaigns, educational events, and active participation in national and international networks, the association raises public understanding of rare diseases and fights stigma and isolation

KEY PROJECTS/ACTIVITIES

- Zoya's law - Zoya's law mandates that if a diagnosis cannot be established within 6 months in Serbia, patients are sent abroad for testing at government's expense. It also provides families with history of genetic diseases access to free prenatal testing and preimplantation genetic testingwith genetic counselling to other family members. Zoya's law has been instrumental in saving over 4000 children by facilitating early and accurate diagnosis in rare diseases.
- Rare Diseases Database Launched incollaboration with Orphanet, this multilingual regional database provides comprehensive information on rare diseases, helping to reduce time to diagnosis and support patients, families, and professionals.
- Magazine "Word for Life" (Reč za život) A dedicated publication sharing personal stories, expert insights, and healthcare updates related to rare diseases, aiming to inform, inspire, and connect the rare disease community

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- LIFE" played a pivotal role in initiating of this law named in honour of Zoya, daughter of founder and president of "LIFE" who passed of Batten disease when she was only 9 years.
- The association has connected over 500 members across Serbia, creating a safe space for sharing experiences, resources, and emotional support. It regularly organizes workshops, conferences, and family gatherings that foster a sense of community and reduce the isolation often faced by rare disease patients.
- By collaborating with healthcare professionals, social services, and international organizations, "LIFE" ensures that families receive not only medical guidance but also psychological and social support.

What do we expect from CREA?

we expect a collaborative platform that strengthens support for individuals and families affected by rare and complex epilepsy syndromes. The alliance should facilitate earlier and more accurate diagnoses, promote access to expert care and innovative treatments, and foster research on underlying genetic and neurological causes. We also anticipate advocacy for patient-centered healthcare policies, increased awareness among professionals and the public, and the development of educational tools for caregivers. It is also necessary to define collaboration with industry.

What can we offer to CREA?

My expertise

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European Patient Advocacy Group

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CDKL5 UK

CDKL5 UK

Empowering Families
Embracing Hope

Vision

CDKL5 UK strives to be a advocate for the CDKL5 community across the UK, ensuring that the needs, voices, and experiences of those affected are heard and addressed. Together, we unite with a shared purpose: empowering children and adults with CDKL5 disorder to embrace their lives to the fullest, celebrating their individuality and achievements while providing the support they need to thrive.

Mision

The mission of CDKL5 UK is to advance the understanding, treatment, and awareness of CDKL5 disorder. While breakthroughs have been made, there is still much to learn. The organization raises funds to support global research efforts, focusing on symptom treatment, genetic causes, and epilepsy. CDKL5 UK also emphasises educating the public and professional communities, advocating for children and young adults, and challenging societal assumptions about disabilities. Funds contribute to clinical trials, research dissemination, and enhancing health services, aiming to improve the lives of those affected while fostering public and professional awareness of the condition.

KEY PROJECTS/ACTIVITIES

- Strengthening Families - Offering access to counselling services for parents and caregivers
- Fostering Family Well-Being – Dedicated support services for siblings of all ages.
- Empowering Growth – Creating specialised training for caregivers, healthcare professionals, and medical providers to help children and adults with CDKL5 thrive

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- Collaboration - Establishment of the CDKL5 Centre of Excellence in Bristol: Recognized by NHS England, this center at the Bristol Royal Hospital for Children provides specialized clinical care and holistic support for children and adults with CDD. Led by
- Strong Partnerships - CDKL5 UK collaborates with Rareminds, the UK's first specialist non-profit rare disease counselling and psychotherapy service. This partnership offers mental health support tailored to the unique challenges faced by individuals with CDD and their families
- Research-Focused – CDKL5 UK has collaborated extensively with the CDKL5 medical community to advance understanding of the condition, contributing to the development of clinical guidelines, real-world evidence, and the broader research landscape

What do we expect from CREA?

Through collaborative efforts, we hope to see real momentum in raising awareness, influencing policy, and driving progress toward better treatments. We trust that CREA will not only support our cause but also amplify our stories to inspire meaningful change.

What can we offer to CREA?

CDKL5 UK can support CREA by bringing the lived experiences of families affected by rare epilepsy, sharing advocacy and policy expertise, contributing to research collaboration, and offering models for mental health and family support. Together, we can drive meaningful change for the wider rare epilepsy community.

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European Patient Advocacy Group

EpiCARE

ASTB FRANCE

Vision

Living and curing Tuberous Sclerosis Complex

Mision

Mobilize around the STB to:
-Inform, share and connect
-Optimising medical, educational and social care
-Participate in the research effort in France and abroad

KEY PROJECTS/ACTIVITIES

- Research :mitochondrial role in Tuberous sclerosis complex epIlepsy
- Research: a connected patch that detects Warning signs of epileptic seizures and alerts the patient
- Develop care pathway coordination throughout France (directory of referring physicians)

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- Organization of a meeting for families and patients on March 15, 2025
- Collaboration on a webinar series on the affective and sexual lives of people with mental disabilities
- Simulated consultations for genetics interns. Training for all future geneticists in the diagnosis of genetic disease.

