



RARE DISEASES

AN OPPORTUNITY FOR PRIMARY CARE

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RARE & MANY

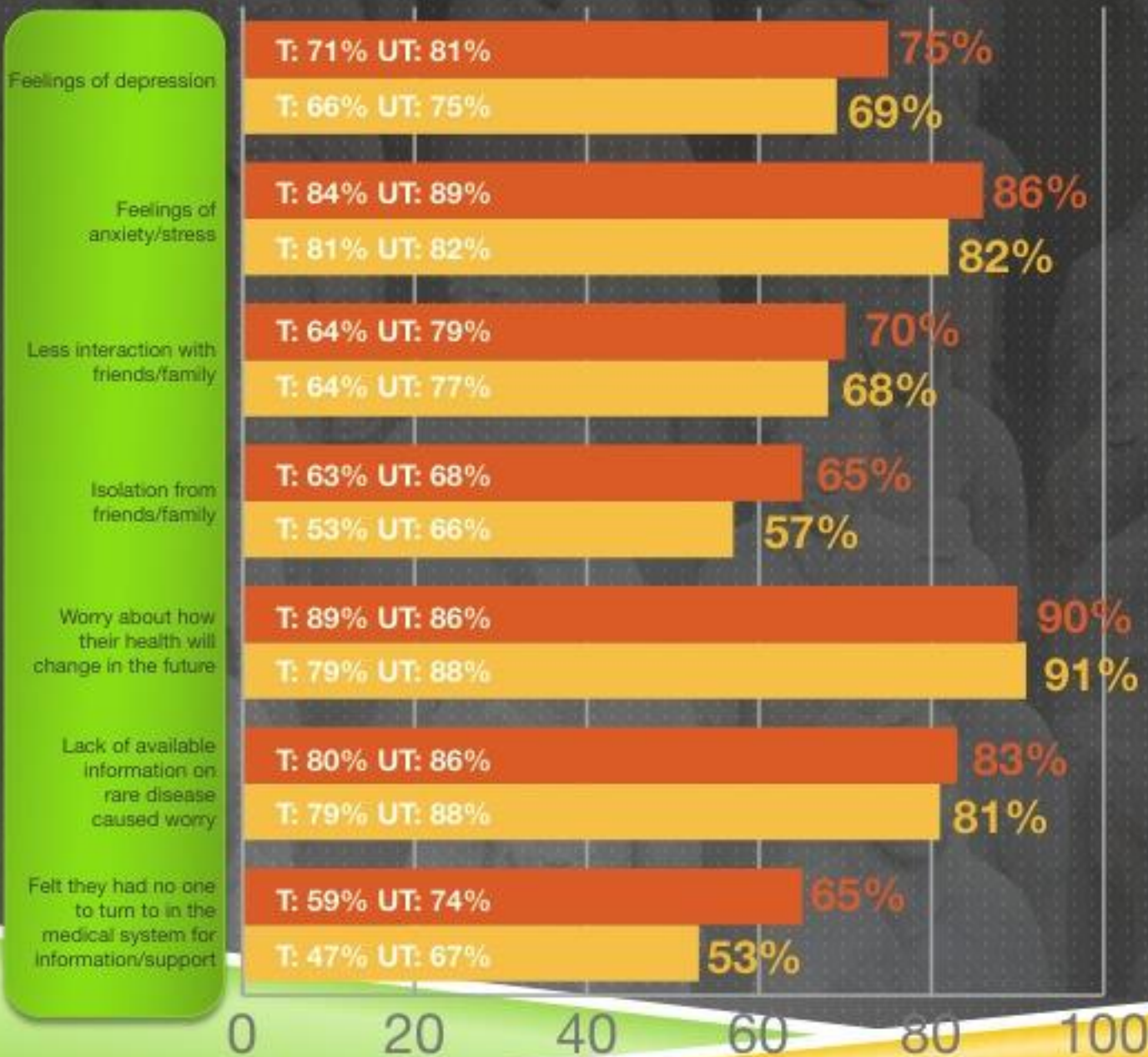


- ▶ The RD Community success story
- ▶ The Lessons Learned
- ▶ The State of the Art
- ▶ New Landscape of Primary Care Practice
- ▶ Facing New Challenges

UNMET NEEDS

- ▶ Problems listed by patients (EURORDIS)
 - ▶ Lack of access to correct diagnosis
 - ▶ Lack of information
 - ▶ Lack of scientific knowledge
 - ▶ Social consequences
 - ▶ Lack of appropriate quality of care
 - ▶ Inequities in treatment and care
 - ▶ High cost of existing drugs and care

IMPACT



DIAGNOSIS PROCESS

According to patients surveyed, it takes:



on average **7.6 years in the US**



on average **5.6 years in the UK**
for a patient with a rare disease to
receive a proper diagnosis

According to patient/caregiver respondents, in order to
get a proper diagnosis, a patient typically visits up to

8 physicians: 4 primary care and 4 specialists



and receives **2** to **3** misdiagnoses

Fabry disease: : average delay of 15 y after clinical presentation

EURORDIS survey : 25 % of respondents (n=5980), 5 to 30 y from
onset of symptoms and diagnosis confirmation, 40% reported an initial
wrong diagnosis

BARRIERS

Barriers	Percentage of US physician respondents that agreed with statement	Percentage of UK physician respondents that agreed with statement
More difficult to address the needs of a rare disease patient in typical office setting	92%	88%
More office visits are required to diagnose	98%	96%
More office visits needed to adequately address symptoms	92%	88%
Medical professional organizations do not give enough attention to rare diseases	46%	50%
Aren't enough opportunities to network with other physicians who treat rare diseases	54%	62%
Difficult to coordinate with other physicians when managing a patient with a rare disease	76%	88%
Adequate and effective treatments are less available once patient is diagnosed	86%	90%

THE FINANCIAL BURDEN

Assigning a monetary value to the loss in quality of life of \$75 000 per QALY, the mean per-patient annual intangible cost of DMD was estimated at between \$37 980 and \$46 080

