INTRO To ERNs & ERN-EYE

European Reference Network Connecting healthcare providers
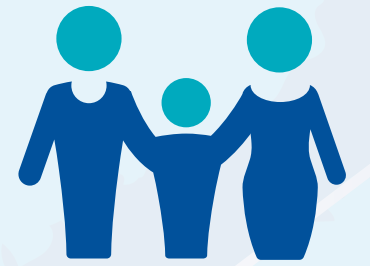
Complex or rare diseases and conditions wich require:
- Highly specialised treatment
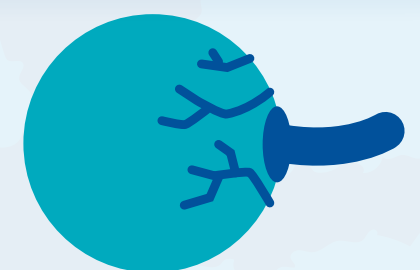- A pooling of knowledge and resources

08 affiliated partners
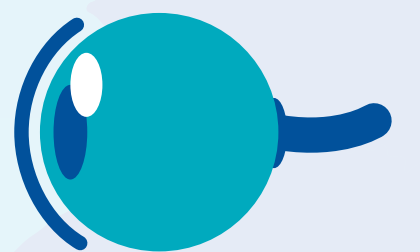
24 EU countries

52 full members

ERN-EYE tools and activities for patient-centered care

- CPMS Virtual clinic
- Guidelines best practices
- Training on RED
- Patient registries
- Support for research

ERN-EYE main axes dedicated to RED

- Paediatric Ophthalmology
- Retinal
- Neuro-Ophthalmology
- Anterior segment
- Genetic Testing
- Low vision

ERN-EYE

European Reference Network

"Integrating ERNs into Member States' healthcare systems is a major challenge that we are all eager to achieve."

Pr Dollfus, ERN-EYE coordinator

ERN-EYE

European Reference Network for Rare Eye Diseases

ERN-EYE

European Reference Network

Paediatric Ophthalmology
Retinal
Neuro-Ophthalmology
Anterior segment
Genetic Testing
Low vision

E-LEARNINGS

Provide a wide access to knowledge on basic clinical and scientific background as well as cutting edge research and therapy developments

GUIDELINES

WEBINARS

Enhance the knowledge on diagnosis and care for Rare Eye Diseases in Europe

Anterior segment RED

Paediatrics RED

Retinal Dystrophies

Neuro-ophthalmology

REDgistry

*Collect uniform data on RED population for epidemiological and/or clinical research
Improve the care and treatment of patient suffering from RED*

☑ **Patient consent**
☑ **Strict rules**
☑ **Pseudonymised**

MOGAD
Jette Lautrup Battistini

WG2

WG1

TWG5

LOW VISION ASSESSMENT OF CORE VISUAL FUNCTIONS CCS
David Keegan

IRON-RELATED RETINOPATIES
Michael Larsen

WG1

RPE65-IRDs
Bart Leroy

WG1

LOW VISION ASSESSMENT OF CORE VISUAL FUNCTIONS CCS
Agnese Suppiej

BARDET-BIEDL SYNDROME
Hélène Dollfus

WG3

TWG5

VIRTUAL CLINIC MEETINGS

The virtual clinic facilitates crossborder dissemination of expertise through the organisation of meetings

ANIRIDIA
Dominique Brémond-Gignac

WG4

KERATOCONUS IN CHILDREN
Luca Buzzonetti

Patient Data Collection

ERN-EYE members are the primary data sources

Submit phenotypic information & genetic test results

• Presentation of complex cases
• Cinical trials
• Educative
• Multidisciplinary
• Novel therapies

