ERN – INFORMATION MEETING FOR PATIENTS

Dublin, April 7, 2017

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A rare disease in Europe is a disease affecting less than 1 in 2,000 citizens

- Over **6,000** distinct rare diseases
- 6% of the population in lifetime
- Many are of **genetic** origin (estimated 70-80%)
- Many **children** are affected by rare diseases. The onset of the disease occurs in childhood for 50% of rare diseases. Increasing over 60s.
- 30 million people living with a rare disease in Europe, more than 300 million world wide
Rare diseases are severe, chronic, progressive, degenerative, disabling and frequently life-threatening diseases, characterised by:

- **Patients and Experts are few, isolated** and geographically scattered, making it difficult to connect with experts and peers
- **Delay to diagnosis** and a significant undiagnosed population
- **Lack of treatment** and challenges to access adequate care
- **Fragmented research, data and information**
- **High social impact and marginalisation within society** at large and within healthcare systems designed for common diseases
- **Heavy psychosocial burden**: hope and society support are essential to patients and families to cope, to be resilient, to be a carer, to be empowered
Clara’s Story, when coming to world

• Inês gave birth to her second child, but something was wrong
• She **searched to find a specialist** privately, due to significant waiting time in local hospital
• Finally, Clara **was diagnosed with Achondroplasia** - a rare bone disease that no one knew about in the local hospital.
• Clara would need close follow-up medical support in a specialist pediatric hospital, which was **120km away**

  • Clara and Inês had to face unthinkable barriers, and were even **declined care** due to being ‘out of area’.
  • The **isolation was unbearable**, Inês was required to fight to access
  • Care came in the form of **multiple medical specialist assessments**, each trip was 120km ... as there was **no expertise locally**
Clara, 9 month old

- With no experts in the local area, Inês became the expert, directing the medical teams
- All the time weakened with worry, sadness and despair, with no psychological support for the family
- Months of countless exams and medical appointments
- Clara had three surgeries, one which was life threatening and possibly avoidable

- Living in a hospital arm chair, taking lots of days off work, leaving behind her 3 year-old son with his grandmother
- No patient groups that could do advise or support them.
- She set up the national Achondroplasia patient group and helped develop the “skeletal dysplasia” clinic in a pediatric hospital

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The Impact of Rare Diseases in the World

No one country, no one continent, can solve alone the problems posed by rare diseases

300 MIO / 7'052 MIO

RARE Diseases

7,000 RARE DISEASES exist, with less than 500 FDA-approved treatments.

ONLY 5% of RARE DISEASES have treatments.

Patients with RARE DISEASES are frequently misdiagnosed or undiagnosed.

80% of RARE DISEASES ARE GENETICALLY BASED.
No one country can solve alone the problems posed by rare diseases!

We need to bring together a critical mass of patients and medical experts, scientists and public health authorities – which does not exist in one single country.

Europe can take pride in what it has done to advance the recognition of rare diseases...

... and rare diseases shows how EU-wide collaboration can deliver added value!

"At times when the European project is under serious scrutiny or even threatened, there are few areas of undisputed added value of EU-wide collaboration such as rare diseases."

Luxembourg Health Minister Mars di Bartolomeo, October 2011

Rare diseases is a case of high added value from European collaboration
European Reference Networks are:

- **Patient-centred**, involving patients from the start and as equal partners in all ERNs
- **Optimal framework** for multisystem rare diseases, enables that every patient with a rare disease to have a home under an ERN, to leave no one behind
- **Anchored into national health systems**, connecting patients, clinicians and researchers
- Vehicle that will pave the way for **faster diagnosis and access to expert care**, by making experts visible and expertise travelling, not the patient
ERNs are a game changer for Rare Diseases: There will be “before” and “after”

ERNs is already a ‘game changer’ in healthcare:

- **European wide collaboration** with nearly 1000 HCP from 26 Member States in 24 ERNs
- **Create a critical mass of patients and data**, push the pace of research and clinical practice
- **Magnetise patients needs to the right expert**, leading to faster diagnosis and treatment
- **Transparency on patient health outcomes from care received** as a driver for clinical excellence
- **Connect and implement existing knowledge and experience** of leading clinicians, researchers and patients
- **Generate new knowledge**
Clara, 4 years old

Inês’ hope is that European Reference Networks offer:

- **better healthcare** opportunities and specialist advice where she lives
- accurate diagnosis as her disease evolve and timely treatment
- better designed clinical trials for rare (bone) diseases, stimulate and activate medical advances, new medicines R&D
- A brighter future

Ensuring that children, like Clara, those in transition to adulthood and adults too avoid multiple repeated medical treatments and unnecessary life threatening surgeries
Our hope is that 30 million lives affected by rare diseases in the EU will be improved through:

- **National healthcare system coverage** of all rare diseases and across all EU countries
- **Open access** to expert advice for timely, accurate diagnosis and treatment
- **Increased evidence and adherence** to clinical guidelines
- **Accelerated pace of research**, integration healthcare and research infrastructures
- **Linked patient data** that are ‘Findable, Accessible, Interoperable and Reusable’
- **Increased access to better therapies**, with faster development of new therapies for ALL RD
Thank You

And thank you to Inês and Clara for sharing their story.