



EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

Rare Diseases Day 2024

Ask the regulator

Webinar

Presented by Kristina Larsson on 29 Feb 2024
Head of Orphan Medicines Office, Human Evidence Generation, EMA

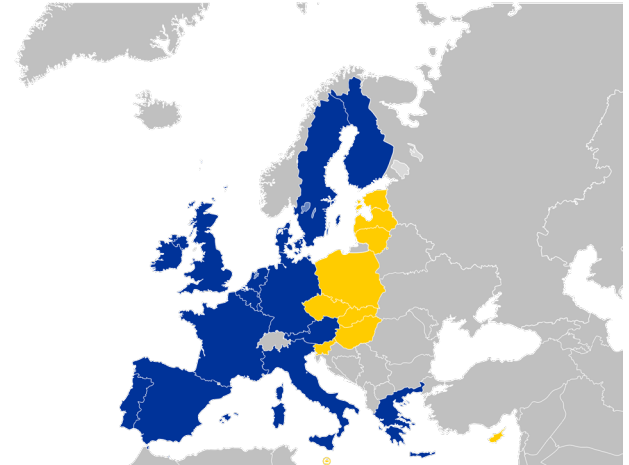
An agency of the European Union



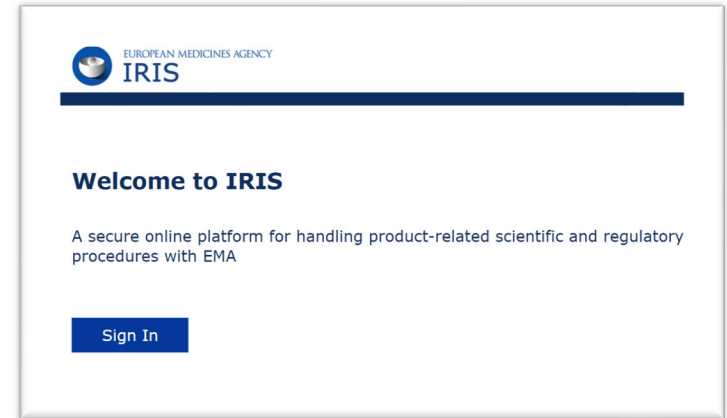
2000 vs now...



designed by freepik



Orphan designation submissions



2000

2024

Designation criteria, regulation (EC) No 141/2000

RARITY (prevalence) / RETURN OF INVESTMENT, Art 3.1 (a)

- Medical condition affecting not more than 5 in 10,000 in the Community (around 250,000 people)
- Without incentives it is unlikely that the marketing of the product would generate sufficient return to justify the necessary investment

SERIOUSNESS

- Life –threatening or chronically debilitating

ALTERNATIVE METHODS AUTHORISED, Art 3.1(b)

