

A NETWORK TO
IMPROVE THE LIVES OF

***PEOPLE AFFECTED BY
A RARE DISEASE***



Share4Rare

**RESEARCH SERVICES
TARGETED TO:**

Patient organisations

Scientific societies

Research groups

Scientific services companies

A collaborative project designed by and for the patients

to advance rare disease research

www.share4rare.org

RESEARCH SERVICES

1

Patients'
registries

2

Natural history
of the disease

3

Burden of the
disease

4

Scales and
questionnaires:
creation and validation

5

Quality of life
studies

6

Clinical image data and
patient-reported data
correlation

RESEARCH SERVICES

1

Patients' registries

Collection of information on health, perception, and patient experience to create robust and useful databases.

These data allow for a better understanding of the disease and patients, to detect needs and to monitor patients prospectively.

2

Natural history of the disease

Studies that allow us to understand how a rare disease evolves over time, from the first symptoms to its advanced stages.

3

Burden of the disease

Assessment of the physical, emotional, social and economic impact that a rare disease has on patients and their families.

4

Scales and questionnaires: creation and validation

Development of specific tools to measure and assess symptoms, progression or impact of the disease with scientific rigor.

Validation of the tool in a sample of patients segmented by diagnosis, age, country or language.

5

Quality of life studies

Analysis of how a disease affects the general well-being and health perception of patients.

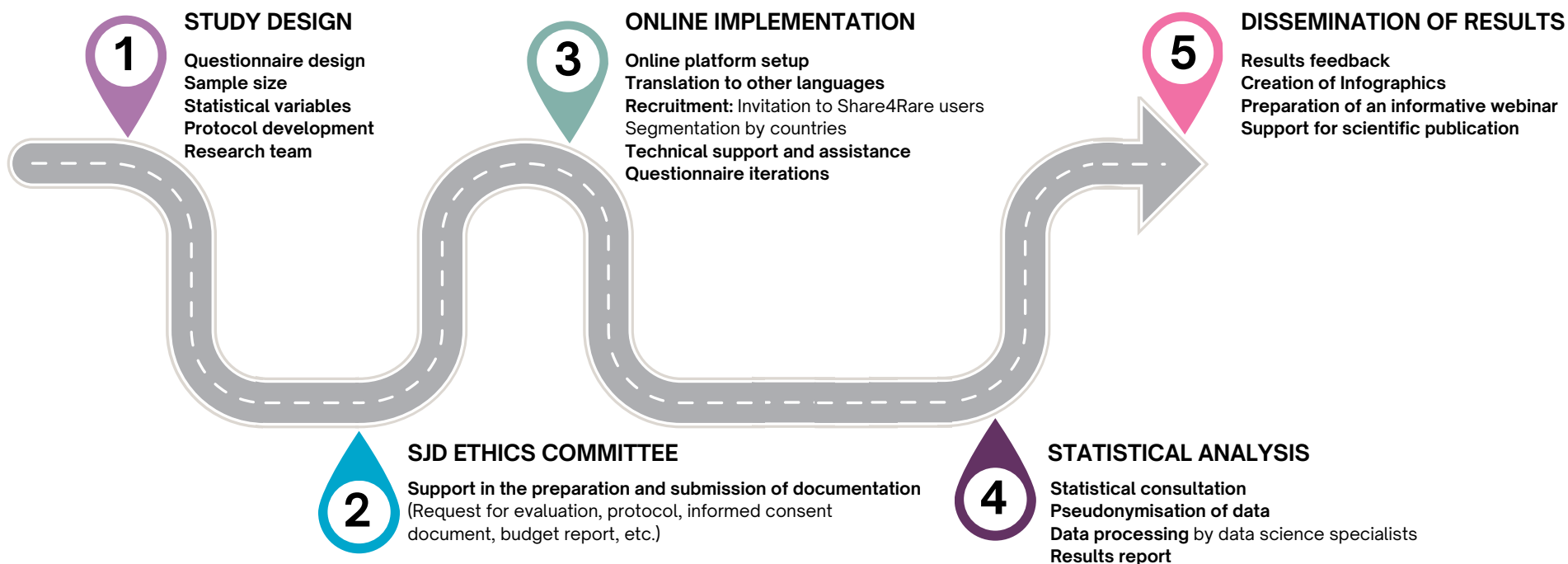
These may be cross-sectional studies of all the rare disease or of a specific diagnosis.

6

Clinical image data and patient-reported data correlation

Linking medical imaging data (such as MRIs) with information provided directly by patients for a comprehensive understanding.

RESEARCH SERVICES



RESEARCH SERVICES – Examples

Patients' registries

- Observatory of patients with **AATD**: Prospective study. Duration: 3 years. Driven by the **Alfa-1 Spain Association** (Alpha-1 Antitrypsin Deficiency), the study aims to assess the evolution of specific aspects of their health and the perception of the care received.

Quality of life studies

- Observational study on the benefits of using *barefoot* footwear in children with **epidermolysis bullosa**. Driven by **DEBRA Piel de Mariposa** Association.

Scales and questionnaires: creation and validation

Repository of available validated scales:

- PROMIS Pediatric Item Bank v2.0 – Pain Interference;
- Neuro-QOL Item Bank v1.0 –Pediatric Social Relations
- NeuroQoL scale on paediatric mobility;
- Neuro-QOL Scale v1.1 –Ped. Upper Extremity Function
- Neuro-QOL Scale v1.1 –Ped. Lower Extremity Function
- Neuro-QOL Item Bank v1.0 –Ped. Social Relations
- PROMIS® Pediatric Item Bank v2.0 – Depressive Symptoms
- PROMIS® Pediatric Item Bank v2.0 – Anxiety – ;
- OVAMA questionnaires on symptoms/appearance;
- OVAMA questionnaires on satisfaction with treatment
- SF-36 Health measures;
- Sleep Disturbance Scale for Children: SDSC;
- Pediatric Sleep Questionnaire (PSQ);
- Brief Infant Sleep Questionnaire (BISQ);
- Morisky Medication Adherence Scale (MMAS).

Clinical image data and patient-reported data correlation

- Pilot study on the genotype-phenotype correlation of brain structure in **Rett syndrome**. Radiological footprint of *MECP2*. **Main objective:** To identify radiological patterns in pediatric patients with RTT, observed in magnetic resonance imaging (MRI), in order to accelerate clinical suspicion of RTT. **Secondary objectives:** To determine if the radiological phenotype correlates with the severity of patients through the RSSS and RSBQ questionnaires.

Value proposition

- **Private** and **secure** environment
- **Multiple-language** studies
- Leadership from a **worldwide-reference** research centre
- Approval from the Sant Joan de Déu Research Foundation **Ethics Committee**
- **Transferable** to non-rare diseases
- **Statistical analyses** of the results
- **Return** of results to participants





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