What do we expect from CREA?

Pooling medical and associative forces to fight rare epilepsies

What can we offer to CREA?

Scientific experience

Contact of the organization

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DRAVET SYNDROME CROATIA

Vision

Raise the quality of life, accept differences.

Mision

Reduce the impact of the disease by improving diagnostics, medical treatments and a holistic approach. Work together to improve the quality of life by increasing the availability of medicines, providing support to persons with rare epilepsy syndromes and their families, and connecting with organizations nationally and internationally.

KEY PROJECTS/ACTIVITIES

- EpiPrehrana: education and support program for implementing a ketogenic dietary treatment for patients with rare and complex epilepsies.
- Event organisation: for providing education and support to affected families and health and non-healthcare professionals.
- Patient advocacy: participation in legislative processes and working groups of the Ministry of Health, including the National Plan for Rare Diseases, and the Health Care Law.

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- Advocating for market access and reimbursement of critical therapies. Dravet syndrome emergency protocol implemented in the National Plan for Rare Diseases.
- An online counselling centre has been launched for families for ketogenic dietary treatment. Empowering caregivers through tailored nutritional advice and education.
- The organization has built a strong community offering educational programs, resources, and emotional support to families affected by rare and complex epilepsy.

What do we expect from CREA?

Networking and knowledge exchange with other organizations and researchers, support in the internationalization of our advocacy efforts and the opportunity to be involved in the development of research and clinical projects related to rare epilepsies, and collaboration on projects of common interest.

What can we offer to CREA?

Experience in advocating for the patients' rights, contributing in shaping policies and clinical trials from the perspective of patients

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ASOCIACIÓN KCNQ2 ESPAÑA

Vision

Raise awareness about the disease and help find improvements in the quality of life of those affected.

Mision

Learn more about other epilepsies

KEY PROJECTS/ACTIVITIES

- Transition to Foundation
- Raise funds to try to reach research into gene therapy
- Improve disease registration throughout the country

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- Financial support for the KCNQ2 Registry, developed by Sant Joan de Deu
- Better relationship with the doctors involved in KCNQ2
- Improved communication and connection with more patients with the same condition

What do we expect from CREA?

We expect to learn as much as possible about different forms of epilepsy, their relationship with KCNQ2, and find synergies regarding therapies, projects, etc.

What can we offer to CREA?

Provide all the information on epileptic encephalopathy caused by genetic alteration of KCNQ2 that can help improve its registration

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European Patient Advocacy Group

EpiCARE

ITALIAN STURGE WEBER SYNDROME ASSOCIATION

Vision

We work every day to support, inform and connect those affected by Sturge-Weber Syndrome throughout Italy. From raising awareness to research, from connecting families to daily support, our association is committed on multiple fronts to fight and give voice to Sturge-Weber Syndrome.

Mision

The Sturge Weber Italia Association began its journey in 2016, with a clear vision and a precise mission: to accompany, support and illuminate the path of families and patients affected by Sturge-Weber Syndrome. Since that date, we have grown both in number and impact, transforming a simple vision into a tangible commitment. In a space dedicated to the Association you will find an overview of our roots, the members who are part of it, how to contribute and take part in our activities, as well as important updates and bureaucratic resources.

KEY PROJECTS/ACTIVITIES

- National register for Sture Weber Syndrom in Italy
- Supporting Families on the way to tratements
- 1°Report of Sturge Wbere Syndrom situation in Italy

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- Guide Through Laser Tratement centers in Italy;
- Supported by our Stientific Comitee and all the laser tratement excellence center involved;
- Research Collaboration – The association invests in research projects to improve the quality of life of patients and their families

What do we expect from CREA?

I expect that the CREA Project will be able to create synergies between the various patient associations at European level, improving collaboration and ensuring better alignment in order to increase the impact in discussions at political and decision-making level.

What can we offer to CREA?

we can offer our experiences developed within our association starting from 2016 onwards

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European Patient Advocacy Group

EpiCARE

UK INFANTILE SPASMS TRUST

UKIST
THE UK INFANTILE SPASMS TRUST

Vision

UK Infantile Spasms Trust (UKIST) raises awareness, improves speed of diagnosis and supports families affected by infantile spasms in the United Kingdom

Mision

The UK Infantile Spasms Trust (UKIST) provides vital support to families with children affected by the rare seizure disorder Infantile Spasms (IS), also known as West Syndrome. The charity offers a lifeline to anxious parents via its online community, and works alongside medical professionals to raise awareness of this little known condition, which can be very difficult to recognise

KEY PROJECTS/ACTIVITIES

- Medical Education Platform launch - A course to train medical professionals about learning to recognise the symptoms of infantile spasms
- Health & Wellbeing guides - Online therapy sessions to support parents and carers during the infantile spasms journey
- Research Project - Participating in the brain development in early development (BEE) study, Kings College London

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- NICE guidance - UKIST is honoured to have been a stakeholder in developing and reviewing the new guidelines, which recognise that suspected infantile spasms are a unique category requiring urgent assessment by a specialist within 24 hours of presentation
- Support Network - UKIST has built an online Facebook network for families affected by infantile spasms and families are encouraged to meet up twice a year to celebrate our children.
- Grants - Funding equipment, therapy or hardship support for families affected by infantile spasms

What do we expect from CREA?

