What is a Patient Registry? Case discussion: Retrospective vs Prospective studies

Session : Understanding Registries & Orphan Drugs

Day 3 - Saturday, 25 April 2026

Dr Nicole CHEMALY (Necker APHP), Angelina BOURBON (Clinical project manager, Imagine Institut)

Who are we?



Dr Nicole Chemaly
(APHP Necker)



Angelina Bourbon, PhD
(Clinical project manager, Institut Imagine)

Before we begin…

Who has already heard the word…..

REGISTRY ?

RETROSPECTIVE STUDY ?

PROSPECTIVE STUDY ?

Have you seen data collected in a hospital or for a research project?

What kind of information do ***you*** think doctors need to understand a disease better?

Before we begin…

Who has already heard the word…..

REGISTRY ?
→ A simple way to organize patient info

RETROSPECTIVE STUDY ?
→ Looking back at data that already exists

PROSPECTIVE STUDY ?
→ Following patients and data over time

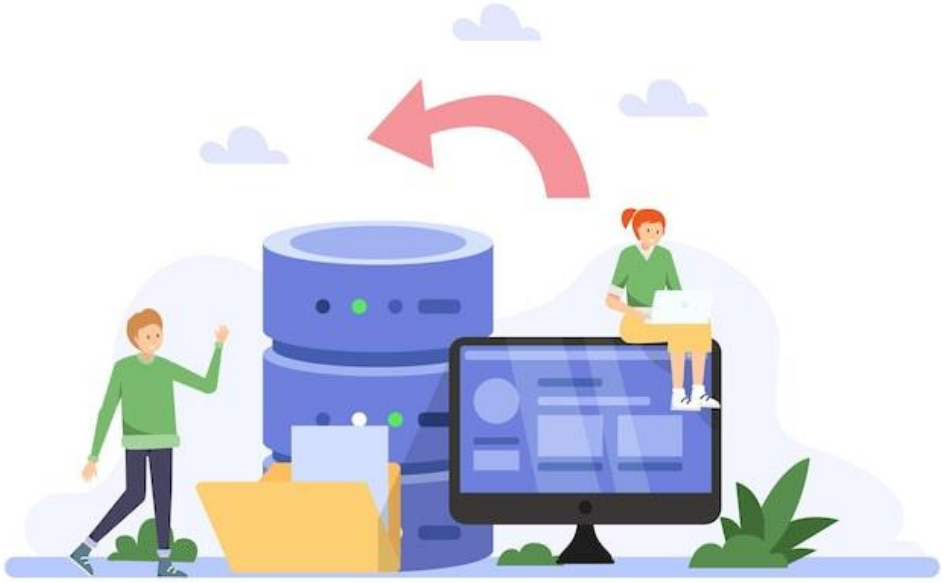
Have you seen data collected in a hospital or for a research project?

What kind of information do ***you*** think doctors need to understand a disease better?

What is a registry ?

Patient registry is a **secure, organized database** with information about patients

Like a *shared online notebook*, made by patients, doctors, researchers and institution *together*

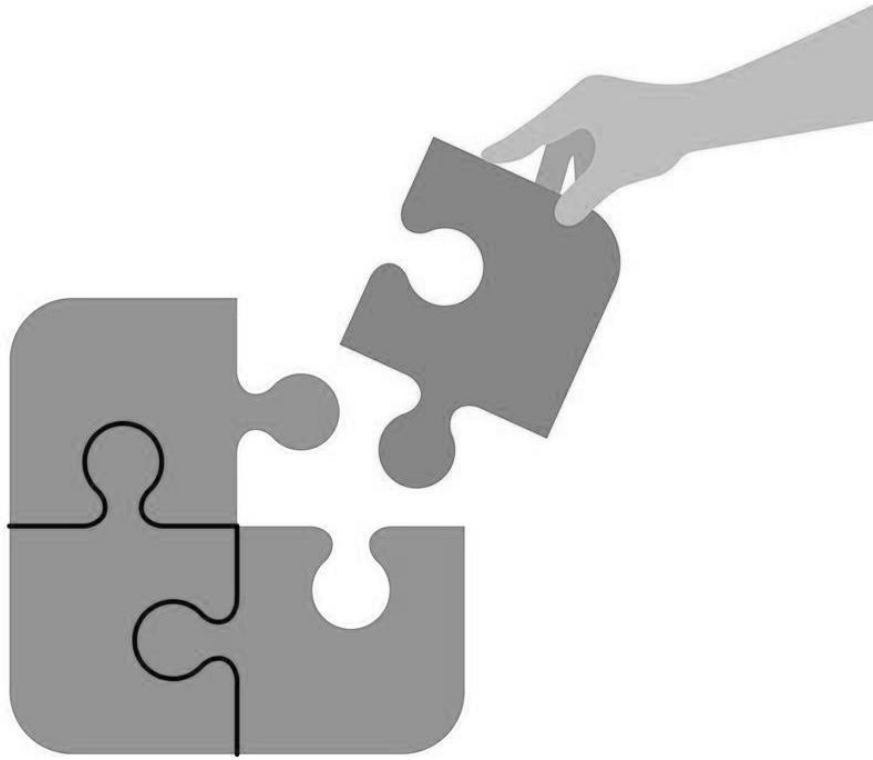


Why do we need registries ?

