Families with a family member suffering rare disease with a particular focus on lysosomal storage disorders: Psychosocial determinants of the medical care and functioning of the family

1. *Establishing of diagnosis of a rare disease: psychosocial impacts on the family*

- How did the caregivers (family members) and patients receive the diagnosis, how do they perceive the medical conditions in terms of having a rare disease
- What is the state of general awareness regarding the rare diseases among the clinicians, based on the family experience
- Number of false diagnoses prior to the establishing of correct one with regards to the age of onset of first symptoms

2. *Psychosocial determinants in the medical care of patients with rare diseases with a particular focus on lysosomal storage disorders*

- Role of medical personnel, interaction with the family
- What is the life quality impact of the current situation/stigmatizing
- ”Daily life” functioning of caregivers: influence on the work situation
- How did the life/quality of life changed after receiving of diagnosis
- Use of antidepressants in caregivers-> would a targeted psychotherapy reflecting the specific needs of a family with a severely ill member be of benefit?
- Quality of sleep of caregivers subjectively and via scores (psychophysiological insomnia and/or RBD?)
- Situation of siblings: explorative, impact on quality of life
- Number of divorced families

Design: Questionnaire-based study. Collection of medical reports, video- and paper patient interviews. Involvement of patient organizations. Specific Questionnaires to be developed in co-operations with patient organizations` representatives.

Further following questionnaires:

- Beck Depression Inventory-II
- European Quality of Life Questionnaire with visual analog scale
- REM-sleep Behaviour Disorder (RBD) questionnaire, 10-item inventory, max. score 13 points
- Bern Sleep Inventory (specific parts, to be specified)
- Fatigue Severity Scale
- Epworth Sleepiness Scale

To be included: Patients worldwide, reached via family organizations (INPDA, Gaucher organization, NPUK, CATS, Hand-In-Hand, Care for Rare, Sdruzeni Meta), US, Brasil and Australia to be reached as well

Aim of the study: Pilot study to evaluate the psychosocial aspects of having a family member with rare disease