For UKIST to be a part of something to make real change for families around the world, whether that is collaborating on future research projects, campaigning for better treatment, support and funding or learning from each other about new opportunities

What can we offer to CREA?

UKIST is committed to CREA and the valuable ongoing work. Personally I can commit some time to be part of a working group to help move projects forward with passion and determination

Contact of the organization

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DE NEU ASSOCIATION

Vision

De Neu Association wants to serve as a central point of connection for everyone involved in neurotransmitter diseases, including affected individuals, families, medical professionals, and related institutions, and improve the quality of life of those affected by Neurotransmitter diseases

Mision

To promote and support medical, scientific, and technical research aimed at improving the quality of life for children with neurotransmitter diseases.
To disseminate knowledge about neurotransmitter diseases among medical professionals, educators, and the general public. To provide support to affected individuals and their families, and to foster international cooperation in research and family relations

KEY PROJECTS/ACTIVITIES

- Help research projects like Characterizing genetic variations in children with SSADH
- Organizing biennial Conference on Neurotransmitter Diseases
- Participation in Natural History Study of SSADH

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- Research Collaboration on international projects and events
- Funding of research projects with Verona University and Sant Joan De Deu Hospital
- Organizing of family meetings and common activities

What do we expect from CREA?

In our Association there are many children with Epilepsy problems. We would like to join efforts with other associations in order to raise awareness of this problem and if possible, tackle this problem from a unite form in front of Europe

What can we offer to CREA?

Support and voluntary work. We are a small association, but the project will be composed of other associations like us

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European Patient Advocacy Group

EpiCARE

RETT SYNDROME EUROPE

rse rett syndrome europe

Vision

A Europe where every individual affected by Rett syndrome is understood, supported, and empowered — through awareness, collaboration, and scientific advancement. We envision a united European community where Rett syndrome is well-known among the public, professionals, and caregivers, and where families never face this journey alone.

Mision

Our mission is to raise awareness of Rett syndrome across all European countries, ensuring that the public, healthcare professionals, carers, and those directly affected are well-informed. We strive to foster strong communication and collaboration within the European Rett community, amplifying our shared voice and promoting unity. As a representative body, we advocate for the rights and interests of individuals with Rett syndrome and their families at the European level. We are committed to supporting the formation and growth of national Rett associations, expanding our network to every European country. In all our efforts, we prioritize the advancement of research, aiming to improve understanding, treatment, and ultimately, the lives of those living with Rett syndrome.

KEY PROJECTS/ACTIVITIES

- RettX - Parent led European Patient Registry for Rett Syndrome
- Numbers - having greater accuracy on number of people with Rett in each European country
- Presume Potential - changing the narrative that everyone with Rett syndrome has a severe learning disability

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- RettX - Establishing a parent led patient registry
- Patient Advocacy - a voice for families advocating for better treatments and new therapies in Europe
- Collaboration - with international patient organisations, rare disease groups and pharma to raise awareness of issues faced by our community, to bring about change and better health outcomes

What do we expect from CREA?

From CREA, we hope to gain collaborative opportunities that strengthen our advocacy and research efforts, particularly in areas where Rett overlaps with other complex epilepsies. We look forward to shared resources, joint awareness campaigns, mutual support in policy and funding initiatives, and a unified platform to influence European healthcare and research agendas. Through CREA, we aim to connect more effectively with other rare epilepsy groups, fostering innovation and solidarity across conditions.

What can we offer to CREA?

As a dedicated European network focused on Rett syndrome, we bring deep expertise in a complex rare epilepsy, strong advocacy experience, and a well-established network of families, professionals, and researchers across Europe. We can contribute to CREA by sharing best practices, raising awareness of Rett-related epilepsy, supporting joint research initiatives, and helping amplify the collective voice of rare epilepsy communities at the European level.

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KCNQ2 FRANCE DEVELOPPEMENT

Vision

At KCNQ2 France Développement, we want every child affected by this rare disease to be seen, loved, and supported. We are here to listen, to explain, and to stand by the families who face this battle every day. Our dream? That research moves forward, that diagnoses become faster, and that every child has the means to thrive. Together, we can make a real difference

Mision

Our mission at KCNQ2 France Développement is to support families affected by the KCNQ2 genetic mutation, a rare and still too little-known disease. We offer moral, administrative, and personal support to parents from the moment of diagnosis, which is often experienced as a shock. The association creates connections between families, provides useful resources, offers opportunities for sharing, and advocates for their rights. We raise awareness among the general public and healthcare professionals to make this disease and its realities better known. We actively support scientific research to better understand the causes of KCNQ2 and promote the development of appropriate treatments. Our fight is to make progress, so that every child carrying this mutation can grow up with dignity, receiving the care, attention, and love they deserve. Because behind every diagnosis, there is a family, a story, a life full of courage and hope.

