European Reference Network
for rare or low prevalence complex diseases

Network Neuromuscular Diseases (ERN EURO-NMD)

Present and future of gene therapy in Neuromuscular Diseases

Satellite Scientific Symposium endorsed by ERN EURO-NMD

February, 22nd 2024

Accessibility To New Therapies: The Patients' View

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Member of the Board of Directors
EURORDIS Rare Diseases Europe

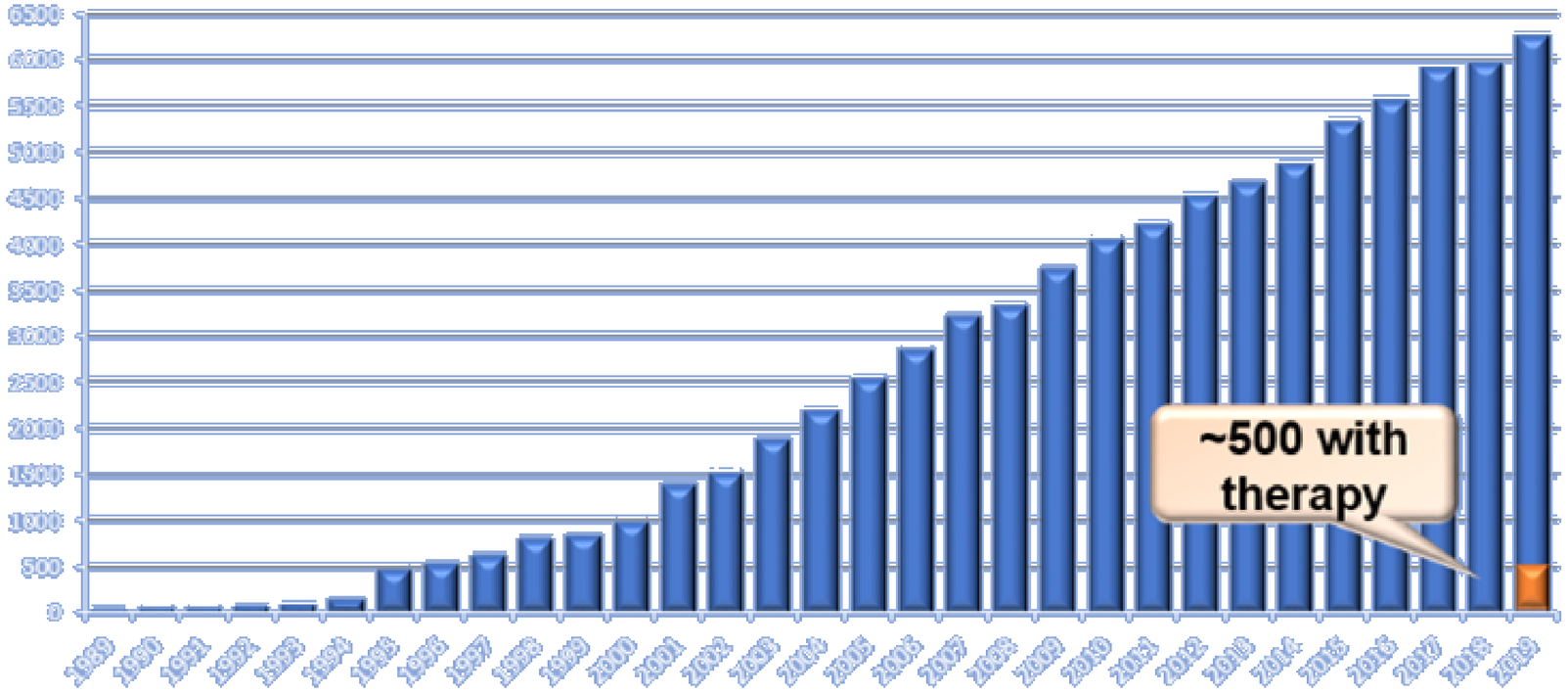
EURORDIS goals

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

Available → Affordable → Accessible

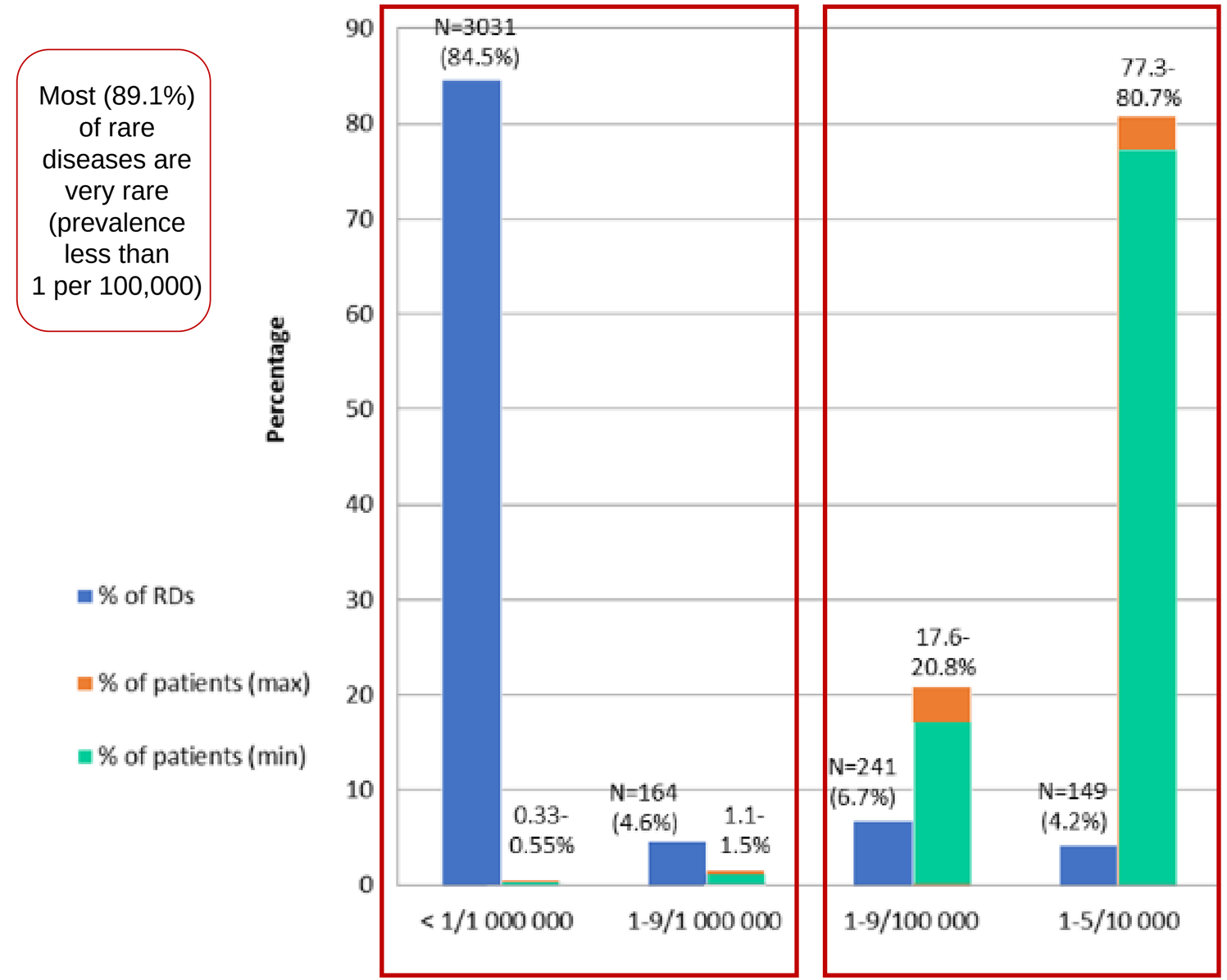
Disorders with known molecular bases



Source: Online *Mendelian Inheritance in Man*, Morbid Anatomy of the Human Genome

