It's Rare Disease Day today!

Importance of rare disease development and patient engagement

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Webinar on Rare World Diseases Day

29 February 2024

WHAT IS RARE DISEASE DAY?

- Taking place on the **last day of February**
  - ✓ This year it will fall on a **leap year** and will take place on **29 February 2024**
- Started in **2008**
- Last year:
  - ✓ **1,300+ events** in **more than 100 countries**
  - ✓ **Video** translated into **49 languages**
  - ✓ **Translated assets** in multiple languages and adapted them to empower you to use in your local context
- This year is the **17th edition**
- Continues to **grow worldwide with Rwanda joining the campaign** for 2024
- Coordinated by **EURORDIS – Rare Diseases Europe, RDI** and **71 National Alliance Partners**

RARE DISEASE DAY

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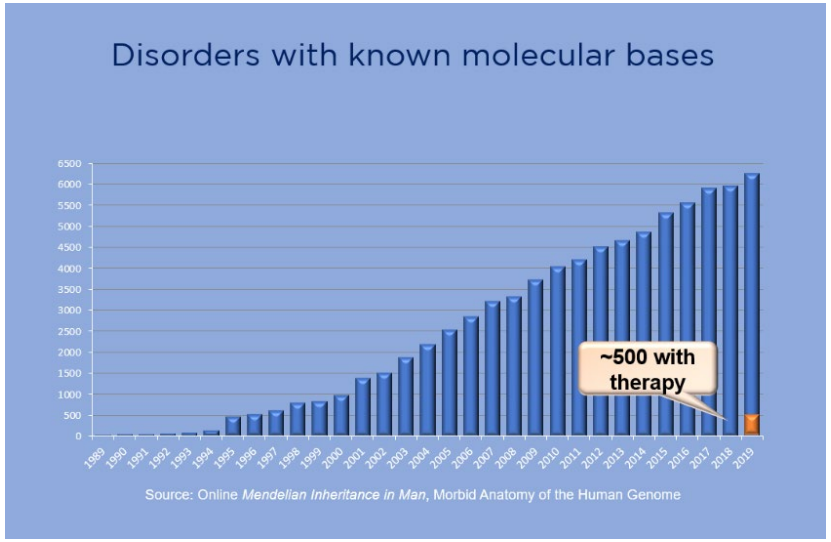
Rare disease development

EURORDIS is committed to deliver on 6 priority areas

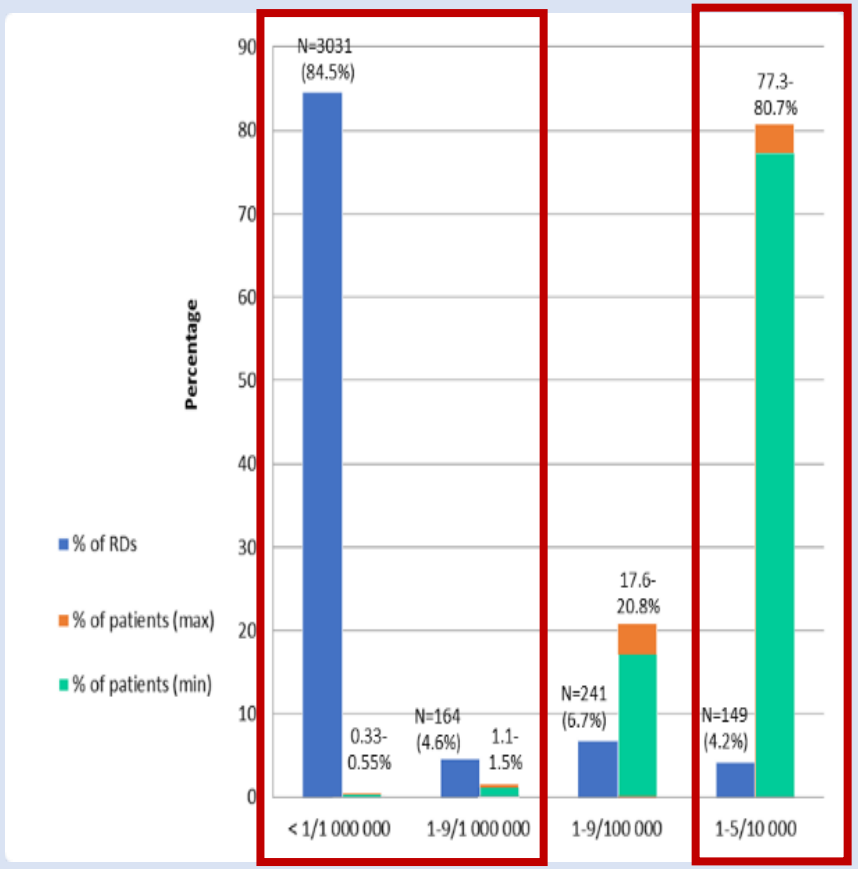
- **By 2030, EURORDIS will have made contributions to the goals of (Based on the Foresight Study Rare 2030):**
  - Earlier, faster and more accurate diagnosis – the goal of diagnosis within 6 months
  - High-quality national and European healthcare pathways, including cross-border healthcare – a goal of improving survival by 3 years on average over 10 years and reducing by one third the mortality of children under 5 years of age
  - Integrated medical and social care with a holistic life-long approach and inclusion in society – a goal of reducing the social, psychological and economic burden by one third
  - Research and knowledge development that is innovative and led by the needs of people living with a rare disease
  - Optimised data and health digital technologies for the benefit of people living with a rare disease and society at large
  - Development and availability, accessibility, and affordability of treatments, particularly transformative or curative therapies – a goal of 1000 new therapies within 10 years