	Direct medical costs and informal care	Indirect costs	Intangible costs	Total annual cost per patient	Total costs per year based on prevalence
Germany	42,360	20,770	45,860	109,000	278 058 000
Italy	23,920	18,220	37,980	80,120	154 465 000
UK	54,160	18,700	46,080	118,950	200 478 000
US	54,270	21,550	45,080	120,910	1 217 373 000

RARE & MANY



▶ Rare

- ▶ 1/1200 in the US
- ▶ 1/2000 in the EU
- ▶ 1/2500 in Japan

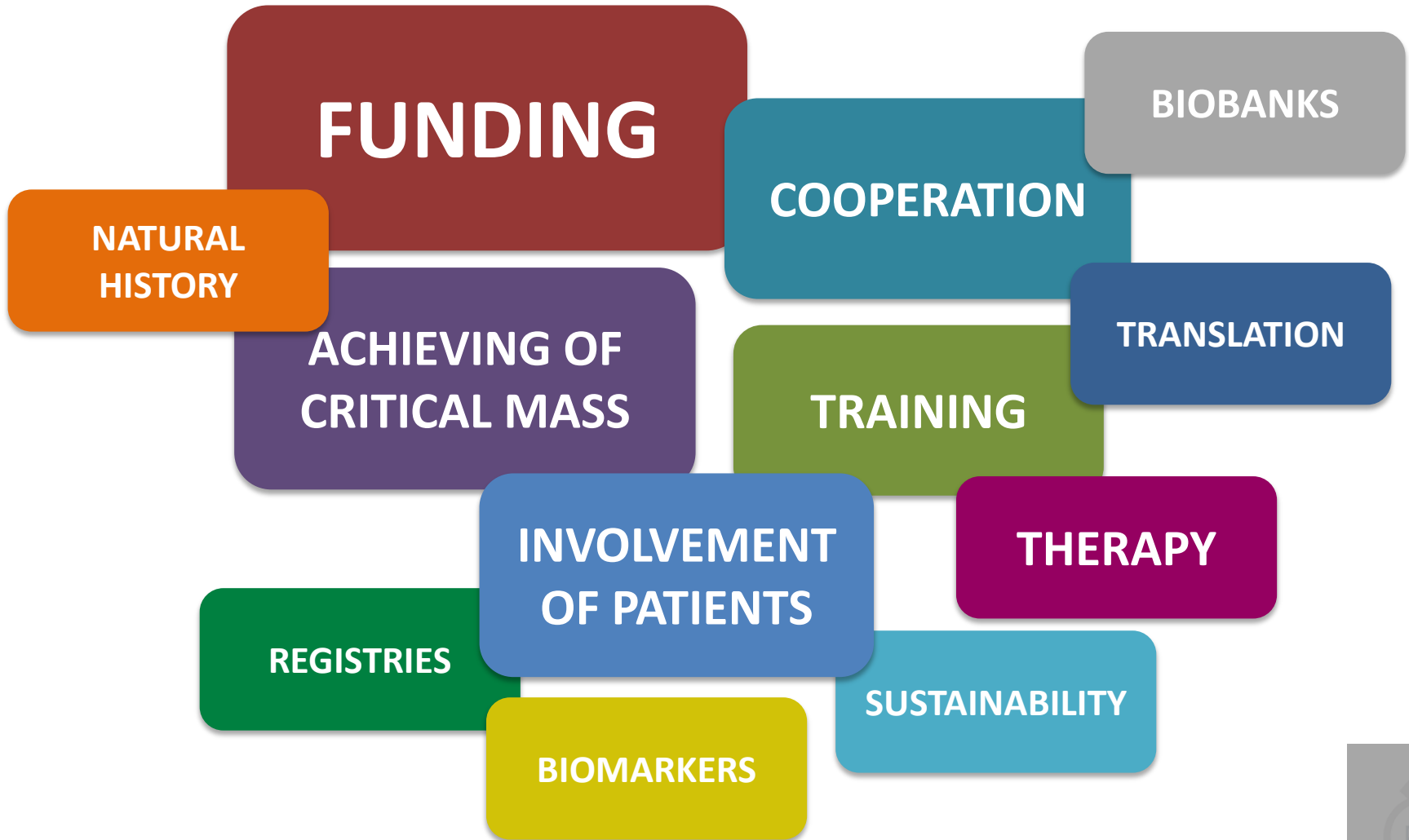
▶ Many

- ▶ 7000 diseases
- ▶ 8% of the population , 25 M in the US
- ▶ 6-8% and 30 M in Europe

RARE & MANY = COLLABORATION

- ▶ Respond to patients' expectations
- ▶ Mobilisation of critical mass of expertise and resources
- ▶ Avoid overlap
- ▶ Deliver new cures and diagnoses
- ▶ Bridging gaps
- ▶ Collective intelligence
- ▶ Gathering all stakeholders***

RESEARCH ISSUES



EU FUNDING

Over two decades of investment in the area

7th EU Research Framework Programme (2007-2013):

- Over € 620 million invested in close to 120 collaborative projects
- Plus more than 100 individual fellowships, grants and training networks