KEY PROJECTS/ACTIVITIES

- Opening of an Inclusive Respite Center: We are currently seeking premises to create a new welcoming and secure space for children with neurodevelopmental disorders.
- This center will offer comprehensive care (medical support, activities, and attentive listening) to provide relief to families and promote the social and educational inclusion of every child.
- Implementation of an Adapted Sports Program: Because movement is also progress, we are developing an adapted sports project for children living with KCNQ2.
  This program will help stimulate their motor skills, boost their self-confidence, and create social connections through fun and accessible sporting activitie.
- Training in Alternative and Augmentative Communication (AAC): We are preparing training sessions for professionals who will be welcoming children at the center.
  The objective? To train them in alternative and augmentative communication, in order to promote each child's expression, autonomy, and understanding — even for non-verbal children

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- Creation of the KCNQ2 Scientific Committee: Under the leadership of Prof. Mathieu Milh, this committee brings together renowned experts in the field of epileptic encephalopathies. A key step forward to structure and accelerate research around the KCNQ2 mutation.
- 1st National Conference on KCNQ2: A unique event that, for the first time, brought together families and researchers around the KCNQ2 gene. A powerful moment of listening, sharing, and hope, strengthening the bonds between science and those directly affected.
- Opening of the Very First Respite Center: An innovative and caring space for children with neurodevelopmental disorders, offering support to families, tailored care, and a true place of inclusion.

What do we expect from CREA?

For me, CREA represents the opportunity to unite the forces of all European associations around a common cause.
I hope this will lead to greater visibility, more resources for research, and, above all, more support and listening for families.

What can we offer to CREA?

We can offer our energy, our creativity, and our heartfelt commitment. Our community is ready to collaborate, to share its experiences, successes, and struggles, in order to advance the cause of rare epilepsies together, on a European scale.

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European Patient Advocacy Group

EpiCARE

SÍNDROME DE DRAVET
FUNDACIÓN
www.dravetfoundation.eu

DRAVET SYNDROME FOUNDATION SPAIN

Vision

At the Dravet Syndrome Foundation Spain, we believe in the power of patient organizations to change the world. We are "impatient patients", and we are proud to represent this movement. Our foundation is based on the principles of collaboration, transparency, and patient-centeredness. We believe that by placing patients at the center of our work, we can drive significant progress in the fight against Dravet syndrome.

Mision

The Dravet Syndrome Foundation Spain is a patient-led scientific organization at the forefront of Dravet syndrome (DS) research and advocacy. Our mission is to accelerate the pace of research, increase our understanding of the syndrome, and help improve the quality of life for patients and families living with this disease. We strive to create a nurturing ecosystem that promotes open communication, mutual respect, and a culture of continuous learning and improvement. Through our efforts, we aspire to be a beacon of hope and a source of tangible advancements in the field of DS. By establishing a network of reserachers, healthcare professionals, and patients and caregivers, we seek a future where a cure for Dravet syndrome exists.

KEY PROJECTS/ACTIVITIES

- FSD Laboratory - Opening of a preclinical laboratory in the Universitas Miguel Hernández of Elche, with the aim of advancing research and treatment for DS
- DS RNA-seq - Transcriptomic analysis of peripheral blood in individuals with DS and healthy controls, uncovering the differential gene expression
- Impact of the risk of SUDEP - A questionnaire to evaluate the impact of the risk of SUDEP on caregivers of patients with DEEs

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- Education - The Foundation organized webinars, outreach events, and collaborated with schools and universities to educate the community on Dravet syndrome. These efforts have significantly raised awareness and understanding of the condition.
- Support Network - The Foundation has built a strong network among families affected by Dravet syndrome, offering social programs and initiatives aimed at improving their quality of life. Additionally, every year, the Foundation hosts an annual family meeting, providing a platform for families to connect, find support, share their experiences, and learn about the latest advancements in Dravet syndrome.
- Research - The Foundation operates its own preclinical laboratory and collaborates with international researchers on both preclinical and clinical scientific projects. Additionally, the Foundation actively participates in scientific and medical conferences, including organizing the European Dravet Syndrome Conference to share the last scientific advances and further enhance collaboration among those dedicated to advancing research and understanding of Dravet Syndrome across Europe and globally.

What do we expect from CREA?

I expect the CREA Project to highlight the pivotal role of patient organizations in advancing research, shaping policies, and improving education by advocating for the real-world issues faced by patients and their caregivers. By bringing attention to these critical aspects and enhancing collaboration, I hope the project will demonstrate how patient organizations contribute to more effective and inclusive healthcare solutions.

What can we offer to CREA?

The Dravet Syndrome Foundation can contribute by sharing our expertise in education, research, and family support, and by actively participating in project activities to advocate for the needs of families affected by Dravet syndrome.

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European Patient Advocacy Group

EpiCARE

SCN2A GERMANY E.V.

