ERDERA
European Rare Diseases
Research Alliance

INNOVATIVE THERAPIES AND THE TRANSLATIONAL RESEARCH JOURNEY WITH EPTRI

SUPPORTING BETTER MEDICINES FOR CHILDREN

Donato Bonifazi

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the European Union

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This presentation will require active participation from all the training candidates.
Please remain attentive throughout.

INTRODUCTION TO EPTRI

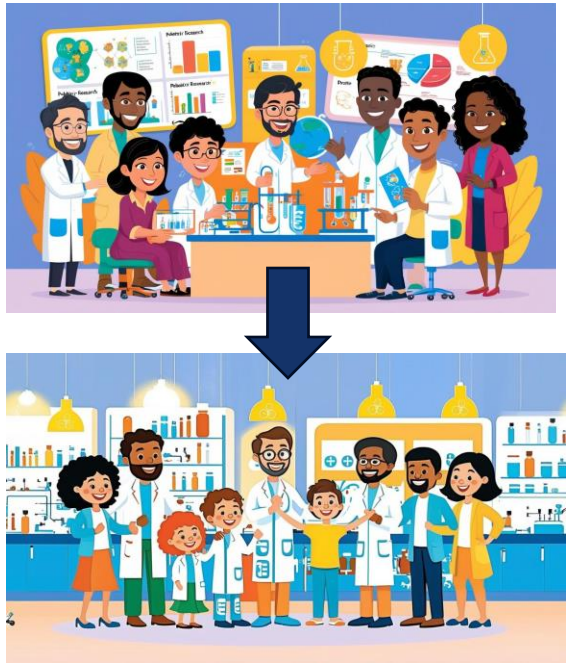
Understanding who we are and why children's research matters

EPTRI is a big team of doctors, scientists, families, and even kids!

We work together to make special medicines just for children. Most medicines are made for grown-ups, we want to change that!

ePTRI
European Paediatric Translational Research Infrastructure

EPTRI



The European Paediatric Translational Research Infrastructure (EPTRI) is like a big open space for science. It allows researchers from different places and backgrounds to share ideas and work as a team.

It started with support from the European Union.

The goal is to help turn scientific discoveries into real medicines that can be used to treat children.

EPTRI also listens to **many different voices.** It brings together not just scientists, but also children and families, doctors, health companies, charities, and government workers. Everyone works together to make sure the medicines being developed are safe, useful, and made with **kids' needs in mind**.

EPTRI

Children and young patients are still left behind an acceptable levels of available treatments. Most of the drugs used today have not been specifically developed and tested for them. Children and young people have different anatomy and metabolism, and their drugs reactions change radically with growth and development.

EPTRI's mission is to **contribute to solving this gap by supporting paediatric research** and creating a new EU political framework focused on children and their health.

It is time to change!

Donato Bonifazi, EPTRI Coordinator

#childrenfirst

THE NEED FOR PAEDIATRIC RESEARCH

Why today's medicines are not enough for children

Children deserve treatments specifically tailored to their needs.

Today, we still have a long way to go — and EPTRI is helping lead the way.

Children and teenagers are a big part of the people living in Europe

Even though they have the right to good healthcare and safe medicines, **only about 35 out of 100 medicines in Europe are made for kids** of different ages or with different illnesses.

The **EPTRI Manifesto** says we need more research to help children get the right medicines.

It asks leaders in Europe to make children's health a big part of their plans and to support the creation of medicines for kids, especially those with rare diseases.

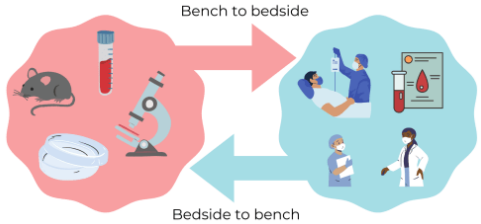
MANIFESTO

ON PAEDIATRIC RESEARCH

THE TRANSLATIONAL RESEARCH JOURNEY

Turning discoveries into real treatments faster

Turning new discoveries into real treatments is a journey — from the lab to the clinic — to help children faster and better.



TRANSLATIONAL RESEARCH JOURNEY

Translational research is like being a science helper who takes cool discoveries from the lab and finds ways to use them to **help people feel better.**

Imagine scientists are like explorers, they discover how our bodies work and what makes us sick. But those discoveries do not help people right away.

Translational research is the **bridge that takes those discoveries and turns them into new medicines, better treatments, medical devices, or approaches** that can directly help children and young people to stay healthy.

It's like turning science into superpowers for doctors and nurses!