Rare disease development



Most (89.1%) of rare diseases are very rare (prevalence less than 1 per 100,000)

45% of all MAs, 40% of all ODs (up until 2020)



Almost all of the people with rare disease (>98%) have one of the 390 most prevalent diseases (more common than 1 per 10,000)

55% of all MAs, 40% of all ODs (up until 2020)

SOURCE: Wakap et al. (2019) ; EMA (2021)

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A tool for developers: the ODDG

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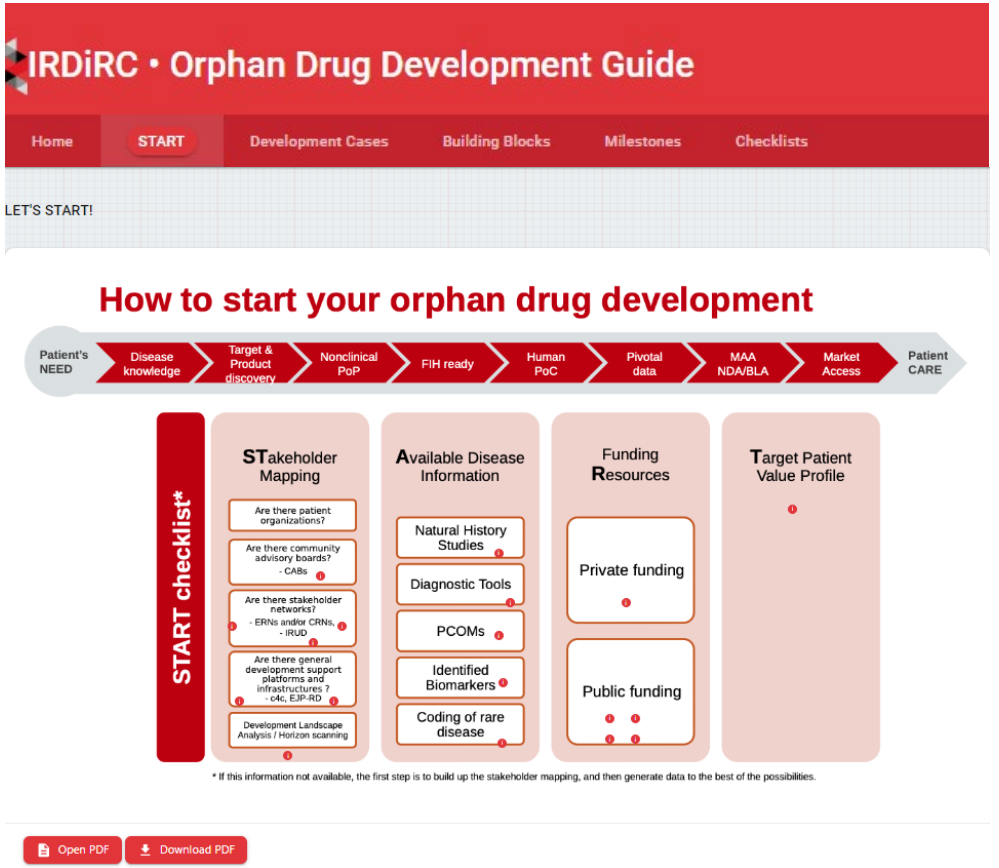
COMMENT | 20 April 2020

Boosting delivery of rare disease therapies: the IRDiRC Orphan Drug Development Guidebook

The International Rare Diseases Research Consortium (IRDiRC) has created a Guidebook to facilitate drug development for rare diseases by organizing available tools into a standardized framework.

By Anneliene Hechtelt Jonker, Virginie Hivert, Michela Gabaldo, Liliana Batista, Daniel O'Connor, Annemieke Aartsma-Rus, Simon Day, Ken Sakushima & Diego Ardigo