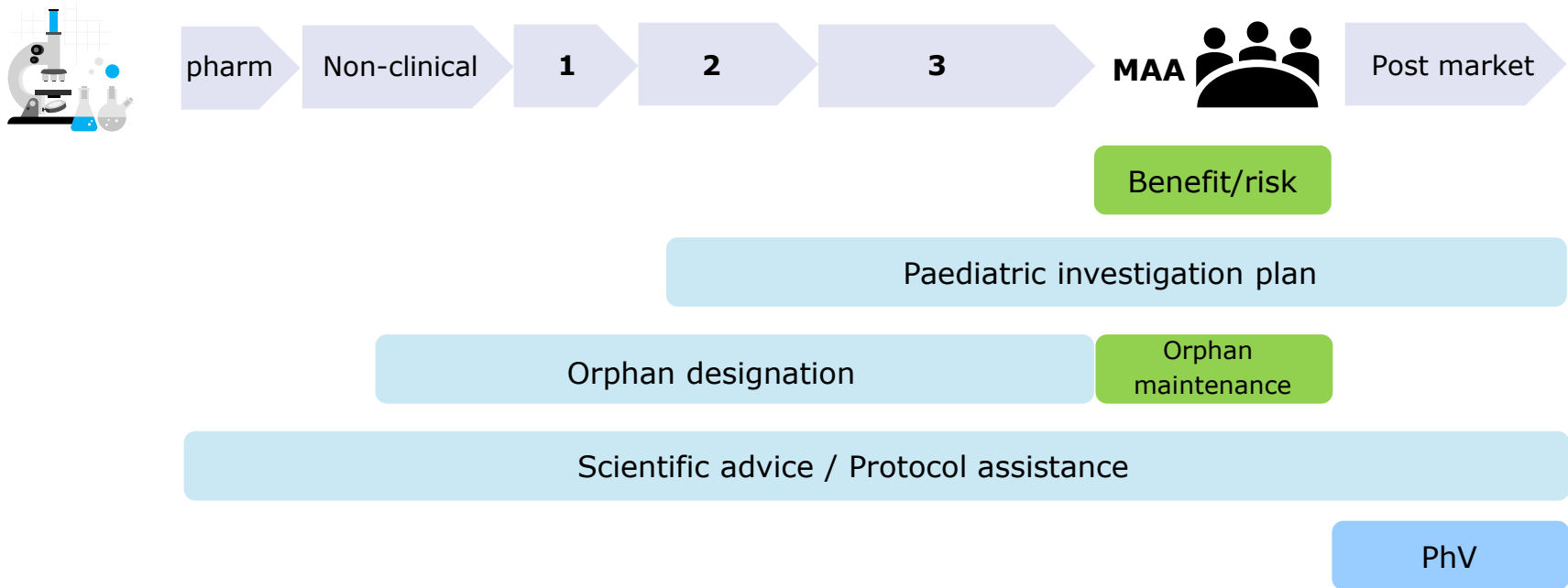
- If satisfactory method exist the sponsor should establish that the product will be of significant benefit

Incentives for orphan medicines

- ❑ Fee reduction / exemptions
 - Extended incentives for Small and Medium sized Enterprises (SMEs)
- ❑ 10-year market exclusivity (+ 2 if paediatric)
 - Protection against similar products structure mech of action same indication
 - Three derogations: Sponsor's consent, lack of supply, clinical superiority
- ❑ Product development
 - Protocol assistance, reduced fee
- ❑ Community marketing authorisation (all EU and EEA member states)



European regulatory input along drug life cycle



Communication

- EMA orphan designation website
 - Q&A (general public)
 - Guidance documents
- COMP minutes
- Scientific publications
- Orphan Maintenance Assessment Report (OMAR) - published with EPAR

Rare diseases, orphan medicines
Getting the facts straight



Drug Discovery Today - Volume 00, Number 00 - June 2017

Animal models for metabolic, neuromuscular and ophthalmological rare diseases

Guillaume Vaquer¹*, Frida Rivière², Maria Mavris³*, Fabrizia Bignami^{4,5}, Jordi Linares-García⁶, Kerstin Westermark^{6,8} and Bruno Sepodes^{1*}

Abstract | Animal models are important tools in the discovery and development of treatments for rare diseases, particularly given the small populations of patients in which to evaluate therapeutic candidates. Here, we provide a compilation of mammalian animal

Stelios Tsigkos¹, Matthias Philipp Hofer¹, Maria Elzbieta Sheehan^{1,2}, Segundo Mariz¹, Kristina Larsson¹, Frauke Naumann-Winter^{3,4}, Laura Fregonese¹ and Bruno Sepodes^{3,5}

¹Orphan Medicines Office, European Medicines Agency, 30 Churchill Place, Canary Wharf, London E16 4EU, UK; ²Max Delbrück Center for Molecular Medicine in the Helmholtz Association, Robert Roske Straße 1, 13125 Berlin, Germany; ³Committee of Orphan Medicinal Products, European Medicines Agency, 30 Churchill Place, E16 4EU, UK; ⁴Handelsinstitut für Arzneimittel und Medizinprodukte Kurt-Georg-Kesinger-Allee 3, Bonn 53117, Germany; ⁵University of Lisbon, Faculty of Pharmacy, Avenida Prof. Gama Pinto, Lisboa 1649-003, Portugal

In the European Union (EU) legislative framework for orphan medicinal products (OMP), establishing that a condition affects not more than five in 10,000 people in the EU is based on rarity. Demonstrating this requirement to the Commission (COMP) can be a particularly challenging task for sponsors. This task is often complicated by common issues with the estimation of prevalence in orphan diseases, such as the lack of explicit contemporary conclusion as critical factors for access to orphan status. These concerns are discussed in detail based on recent examples of published European Medicines Agency (EMA) documents.

