National Clinical Programme for Rare Diseases
Prof. Eileen Treacy
April 7th, 2017
European Reference Networks

http://ec.europa.eu/health/ern/toolkit_en
A European Framework for Rare Disease Services: EU 2009

Member States should:

- Identify Centres of Expertise (CoE)
- Multi-disciplinary approach to care
- Organise healthcare pathways for patients (through Centres of Expertise or Treatment abroad)
- Support the use of information and communication technologies such as telemedicine
- Foster participation in European Reference Networks
“You have to learn about thousands of diseases, but I only have to focus on what’s wrong with ME! Now which one of us do you think is the expert?”
EURORDIS European Reference Network Vision

‘Every person with a rare disease to have a home, a treatment pathway’

Networks should:

• Be developed in a step-wise approach, building on existing capacities
• Improve access to quality care, clinical excellence and patient health outcomes
• Improve integration and interoperability for RDs within Europe
• Define Patient Healthcare Pathways
• Identify experts, connect scarce expertise
• Create the critical mass of rare disease data
• Increase the pace of rare disease research
• Foster translational research and clinical trials
• Engage patient advocates
Centre of Expertise (CoE)

• These are Health Care Providers that provide multidisciplinary, patient centred care with clinical teams that manage and care for rare disease patients
Rare Disease European Reference Networks
Core Members: Nationally Designated HCPs (CBD)

- Improve high-quality specialised care for RD patients
- Promote exchange of expertise and clinical data
- Telemedicine and IT strategies
- Pool disease, diagnosis and treatment knowledge
- Provide a multi-disciplinary approach

- Share clinical guidelines and patient pathways
- Registries
- Serve as focal points for medical training and research
- Cooperation and economy of scale/cross border referral
- Collaboration with Patient Organisations

www.rd-action.eu
Criteria and conditions for all Centres of Expertise in ERNs

a) Patient empowerment and patient centred care

b) Organisational, management and business continuity

c) Research and training capacity

d) Exchange of expertise, information systems and e-health tools

e) Expertise, good practice, quality, patients safety and evaluation
For ERN membership, Centres of Expertise are Evaluated on:

✓ Documented competence, experience and activity
✓ Evidence of good clinical care and outcomes
✓ Organisation and delivery of services
✓ Skill set within the multidisciplinary healthcare team
✓ Accessibility of specialized equipment within the centre
✓ Remote communication (eHealth and telemedicine)
# ERN first call outcomes – December 2016

<table>
<thead>
<tr>
<th>Acronym</th>
<th>Proposal Title</th>
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<tbody>
<tr>
<td>BOND ERN</td>
<td>European Reference Network on BONe rare Diseases</td>
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<tr>
<td>CRANIO</td>
<td>Rare craniofacial anomalies and ENT disorders</td>
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<tr>
<td>Endo-ERN</td>
<td>Rare Endocrine Conditions European Reference Network</td>
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<td>EpiCARE</td>
<td>A European Network for Rare and Complex Epilepsies</td>
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<td>ERKNet</td>
<td>EUROPEAN RARE KIDNEY DISEASE REFERENCE NETWORK</td>
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<td>ERN GENTURIS</td>
<td>European Reference Network for GEnetic TUmour RIsk Syndromes - GENTURIS</td>
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<tr>
<td>ERN-EYE</td>
<td>European Reference Network for Rare Eye Diseases</td>
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<td>ERNICA</td>
<td>European Reference Network on Rare inherited and congenital anomalies</td>
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<tr>
<td>ERN-LUNG</td>
<td>ERN-LUNG Rare Respiratory Diseases</td>
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<tr>
<td>ERN-RND</td>
<td>European Reference Network on Rare Neurological Diseases</td>
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<td>ERN-Skin</td>
<td>ERN on Rare and Undiagnosed Skin Disorders</td>
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<tr>
<td>EURACAN</td>
<td>European Reference Network on Rare Adult Cancers</td>
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<tr>
<td>EuroBloodNet</td>
<td>ERN in Rare Hematological Diseases</td>
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<tr>
<td>EURO-NMD</td>
<td>Rare Neuromuscular Diseases European Reference Network</td>
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<tr>
<td>GUARD-HEART</td>
<td>Gateway to Uncommon And Rare Diseases of the HEART</td>
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<td>ITHACA</td>
<td>European Reference Network For Rare Congenital Malformations and Rare Intellectual</td>
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<td>MetabERN</td>
<td>A proposal for a European Reference Network on rare hereditary metabolic diseases</td>
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<tr>
<td>PaedCan-ERN</td>
<td>European Reference Network on Paediatric Cancer</td>
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<tr>
<td>RARE-LIVER</td>
<td>RARE-LIVER: A European Reference Network in Rare Hepatological Diseases</td>
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<tr>
<td>ReCONNET</td>
<td>Rare Connective Tissue and Musculoskeletal Diseases Network</td>
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<td>RITA</td>
<td>Rare Immunodeficiency, AutoInflammatory and Auto Immune Diseases Network (RITA)</td>
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<tr>
<td>TRANSCHILD</td>
<td>TRANSCHILD - Transplantation (SOT HSCT) in Children</td>
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<tr>
<td>VASCern</td>
<td>ERN Rare Multisystemic Vascular Diseases (VASCern)</td>
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24 ERN proposals
937 centres of expertise
## European Reference Networks