Vision

SCN2A Germany e.V. is made up of SCN2A families for SCN2A families, with the objective of developing effective treatments for any type of SCN2A related disorder. While focusing on all German-speaking families and professionals, SCN2A Germany collaborates with the rest of the world to learn from each other, share data, foster research and make clinical trials available to every SCN2A patient.

Mision

SCN2A Germany e.V. welcomes all German-speaking families and professionals. We are parents of affected children and aim to educate others about this complex disease, which is related to various genetic defects in the SCN2A gene, and to accompany families and relatives on their 'SCN2A journey'. The spectrum of effects of an SCN2A gene defect is enormous. It ranges from initial epileptic seizures that disappear over time, allowing normal development, to autism, severe multiple disabilities, treatment-resistant epilepsy, and even families who have already lost their 'SCN2A child'. SCN2A Germany e.V. is here for everyone. SCN2A was only discovered in 2012 and, since the number of diagnosed patients is still small, we are committed to raising awareness about this disorder among physicians, therapists, and educators. We want to promote research, and provide parents with the latest scientific findings so that their child can receive optimal care.

KEY PROJECTS/ACTIVITIES

- "Rare As One Network" - Chan Zuckerberg Initiative (CZI) – Starting this 5-year collaboration with a focus on conferences and research
- First funding of research project – At UK Bonn (Germany), with Dr. Fazeli
- Organisation of the 3rd European SCN2A/SCN8A family conference – Leadership in the organisation of this biennial conference

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- Yearly family conferences for the German speaking families – Get together for all SCN2A families speaking German, incl. medical and scientific presentations
- Visibility of SCN2A on the Internet and social media – Website, Facebook, Instagram, WhatsApp groups, YouTube channel, first webinar, articles in specialized magazines, launch of a flyer...
- Participation in conferences and funding activities – Showing presence all over the world and collecting funds for the association

What do we expect from CREA?

We expect the CREA Project to make the area of complex and rare epilepsies more visible at all levels, hoping that a unified approach showing bigger figures will help our communities. We hope that the exchanges with other patent organisations, sharing very common issues and challenges, will help us improve the services we offer to our particular community. We assume that identified synergies will avoid duplicated efforts and help each patient organisation better allocate its resources.

What can we offer to CREA?

We can offer our figures as well as our experience gathered over the last three years, especially in terms of organisation of conferences, visibility in Internet and Social Media and relationship with stakeholders.

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OR.S.A. ORGANIZZAZIONE SINDROME DI ANGELMAN

Vision

The OR.S.A. Association (Organization for Angelman Syndrome Onlus) pursues a vision of social solidarity and the protection of the civil rights of individuals with Angelman Syndrome.

Vision of OR.S.A.

- *Enhance, assist, and protect the civil rights of individuals with Angelman Syndrome*
- *Promote and participate in research initiatives*
- *Promote and advocate for prevention and rehabilitation*
- *Represent and protect disadvantaged individuals*

Mision

The associations dedicated to Angelman Syndrome have the mission of improving the quality of life for individuals affected by this condition and supporting their families.

Mission of OR.S.A.

- *Support families with children affected by Angelman Syndrome*
- *Raise awareness about Angelman Syndrome*
- *Promote understanding, research, and clinical trials*
- *Create new opportunities for individuals with Angelman Syndrome*

KEY PROJECTS/ACTIVITIES

- Genetic Research: Study the role of protein degradation in cognitive defects, Study the alterations in sumoylation in Angelman Syndrome, Study the potential functions of the ubiquitin ligase protein UBE3A
- Registries: The Global Angelman Syndrome Registry is a project that collects data on Angelman Syndrome to understand the progression of development and to advance the research for therapies.
- The creation of a National Multidisciplinary Center to coordinate and develop rehabilitation efforts

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- In the field of information:Improve awareness of Angelman Syndrome in clinical settings, from diagnosis to rehabilitation, as well as in the socio-healthcare and educational sectors.
- In Rehabilitation: Promote and fund training courses for rehabilitation therapists on Angelman Syndrome across the entire national territory.
- Active collaboration in various genetic research projects, becoming an international interlocutor.

What do we expect from CREA?

I expect that the CREA project will help strengthen the role of patient organizations at the European level, ensuring that their voice is truly represented in political debates and research initiatives. I hope it will foster new collaboration opportunities, the development of skills, and more active and meaningful involvement of patients in decision-making processes.

What can we offer to CREA?

Each of our experiences is an added value, enriching every aspect and facet, bringing a unique perspective made of direct experiences, resilience, and creativity. Sharing projects, inclusive activities, educational or autonomy pathways already tested in Italy. Building bridges between different countries, promoting cultural exchanges, joint projects, and a stronger, more representative European or global network

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DRAVET SYNDROME HUNGARY

Dravet szindróma Magyarország

Vision

Build a community around people with Dravet Syndrome. Create opportunities for better care, education and quality of life.