47 projects

€ 64 million



59 projects

€ 230 million



~ 120 projects

> € 620 million

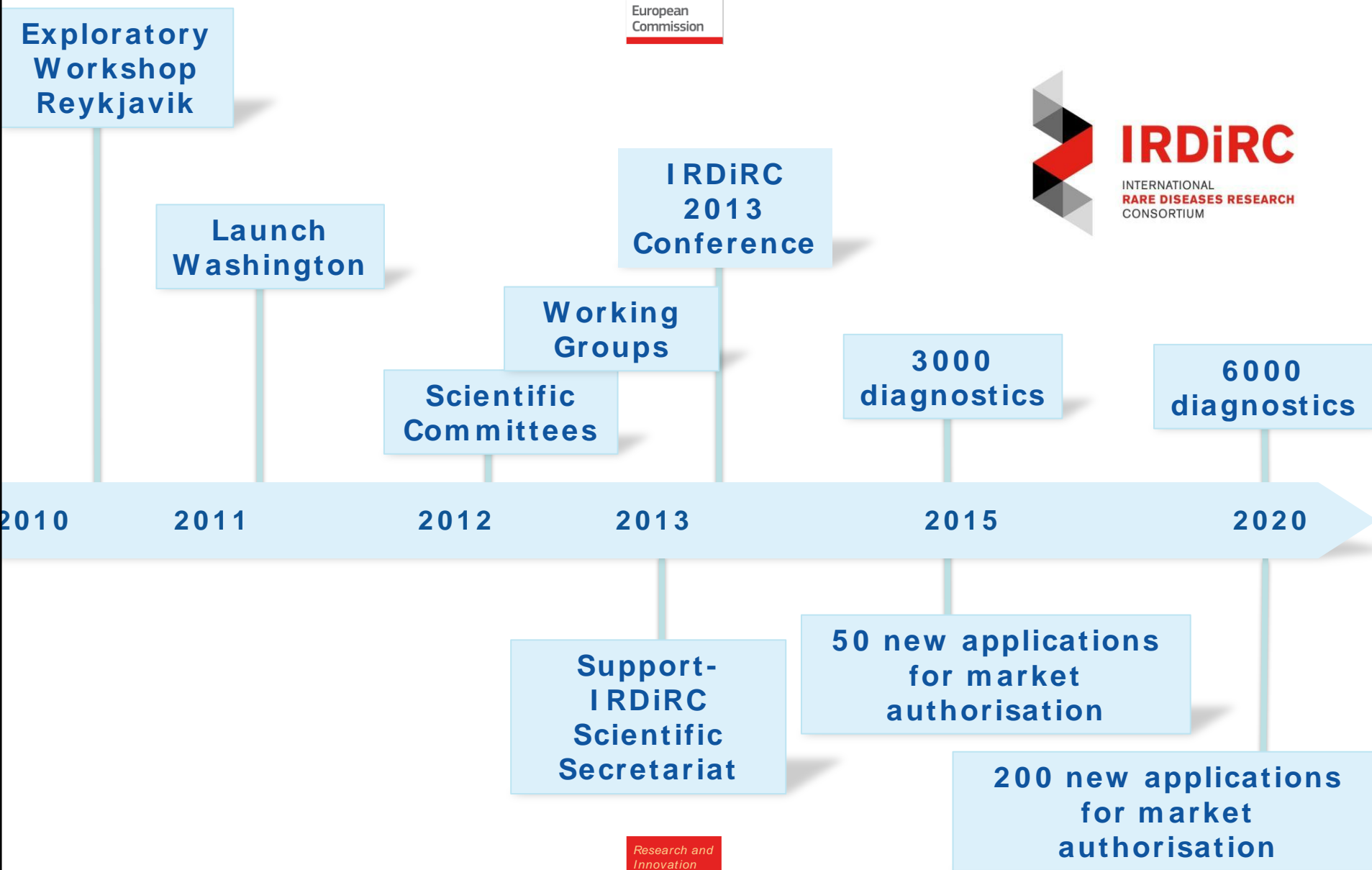
EU funded collaborative research in rare diseases

- Europe wide studies of natural history and pathophysiology
- In vitro/in vivo models
- Registries & bio-banks
- Identification of biomarkers
- Clinical trials methodologies for small populations
- -omics for rare diseases and linking data
- Development of preventive, diagnostic and therapeutic interventions

WORLDWIDE



COLLABORATION

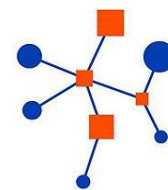


E RARE 3: COLLABORATION INFRASTRUCTURES

EUROPEAN RESEARCH INFRASTRUCTURES:

BIOBANKS, CLINICAL TRIALS, TRANSLATIONAL
MEDICINE, MOLECULE SCREENING, MOUSE
MODELS

- Facilitated access
- Exchange of best practices
- Models for sustainability
- Communication & training



BBMRI

Biobanking and
Biomolecular
Resources Research
Infrastructure



INFRAFRONTIER

mouse disease models

2014 REPORT ON THE STATE OF THE ART OF RARE DISEASE ACTIVITIES IN EUROPE



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PART IV: EUROPEAN MEDICINES AGENCY ACTIVITIES AND OTHER EUROPEAN ACTIVITIES IN THE FIELD OF RARE DISEASES

