Mitocon
Insieme per lo studio e la cura
delle malattie mitocondriali ODV

Registration is now open for the TK2d International Webinar 2026 .

TK2d across countries: diagnosis, multidisciplinary care and therapeutic pathways

Registration is now open for the international webinar "**TK2d Across Countries: Diagnosis, Multidisciplinary Care and Therapeutic Pathways"**, organised by Mitocon in collaboration with International Mito Patients (IMP) and partner patient organisations. The webinar will take place online on **Wednesday, 9 September 2026 at 3pm (CEST)**.

For the first time, the annual TK2d educational webinar will bring together leading clinicians and researchers from different countries to share clinical experience, discuss current challenges and explore the latest advances in the diagnosis, multidisciplinary management and treatment of thymidine kinase 2 deficiency (TK2d).

The initiative is specifically designed for **physicians and healthcare professionals** involved in the diagnosis and care of patients with mitochondrial and neuromuscular diseases. The webinar is organised as part of the international initiatives for **TK2d Awareness Day**, promoting greater awareness and earlier diagnosis of TK2 deficiency.

TK2 deficiency is a rare, progressive and multisystem mitochondrial disorder that may present from infancy through adulthood with highly heterogeneous clinical manifestations. The recent approval of **deoxythymidine (dT)** and **deoxycytidine (dC)** therapy by both the European Commission and the U.S. Food and Drug Administration has transformed the therapeutic landscape, making timely diagnosis, early treatment and coordinated multidisciplinary care more important than ever.

Developed under the scientific coordination of prof. **Caterina Garone**, the programme will feature internationally recognised experts discussing the issues currently shaping clinical practice, including early diagnosis, genotype–phenotype correlations, therapeutic advances and implementation challenges, multidisciplinary care and long-term management. The webinar will conclude with an international discussion on harmonising standards of care, newborn screening, emerging gene therapies and future directions in TK2 deficiency.

The programme will open with the story of a family living with TK2 deficiency, providing healthcare professionals with the patient and family perspective. This session aims to highlight the real-world impact of diagnostic delay and the importance of early recognition in improving patient outcomes.

Who should attend?

The webinar is intended for neurologists, paediatricians, child neurologists, metabolic disease specialists, medical geneticists, pulmonologists, rehabilitation specialists, physiotherapists, nutritionists, dietitians, general practitioners, researchers and all healthcare professionals involved in the diagnosis and management of rare mitochondrial and neuromuscular diseases.

The webinar will be held in English, with simultaneous translation available.

Participation is free of charge.

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Scientific Programme

TK2d across countries: diagnosis, multidisciplinary care and therapeutic pathways

3:00 pm – **Patient and Family Perspectives: The Journey Towards Diagnosis and Access to Care**

3:10 pm **- TK2d: From Gene Discovery to Therapy**

Caterina Garone, University of Bologna; IRCCS Institute of Neurological Sciences of Bologna, Italy

3:15 pm - **Diagnostic challenges and early recognition in TK2d: from genotype to phenotype and vice versa**

Caterina Garone, University of Bologna; IRCCS Institute of Neurological Sciences of Bologna, Italy

Vincent Procaccio, MitoVasc Institute, Angers University Hospital (CHU), France

4:00 pm - **Therapeutic Advances and Implementation Challenges in TK2 Deficiency**

Cristina Dominguez, CIBERER, Instituto de Investigación imas12, Hospital Universitario 12 de Octubre, Madrid, Spain

4:20 pm - **Multidisciplinary care and clinical management**

Victoria Nesbitt Oxford University Hospitals NHS Foundation Trust, Oxford, United Kingdom

Costanza Lamperti, Fondazione IRCCS Istituto Neurologico "Carlo Besta" Milano, Italy

5:00 pm - **International Perspectives: Harmonization of Protocols, Newborn Screening and Gene Therapies**

Michio Hirano, H. Houston Merritt Neuromuscular Research Center, Department of Neurology, Columbia University Irving Medical Center, New York, USA

5:45 pm - **Final Discussion and Closing Remarks**

IN COLLABORATION WITH

imp international mito patients

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AMMi Association contre les Maladies Mitochondriales

The Jeremiah Gracen TK2D FOUNDATION

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