More figures

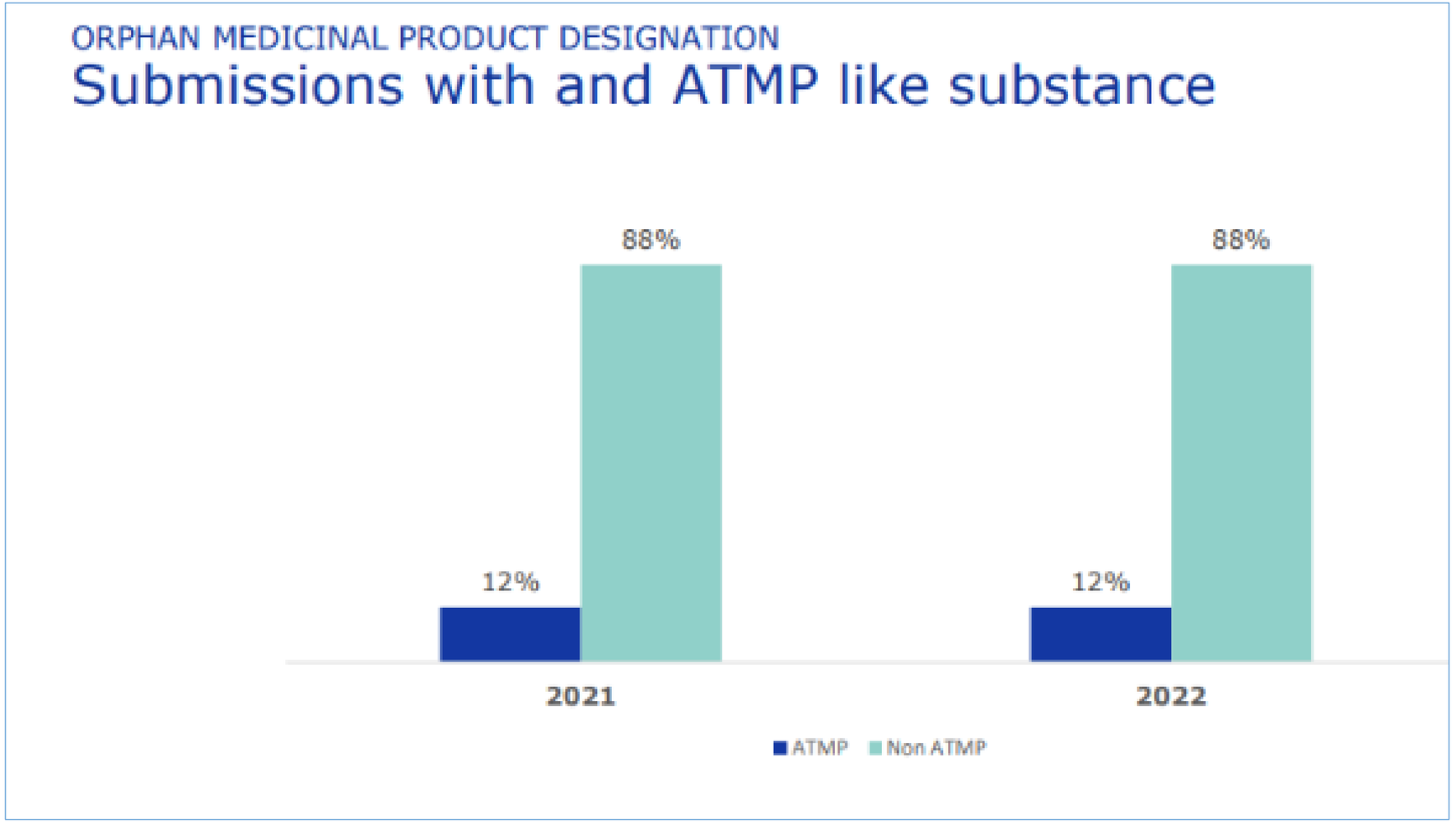
Most (89.1%) of rare diseases are very rare (prevalence less than 1 per 100,000)



