EURORDIS
Rare Diseases Europe

WORKSHOP 5

Registries, Bio banks, Research: patients contribution to operational models of national and international initiatives

EURORDIS
Rare Diseases Europe

The International Rare Diseases Research Consortium (IRDiRC) – a collaboration effort at global level

Béatrice de Montleau

Board Member, EURORDIS
Member of the Executive Committee of IRDiRC

STARTING POINTS

- **Rare Diseases - High unmet medical needs:**
- **Vast majority of rare diseases remain untreated**
- **Delays in diagnosis – can vary from several months to even 30 years ("*The Voice of 12 000 Patients*")**
- **Rare Diseases – The facts:**
- **Each disease patients' population is small**
- **Expertise is scarce and widespread BUT exists!**

EURORDIS
Rare Diseases Europe

RARE DISEASES: HIGH COMMUNITY ADDED VALUE

- **Rare diseases – High community added value**

Rarity requires to:

- **Pool patients and expertise**
- **Share common tools: registries, data, bio samples, guidelines**
- **Harmonise data**
- **Enhance dialogue amongst researchers, healthcare professionals, patient groups & policy makers**

EURORDIS
Rare Diseases Europe

BUILDING COOPERATION: STRUCTURED & SUPPORTED NETWORKS

- **EURORDIS (> 1997)**
- **ORPHANET (>1998)**
- **European Reference Networks for Rare Diseases (pilots since 2008 - expected expansion from 2014 until 2020)**
- **E-RARE 1 & 2 -Transnational funding for RD collaborative, multidisciplinary projects**
- **Clinical Research Networks (e.g. ECRIN 2006)**
- **Common Infrastructures on Biobanks (e.g. EuroBioBanks > 2001) and Registries (EPIRARE, 2012-2018)**
- **EUCERD Joint Action (2012-2015 +)**
- **IRDiRC: International Rare Diseases Research Consortium (>2011)**

EURORDIS Rare Diseases Europe

The EU: A major player in funding health research in rare diseases

- Over two decades of investment in the area
- Over € 430 million invested in current programme
- More than 60 collaborative research projects ongoing
- Continued strong investment through the next funding programme foreseen

BUILDING a VISION for RARE DISEASE RESEARCH

The International Rare Diseases Research Consortium (IRDiRC)

A mechanism to catalyse collaboration on RD research on a global scale

Two main objectives by 2020:

200 new therapies for rare diseases

Diagnosis for most rare diseases

EURORDIS
Rare Diseases Europe

WHY Research on RARE DISEASES?

- **The ethical and social justice imperative: “extra” vulnerability demands “extra” ordinary measures**
- **Public intervention to overcome a perceived lack of attractiveness**
- **Research on rare diseases brings wider benefits**
  - Research on RDs has proven to be very useful to **better understand the mechanism of common conditions**
  - RDs are at the forefront of **personalised medicine**
  - RDs are also a **laboratory for new health care policies**

EURORDIS position paper WHY Research on Rare Disease:
http://www.eurordis.org/sites/default/files/publications/why_rare_disease_research.pdf

EURORDIS
Rare Diseases Europe

WHY NOW?

The time to collaborate in rare diseases research is now !

- New knowledge thanks to advances in research
- Increased number of orphan drug designations need further research
- Drawing upon successful examples of international research collaborations

EURORDIS
Rare Diseases Europe

HOW? Patients Priorities and Needs for RD Research

- **Allocate more funds to basic, translational and clinical research**
- **Support disease registries and biobanks as preconditions for research; harmonise data collection**
- **Understanding underlying mechanisms (genetic basis, molecular and pathophysiological mechanisms and the natural history)**
- **Reinforce multidisciplinary networks and integrated action (ERNs, international platforms e.g. IRDiRC)**
- **Long-term sustainability of RD research projects**
- **Empowering patients as full research partners**

Ultimate priority : translate research into therapies!

EURORDIS position paper on Patients' Needs and Priorities:
http://www.eurordis.org/sites/default/files/publications/what_how%20_are_disease_research_0.pdf

EURORDIS
Rare Diseases Europe

HOW?

