THE FUTURE OF HEALTH RESEARCH AND INNOVATION IN EUROPE: THE NEED FOR STRATEGIC ACTION

23 May 2012, Brussels
RARE DISEASES: EUROPEAN COORDINATION AT A GLANCE

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Rare Diseases are a EU Public Health challenge with high unmet medical needs and high community added value.
RARE DISEASES: HIGH UNMET MEDICAL NEEDS

• 1 Rare Disease = < 1 in 2000 persons

• Over 5 000 rare diseases

• Estimated 25-30 million people in the EU*

• 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”)

*Source: Commission Communication on “Rare Diseases: Europe’s challenges”
RARE DISEASES: HIGH COMMUNITY ADDED VALUE

• Each disease patients’ population is small

• Expertise is scarce and widespread BUT exists!

• 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

“The Future of Health Research and Innovation in Europe, 23 May 2012, Brussels
A STRATEGIC VISION AND STRATEGY FOR EUROPE

A unique EU strategy

- integrated
- comprehensive
- long term
- to address RD patients’ needs

EU and national level
- research, drug development & access,
- health care provision, codification,
- information, patient empowerment

until 2020
- trying to build in sustainability

driven by patient advocacy & expert
- based on patients’ and families’ needs
- in partnership with stakeholders
BUILDING THE EU
REGULATORY and POLICY FRAMEWORK

• EU Regulation on Orphan Medicinal Products (1999)
  In 2012 - 975 Orphan Drugs Designated + 63 Products Approved, benefiting approximately 3 million EU patients

• EU Regulation on Medicinal Products for Paediatric Use (2006)

• EU Regulation on Advanced Therapies (2007)

• Communication from the European Commission: “Rare Diseases: Europe’s challenges” (2008)

• Council Recommendation on an Action in the field of Rare Diseases (2009) - Recommends national plans for rare diseases in all EU Member States by 2013: “Research on Rare Diseases”: one of the 7 Pillars of the Recommendation

• Cross Border Health Care Directive (2011) - Mobility across EU of patients, data and expertise

• Public Health Programmes and Framework Research Programmes (HORIZON 2020)
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)
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
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
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
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%20are_disease_research_0.pdf
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 $ 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

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)
- Proensa (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

23 committed members -
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
HOW will it work? The IRDiRC governance

Executive Committee

− Funding members
− 3 Patients’ representatives: EURORDIS, NORD, Genetic Alliances
− Interim Executive Committee chaired by Dr. Ruxandra Draghia Akli

Scientific Committees

Diagnostics
including sequencing and characterisation

Interdisciplinary
incl. ontologies, natural history, biobanking, registries

Therapies
incl. pre-clinical and clinical development

Working Groups

Sequencing
Ontologies
Model systems
Clinical

Registries
Natural history
Biomarkers
etc.

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

− Representatives of funded projects

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IRDiRC timeline

Launch of IRDiRC

Scientific Committees

3000 diagnostics

6000 diagnostics

YEAR

2012

Working Groups

2015

50 new applications for market authorisation

2020

200 new applications for market authorisation
“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)