Mision

We collect, organize and disseminate information about the disease to families and medical professionals.We organize yearly gatherings to strengthen the sense of community and share key knowledge. In the long term, we would like to provide oppotunities for daycare and if possible, boarding for those that cannot lead an independent life.

KEY PROJECTS/ACTIVITIES

- Clinical Trial Preparation - Prepare for upcoming disease modifying clinical trials
- Involve new families - Involve the parents of young children.
- Care about siblings - Give help to families better support the siblings.

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- First Aid Course - We have organized first aid course to prepare for BLS in epilepsy status with great success.
- Oxygen for Travel - we have provided small oxygen tanks for families who would not be able to travel without them.

  Bring Information home - We have paid for the conference participation of a young neurologist to get first hand information about novelties in the syndrome.

What do we expect from CREA?

I expect shared resources and best practices.

What can we offer to CREA?

Our insight, knowledge, but very little resources.

Contact of the organization

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APS FAMIGLIE SYNGAP1 ITALIA

Vision

The Association aims to:
- Create awareness of this rare genetic disease;
- Promote and support research;
- Disseminate issues related to SYNGAP1;
- Promote and expand a network of relationships between families, schools and health professionals;
- Facilitate genetic diagnoses to identify other people with SYNGAP1.

Mision

The missions of our Association are:
- activities to disseminate education for inclusion in school, training and work;
- activities to disseminate medical health information derived from research carried out in Italy, Europe and the world, through the website and social pages;
- activities of collaboration between families such as exchange of information and support,
hospitality and contribution to travel within maximum limits and under the conditions previously established by the Board of Directors;
- dialogue with institutions to improve the lives of all people affected by this genetic mutation and their families with social and administrative support or subsidies;
- agreements with other associations, bodies, federations;
- search for funding for scientific research and specific medical treatments;
- activities of socialization, recreational and motor activities and enhancement of cognitive functions in structures or realities dedicated and/or close to the school or residential locations of people affected by this genetic mutation.

KEY PROJECTS/ACTIVITIES

- National meeting on SYNGAP1: this event will allow families of affected subjects to meet and will also be an opportunity for discussion with doctors and researchers.
- Support for basic research in collaboration with "La Sapienza" University of Rome
- Support for clinical research in collaboration with the Child Neuropsychiatry Unit of Verona

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- New diagnoses - the association has also disseminated information on this pathology in provincial medical centers and general pediatricians.
- Support Network - The organization has built a strong community, offering educational programs, resources, and emotional support to families affected by SYNGAP1 gene mutation.
- Research Collaboration - New diagnoses - the association has also disseminated information on this pathology in provincial medical centers and general pediatricians.Actively involved in research, the Association supports patient registries and collaborates internationally to improve knowledge of this condition.

What do we expect from CREA?

I expect the CREA project to consolidate the importance of networking between patient associations. Sharing care needs is essential to formulate concrete proposals to political organizations.

What can we offer to CREA?

We can share with CREA our experience as a young association.

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RARE EPILEPSY NETWORK (REN)

Vision

The vision of REN is to support, grow and maintain a network of rare epilepsy groups formed around a living overarching structure tasked with defining REN projects, in which REN groups are able to choose to participate.

Mision

The mission of Rare Epilepsy Network (REN) is to work with urgency to collaboratively improve outcomes of rare epilepsy patients and families by fostering patient-focused research and advocacy.

KEY PROJECTS/ACTIVITIES

- Identifying innovative clusters across rare epilepsy disorders to drive deeper understanding, accelerate research, foster therapy development, and improve patient outcomes.
- Identifying innovative clusters across rare epilepsy disorders to drive deeper understanding, accelerate research, foster therapy development, and improve patient outcomes.
- Driving initiative to better characterize SUDEP and other common causes of mortality in individuals with rare epilepsies, including the development of resources for caregivers and clinicians.

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- REN hosts a one of its kind annual workshop including PAG leaders, clinicinans, researchers, government and industry stakeholders to collaboratively address cross-cutting issues - demonstrating signficcant year over year growth.
- Co-Developed and led a 2.5 day Rare Advocacy Development Workshop bringing together 28 REN disorder organizations to explore research and therapy development neurogenetic disorders through an immersive small-group format.
- Expanded REN's network by 34% in 2024, onboarding 38 new Members and Partners – growing the community to over 155 Organizations strong.

What do we expect from CREA?

Rare epilepsies diagnoses transcend geographic boundaries, andREN proudly serves a global community. While REN is rooted in the US and brings deep experience in American healthcare, policy and regulatory systems, CREA – based in Europe- offers valuable perspetives and opportunities grounded in a different healthcare framework. Together, we create space for shared learning and global collaboration.

What can we offer to CREA?

With over 12 years of uniting rare epilepsy organizations into a collaborative network, REN is excited to partner with CREA to amplify our shared impact. Together we aim to raise awareness of rare epilepsies, advocate for increased research and investment, disseminate best practices, and foster cross-network learning. We welcome the opportunity to explore joint research initiatives, co-develop educational materials and programs, create toolkits on shared priorities, and promote aligned data standards and shared access to advance the field collectively.