https://orphandrugguide.org/

(Tutorial) https://www.youtube.com/watch?v=QMJW85VP3Y8

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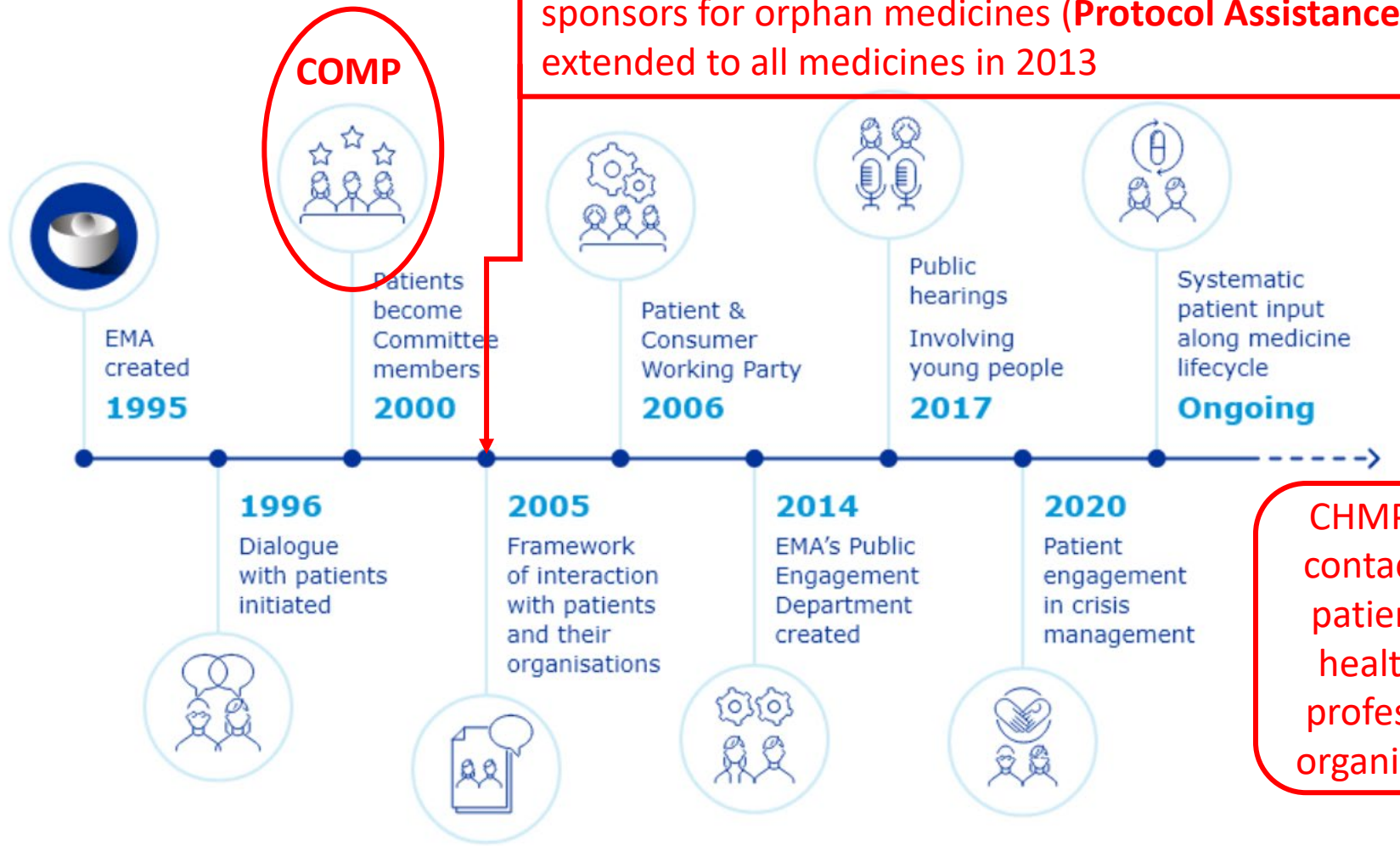
Patient engagement

Key milestones of EMA interaction with patients and consumers

Patients included in Scientific Advice discussions with sponsors for orphan medicines (**Protocol Assistance**) – extended to all medicines in 2013



CHMP early contact with patient and healthcare professional organisations

Patient engagement

- EURORDIS works with the **3 patient representatives** on the Committee for Orphan Medicinal Products
- Collaboration to identify PLWRD for **Protocol Assistance dossiers** requiring patient input + **Scientific Advice Groups (SAGs)** as well as other activities
- Contribute to **CHMP early contact** with patient and healthcare professional organisations by getting in touch with **Patient Organisations** and gathering their input

Questions from our members/patients contacting EURORDIS

- How to contact the EMA? (Protocol Assistance, CHMP early contact, other)
- How to deal with Competing interests? (EMA Policies, PARADIGM recommendations)
- What to do when a product got a MA from the FDA? How to contribute to discussions at the EMA?
- How to share information with the regulators when there are several products in the pipeline? On the market? (e.g. Assessment of significant benefit)
- How to deal with situations where all the clinical programs fail/stop?
- How to contribute on updating guidelines for a specific rare disease?

What we need from the developers

- Invest in ultra-rare diseases and rare diseases with no therapeutic options
- Keep investing in rare diseases with therapeutic options: remaining unmet medical needs, treatments approved for third line which would benefit patients in 1st line (rare cancers)
- Invest in Europe and anticipate on the access requirements (early interactions with HTA bodies and payers)