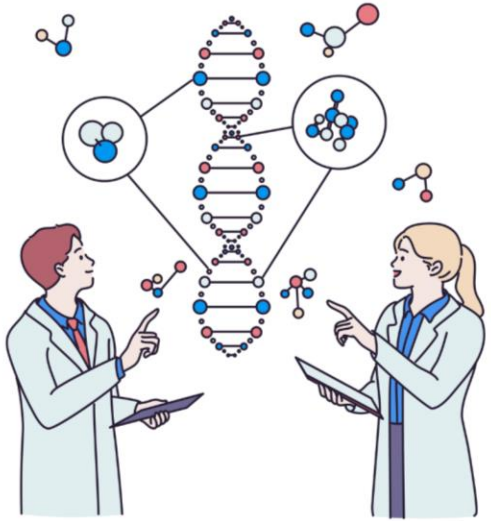
Translational research is like a bridge.

It connects lab discoveries into real-life solutions.

TRANSLATIONAL RESEARCH JOURNEY

Translational research helps **reduce the time** between discovering something new in science and using it to treat patients.

That means people can get **better treatments faster and live healthier lives**.



TRANSLATIONAL RESEARCH JOURNEY

1. Discovery – "The Big Idea"

Scientists start by trying to understand how the body works and what causes a disease. They look for clues, like how a virus spreads or what makes a cell sick. This is where they get the idea for a new medicine.

Like a detective solving a mystery!

2. Testing in the Lab – "Is It Safe?"

Next, scientists test the idea in the lab, using cells or tiny animals to see if the medicine might work and if it's safe.

Like trying out a new recipe on a small scale before making it for everyone.

TRANSLATIONAL RESEARCH JOURNEY

3. Clinical Trials – "Try It with People"

If it looks safe, doctors start testing it with real people in steps called clinical trials:

- Small group: Is it safe for humans?
- Medium group: Does it seem to work?
- Large group: Does it work better than other treatments?

Like checking if a new toy is fun, safe, and works for lots of kids before selling it in stores!

TRANSLATIONAL RESEARCH JOURNEY

4. Approval and Use – "Medicine for Everyone"

If it passes all the tests, the medicine gets approved and can be used in hospitals and clinics to help people feel better.

Like the new toy getting on store shelves!

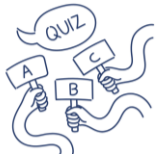
5. Keep Learning – "Always Improving"

Even after a medicine is approved, scientists keep studying it to make sure it stays safe and works well over time.

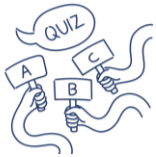
Like updating a game or fixing bugs after it's released.

A REAL-LIFE EXAMPLE

Let's say doctors notice that some people with a certain type of cancer respond very well to a specific diet.	➡️	1. **Clinical Observation** 🧍➡️🍽️➡️📈 Doctors see some kids get better with a special diet.
Scientists get curious and start researching why that happens. Through lab studies, they discover that the **cancer cells** rely on a particular type of **sugar** to grow.	➡️	2. **Basic Research** 🔬🧪 Scientists discover cancer cells love a certain sugar.
Thanks to translational research, they develop **a new drug** that blocks the cancer's ability to use that sugar.	➡️	3. **Drug Development** 💊🧿 They create a medicine that stops cancer from using sugar.
They test it in the **lab**, then in **animals**, and finally in clinical trials with **patients**.	➡️	4. **Preclinical Testing** 🐁 They test it on cells and animals to make sure it's safe.
If it works and is safe, it becomes a **real treatment** that helps people with that kind of cancer live longer and better.	➡️	5. **Clinical Trials and Approval** 👥💾✅ They tested it in people through phased clinical trials, and if proven safe and effective, it becomes an approved treatment.



QUICK QUIZ TIME!



1. What is the first step in the translational research process in this example?

a) Start clinical trials immediately

b) Research the relationship between diet and cancer response ✅

c) Publish findings without further testing

2. After identifying that sugar is important for cancer cell growth, what do the researchers do next?

a) Develop a drug to block the cancer cells' ability to use sugar ✅

b) Conduct more animal studies without developing a drug

c) Stop the research because it's too complicated

3. What is the next step after developing the drug?

a) Test the drug in the lab, then on animals, and finally in clinical trials with patients ✅

b) Immediately start clinical trials without lab testing

c) Abandon the drug after lab testing

4. If the drug works and is safe, what happens next?

a) The drug becomes a real treatment to help cancer patients ✅

b) The drug is discarded, and no further research is done

c) The drug is tested on a different disease

EPTRI IN TRANSLATIONAL RESEARCH

Bridging lab discoveries and real treatments for children

New ideas like gene and cell therapy are changing children's healthcare.
EPTRI is making these innovations possible.