REPORT ON THE STATE OF THE ART OF RARE DISEASE ACTIVITIES IN EUROPE

PART I: OVERVIEW OF RARE DISEASE ACTIVITIES IN EUROPE

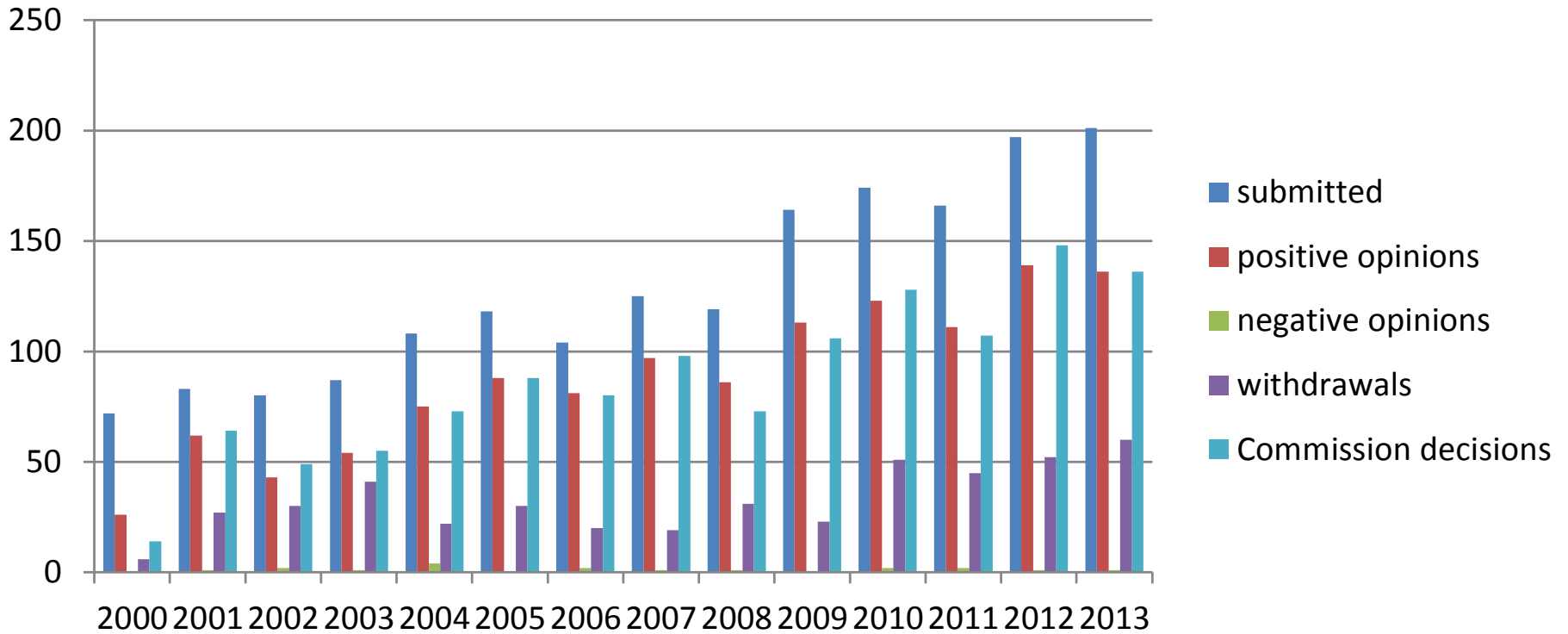


PART II: KEY DEVELOPMENTS IN THE FIELD OF RARE DISEASES IN EUROPE IN 2013

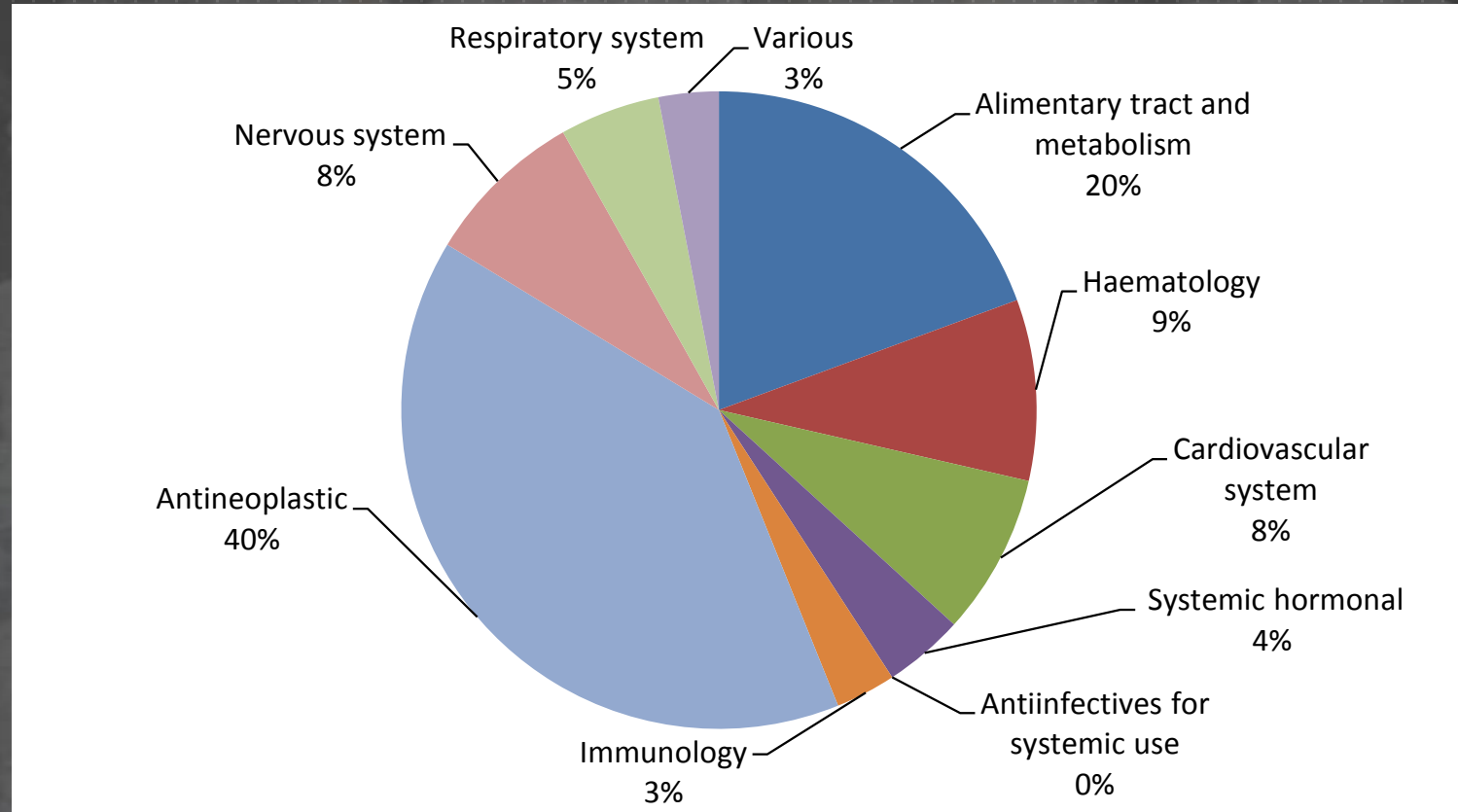
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ORPHAN DRUG: STATUS DESIGNATION



OD POSITIVE OPINION BY THERAPEUTIC AREA



PATIENT TRAINING



EURORDIS in brief

EURORDIS is a patient-driven alliance of patient organisations and individuals active in the field of rare diseases

EURORDIS' mission is "to build a strong pan-European community of patient organisations and people living with rare diseases, to be their voice at the European level, and – directly or indirectly – to fight against the impact of rare diseases on their lives"

Today, EURORDIS has 423 member organisations in 43 countries, of which 29 are European Member States and represents more than 1000 different rare diseases.

