



Introduction

Project focused on patient-centered resources and evidence synthesis for *GNAO1*-related disorders (*GNAO1*-RD).

Main topics: dyskinetic crises and deep brain stimulation (DBS) decision-making.

Aims to:

- Describe disease burden, management strategies, and caregiver needs.
- Co-create plain-language, multilingual leaflets for families and clinicians.
- Inform ERN-RND care pathways and future consensus recommendations.

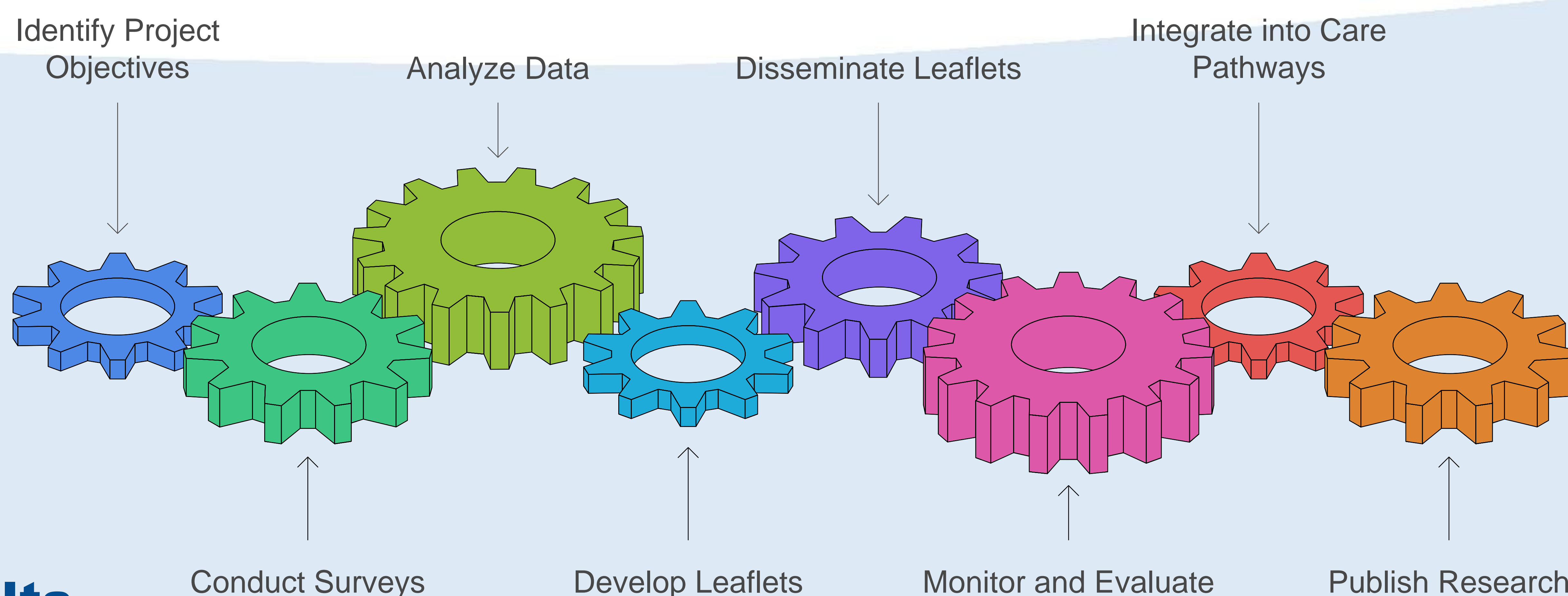
Work Plan

Design: two multi-country, cross-sectional surveys across EU centres.

Participants:

- Dyskinetic crises survey: 26 parents/caregivers of children with confirmed *GNAO1*-RD.
- DBS decision-making survey: 12 caregivers from 10 countries whose relatives underwent DBS.

Patient involvement: active collaboration with caregiver representatives in design, question wording, and leaflet review for readability.



Results

Dyskinetic crises:

- Reported by 80% of families.
- Mean onset: 4.2 years; variable frequency and duration.
- Common triggers: infections and emotional stress.
- Major impact on quality of life, motor function, and family burden.
- Responses to medications were heterogeneous.
- 10 children underwent DBS with mixed outcomes.
- Identified unmet needs in crisis planning and access to specialized care.

DBS decision-making:

- Median age at surgery: 10.7 years.
- Main indication: status dystonicus/dyskinetic crises (9/12 cases).
- Decision often urgent (9/12).
- 8/12 families reported marked reduction in crises after DBS.
- Highlighted gaps in pre-operative information and psychological support.
- Outputs: two multilingual (EN, ES, DE, FR, IT) caregiver leaflets drafted and formatted.

Outlook

Dissemination: publish on ERN-RND, Bow Foundation, Famiglie *GNAO1* Italy, and *GNAO1* España websites; share via newsletters and social media.

Integration: incorporate into hospital neurology department crisis plans and DBS counselling checklists; align with ERN care pathway nodes.

Monitoring: track downloads and collect short feedback (usefulness, clarity, actions taken).

Clinical use: distribute PDFs to ERN centres and patient associations.

Publications

1. Domínguez-Carral J et al., Caregivers' Perspectives and Decision-Making on Deep Brain Stimulation in *GNAO1*-Related Disorders. *Neuromodulation*. 2025 Jun 22;S1094-7159(25)00192-8.
2. Domínguez-Carral J et al., Exploring the Impact of Dyskinetic Crises in *GNAO1*-Related Disorders: A Survey for Parents and Caregivers. *Mov Disord Clin Pract*. 2025 Apr 25. doi: 10.1002/mdc3.70102.