REDVAR
ERN-EYE

Rare Eye Disease VARiants review

Pr De Baere

Pr Leroy

Evaluate the results
Assess the pathogenicity of variants

Online presentations & ranking variants discuss

Official results document available on the CPMS

REDgistry
Rare Eye Diseases European Registry
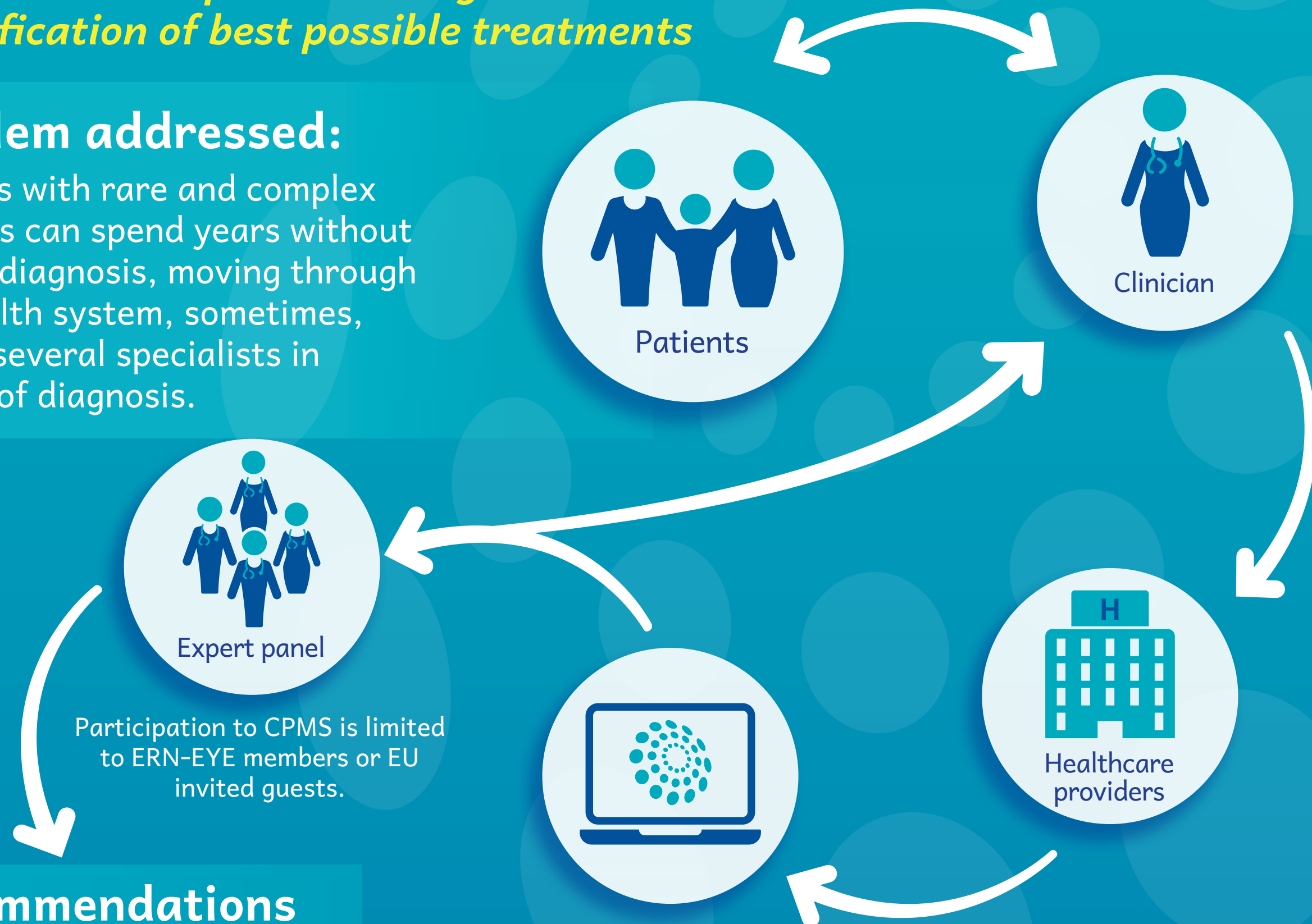
VIRTUAL CLINIC

ERN-EYE CPMS

Improvement in patient's diagnosis and identification of best possible treatments

Problem addressed:

Patients with rare and complex diseases can spend years without a clear diagnosis, moving through the health system, sometimes, seeing several specialists in search of diagnosis.

Patients

Clinician

Healthcare providers

Expert panel

Participation to CPMS is limited to ERN-EYE members or EU invited guests.