EURORDIS and Orphan Drug Development

EURORDIS played a front-line advocacy role in obtaining the EU Regulation on Orphan Drugs (1999), some key measures for patients in Revision of EU Pharmaceutical legislation (2003), EU Regulation on Paediatric Use of Medicines (2006) and the EU Regulation on Advanced Therapies (2007).

Since 2000, EURORDIS officially represents the 30 million rare disease patients at the European Medicines Agency (EMA) as members of various committees and working parties responsible for different regulatory steps of orphan drug development.

EURORDIS is also active in improving quality of information on and access to therapies for rare diseases.

ORGANISERS

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Therapeutic Development Director

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Tel: +33 1 56 53 52 19



EURORDIS SUMMER SCHOOL 2010



2008



2009

EURORDIS Summer School in Regulatory Affairs and Health Technology Assessment (HTA) for Advanced Patient Advocates



Fundació Doctor Robert
UAB

Inserm

EUROPEAN COMMISSION

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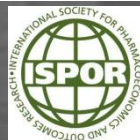
EUROPEAN COMMISSION

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www.eurordis.org

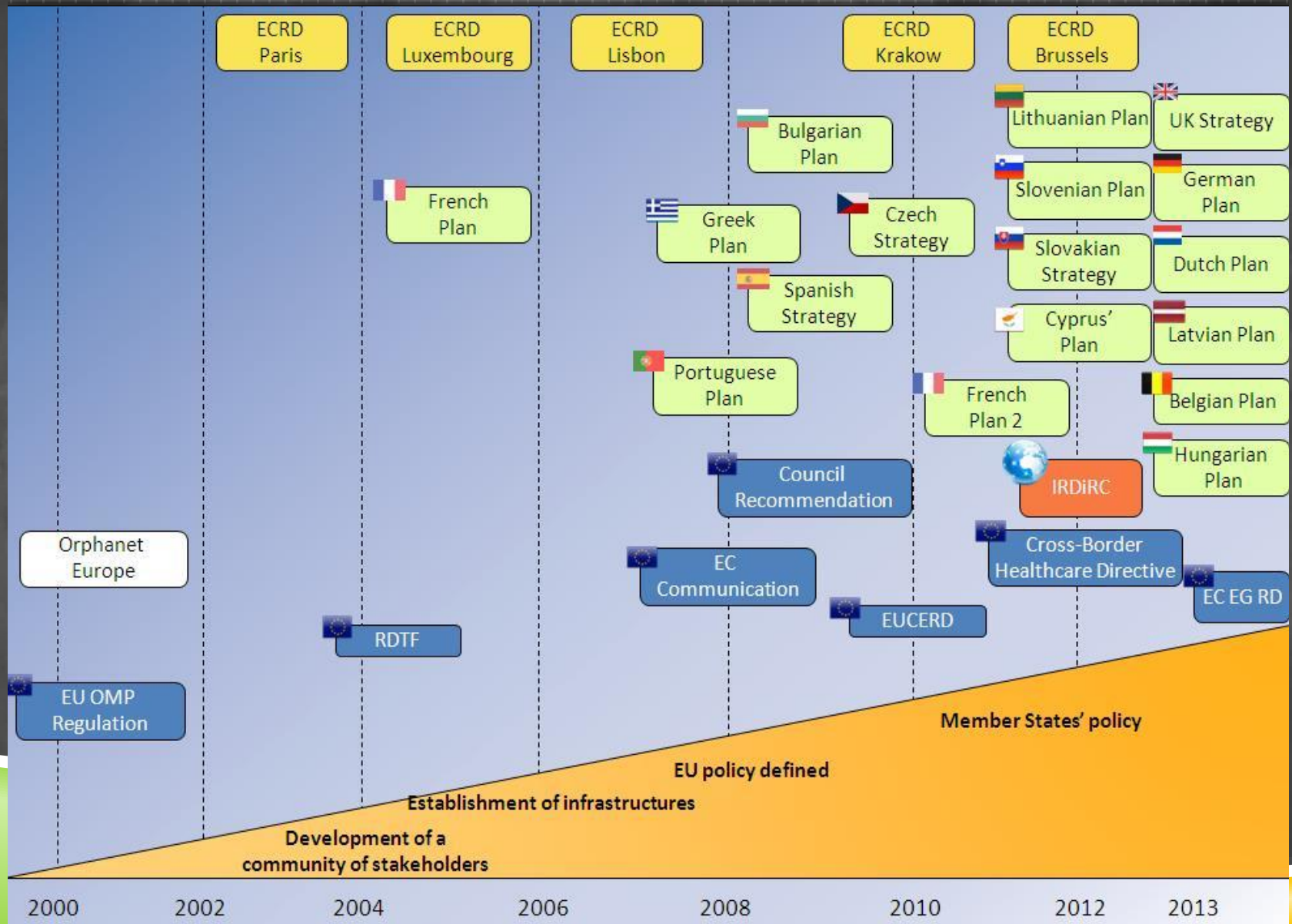


Web:
www.patientsacademy.eu

Twitter: @eupatients
as well as:



NATIONAL RD PLANS



SOCIAL HEALTH

Every year, 28% of the PLWRD needed the assistance of a social worker.

92% of PLWRD consider that «informing patients about their rights and guiding them towards social services, schools, leisure activities or vocational guidance» is necessary

Social assistance services respond inadequately to the expectations and needs of PLWRD

On average, 16% of PLWRD (up to 24% for the low income group) were forced to move house because of their disease;

FRANCE RD NATIONAL PLAN (I)

131 National centres of expertise were designated in University hospitals (2005-2007), then 500 centres of competence in regional hospitals organised in disease specific networks linked to the centres of expertise (2007-2008)





Missions of the centres of expertise (« centres de référence » in French) :

- Improve diagnosis, organise pluridisciplinary care from birth to end of life
- Expertise and second opinion
- Research, epidemiological surveillance and clinical trials
- Production of National protocols for diagnosis and care, participation in European guidelines if possible
- Information and training of health and social professionals, patients and their family,
- **Coordination with provision of primary care, medical and social care**

During the 1st Plan, two pilot networks in the field of social services were launched by centres of expertises in the regions of Pays de la Loire and Languedoc Roussillon, as well as a few other initiatives in other regions, (in particular therapeutic education programmes).

