Actualización en el diagnóstico, tratamiento y manejo del Síndrome de Dravet

Esta enfermedad va más allá de ser solamente una forma de epilepsia: su tratamiento necesita un enfoque multidisciplinario que involucre a profesionales de diversas áreas de la salud. Este Curso de Verano abordará aspectos fundamentales para el enfoque adecuado del Síndrome de Dravet.

26.Jun - 10.Jul 2025

Cod. O09-25

Mod.:
Streaming

Edition
2025

Activity type
Summer course

Date
26.Jun - 10.Jul 2025

Location
Online, 26 de junio, 3 de julio y 10 de julio

Languages
Spanish

Academic Validity
50 hours

Organising Committee

Description

El síndrome de Dravet (SD), también conocido como Epilepsia Mioclónica Severa de la Infancia (SMEI), es una enfermedad neurológica de origen genético. Aproximadamente entre el 80% y el 90% de las personas afectadas muestran una mutación en el gen SCN1A. Esta enfermedad es considerada rara, con una incidencia de 1 entre 16.000 nacimientos (1/2.500 para enfermedades poco comunes). Se estima que en España hay alrededor de 450-500 pacientes con un diagnóstico correcto de SD, aunque los datos de prevalencia sugieren que el número real podría superar los 1.500.

Uno de los síntomas notables del SD es la epilepsia, que suele aparecer entre los 4 y 12 meses de edad. Inicialmente, las crisis epilépticas pueden ser confundidas con crisis febriles comunes en la infancia, pero a diferencia de estas, las crisis iniciales en el SD tienden a ser prolongadas, difíciles de controlar y pueden desencadenar estados epilépticos que requieren hospitalización en unidades de cuidados intensivos. A medida que avanza la edad, es común la aparición de otros tipos de crisis, acompañados de retraso cognitivo, trastornos neurológicos y graves alteraciones de conducta.

Un porcentaje entre el 15% y el 20% de las personas con SD fallece prematuramente debido a la enfermedad. En España, existen actualmente ocho centros, servicios y unidades de referencia del Sistema Nacional de Salud en epilepsia refractaria, los cuales podrían tratar el SD. Sin embargo, dado que el SD va más allá de ser solamente una forma de epilepsia, su tratamiento necesita un enfoque multidisciplinario que involucre a profesionales de diversas áreas de la salud. Por esto, no hay un centro específico completamente capacitado para abordar la complejidad y la necesidad multidisciplinaria que conlleva el manejo y tratamiento del SD.

Este Curso de Verano abordará aspectos fundamentales para el enfoque adecuado del SD:

- Descripción de los signos y síntomas del SD y epilepsias relacionadas que pueden facilitar un diagnóstico clínico y genético.
- Discusión sobre las opciones de tratamiento emergentes para el SD en el contexto del paradigma de tratamiento actual.
- Descripción del papel del equipo multidisciplinar en el manejo de pacientes con SD.

Objectives

Introducir al alumnado en el conocimiento de una enfermedad que, por su condición de poco frecuente, difícilmente estudiarán durante sus años de entrenamiento como futuros profesionales médicos.

Adelantarse a la educación que el estudiantado del ámbito sanitario recibirá durante futuras residencias formativas en centros y unidades docentes acreditadas para la adquisición de competencias profesionales propias de su especialidad.

Introducir o ampliar los conocimientos del alumnado profesional acerca de una enfermedad severa infradiagnosticada y no siempre bien atendida.

Generar el interés del alumnado por trabajar en un futuro con un grupo de pacientes y familias que necesita de profesionales preparados que le proporcione todos los cuidados a su alcance. Generar a su vez el interés del alumnado por investigar en SD.

Organised by

SÍNDROME DE DRAVET
FUNDACIÓN
www.dravetfoundation.eu

Program

26-06-2025

16:00 - 16:15 “Registro“Presentation by the Director of the activity

José Ángel Aibar --- | Fundación Síndrome de Dravet - Presidente

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16:15 - 17:00 “Signos y síntomas del SD. Diagnóstico diferencial con respecto a otras epilepsias relacionadas“

Antonio Gil-Nagel Rein | Hospital Ruber Internacional - Jefe de Servicio, Director Unidad de Epilepsia

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17:00 - 17:45 “Diagnóstico genético del SD y epilepsias relacionadas“