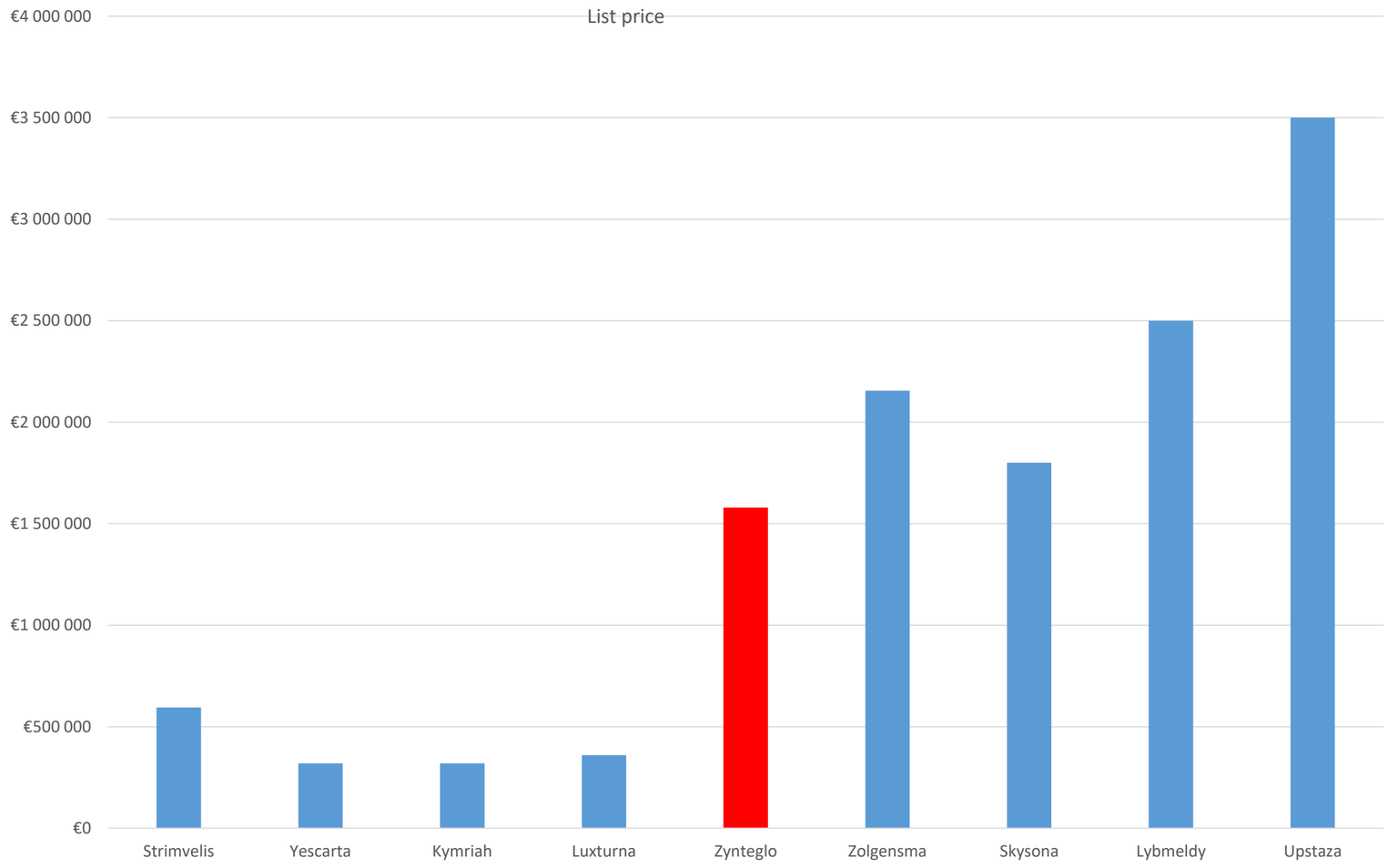
Almost all of the people with rare disease (>98%) have one of the 390 most prevalent diseases (more common than 1 per 10,000)

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

More figures



Available → Affordable → Accessible



Is the only way up?

