Diagnóstico, tratamiento y manejo del síndrome de Dravet

07.Jul - 09.Jul 2021

Cod. O09-21

Mod.:
Streaming

Edition
2021

Activity type
Summer course

Date
07.Jul - 09.Jul 2021

Location
Live online

Languages
Spanish

Academic Validity
30 hours

Organising Committee

Description

Enrollment is free thanks to the sponsorship of Dravet Syndrome Foundation.

Dravet syndrome (DS), also known as Severe Myoclonic Epilepsy in Infancy (SMEI), is a severe and disabling neurological disease of genetic origin. Between 80 and 90 percent of affected patients have a mutation in the *SCN1A* gene. With an incidence of 1 in 16,000 births, DS is included in the group of rare diseases (1/2,500). In Spain it is estimated that there must be around 450-500 patients correctly diagnosed with DS. However, data on the prevalence of the disease suggest that this number should be higher than 1,500.

One of the most visible symptoms in DS is epilepsy, which begins to manifest itself between 4 and 12 months of age. Seizures may initially be mistaken for febrile seizures, which are common in many children. However, unlike febrile seizures, the initial seizures in DS are usually prolonged and difficult to control, leading to status epilepticus and requiring admission to the ICU. At older ages, other types of seizures frequently appear, cognitive delay becomes evident and other neurological disorders and severe behavioral problems appear. Between 15 and 20 percent of people with DS die as a result of the disease.

In Spain there are currently eight Centers, Services and Reference Units of the National Health System for refractory epilepsy, where DS could be included. However, DS is much more than an epilepsy, and its approach must go beyond treating only seizures; DS must be managed from a multidisciplinary point of view, involving health professionals from different areas. Therefore, no center is fully prepared for the complexity and multidisciplinarity involved in the management and treatment of DS. Moreover, according to current Dravet Syndrome Foundation Spain data, the Basque Country is one of the five Spanish Autonomous Communities with the highest rate of underdiagnosis of DS, especially in the group of adult patients.

The Summer Course 'Diagnosis, treatment and management of Dravet syndrome', aimed mainly at students and professional staff in the field of health and life sciences, and organized by the Dravet Syndrome Foundation Spain, raises essential aspects for the correct approach to DS: (1) Description of the signs and symptoms of DS and related epilepsies that can facilitate a clinical and genetic diagnosis, (2) Discussion of emerging treatment options for DS in the context of the current treatment paradigm, and (3) Description of the role of the multidisciplinary team in the management of patients with DS.

Objectives

To introduce students to the knowledge of a disease that, due to its infrequent nature, will be difficult to study during their years of training as future physicians.

To anticipate the education that these students will receive during future training residencies in accredited centers and teaching units for the acquisition of professional competencies specific to their specialty.

To introduce or broaden the knowledge of professional students about a severe disease that is underdiagnosed and not always well attended.

To generate the students' interest in working in the future with a group of patients and families who need prepared professionals to provide them with all the care available.

To generate as well the interest of students to research in DS.

Program

07-07-2021

16:00 - 16:05 “Registro“Presentation by the Director of the activity

Elena Cardenal Muñoz Fundación Síndrome de Dravet - Directora Científica FSD

16:05 - 16:10 Institutional Opening session

José Ángel Aibar Moreno Fundación Síndrome de Dravet - Presidente FSD

16:10 - 16:55 “Signos y síntomas del SD. Diagnóstico diferencial con respeto a otras epilepsias relacionadas “

Susana Boronat Hospital de la Santa Creu i Sant Pau - Directora del Servicio de Pediatría

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16:55 - 17:15 Break

17:15 - 18:00 “Diagnóstico genético del SD y epilepsias relacionadas“

Susana Boronat Hospital de la Santa Creu i Sant Pau - Directora del Servicio de Pediatría

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18:00 - 18:10 Break

18:10 - 18:50 “Retos clínicos a la hora de realizar un diagnóstico“

Susana Boronat Hospital de la Santa Creu i Sant Pau - Directora del Servicio de Pediatría

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18:50 - 19:00 Synthesis

08-07-2021

16:00 - 16:05 Presentation by the Director of the activity

Elena Cardenal Muñoz Fundación Síndrome de Dravet - Directora Científica FSD

16:05 - 16:50 “Enfoques actuales en el tratamiento del SD“

Vicente Villanueva Haba Hospital Universitario y Politécnico La Fe - Jefe de la Unidad de Epilepsia Refractaria y del Programa de Cirugía de Epilepsia