Eulàlia Turón Viñas

17:45 - 18:15 Break

18:15 - 19:00 “Retos clínicos a la hora de realizar un diagnóstico“

Ángel Aledo Serrano | Hospital Vithas Madrid La Milagrosa - Neurólogo y Epileptólogo

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19:00 - 19:10 Synthesis

27-06-2025

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

José Ángel Aibar --- | Fundación Síndrome de Dravet - Presidente

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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 - 17:35 “Terapias avanzadas en investigación para SD. Papel del paciente en el desarrollo de nuevos tratamientos“

Simona Giorgi --- | Fundación Síndrome de Dravet - Directora Científica

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17:35 - 18:05 Break

18:05 - 18:50 “Desafíos clínicos y tratamiento de emergencia“

Manuel Toledo | Hospital Vall d'Hebron - Neurólogo y epileptólogo

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18:50 - 19:00 Synthesis

28-06-2025

15:30 - 15:35	Presentation by the Director of the activity **José Ángel Aibar ---** | Fundación Síndrome de Dravet - Presidente
15:35 - 16:20	“Impacto del enfoque de equipo multidisciplinar en la calidad de vida y bienestar del paciente y su familia“ **Esther Moraleda Sepulveda** | Nueroped - Directora
16:20 - 16:40	“El papel de las organizaciones de pacientes en el apoyo a las familias“ **José Ángel Aibar ---** | Fundación Síndrome de Dravet - Presidente
16:40 - 17:10	Break
17:10 - 17:55	“Función de los miembros del equipo central y otros profesionales“ **Victoria Ros Castelló** | Hospital de la Santa Creu i Sant Pau - Coordinadora Unidad de Neuropediatría
17:55 - 18:05	Closing session **José Ángel Aibar ---** | Fundación Síndrome de Dravet - Presidente

03-07-2025

16:00 - 16:05	“Presentación por parte de la Dirección de la actividad José Ángel Aibar “Presentation by the Director of the activity **José Ángel Aibar ---** | Fundación Síndrome de Dravet - Presidente
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
16:50 - 17:35	“Datos más recientes sobre la eficacia y seguridad de nuevos fármacos y papel del paciente en el desarrollo de tratamientos“ **Simona Giorgi ---** | Fundación Síndrome de Dravet - Directora Científica
17:35 - 18:05	Break
18:05 - 18:50	“Desafíos clínicos y tratamiento de emergencia“ **Manuel Toledo** | Hospital Vall d'Hebron - Neurólogo y epileptólogo
18:50 - 19:00	Synthesis

10-07-2025

15:30 - 15:35	Presentation by the Director of the activity **José Ángel Aibar ---** | Fundación Síndrome de Dravet - Presidente
15:35 - 16:20	“Impacto del enfoque de equipo multidisciplinar en la calidad de vida y bienestar del paciente y su familia“ **Esther Moraleda Sepulveda** | Nueroped - Directora
16:20 - 16:40	“El papel de las organizaciones de pacientes en el apoyo a las familias“ **José Ángel Aibar ---** | Fundación Síndrome de Dravet - Presidente
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17:55 - 18:05	Synthesis
18:05 - 18:15	Synthesis

Directed by

José Ángel Aibar ---

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.

Simona Giorgi ---

Fundación Síndrome de Dravet

Simona Giorgi holds a PhD in Health Biotechnology from Miguel Hernández University and a MS in Pharmaceutical Biotechnologies from the University of Bologna. Her career is marked by active collaboration in both basic and translational research projects, with a particular focus on the study of neurons and ion channels. During her doctorate, she led projects centered on the development of a pioneering in vitro model of human sensory neurons, achieving notable advances in the compartmentalized culture of neurons. Additionally, she expanded her experience with a stay at Semmelweis University in Budapest, where she delved into the study of in vitro models of human neurons. Currently, Simona serves as the Scientific Director at the Dravet Syndrome Foundation, where she centralizes and coordinates preclinical and clinical research projects. Her work is aimed at expanding knowledge about this rare disease, with the goal of improving the quality of life for patients and their families. Moreover, Simona is dedicated to scientific dissemination, combating the stigma associated with epilepsy and disability, and providing support to families through the promotion of research.

Teachers

José Ángel Aibar ---

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.