Recommendations

Submit phenotypic information & genetic test results

Evaluate the results Assess the pathogenicity of variants

Online presentations & ranking variants discuss

Official results document available on the CPMS

Virtual clinic meetings

The virtual clinic facilitates crossborder dissemination of expertise through the organisation of meetings

○ **Presentation of complex cases**
○ **Cinical trials**
○ **Educative**
○ **Multidisciplinary**
○ **Novel therapies**

GREET
ERN-EYE

Grand **R**ounds **E**RN-**E**YE **T**ransnational meeting

ERN-EYE Paediatrics

ERN-EYE Retina

Dr Noval   Dr Mauring   Pr Brémond -Gignac

ERN-EYE Neuro ophthalmology

Pr Herrmann   Pr Leroy

Dr Hamann   Dr Vignal -Clermont

REDVAR
ERN-EYE

Rare **E**ye **D**isease **VAR**iants review

Pr De Baere   Pr Leroy

ERN-EYE

European Reference Network

EDUCATIONAL TOOLS

ERN-EYE TRAININGS

WEBINARS

Enhance the knowledge on diagnosis and care for Rare Eye Diseases in Europe

Open to all

- High-level & hot topics discussions
- Debates by expert speakers

Past topics

- Foveal hypoplasia
- Marfan Syndrome
- Neurofibromatosis
- Coats Disease
- Idiopathic Intracranial Hypertension
- Genetic testing

SHORT TERM EXCHANGE
for professionnals

👤 Trainees

🏢 Hosts

How long?
1 to 2 week(s)

For whom ?
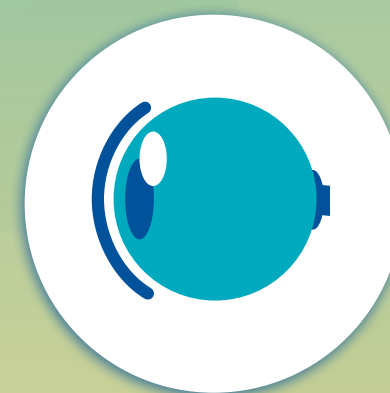Medical specialists, nursing staff, lab staff, orthoptists...

Thematic Coverage
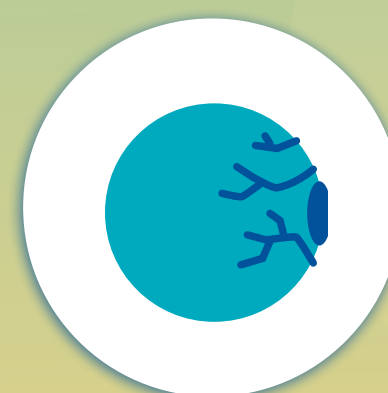Clinical aspects, Psychological support, Organisational strenghtening...

E-LEARNINGS

Provide a wide access to knowledge on basic clinical and scientific background as well as cutting edge research and therapy developments

Different courses offered

Anterior segment RED

Retinal Dystrophies

Neuro-ophthalmology

Paediatrics RED

ERN-EYE GUIDELINE

CLINICAL CONSENSUS STATEMENTS

Improving standards of care throughout Europe

Clinical Consensus Statements developed & their Coordinators

Topic suggestion
ERN-EYE Member

Prioritization of CCS topics
ERN-EYE WG leaders

START OF CONSENSUS

Kick off meetings

Clinical questions identification

Literature research

Delphi process

Manuscript redaction

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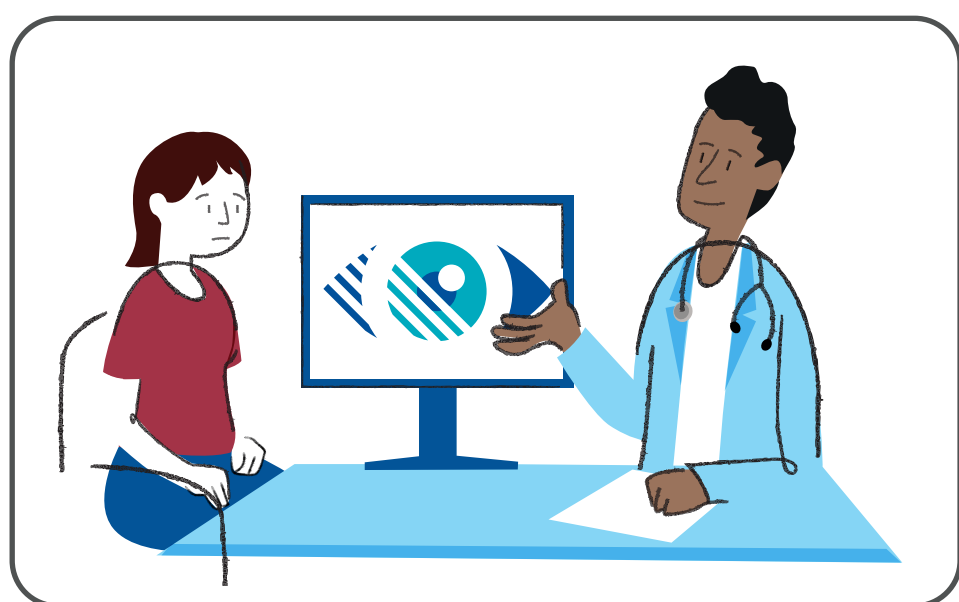
REDgistry