EPTRI INNOVATION IN PAEDIATRIC CARE

PAEDIATRIC MEDICINES DISCOVERY

Helping discover new drugs made just for kids

PAEDIATRIC BIOMARKERS AND BIOSAMPLES

Finding clues in kids' cells to treat diseases better

DEVELOPMENTAL PHARMACOLOGY

Studying how children's bodies grow and react to medicine

PAEDIATRIC MEDICINES FORMULATIONS

Making child-friendly pills and syrups that are easy to take

PAEDIATRIC MEDICAL DEVICES

Designing special tools made just for kids' bodies

EPTRI INNOVATION IN PAEDIATRIC CARE

ADVANCED THERAPY MEDICINAL PRODUCTS

These special medicines help fix genes and are made to be safe, even for a long time. Scientists work hard to make sure they're not too tricky or too expensive to use for kids.

DIGITAL HEALTH TECHNOLOGIES

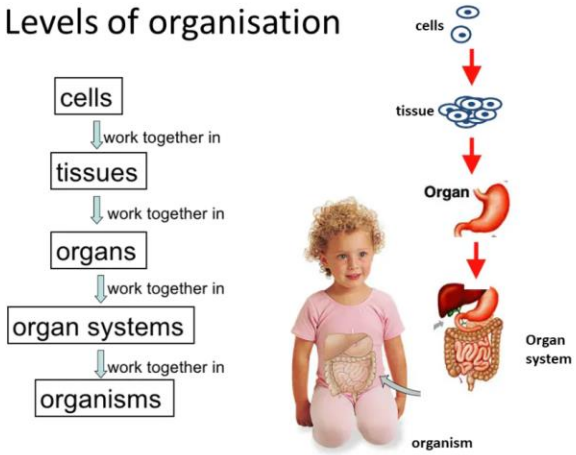
Digital health tools help track kids' health, share info, and make data easier to access, leading to better care and healthier kids.

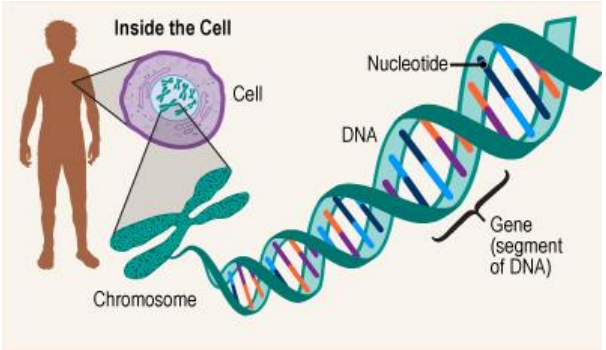
ADVANCED THERAPY MEDICINAL PRODUCTS

MEET THE HERO MEDICINES!

ATMPs are a category of **innovative therapies** for human use based on genes, cells or tissues.

They are used especially for **treating rare or genetic diseases**.





Advanced Therapy Medicinal Products (ATMPs)

ADVANCED THERAPY MEDICINAL PRODUCTS

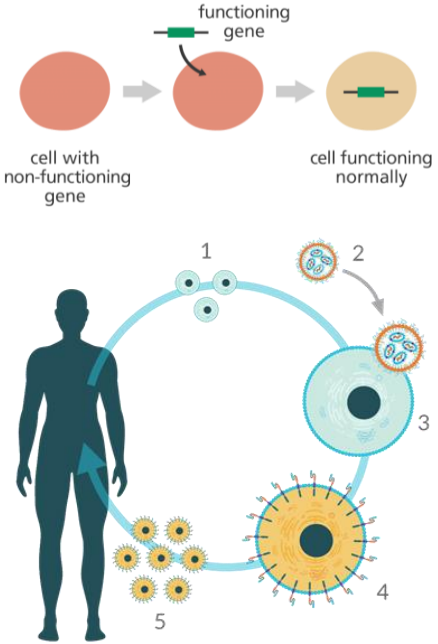
MEET THE HERO MEDICINES!

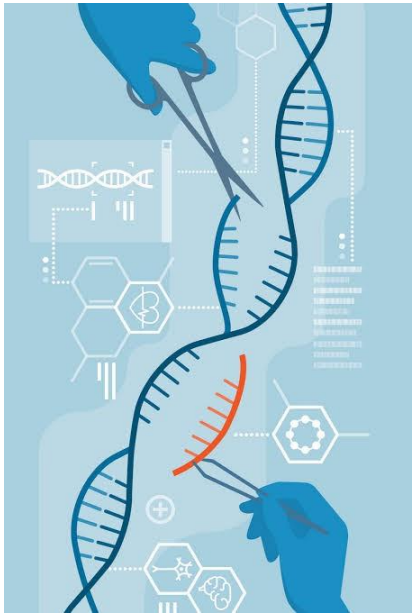
ATMPs can be classified into three main types:

1. Gene Therapy: is a medical technique that involves the introduction, alteration, or removal of genetic material within an individual's cells to treat or prevent disease by restoring the cell functionality.

2. Cell Therapy: is a medical technique that use modified cells to restore the functionality of tissues and organs that are damaged.