RESEARCH **Open Access**

Use of biomarkers in the context of orphan medicines designation in the European Union

Stelios Tsigkos¹, Jordi Linares¹, Segundo Mariz¹, Stiina Aarum¹, Laura Fregonese¹, Bozena Dembowska-Baginska², Rembert Elbers⁴, Pauline Evers², Tatiana Foltanova³, Andre Lhoir², Ana Corrêa-Nunes², Daniel O'Connor²

Drug Discovery Today

Available online 9 October 2017
In Press, Corrected Proof

Review
Keynote

Demonstrating significant benefit of orphan medicines: analysis of 15 years of experience in Europe

Laura Fregonese¹, Lesley Greene², Matthias Hofer¹, Armando Magrelli³, Frauke Naumann-Winter⁴, Kristina Larsson¹, Maria Sheehan¹, Violeta Stoyanova-Beninska⁵, Stelios Tsigkos¹, Kerstin Westermark⁶, Bruno Sepodes⁷

Orphan Medicines Office (OMO) of the European Medicines Agency (EMA) is responsible for the assessment of orphan medicinal products (OMP) of the European Union (EU) and for the assessment to establish that a condition affects not more than 5 in 10,000 people in the EU. This assessment is particularly challenging when therapies already exist, the

Support to development

[PRIME](#)

[Innovation Task Force](#)

[Orphan designation](#)

[Paediatric development](#)

[Scientific advice and Protocol assistance](#)

[Qualification advice and opinion](#)

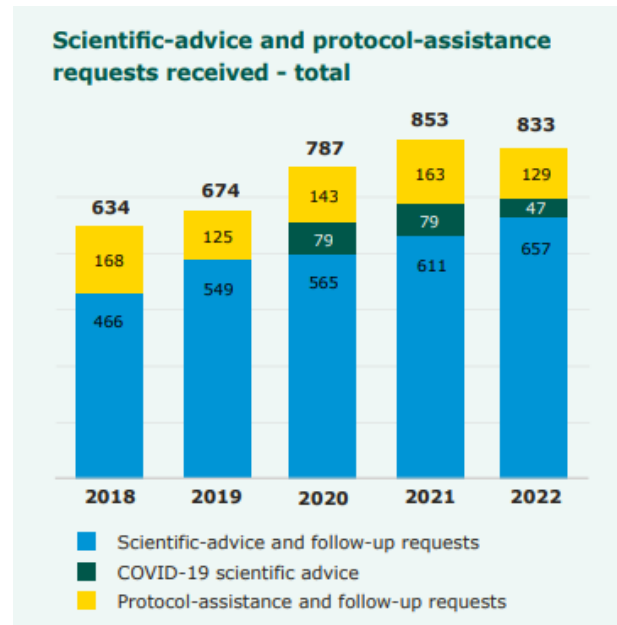


Protocol assistance

Scientific Advice Working Party.

Advising companies on specific questions they have during development of medicines to meet regulatory and scientific requirements:

- how to manufacture them;
- how to test them first in the tube and in experimental animals; and
- most importantly: how to test them in humans in clinical trials.
- COMP responds to the question on Significant Benefit



Contact points

[Patients and carers](#)

[Academia](#)

[SME office](#)



Some things do not change

- Our devotion and support to developments of rare diseases
- Our work to improve the experience for stakeholders working with EMA

Remember:

- The importance of early interaction with EMA and the benefits of early orphan designation
- Ask for protocol assistance (or scientific advice)
- Use the suitable contact points, we are here to help you!