Contact of the organization

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AAPE ASSOCIATION D'AIDE AUX PERSONNES EPILEPTIQUES

Vision

The vision of AAPE is:

- *Achieving the best possible treatment for all forms of epilepsy in Luxembourg or in specialized centers abroad without unnecessary hurdles*
- *Having multidisciplinary care in a team available for everyone*
- *Achieving psycho-educational and psycho-social support as a care norm for people with epilepsy and their families in Luxembourg*
- *No discrimination of persons with epilepsy*
- *A population well informed about epilepsy*
- *Openness of persons with epilepsy to not hide their chronic illness anymore*

Mision

The mission of AAPE is to:

- *give those concerned by all forms of epilepsy the opportunity to speak about their experience with epilepsy and their problems*
- *inform those affected by epilepsies and the persons in contact with them*
- *raise public awareness on epilepsies to reduce prejudice and facilitate integration of people with epilepsy*
- *exchange, provide and receive information likely to help people with epilepsy and their families*
- *train those in contact with a child or person with epilepsy on the needs of persons with epilepsy, safety and first aid interventions*
- *work together to find solutions for complex problems posed by epilepsies*
- *encourage dialogue with all professional categories confronted with epilepsies*
- *provide moral, psycho-educational and social support to people with epilepsy and their families*
- *advocate for the rights of persons with epilepsy and for best possible multidisciplinary care*
- *combat stigmatization*

KEY PROJECTS/ACTIVITIES

- New information material developed and project for information material on SUDEP
- Keep our homepage up to date
- Advocate for neurologists specialized in epilepsy to work in Luxembourg

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- The positive feedback after our individual exchange and counselling sessions
- Recognition by Ministry of Education to offer education on epilepsy for teachers and children
- Advocate for neurologists specialized in epilepsy to work in Luxembourg

What do we expect from CREA?

Enhanced competencies to counsel and orient on complex and rare epilepsies

What can we offer to CREA?

Our experience in counselling

Contact of the organization

Contact us
aape@pt.lu

Web
www.aape.lu

DUP15Q FRANCE

Vision

Representation of people and families living with 15q duplication syndrome in France.
(and francophone countries such as Belgium, Switzerland and New Caledonia) in a spirit of solidarity and respect

Mision

Putting into contact and creating relationship between Dup15q families.
Offering them information about Dup15q syndrome.
Make Dup15q syndrome known to the medical and scientific community, in particular Dup15q epilepsy and behaviour.
Spread awareness in public about Dup15q syndrome.
Work together with other rare disease organisations on a national, European and international level.

KEY PROJECTS/ACTIVITIES

- Make known Dup15q syndrome (with AnDDI-rares and DefiScience
- Make known Dup15q epilepsy (with LFCE)
- Make known Dup15q behaviour (with psychiatry)

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- Advocacy for having a national protocole (pnds) (2022)
- Offering to families (in)formation (ex. website) and emotional support
- Make known Dup15q behaviour (with psychiatry)

What do we expect from CREA?

Make the voices heard of rare and complex epilepsies' organisations on an European level .
Promote research in genetic epilepsies.
Get into contact with research intitiatives being interested in Dup15q epilepsy or behaviour problems.

What can we offer to CREA?

Share CREA initiatives on our FB page and with EFAPPE. Represent our French community.

Contact of the organization

Contact us
info@dup15qfrance.fr

Web
www.dup15qfrance.fr

Social media
www.facebook.com/dup15qFrance/

European Patient Advocacy Group

EpiCARE

EPILEPSIENL

EpilepsieNL

Vision

A world without epilepsy: Until that world exists, EpilepsieNL aims to be the leading source of inspiration, support, and connection for everyone affected by epilepsy in The Netherlands.

Mision

EpilepsieNL is committed to improving the quality of life for people with epilepsy and those around them, and to finding a cure for epilepsy. We do this by fund raising, funding innovative scientific research, sharing reliable information, advocating for patients' interests, and working closely with and for our community to create lasting impact.

KEY PROJECTS/ACTIVITIES

- R&D driven by the needs of people with epilepsy
- Sharing realiable information
- Patient advocacy

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- Establishment of the first Dutch Epilepsy Research Agenda- Together with people with epilepsy and their surroundings and helth care professionals we established a research agenda
- Accurate and reliable information- The contact of our website is up to date and accurate, checked by experts involved in the epilepsy guide lines. Seizure detection and SUDEP are included
- Patient advocacy

What do we expect from CREA?

We hope C.R.E.A. will:

1. Advocate for structural European funding for comprehensive care for people with complex and rare epilepsy—not just medical treatment, but also mental resilience and social support, which are equally vital for quality of life.

2. Promote investments in research and the development of innovative therapies, including gene-modifying therapies that offer the prospect of a cure. This includes ensuring that European policy and regulatory frameworks actively support such innovation.

3. Raise awareness among European policymakers about the urgency and impact of complex and rare epilepsies, and the need for coordinated, cross-border solutions to ensure access to diagnosis, treatment, and long-term care.