Rare Eye Diseases European Registry

*REDgistry is a European patient registry dedicated to Rare Eye Diseases (RED)*

○ Collect uniform data on RED population for epidemiological and/or clinical research
○ Improve the care and treatment of patients suffering from RED
○ REDgistry follows the FAIR principles and respects the GDPR

1 Patient Data Collection

In addition to the Eye Dataset specific for RED, REDgistry contains a dataset, the JRC (Joint Research Centre) Common Data Elements (CDE), which makes it interoperable with other registries of the European Rare Disease Registry Infrastructure (ERDRI).
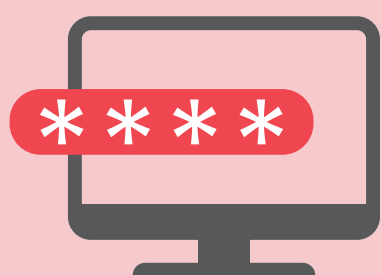
☑ **Patient consent**
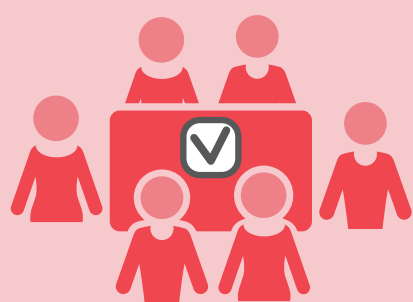☑ **Strict rules**
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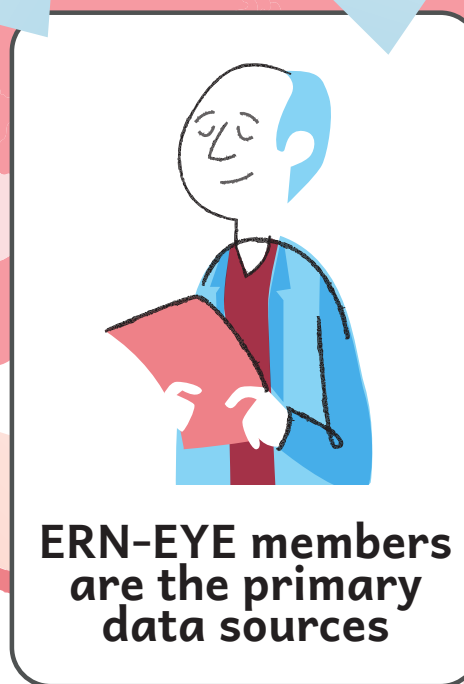
ERN-EYE members are the primary data sources