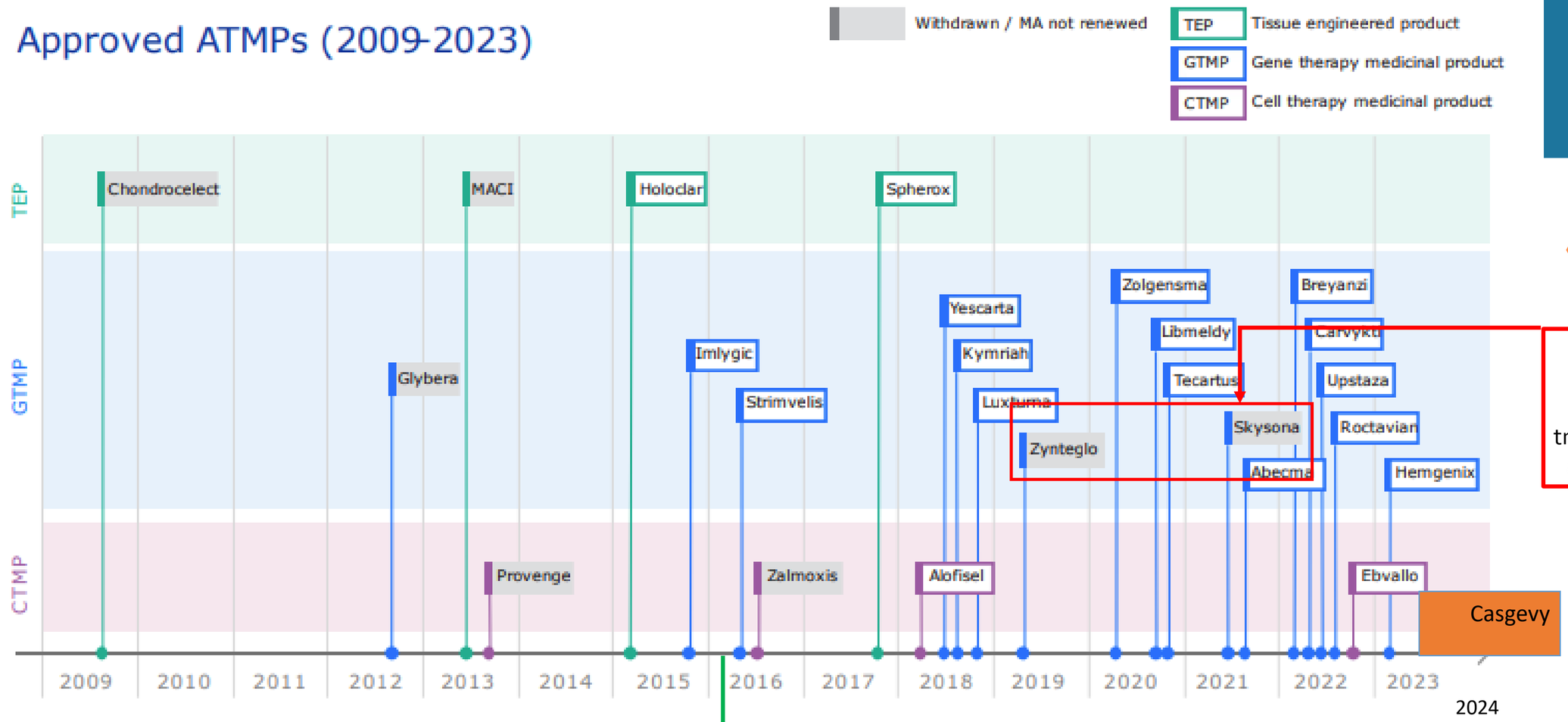
Sources: List prices as indicated in the Italian Gazzetta Ufficiale

Available

Affordable

Accessible

Approved ATMPs (2009-2023)



2022: Systemic downstream failure leads to fewer transformative options available to patients

2016: PRIME scheme introduced

(Source: EMA CAT Quarterly update October 2023)

Foresight study - Access

- A robust regulatory science agenda (building on the existing EMA agenda) should be developed and financially supported at European level, with particular attention to the specificities of rare diseases, emerging technologies and advanced therapies
- Investments into public private partnerships operating in the pre-competitive space should be increased, with greater coordination and collaboration between funding sources and across sectors, and with particular attention to tech-intensive and other advanced approaches
- an EU-Fund to co-finance the generation of evidence across EU Member States and reduce uncertainties during the first years following approval, for advanced therapies for the rarest diseases (affecting less than 1/100 000.)
- In the case of advanced therapies for the rarest diseases (those affecting fewer than 1 /100,000), an EU-Fund should be established to co-finance the generation of post marketing authorisation evidence across EU Member States during the years initially following approval, in order to reduce uncertainties
- A workable system should be developed at European level to economically regulate the relationship between public buyers and companies, via a European Table on pricing and negotiations: this is particularly urgent in order to ensure access to advanced therapies such as gene therapies