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16:50 - 17:10 Break

17:10 - 17:55 “Datos más recientes sobre la eficacia y seguridad de nuevos fármacos. Terapias avanzadas en investigación para SD y papel del paciente en el desarrollo de nuevos tratamientos“

Patricia Smeyers Durá Hospital Universitario y Politécnico La Fe - Neuropediatra de la Unidad de Epilepsia Refractaria
José Ángel Aibar Moreno Fundación Síndrome de Dravet - Presidente FSD

17:55 - 18:05 Break

18:05 - 18:50	“Desafíos clínicos y tratamiento de emergencia“ **Patricia Smeyers Durá** Hospital Universitario y Politécnico La Fe - Neuropediatra de la Unidad de Epilepsia Refractaria
18:50 - 19:00	Synthesis

09-07-2021

15:30 - 15:35	Presentation by the Director of the activity **José Ángel Aibar Moreno** Fundación Síndrome de Dravet - Presidente FSD
15:35 - 16:20	“Función de los miembros del equipo central y otros profesionales“ **Eulàlia Turón Viñas** Hospital de la Santa Creu i Sant Pau, Barcelona - Coordinadora de la Unidad de Neuropediatría
16:20 - 16:40	Break
16:40 - 17:25	“Impacto del enfoque de equipo multidisciplinar en la calidad de vida y bienestar del paciente y su familia“ **Carlos Ruiz García** Hospital de la Santa Creu i Sant Pau, Barcelona - Médico de Atención a la Cronicidad y Cuidados Paliativos Pediátricos
17:25 - 17:35	Break
17:35 - 17:55	“El papel de las organizaciones de pacientes en el apoyo a las familias“ **José Ángel Aibar Moreno** Fundación Síndrome de Dravet - Presidente
17:55 - 18:05	Synthesis
18:05 - 18:15	Closing session **José Ángel Aibar Moreno** Fundación Síndrome de Dravet - Presidente

Directed by

José Ángel Aibar Moreno

Fundación Síndrome de Dravet

Jose Ángel Aibar has an international background in both electronics and aerospace engineering, and holds a management position in a leading technology company. One of his children has Dravet syndrome, which gave him the motivation to become involved with the Dravet Syndrome Foundation Spain, where he serves as president and chief executive officer since June 2018.

Elena Cardenal Muñoz

Dravet Syndrome Foundation Spain, Directora Científica

Elena Cardenal Biologia Molekularrean eta Biomedikuntzan doktorea da. Urte askoz Espainian eta Suizan zientzilari lanean aritu da. Azken urtetan, Elenaren karrera kudeaketa eta komunikazio zientifiko aldera jo du, Life Siences Switzerland (LS2) erakunderako zientzia-zuzendari lanetan, baita erakunde zientifiko anitzetan sare sozialen editore eta kudeatzaile lanetan ere. Gaur egun, Elena Dravet Sindrome Fundazioko Ikerketa Zuzendaria da, gaixo erakunde honek martxan jarritako zientzia eta medikuntza jarduera guztien plangintza, antolakuntza, sustapena eta zuzendaritzaz arduratuz.

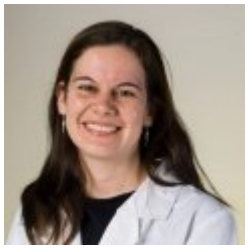
Teachers

Susana Boronat

Susana Boronat MD, PhD, was trained as a pedriatic neurologist in Vall d-Hebron hospital, in Barcelona, where later she did most of her neuropedriatic clinic work, specializing herself in dismophologic syndroms and genetic based neurologic illnesses. She undertook her PhD on type 1 Chiari malformation at the Universitat Autònoma de Barcelona. From 2011 to 2013 she carried out a clinic investigation fellowship in Massachusetts General Hospital, in Boston, at the children epilepsy and tuberous sclerosis department, directed by Dr. Elisabeth Thiele, and completed her education in research, neurogenetics and dysmorphology in Harvard University. From 2016 to 2018, she worked as a clinic geneticist at the Vall d'Hebron hospital Genetics Unit. Since 2018 she is the pediatric director of la Santa Creu i Sant Pau hospital in Barcelona. She keeps working as a neuropedriatist and clinic geneticist, focusing on genetic based epilepsy.