The International Consortium for RD Research: An initiative for global cooperation launched in April 2011

- **Organisations investing in rare diseases research - each member commits to invest 10 million US$ over 5 years**

  public health research institutes, universities, public health bodies, pharmaceutical companies, umbrella patient organisations, foundations

- **Each organisation funds research its own way …**
- **…BUT funded projects adhere to a common framework**

25 members in May 2012

23 committed members

Europe

European Commission

German Federal Ministry of Education and research

Italian Higher Institute of Health Research

Italian Telethon Foundation

French Association against Myopathies

French National Research Agency

Netherlands Organisation for Health Research and Development

Lysogene (FR)

Prosensa (NL)

Spanish Carlos III Health Institute

UK National Institute for Health Research

North America

Canadian Institutes for Health Research (CA)

Genome Canada (CA)

Sanford Research (US)

Mendelian Disorders Genome Centres(US)

National Centre for Translational Therapeutics (US)

National Cancer Institute (US)

National Institute of Neurological Disorders and Stroke (US)

National Institute of Arthritis and Musculoskeletal and Skin Diseases (US)

National Institute of Child Health and Human Development (US)

National Eye Institute (US)

Office of Rare Diseases (US)

Australia

Western Australian Department of Health

HOW?

An International Consortium for RD Research: The strength of collaboration

- Mobilise the necessary **critical mass** of expertise and resources
- **Avoid overlaps** in research allowing for more diseases to be tackled
- Speed up the **uptake** of research efforts into **clinical practice**
- Deliver new **cures and diagnoses** to treat patients world-wide

EURORDIS
Rare Diseases Europe

HOW will it work? The IRDiRC governance

Executive Committee

– Funding members

– 3Patients'representatives: EURORDIS, NORD, Genetic Alliances

– Interim Executive Committee chaired by Dr. Ruxandra Draghia Akli

Scientific Committees

Diagnostics	Interdisciplinary	Therapies
including sequencing and characterisation	*incl. ontologies, natural history, biobanking, registries*	*incl. pre-clinical and clinical development*

– Top experts in various fields
–All stakeholders (academics, industry, patients, research bodies)
–Advise on research priorities

Working Groups

– Representatives of funded projects

Sequencing	Ontologies	Model systems	Clinical
Registries	Natural history	Biomarkers	etc.

EURORDIS Rare Diseases Europe

Patients' contribution in IRDiRC

- **EURORDIS involved in the IRDiRC since its conception and first meetings**
- **Active contribution to the definition of IRDiRC policy and governance**
- **EURORDIS has 1 representative at the Executive Committee (Béatrice de Montleau) – other patients' reprsentatives are from GeneticAlliance (US), NORD (US), AFM (France)**
- **1 EURORDIS representative appointed at the THERAPIES Scientific Committee (Maria Mavris)**

IRDiRC timeline

Launch of IRDiRC

Scientific Committees

3000 diagnostics

6000 diagnostics

YEAR **2012** **2015** **2020**

Working Groups

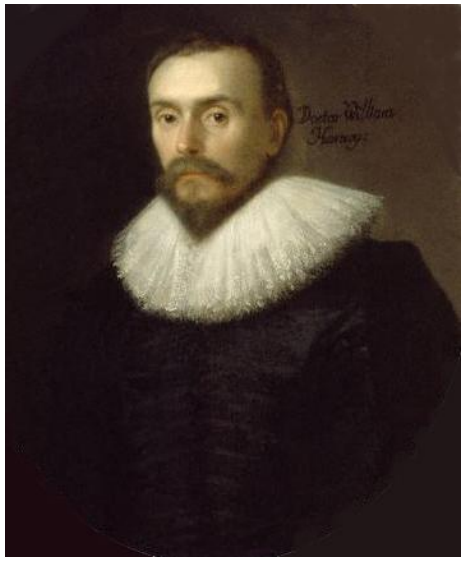
50 new applications for market authorisation

200 new applications for market authorisation

THANK YOU !

"Nature is nowhere accustomed more openly to display her secret mysteries than in cases where she shows tracings of her workings apart from the beaten paths; nor is there any better way to advance the proper practice of medicine than to give our minds to the discovery of the usual law of nature, by careful investigation of cases of rarer forms of disease"

William Harvey, English physician (1578-1657)



EURORDIS
Rare Diseases Europe