Rare diseases are… **rare** 😅

- ➔ Few patients
- ➔ Data is scattered
- ➔ Hard to understand the disease

*Registries bring « **everything** » together*

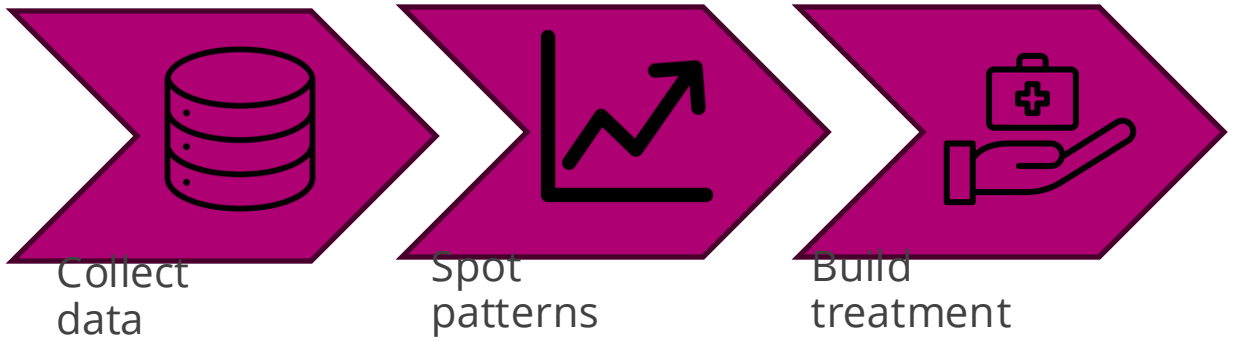


How do registries help research?

When scientists have more (organized) data, they can:

➔ understand the disease better
➔ discover *what* **works** (or *doesn't*)
➔ develop new treatments *faster*

More (organized) data = a better shot at new treatments



Why do registries matter to *You* ?

Patients registers **help** the *rare disease community*

European Rare Diseases Research Alliance

Registries give scientists the proof they need to plan **clinical trials**, apply for **orphan drug status**, and talk with groups like the **EMA** and **FDA**.

What does a registry collect ?

A registry **collect** *informations* and *all sorts* of data

Symptoms & Diagnosis
When symptoms start, how strong they are, and how they change over time



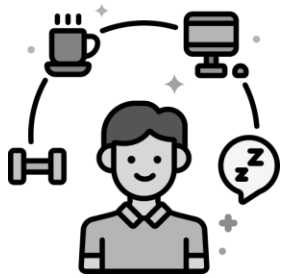
Treatments
Medicines, therapies, doses, and how well they work



Tests results
e.g. blood tests, scans, genetic info, and other clues from the body



Daily life experiences
How you feel day to day, what life is like, and the challenges you face



Key vocabulary

Registry
a systematic collection of data on a defined population, for a specific purpose

Cohort
a group of patients who share a common characteristic, often followed over time.

Study
a research project designed to answer a scientific question;

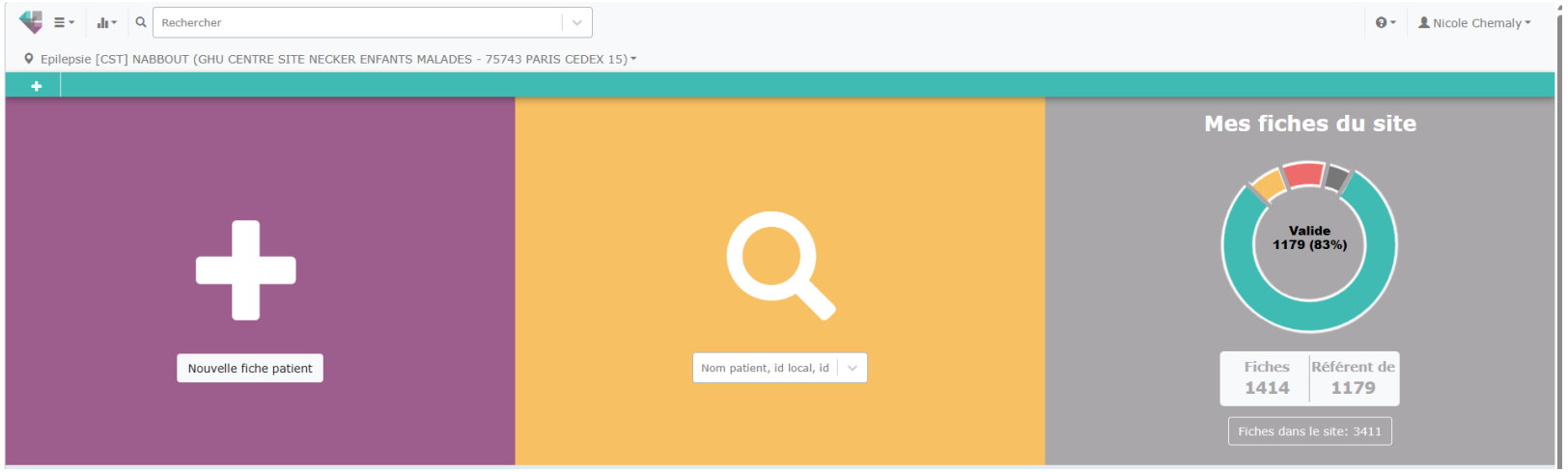
Trial
a study that tests an intervention.

