



ΕΛΛΗΝΙΚΗ ΔΗΜΟΚΡΑΤΙΑ  
Εθνικόν και Καποδιστριακόν Πανεπιστήμιον Αθηνών  
Ιατρική Σχολή  
Α΄ Νευρολογική Κλινική  
Διευθυντής: Καθηγητής Λεωνίδας Στεφανής



# Transforming Care for Rare Diseases *Centers of Expertise in Greece*

# ***Building the path from diagnosis to access***

- ....through National and European Reference Networks
- ERNs
  - initially launched in March 2017
  - > 900 highly-specialized healthcare units  
from > 300 hospitals in 26 EU countries
  - 24 ERNs

# Aims of ERNs

- **Implement good clinical practice for the diagnosis and care for patients suffering from rare or complex diseases or diseases of low prevalence**
- **Ensure that patients have access to multidisciplinary teams of health professionals who can cover different aspects of their health problem**
- **Strengthen research, epidemiological surveillance, education and training for health professionals**
- **Promote the exchange of expertise, knowledge and best practices**
- **Cooperate and network with other Centers of Expertise at national, European or international level, as well as with patient associations in their field of action**

# ***Building the path from diagnosis to access***

- **The Greek experience**

- **1<sup>st</sup> Greek National Committee for Rare Diseases set up in 2018**

- **President: Aris Anastasakis, MD, PhD**

- Vice-President: Adamantia Englezopoulou, MSc, PhD**

- **27 Health Care Units obtained National Endorsement**

- **most of them applied to become a member of an ERN**

# ***Building the path from diagnosis to access***

- **1<sup>st</sup> Greek National Committee for Rare Diseases: Strategic Goals**
  - **Information, Training and Education**
  - **Patient care and Support**
  - **Research and Data Collection**

# ***Building the path from diagnosis to access***

- **1<sup>st</sup> Greek National Committee for Rare Diseases: Initiatives**
  - **Early diagnosis and prevention, including prenatal genetic screening**
  - **Improvement of process of recognition and organization of Centers of Excellence**
  - **Improvement of the delivery of specialized therapies**
  - **Establishment of Research hub for Rare Diseases, including relevant Registries**
  - **Information exchange with patients, families and health professionals**
  - **Educational activities for sensitization for rare diseases at pre- and post-graduate level**
  - **Recognition, Benefits, Compensation and Health Care provision**
  - **Collaboration with patient organizations**

# ***The paradigm of the First Department of Neurology at Eginition Hospital***

- ... was already a centre of reference (by reputation) for many rare neurological diseases (neurodegenerative, neuromuscular etc..)
- 2018-2019: laborious and systematic preparation to be approved as a Centre of Expertise at a National level for many rare diseases
- April 2019: national endorsement for
  - Rare Neurological Diseases
  - Rare Neuromuscular Diseases
  - Rare Epilepsies
  - Rare inflammatory and autoimmune Diseases
- November 2019: application for ERN membership
- September 2020: ERN approval to become full members in:
  - ERN-RND
  - ERN-NMD
  - ERN-RITA



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# An important step in the preparation of the application.....



EURORDIS  
RARE DISEASES EUROPE

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**EUROPEAN REFERENCE NETWORKS**

**Matt Bolz-Johnson,**  
EURORDIS

Athens, Greece  
8 December 2018

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A black and white photograph of a young child smiling broadly, with a green diagonal graphic overlay on the left side of the image.



# ***Building the path from diagnosis to access: the First Dept of Neurology at Eginitio Hospital***

- **Specialization in medical care, including specialized diagnostic tests, experts in rare Diseases, and specialized treatments-concept of personalized Medicine**
- **Multidisciplinary teams, including external collaborators, leading to the holistic management of the patients**
- **Point of reference and assistance to smaller Neurology Units across the country**
- **Development of tele-medicine approaches**
- **Programs of support and empowerment of patients and caregivers**
- **Information dissemination, to enable the diagnostic and therapeutic journey.....**
- **Annual organization of Meetings on rare Neurological Diseases: from pathogenesis to treatment**

# ***Building the path from diagnosis to access: the First Dept of Neurology at Eginitio Hospital: The future within the ERNs***

- **Exchange of knowledge, resources, expertise**
- **Networking, Training of young colleagues**
- **Updating of best clinical practices in diagnosis and management**
- **Diagnosis and Management of challenging individual cases**
- **Updating information systems for the purpose of archiving and exchanging information**
- **Participation in clinical trials**

**WE APPRECIATE THE HELP OF  
EVERYONE INVOLVED!**



<https://eginitio.uoa.gr/> , <https://eginitio-en.uoa.gr>