Carlos Ruiz García

Pediatrician specialized in pediatric palliative care, home hospitalization and neuropediatrics. Member of the Spanish Association of Pediatrics, Spanish Society of Neuropediatrics and Spanish Society of Pediatric Palliative Care. PROFESSIONAL CAREER - Pediatric Palliative Care and Chronic Care Team, Neuropediatrics Section - Hospital de la Santa Creu i Sant Pau, Barcelona (June 2020 - present). - Palliative Care and Complex Chronic Patient Service (C2P2) - Hospital Univ. Sant Joan de Deu, Esplugues de Llobregat (Nov 2020 - Jan 2021) TRAINING - University Master's Degree in Pediatric Palliative Care - Univ. Internacional de la Rioja - INTERNSHIP RESIDENT SPECIALIST Hospital Univ. Clínico San Cecilio, Granada (May 2016 - May 2020) o Sub-specialization in neuropediatrics and pediatric palliative care. o External rotations in U. Comprehensive Palliative Care in HU Infantil Niño Jesús and U. Pediatric Complex Pathology in HU La Paz, Madrid.



Eulàlia Turón Viñas

EDUCATIONAL BACKGROUND Degree: Medicine and Surgery from the University of Barcelona (UB) (1996-2002). Specialist degree: Pediatrics and its specific areas. Hospital Sant Pau (2003-2007). Master

in Neuropediatrics by the University of Barcelona. Sant Joan de Déu Hospital (2008-2010). Doctorate in the Pediatrics, Obstetrics and Preventive Medicine and Public Health program at the Universitat Autònoma de Barcelona (UAB) September 2020. Epilepsy Fellowship at Hospital del Mar (2020 – 2021) PROFESSIONAL EXPERIENCE Pediatrician specialized in Neuropediatrics and Pediatric Critical Care. Management of pediatric neurocritical patient. Coordinator of the Neuropediatrics Unit at Hospital Sant Pau. Coordinator of the Epilepsy Unit's pediatrics section at Hospital del Mar – Hospital Sant Pau Associate Professor of the Degree of Medicine of the UAB, of the Master of Neuropsychology of the UAB - Hospital Sant Pau and of the Master of Pediatric Nursing of the UB - Hospital Bellvitge.

Vicente Villanueva Haba

Vicente Villanueva MD, PhD, is a neurologist in Hospital La Fe, Valencia, since 2004. Since 2005 he also works at the La Fe Multidisciplinary Epilepsy Unit, where he is Head of the Refractory Epilepsy Unit and Epilepsy Surgery Programme. He serves as a representative of the European Reference Network Epi-CARE and is a member of the ILAE Intellectual Disability Task Force. He is an Associate Professor of Neurology at the Univ. of Valencia since 2017. Dr Villanueva undertook his training at Fundación Jiménez Diaz in Madrid (ES), Epilepsy Center of Univ. of Alabama (US), Epilepsy Center of New York Univ. (US) and Hôpital Saint-Vincent de Paul in Paris (FR). His current research interests include refractory epilepsy, video-EEG monitoring, and epilepsy clinical trials and surgery. Vicente is a member of the EEG board and the Epilepsy Guidelines Board of the Spanish Neurological Society, which awarded him in 2014 with the Scientific Prize in epilepsy and author of more than 100 articles about epilepsy.

Patricia Smeyers Durá

Dr. Patricia Smeyers is a Child Neurologist at of Hospital Univ. y Politécnico La Fe in Valencia, being the head of the pediatric section of the Multidisciplinary Functional Unit CESUR of Refractory Epilepsy and Epilepsy Surgery. PhD in Medical Genetics and awarded with the Extraordinary Prize by the University of Valencia for her Doctoral Thesis on Clinical Molecular Genetics of Friedreich's Ataxia, contributing to the isolation of the responsible gene. Neurology, Pediatrics and Clinical Neurophysiology specialist, she made training stays in epilepsy at King's College Hospital in London (UK) and at the University of Göteborg (SE). With a Childcare degree and a Clinic Neuropsychology PhD. In addition to her dedication to a lot of scientific activities of interest in the field of refractory epilepsies of genetic origin, her social involvement stands out with the publication of children's stories to understand epilepsy, a work awarded in 2011 by the Spanish Society of Neurology (SEN).

Registration fees

LIVE ONLINE	UNTIL 07-07-2021
FREE REGISTRATION	0 EUR

Place

Live online

Live online