What can we offer to CREA?

EpilepsieNL can make a valuable contribution—particularly in shaping research and development agendas—by translating the real needs of people with epilepsy into research priorities. Based upon these agenda's, we can build R&D trajectories with concrete and feasible goals. We have the in-house expertise to ensure that people with epilepsy and their families are meaningfully involved in research design and implementation, leading to more relevant, impactful outcomes.

Contact of the organization

Contact us
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Web
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Social media
Facebook: www.facebook.com/EpilepsieNL
Instagram: www.instagram.com/epilepsienl
LinkedIn: www.linkedin.com/company/epilepsienl

EFAPPE EPILEPSIES

Vision

EFAPPE Epilepsy: Federation of Associations for People with Severe Epilepsy. "Together for the Most Normal Life Possible". The disabling nature of severe epilepsy is what makes EFAPPE Epilepsy's actions, knowledge, and expertise unique. EFAPPE Epilepsy brings together French associations:

- National associations for a syndrome in which epilepsy is a major factor. Syndrome associations bring together families affected by these illnesses, represent them to the medical profession, and support and fuel research into these diseases. Some of them are already in a European network for their illness or in contact with other European associations for their illness or are the French-speaking association for their illness.

- Local parents' associations, created to meet local needs in French regions.

Mision

EFAPPE Epilepsy's Missions: Support the work of its member associations for people with severe epilepsy (children and adults).

1. Represent member associations wherever delegated by its members to:

- Ensure recognition and consideration of the specific characteristics of this disease for appropriate compensation for the disabilities it causes: advocacy with public authorities, disability and healthcare stakeholders, conferences, publications, research, etc.

- Develop and adapt the reception of these people in specific or non-specific structures, existing or to be created (accommodation, social life, education, activity, and employment), according to their impairment(s) and autonomy.

2. Organize cooperation between its member associations to create links in France between healthcare personnel, associations, and families.

3. Support associations seeking to become member

KEY PROJECTS/ACTIVITIES

- 1. Représenter en France les personnes handicapées par des épilepsies sévères auprès des partenaires institutionnels
- 2. Participate in the reflection on the impact of severe epilepsy in different contexts to develop institutional responses in partnership with neurologists
- NETWORK, NEWSLETTER EFAPPE epilespy: Creating links in France between healthcare staff (Cure&Care), associations and families

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- 3000 members represented in France
- Recognized expertise on epilepsy-related disability in France: representation of people with severe epilepsy with 10 national institutional partners
- NETWORK, NEWSLETTER EFAPPE epilespy: Creating links in France between healthcare staff (Cure&Care), associations and families

What do we expect from CREA?

- That the strength of the grouping at European level gives a boost to research! (on the xxx pathway or on the aa chromosome) with the involvement of associations representing patients from all European countries where they exist.

- that this research must not only focus on drug or surgical therapies (cure) but also on the quality of life of patients (care) and associated disorders (neurodevelopmental disorders, psychobehavioral disorders, see DSM5 definitions). In this respect, CREA should promote the sharing of good practices between countries by making them known to associations, which will be able to participate in the work of adapting them to the living context of their country.

What can we offer to CREA?

Our knowledge of life with severe disabling epilepsy, the link with rare epilepsy associations that cannot send a representative to CREA but are nevertheless very interested in this opportunity to move things forward together on a European scale. This is particularly important for very rare epilepsies, for recently discovered genetic mutations, but also for our associations that are already in contact with other associations of their syndrome in Europe to have a broader vision. And to see what place to give to research on epilepsies that do not yet have a diagnosis of the cause(s) e.g. Lennox-Gastaut syndrome, NORSE-FIRES syndrome.

Contact of the organization

Contact us
BernardTV@free.fr

Web
www.efappe.epilepsies.fr

Social media
https://www.linkedin.com/company/efappe-epilepsies/

ALLIANCE SYNDROME DE DRAVET

Vision

Not a single family dealing with Dravet syndrome should fight alone. Together we are so much stronger

Mision

To provide help to the families : trainings both presential and on-line,

KEY PROJECTS/ACTIVITIES

- Transition from childhood to adulthood, working with different stakeholders. Urgent need to take also care of the adults
- How to better communicate the diagnosis and possible course of the disease (developed for profesionals)
- Being in close contact with pharmaceutical companies and doctors to inform families about clinical trials etc

KEY ACHIEVEMENTS/SUCCESSES TO DATE

- Wide choice of trainings and leisure we proposed to the families
- Buccolam for adults accepted
- Being in close contact with pharmaceutical companies and doctors to inform families about clinical trials etc

What do we expect from CREA?

To speak in the name of various associations dealing with severe epilepsies // to enable best practice among these associations // to forge strong links between the members with presential meetings

What can we offer to CREA?

More than 15 years of experience. Democratic and tansparent administrative board. Successful annual meeting with families and profesionals. Very good cooperation with various professionals

Contact of the organization

Contact us
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Web
Dravet.fr

Social media
Alliance syndrome de dravet in facebook, Instagram and linkedin