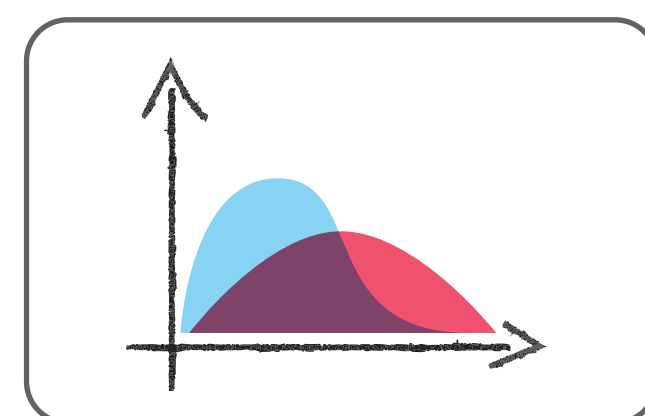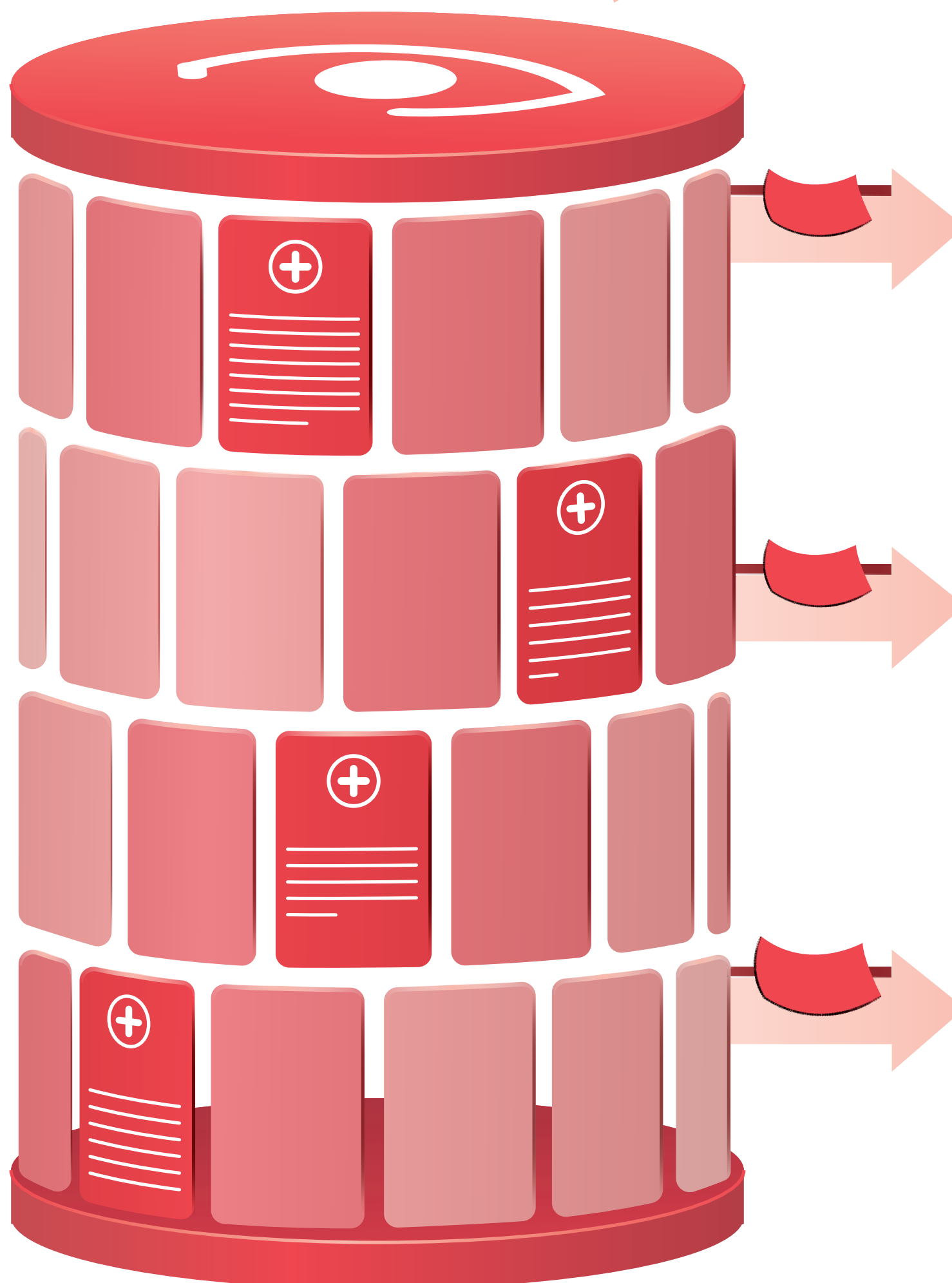### 2 Access to REDgistry data

For stakeholders who request data access

Review by Data access commitee

Agreement between ERN-EYE & stakeholders that have received positive answer

Help researchers carry out epidemiological studies

Support research projects

Identify and include patients in clinical trials and thus grant patients access to new treatments.