Protocol
the detailed plan describing how the research will be done.

A **registry** can provide the data source, the **cohort** defines the patients, the **study** answers the question, the **trial** tests an intervention, and the **protocol** explains how everything is done.

An example BaMaRa

It is a french registry for rare diseases
It compiles data about the symptoms, the diagnosis, the treatment used, the genetic aetiology if applicable
It uses the Orphanet Codes



An example TOSCA



It is a registry of the clinical manifestations of a rare
disease called Tuberous Sclerosis
It collected datas from over 2000 patients for 5 years, making it the biggest cohort worldwide for this pathology

It highlighted the difficulties that the patients and families have everyday

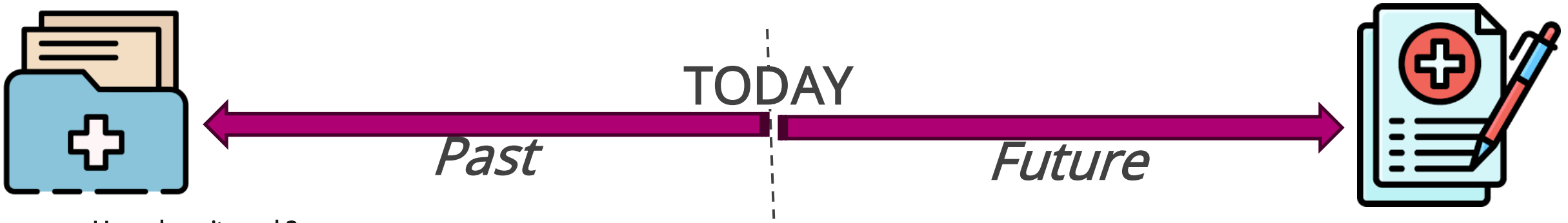
Retrospective vs. Prospective



In the rearview mirror, you see what you've already passed—you can only observe, not change. Through the windshield, you see the choices ahead—you can influence the journey!

Retrospective vs. Prospective

Two perspectives on time



How does it work?
We examine hospital archives and pre-existing medical records

What can we do ? For example :
- Identify trends over extended periods
- Compare groups of patients who received different treatments
- Analyze rare events using large datasets

How does it work?
A precise protocol is established, patients are recruited, and they are followed prospectively through examinations, questionnaires, blood tests, and scheduled visits

What can we do ? For example :
- Observe the effect of a new treatment from the outset
- Measure outcomes and adverse effects with precision
- Collect standardized, high-quality data

An example : RETRO ➔ PRO

In Innov4Epik, we didn't choose between past or future — we used both!
➔ *Project is about rare genetic epilepsies (Developmental and Epileptic Encephalopathies (DEE))*

DEERETRO

Uses **existing medical data**
~400 patients across France
→ **Goal:** understand the disease
= Identify patterns & early biomarkers

DEEPRO

Collects **new data over time**
~150 patients across France
Includes: clinical follow-up, neuropsychological questionnaires, biological samples, specific MRI
→ **Goal:** confirm findings & improve care

DEERETRO helped us understand patterns… and **DEE**PRO is designed using that knowledge

Three things to remember

- ✓ A **registry** = data collection for learning…and more!
- ✓ **Retrospective** = looking back
- ✓ **Prospective** = following forward

… one more thing / slide

~~✓ A registry = data collection for learning… and more!~~
~~✓ **Retrospective** = looking back~~
~~✓ **Prospective** = following forward~~

you are ***SUPER*** important in research! Your stories, your questions, your experiences - they make research better !

You are part of the solution!

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ERDERA
European Rare Diseases Research Alliance

Thank you for your attention!