3. Tissue-engineered Therapy: is a medical technique that uses tissues that have been modified so they can be used to repair, regenerate or replace human tissue;





ADVANCED THERAPY MEDICINAL PRODUCTS

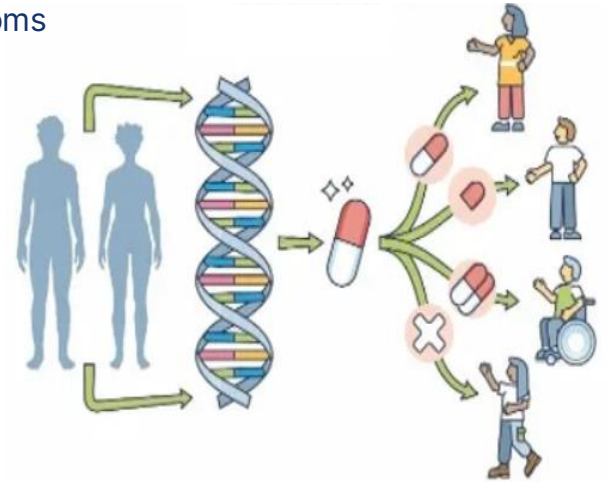
MEET THE HERO MEDICINES!

ATMPs are mainly used to **treat rare diseases especially in paediatric population**.
Almost 70% of rare diseases have genetic origin with paediatric onset.

ATMPs by targeting specific genes can potentially cure or alleviate the symptoms of genetic disorders where traditional treatments may be ineffective.

ATMPs enable **personalised medicine**, an innovative approach in healthcare that tailors treatment to each patient's characteristics.

This allows for **more precise, effective, and targeted interventions**, optimising medical outcomes and minimising potential side effects.



ADVANCED THERAPY MEDICINAL PRODUCTS

1. ATMPs are innovative therapies that include gene therapy, **cell therapy** and tissue-engineered therapy.

2. One primary benefit of ATMPs is their potential to treat diseases that currently do not have **effective treatments** .

3. Gene therapy, a type of ATMP, works by introducing, removing, or altering **genetic materials** within a patient's cells.

4. ATMPs are mainly used to treat **rare diseases** especially in paediatric population.

DIGITAL HEALTH TECHNOLOGIES

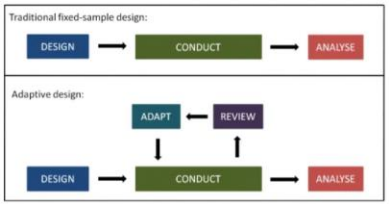
Refers to **tools and services** that use information and communication technology to **improve prevention, diagnosis, treatment, monitoring and management of health and lifestyle**

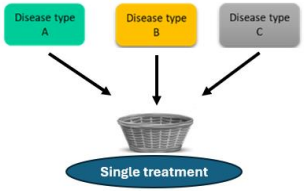
- Mobile Health
- Telemedicine
- Wearable Devices
- Artificial Intelligence (AI)
- Electronic Health Records (EHRs), Consent (eConsent) and Patient-Reported Outcomes (ePRO)

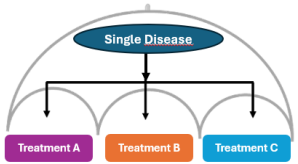
Digital Health Technologies (DHTs)

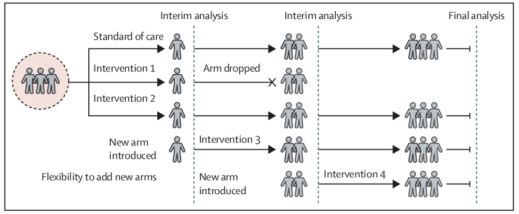
INNOVATIVE TRIAL DESIGNS

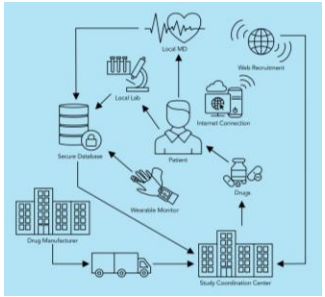
Innovative Trial Designs are **advanced methods for structuring clinical trials**, using approaches that can adapt to real-time data, test multiple treatments or conditions simultaneously, or leverage digital tools to broaden patient access.











Innovative Trial Designs

EPTRI INNOVATION IN PAEDIATRIC CARE

Patient and public involvement (PPI) in research is the way in which patients, families or the general public can help researchers ensure that the design of their research is relevant, friendly and meets the needs of the community. Lay people can work with research funders to prioritise research, offer advice as members of a project steering group, review and develop research materials or undertake interviews with research participants.

CONCLUSION

Together, we can create a healthier future for all children

European **Rare Diseases**
Research Alliance

Thank you for your attention!

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