Foresight study - ATMPs

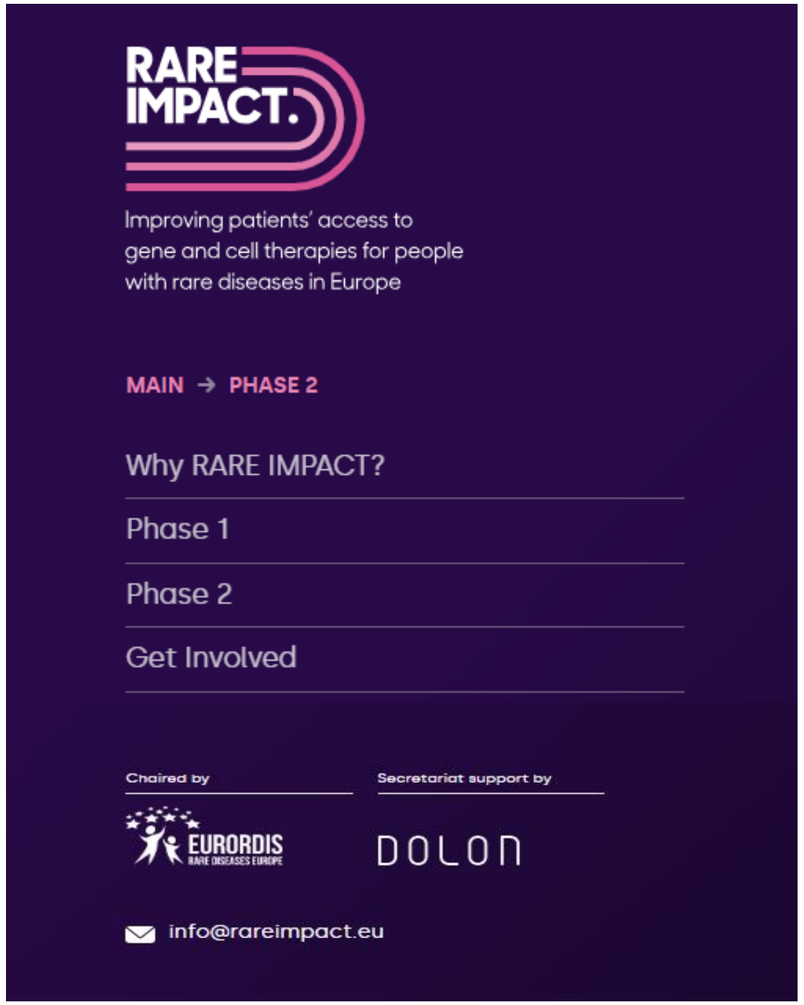
ADVANCED THERAPY MEDICINAL PRODUCTS

The advanced therapy medicinal products hold promise for the treatment of a variety of rare diseases: specific EU-level actions must be taken to support the availability and accessibility of gene, cell and tissues therapies, which will become more numerous in future:

+ Cross-country collaborations should be put in place, to streamline access to advanced therapies for rare diseases, avoiding the requirement for patients to fund significant costs upfront (or else private funding collections/ crowdfunding should be encouraged for the therapies entering the markets)
+ As above, proposals for a shared European fund for advanced therapies' reimbursement should be developed, to support the practicalities of patients receiving care in a different country (ensuring hospitals receive funds from agencies in different Member States in a reliable and timely fashion, for instance)
+ In the case of advanced therapies for the rarest diseases (those affecting fewer than 1 /100,000), an EU-Fund should be established to co-finance the generation of post marketing authorisation evidence across EU Member States during the years initially following approval, in order to reduce uncertainties
+ The precise role European Reference Networks (ERNs) can play in facilitating access to advanced therapies should be explored - and where relevant, enacted - ranging from supporting more experts and informed decision-making concerning which patients would benefit from which therapies, to actually providing advanced therapies in a limited number of centres across Europe, and collecting monitoring data

> "A cure would be fantastic, but failing that, reasonably priced (or government funded) medication is needed"
>
> **Rare Disease Patient**