FRANCE RD NATIONAL PLAN (2)

1. Creation of about 25 disease specific networks

-  Composition : all stakeholders - centres of expertise, diagnosis and research laboratories, patient associations, **social professionals**, care networks -
-  Aim : strengthen them, share resources and tools, and cover all rare diseases and patients with unclear diagnosis in the long term :
-  Missions: to reduce diagnostic delay for all diseases, including the very rare, improve legibility of the health care system for all, develop continuity of medical care, diagnostic and therapeutic innovation, basic, clinical and translational research and **social care**.
-  call for proposals in 2013, 15 networks already identified. Governance and coordination shall be supported by the Ministry of Health in 2014.

FRANCE RD NATIONAL PLAN (2)

AnDDI-Rare developmental anomalies and malformations
CARDIOGEN transmitted heart diseases
DEFI SCIENCE (Challenge for Science) brain development diseases and intellectual disabilities
FAI2R rare auto-immunes and auto-inflammatory diseases
FILFOIE rare liver diseases
FILNEMUS neuromuscular diseases
FIMARAD rare dermatological diseases
FIRENDO rare endocrine diseases
G2M rare hereditary diseases of metabolic origin
MARIH immuno-hématologic rare diseases
MCGRE rare diseases of red cells and of erythropoïesis
MUCO cystic fibrosis and CFTR anomalies
ORKID rare kidney diseases
RESPIFIL rare respiratory diseases
SENSGENE rare sensory diseases
SLA amyotrophic lateral sclerosis

RARE & MANY = COLLABORATION

- ▶ Respond to patients' expectations
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PRIMARY CARE ACTORS ?

NEW LANDSCAPE OF PRIMARY CARE

- ▶ Re-appraisal
 - ▶ Role and Responsibility
- ▶ Rebuilding the ecosystem
- ▶ Re-positioning
 - ▶ Front Line
 - ▶ Go-between actor

PRIMARY CARE

- ▶ Front line
- ▶ Management of undifferentiated problems in unselected patients
- ▶ Management of common problems in common patients
- ▶ And ...extraordinary, **complex** cases in ordinary practice...
- ▶ “Narrative Medicine” Centered approach
 - ▶ Story telling, anecdotes
 - ▶ Diagnosis delivery, announcement
 - ▶ Decision making
 - ▶ ...in Real life...

PRIMARY CARE

- ▶ 12,7% patients with RD
- ▶ The first to identify the problem in 89%
- ▶ To establish the definitive diagnosis in 54%
- ▶ To provide acute care for the problem in 56% and continuing care in 76%
- ▶ ...care for the patients through their final illnesses in 17%...
- ▶ ...sought consultation from specialists for 85%

