European Reference Network
for rare or low prevalence complex diseases

Network Epilepsies (ERN EpiCARE)

WP2 : Genetics and laboratory tests

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Orphanet thesaurus

WG: L Lagae, H Cross, R Nabbout

Reorganization according to etiology with one extra group :

- Structural
- Genetic
- Metabolic
- Immune/infectious
- Unknown and epilepsy syndromes without a clear etiology or with presumed genetic cause (ex Lennox Gastaut, rolandic)

2. If for a syndrome/disease , seizures are only a ‘minimal’ phenomenon, they should not be included
3. Duplicates (especially the genetics) will be removed as much as possible
4. Association reported in one paper about one family and one genetic finding will be removed

In collaboration with the registries WP (L Lagae)

Participation of EPICARE WP2

- Solve –RD : solving the unsolved rare diseases

Coordinator: Olaf Reis, Tubingen, Germany

Participants: ERN-RND, ITHACA, Euro-NMD, eUROGEN (4 ERNs)

  - genetic testing for unresolved cases
  - No specific budget for Epicare
  - Proposal (dead line Feb 15, 2019) within the EJP-RD 2019 to enhance collaboration between EPICARE and solve –RD (and other initiatives like Epi 25…) for possible additional access to genetic testing and analysis for countries within EPIcare network with no access to WES and to perform WGS for unresolved cases for all partners of EPICARE and functional testing for some selected mutations (Project coordinator, F Zara)

EJP RD (EPICARE IN PILLARS 2,3 AND 4)

FUNDING

- Joint Transnational Calls
- Networking support scheme
- RD Challenge (PPP collaborations)
- Monitoring of granted projects

1

COORDINATED ACCESS

- Next generation data infrastructure
- Multi-omics strategies to unravel new disease mechanisms
- Brokerage system

2

COORDINATION
& TRANSVERSAL ACTIVITIES

4

- Support to accelerate transnational and clinical research
  - access to translational research
  - Support to design CTs
  - Innovative strategies for development of therapies and CTs

Accelerating therapies

3

- Training on data management & quality
- Capacity building and training of patients
- Online academic education courses
- ERN RD research training and support programmes

CAPACITY BUIDLING

WP current and future activity

WG for genetics and laboratory diagnostics

- Consolidate the WG (one webex meeting/2months)
- Non surgical and surgical case presentation FU
  (representative of the group FU to discuss lab testing needs)
- Boost national and network meetings (virtual) for difficult to analyse cases
- Elaborate guidelines for genetic, autoimmune and metabolic testing in rare and complex epilepsies (when and how to perform the tests ?)
- Providing help and expertise in the interpretation of “difficult to analyse” results.
- Implementation with Orphanet of the new thesaurus

- We still seek for new candidates for the WG work and interest in the guidelines work