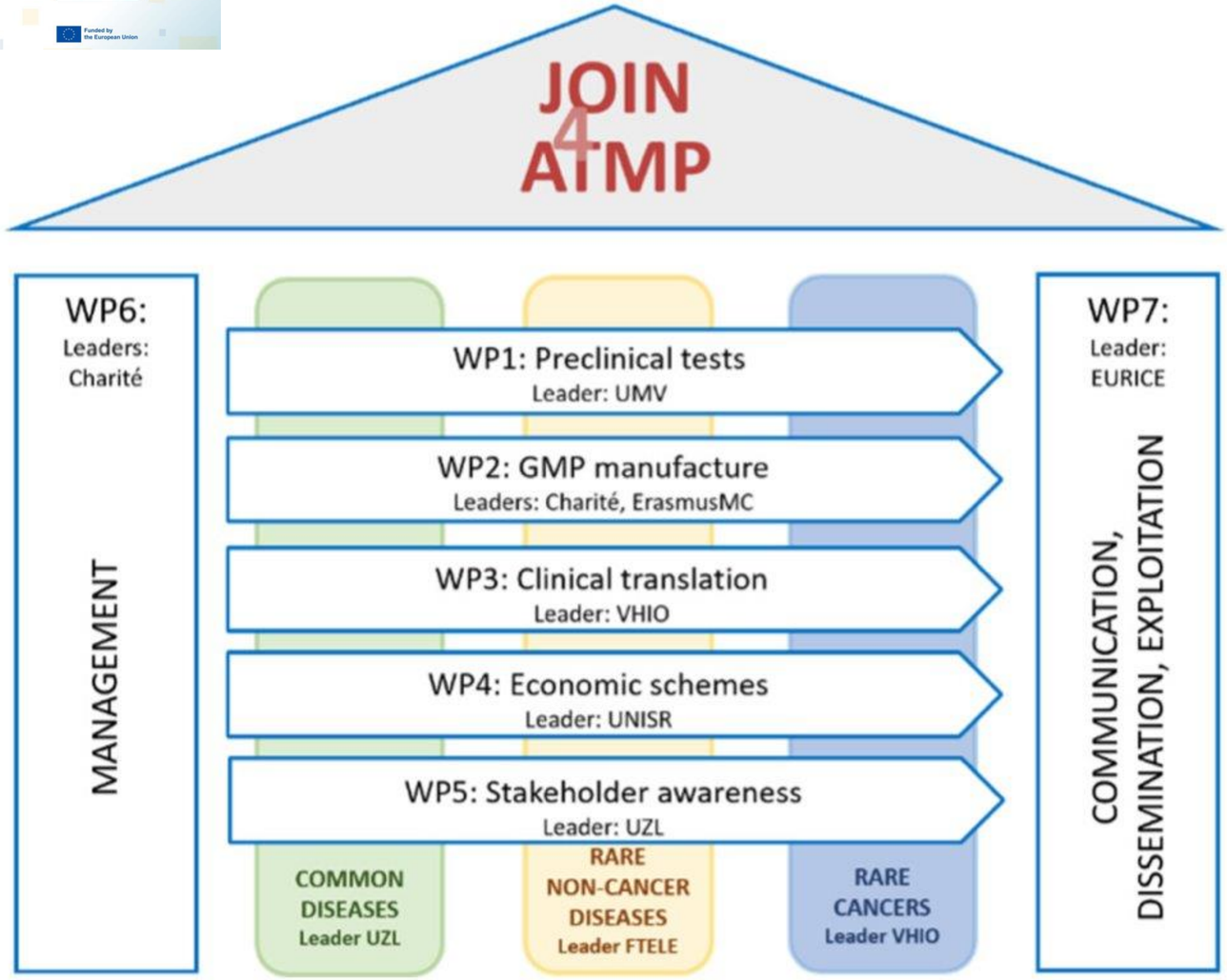
Projects and initiatives



JOIN 4 ATMP

Map, join and drive European activities for advanced therapy medicinal product development and implementation for the benefit of patients and society

Join 4 ATMPs



WP1: How to increase the **predictability of preclinical safety and efficacy data**.

WP2: Road to standardized **Good Manufacturing Practice** ATMP production across the EU.

WP3: **Streamlining clinical ATMP development**: From early phases to marketing authorization.

WP4: New frameworks for **value assessment, pricing and reimbursement schemes.**

WP5: **Increasing ATMP regulatory awareness** in producers and ATMP acceptance in patients and healthcare professionals.

WP6: **Project management and scientific coordination**.

WP7: Innovation Management: **Communication, Dissemination, Exploitation.**

EMA Pilot

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Progress update on pilot for academic and non-profit developers of advanced therapy medicines

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8 February 2024

Following the launch in September 2022, EMA has accepted three academic and non-profit organisations developing advanced therapy medicinal products (ATMPs) into a pilot scheme, in which they benefit from enhanced support from the Agency.

News | Human | Advanced therapies | Data on medicines

Key considerations

- Hospital exemption – role of Hospital pharmacies in the production/manufacturing of ATMPs – accessibility/CBHD ? GMP standards ? reproductibility ?
- Harmonisation on HE rules, and in particular on safety and data gathering and (long-term) reporting
- Even in countries where an industrial ATMP is available, with an approved price and reimbursement, access also depends on the hospital pharmacy budget and other criteria
- HE-ATMPs versus industrial product: choice of the national authorities? (Lower price? Rewarding the innovation? Payment models?)
- N=1 approaches versus larger indication
- Platform approaches and regulatory acceptance
- Dependence vis-à-vis of the US or other countries – shortages?
- Autonomy capacity is linked to training of high-quality technicians and engineers

Thank you!