<table>
<thead>
<tr>
<th>No.</th>
<th>Network Code</th>
<th>Description</th>
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</thead>
<tbody>
<tr>
<td>1.</td>
<td>Endo-ERN</td>
<td>European Reference Network on endocrine conditions</td>
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<tr>
<td>2.</td>
<td>ERKNet</td>
<td>European Reference Network on kidney diseases</td>
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<tr>
<td>3.</td>
<td>ERN BOND</td>
<td>European Reference Network on bone disorders</td>
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<td>4.</td>
<td>ERN CRANIO</td>
<td>European Reference Network on craniofacial anomalies and ENT disorders</td>
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<td>5.</td>
<td>ERN EpiCARE</td>
<td>European Reference Network on epilepsies</td>
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<td>6.</td>
<td>ERN EURACAN</td>
<td>European Reference Network on adult cancers (solid tumours)</td>
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<td>7.</td>
<td>ERN EuroBloodNet</td>
<td>European Reference Network on haematological diseases</td>
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<tr>
<td>8.</td>
<td>ERN eEUROGEN</td>
<td>European Reference Network on urogenital diseases and conditions</td>
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<td>9.</td>
<td>ERN EURO-NMD</td>
<td>European Reference Network on neuromuscular diseases</td>
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<td>10.</td>
<td>ERN EYE</td>
<td>European Reference Network on eye diseases</td>
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<tr>
<td>11.</td>
<td>ERN GENTURIS</td>
<td>European Reference Network on genetic tumour risk syndromes</td>
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<td>12.</td>
<td>ERN GUARD-HEART</td>
<td>European Reference Network on diseases of the heart</td>
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<tr>
<td>13.</td>
<td>ERNICA</td>
<td>European Reference Network on inherited and congenital anomalies</td>
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<td>14.</td>
<td>ERT ITHACA</td>
<td>European Reference Network on congenital malformations and rare intellectual disability</td>
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<td>15.</td>
<td>ERT LUNG</td>
<td>European Reference Network on respiratory diseases</td>
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<td>16.</td>
<td>ERN PaedCan</td>
<td>European Reference Network on paediatric cancer (haematology-oncology)</td>
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<td>17.</td>
<td>ERT RARE-LIVER</td>
<td>European Reference Network on hepatological diseases</td>
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<td>19.</td>
<td>ERT RITA</td>
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<tr>
<td>22.</td>
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<td>VASCERN</td>
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View the list and individual information leaflets on each ERN at [https://ec.europa.eu/health/ern/networks_en](https://ec.europa.eu/health/ern/networks_en)
To support ERN collaboration activities:

- online communication
- document management
- event organisation.

~ NOT to exchange clinical patient data ~
Clinical Patient Management System is critical for:
- patient care, diagnosis and treatment; and
- future clinical research.

The focus is first on diagnosis and treatment for patients by providing tools for collaboration, virtual consultations.
Irish participation in Rare Disease ERNs

How can Irish experts participate in ERNs?
• **Voluntary** ERN participation
• Offer **multidisciplinary management** of rare diseases
• Registered on Orphanet
• Letter of approval from HSE to join an ERN

2 levels of membership:
• Full Members – meet all criteria
• Affiliated members – small centres with some criteria
Structuring the Irish Rare Disease service

‘Full member’ in an ERN (in Ireland)

• Centres of Expertise with a full multidisciplinary service, clinical research and teaching
• EC application form and European audit of service to join

Details of how to register as a Centre of Expertise, affiliated member and more about European reference networks can be found on the Rare Disease Office website:

http://www.hse.ie/eng/services/list/5/rarediseases/ernexpertcentres.html
NRDO
National Rare Diseases Office

www.rd-action.eu
National Rare Diseases Office

What is a rare disease?
A ‘rare disease’ is defined in Europe as a life-threatening or chronically debilitating disease affecting no more than 5 people per 10,000. There are an estimated 6-8,000 known rare diseases affecting up to 6% of the total EU population, (at least 30 million Europeans), and perhaps up to 300,000 Irish people during their lives. This means that 6-8% of the Irish population or 1 person in 12 may have a rare disease at some stage in their lifetime. Approximately 70-80% of rare diseases are genetic and are present throughout the person’s entire life.

Learn more about rare diseases with the Irish Easy Guide to Rare Diseases 2013 which gives you more information and real-people stories.

Who we are
The National Rare Diseases Office provides current and reliable information about genetic and rare diseases to patients, families and health professionals. It was established in June 2015 by the Health Services Executive (HSE). It is staffed by Information Scientists who have significant experience working with individuals and families affected by rare disorders. Find out what we do.

For relevant information click on one of the sections below:
› Patients and Families
› Healthcare Providers
› Service Gaps and Complaints
› ERNs Expert Centres Call for Interest
› Disclaimer

If you have a query, or if your organisation works with rare diseases and you would like to share information with us, please contact us.

www.rarediseases.ie
3. National Rare Diseases Office
Orphanet

Pan-European site
www.orpha.net

Irish section of Orphanet
www.orpha.net/national/IE-EN/index/homepage/
5. Education & research

e-learning modules available on www.rarediseases.ie

1. Recognition & Management
2. EU and Irish Policy
3. Inheritance Patterns
4. Congenital Anomalies & Screening
5. European Reference Networks

Orphanet
If you need information, we are here to help!

NRDO Website & Email

www.rarediseases.ie
rare.diseases@mater.ie

Rare Diseases Information Line

(Mon-Thurs inclusive 9.30am -1.30pm)

• 1800 24 0 365
• 01 854 5065
THANKS TO...

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Ms Jackie Turner, Information Scientist
Ms Rita Marron, Senior Administrator
Ms Grace Turner, Clinical Programme Manager
Ms Maureen Mason, Transition Nurse Co-ordinator
National Clinical Programme for Rare Diseases Working Group
National Clinical Programme for Rare Diseases Clinical Advisory Group
Dr Colm Henry, Acute Services, HSE
Ms Sharon Dwyer, Acute Services, HSE