▶ W Philips, 2004

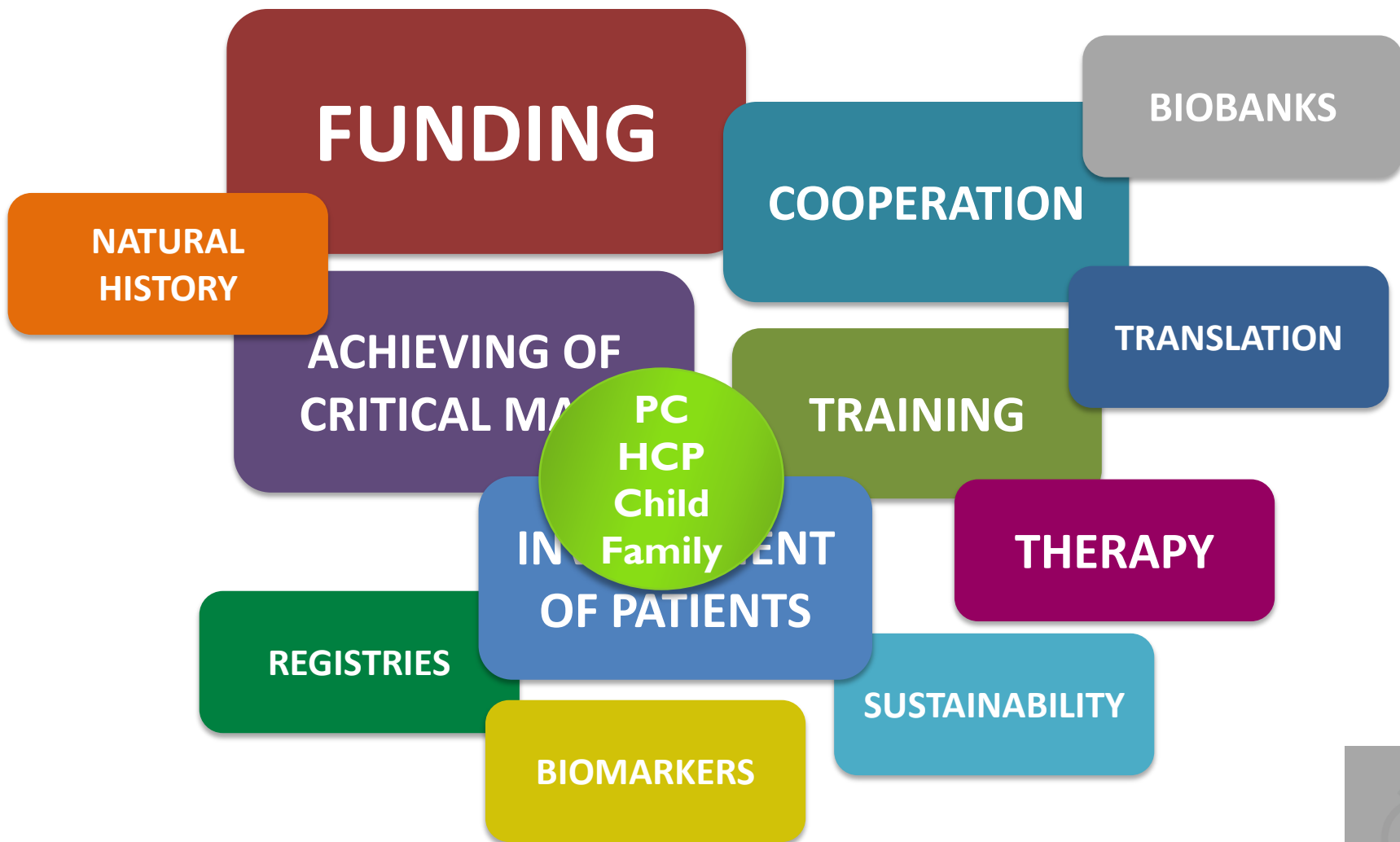
DIAGNOSIS PROCESS IN PRIMARY CARE

- ▶ I suspect something wrong....
- ▶ And consult the literature: 12.3%
- ▶ And consult experts to help me: 23.1%
- ▶ And refer to experts to make the diagnosis: 64.6%

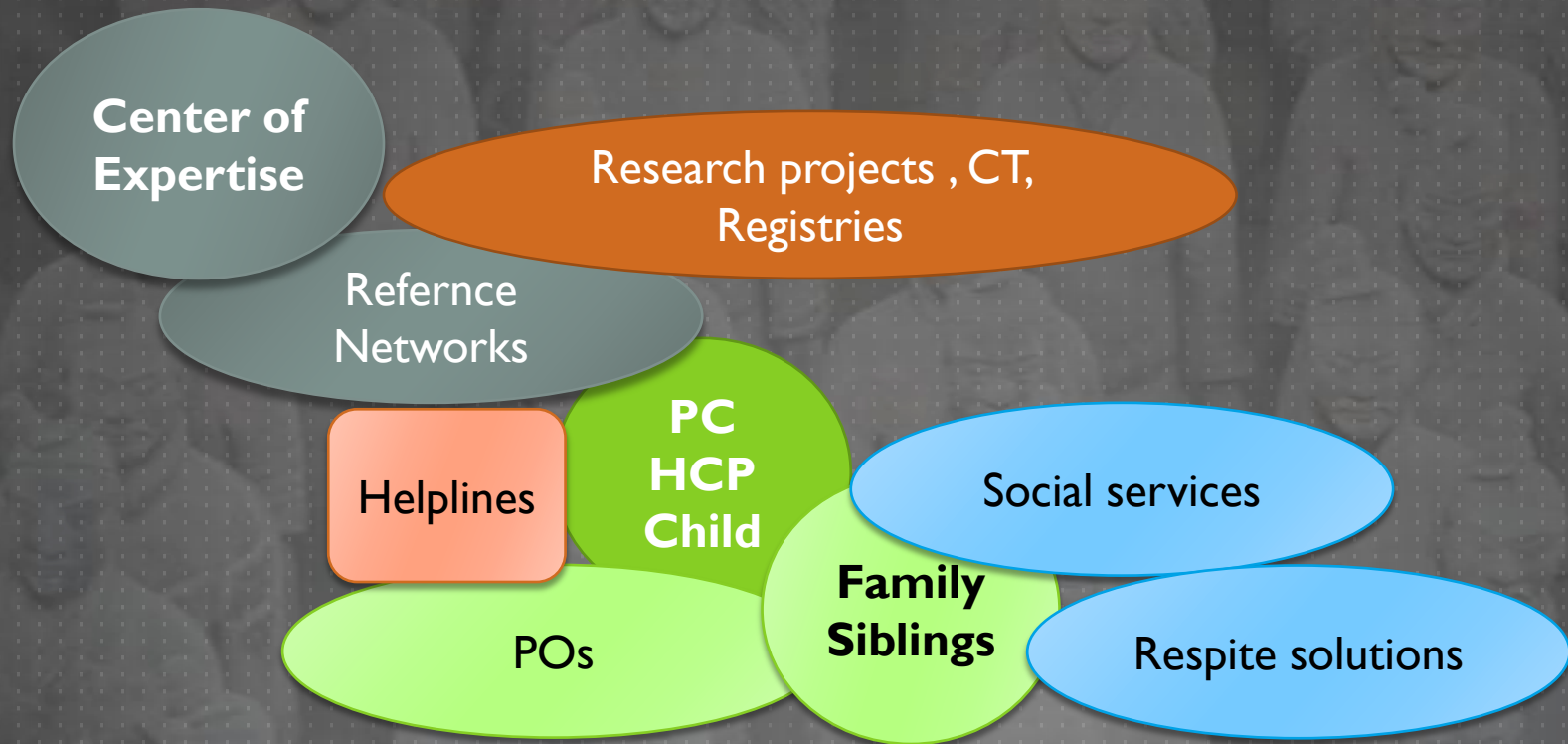
ASPECTS OF DIAGNOSIS

- ▶ Atypical presentation
- ▶ Non specific symptoms
- ▶ Co-morbidity +++
- ▶ Very rare conditions
- ▶ Life threatening
- ▶ Family burden
- ▶ “first time” features
- ▶ “something wrong”

PRIMARY CARE ISSUES



RD PRIMARY CARE ECOSYSTEM



LESSONS LEARNED: NEW CHALLENGES

Patient social media, communities on line

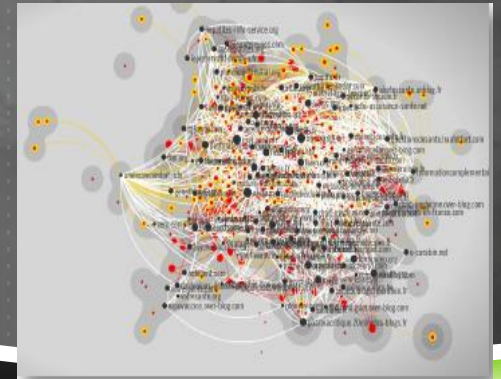
Telemedicine, e patient, patient 2.0

Advanced Therapy, Gene therapy

Consequences of fast track drug development process

Personalised Medicine

Big data



PERSPECTIVES... THE CHILDREN WORLD



Rare Disease Communities helps patients to understand their condition, connect with other patients and provides tools for living with their diseases

Understand.