Ángel Aledo Serrano

Ángel Aledo-Serrano MD, PhD is a neurologist and epileptologist, director of the Vithas Madrid Neuroscience Institute. His main focus of clinical and research work is in the area of neurogenetics, precision medicine and developmental and epileptic encephalopathies, including Dravet syndrome and other sodium channelopathies, MOGHE, CDKL5 deficiency disorder or SYNGAP1 encephalopathy, among others. He is very active in the social and educational aspects of DEEs, with a science dissemination platform in social media (@AledoNeuro).

Antonio Gil-Nagel Rein

Dr. Antonio Gil-Nagel Rein graduated in Medicine and received his PhD from the Complutense University of Madrid. He specialised in Neurology at the Hospital Universitario 12 de Octubre and Fellowships at the Minnesota Comprehensive Epilepsy Centre. He practiced neurology at the University of Minnesota and Gillette Children's Hospital, Minneapolis and at Rush Medical Centre, Chicago. He is Associate Chief of Service and Director of the Epilepsy Unit of the Neurology Service of the Ruber International Hospital and President of the INCE Foundation. He is Director of the Epilepsy Programme at the Ruber Internacional Hospital in Madrid. He is Professor at IE University, Ad Honorem Professor at the Biomedical Technology Centre of the Polytechnic University of Madrid, and directs the Chair of Epilepsy at the Francisco Vitoria University in Madrid. He was one of the founding members of the European Epilepsy Research Network and the European Epilepsy Monitoring Association. In addition to

being a frequent speaker at national and international meetings, he is the author of several books on epilepsy and electroencephalography, and 188 articles.

Simona Giorgi ---

Fundación Síndrome de Dravet

Simona Giorgi holds a PhD in Health Biotechnology from Miguel Hernández University and a MS in Pharmaceutical Biotechnologies from the University of Bologna. Her career is marked by active collaboration in both basic and translational research projects, with a particular focus on the study of neurons and ion channels. During her doctorate, she led projects centered on the development of a pioneering in vitro model of human sensory neurons, achieving notable advances in the compartmentalized culture of neurons. Additionally, she expanded her experience with a stay at Semmelweis University in Budapest, where she delved into the study of in vitro models of human neurons. Currently, Simona serves as the Scientific Director at the Dravet Syndrome Foundation, where she centralizes and coordinates preclinical and clinical research projects. Her work is aimed at expanding knowledge about this rare disease, with the goal of improving the quality of life for patients and their families. Moreover, Simona is dedicated to scientific dissemination, combating the stigma associated with epilepsy and disability, and providing support to families through the promotion of research.

Esther Moraleda Sepulveda

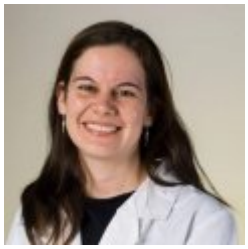
Victoria Ros Castelló

I graduated in Medicine from the University of Barcelona. I specialised in Neurology at the Ramón y Cajal Hospital in Madrid. At the end of my residency, for my quality of care and research, I was awarded the Final Residency prize by the Teaching Commission of the Ramón y Cajal Hospital, and after completing my residency I worked as an assistant in Neurology at the Sagrat Cor University Hospital. I completed the Cleveland Clinic course (Neurophysiology, EEG and Epilepsy Course) in 2020. After obtaining a scholarship from La Caixa for post-graduate studies, I did a two-year fellowship in Epilepsy

and EEG at the Montreal Neurological Hospital (McGill University, Canada). After finishing the fellowship, I took the Canadian Society of Clinical Neurophysiologists (CSCN) exam and obtained the EEG certificate. Since October 2023 I have been working at the Transversal Epilepsy Unit of the Hospital de la Santa Creu i Sant Pau and I combine my healthcare activity with research, carrying out doctoral studies at the University of Barcelona and also collaborating in several clinical trials (Marinus and Jazz Pharmaceuticals). Finally, I am part of the Telemedicine 2.0 programme of the Recover Foundation.

Manuel Toledo

Dr Toledo has more than 20 years of experience treating patients with epilepsy. He is a board member of the Spanish Society of Neurology and the Spanish Epilepsy Society. He is a scientific leader recognised for his contribution to epilepsy treatments.



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.

Registration fees

LIVE ONLINE	UNTIL 26-06-2025
General	10,00 EUR

Place

Online, 26 de junio, 3 de julio y 10 de julio

Gipuzkoa