Daily life with a rare disease.

- Browse patient testimonies,
- Share images & photos,
- Find explanations,
- Contact patient associations

Meet!

Connect with other patients.

- Start conversations,
- Interact with others,
- Ask questions,
- Meet other patients or families

Learn.

Learn more and find resources.

- Contact expert patients,
- Become informed,
- Download,
- Find information adapted to your needs



Visit a community

- Alkaptonuria (AKU)
- Alternating Hemiplegia
- Atypical Hemolytic Uremic Syn.
- Bechet's Syndrome
- CAPS
- CDG
- Cystinosis
- Dravet syndrome
- Ehlers Danlos syndrome
- Epidemiology Bulosa
- Familial Mediterranean Fever
- Ghelt DS
- Hereditary Spastic Paraplegia
- Waldenström macroglobulinemia
- Multiple Myeloma
- Moebius syndrome
- Paraneoplastic Neurological syn.

Custom Rare Disease Search Engine

No online community for your area of interest yet? Use this search engine to search the following websites: eurodis.org, orpha.net, rare Diseases.org and rare Diseases info.nih.gov. More info about these sites. You can also contact us to inform us of your interest in setting up future online patient communities.

Announcements

Child Neurology Society Annual Meeting Dates from 31 October to 3 November 2012 41st Annual Meeting Oct 31 - Nov 2, 2012 Huntington Beach Resort Huntington Beach, California [More info](#)

MULTIMEDIA PATIENT SOCIAL NETWORKS....

What is Social Media ?

- ▶ "...social media collaboration, interaction
- ▶ and sharing – web 2.0 video... some popular tools are blogs, wikis, Twitter and ...?"

Join this community

CAPS community

Search this community

what?
learn to live with the disease

meet!
discuss with other patients

learn.
information and resources

CAPS or Cryopyrin Associated Periodic Syndromes are a group of auto-inflammatory rare diseases (MWS, FCAS & NOMID). Below, you'll find testimonies of patients who live with them.

NOMID Alliance
by Karen Durant 2 months ago

Patient groups

- NOMID Alliance
- AMWS/CINCA
- Canadian CAPS Network
- ASP
- AIFP

See all patient groups

Filters

- All Articles and Stories
- Editor's Articles
- Patient Stories

CAPS Recent Activity

- 11-1 Family Members and the Inflammation Conference topic, published 23 days ago
- Canadian CAPS Network topic, published about 1 month ago
- CAPS patients in the US topic, published 2 months ago
- Getting information on CAPS topic, published 2 months ago
- Canakinumab (Ilaris) muscle warts topic, published 5 months ago
- Muscle warts topic, published 7 months ago
- New Website for Searching for Clinical Trials topic, published 7 months ago
- House MD TV show featured CAPS on the show on Rare Disease Day

Greetings to All,
We are very pleased to be involved in the CAPS community on the www.rarediseasescommunities.org site that is sponsored by EURODIS and NORD. These organizations are de... [Read more](#)

Fifth anniversary AMWS CINCA Patient Group France
by AMWS CINCA 2 months ago

They came from the great West and even the southeast of France to welcome Karen Durant, her husband and their three boys, Nathan, Ethan, Seth and Lucas, arrived from San Francisco (California, East Coast of the United States almost Pacific Coast), for a friendly meeting to mark the fifth...



neoin™
smart health commentary

An exercise health

HOME HEALTH POLICY VIDEO AUDIO INTERVIEWS

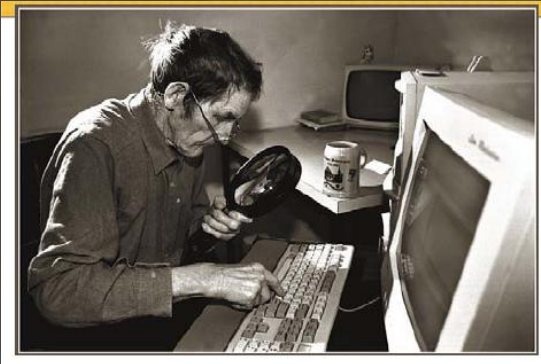
ARTICLE COMMENTS

Evidence-Based Social Media In Medicine
June 24th, 2010 by Bercl in Better Health Network, Health Policy, Research

ScienceRoll
The science of a healthy life and medicine for the people

I've started a series on evidence-based social media in which I share peer-re using social media in medicine or healthcare!

New practices: information search



Orphanet
Eucerd
Irdirc
Horizon 2020
EMA PDCO, COMP
EudraCT
Eurordis

...



**I have a RD X
With a mutation XYZ
Under a OD**

- MA under control
 - PMS
 - RMP
- Registries (Drug and Disease)
 - ATU
 - Off label
- FB : shortage soon
 - Care protocol
 - Biomarker follow
- Reimbursement protocol
 - Informed consent
 - Data privacy
- Transborder directive
- Reference network



RD AN OPPORTUNITY FOR PRIMARY CARE

- ▶ Lessons learned from RD Community activism and activities
 - ▶ How to manage complex situations
 - ▶ How to step in the future
 - ▶ How to move from “disease centered” to “patient centered” care
 - ▶ How to innovate (tools, practices, drug development and drug access, pricing, HTA...)
 - ▶ How to implement Quality of practice (COI)

Disease



Patient

**HCP : Guardianship of
Patient Centered Care
& Ethics of Care**

CONCLUSION: A CALL TO ACTION

- ▶ An empowerment program
 - ▶ Training: RD intensive course
 - ▶ Research in primary care
 - ▶ Becoming active stakeholders in CE, Reference Networks
 - ▶ Implementing information platform
 - ▶ Taking part in registries, surveys
 - ▶ Participating in calls for experts and calls for projects
 - ▶ To advocate
 - ▶ Added value of care
 - ▶ For inclusion of "primary care" in research program (Horizon 2020)
 - ▶ For new organisation of primary care integrating "case manager"