The interdisciplinary and holistic analysis of the functioning of families with child with rare genetical disease

In the group of rare genetical diseases (RGD) there are more than 9,000 recognized ones (Orphanet, 2022). RGD are defined as rare when their prevalence is 1 in 2,500 live births or lower. They are characterized by being chronic and progressive and have an impact on the functioning of the family system in terms of occurrence of depressive symptoms, deterioration in member's needs (displayed in example in self-actualization), affects relationships between parents or overall quality of life. The aim of the project is to investigate the specificity of the functioning of families with children with RGD in terms of psychological, and socio-emotional functioning and also to investigate how these families have been institutionally cared.

The projected study is divided into two parts. The first part will be focused on the explorative analysis of the functioning of 120 families representing 4 different groups divided regarding the type of inheritance of the rare genetical disease: sex-linked genetic disease, monogenic disease inherited in an autosomal recessive manner, microdeletion within one chromosome, and genomic imprinting (30 families for every condition). The participants will fill out paper-pencil questionnaires about their children's adaptive abilities, clinical symptoms, and data about the history of the child's RGD. They will also feel out in research tools regarding their self-actualization, quality of life, depressive symptoms, and satisfaction from many aspects of their life (incl. marriage).

In the second part four interdisciplinary detailed case studies (one for every representative group) will be conducted. For this part, the neurocognitive and socio-emotional assessment of children is planned. Moreover, parents (if presents both) will participate in semi-structured interviews that will provide qualitative data about their functioning. Moreover, the biological data will be analyzed. The blood samples will be collected and analyzed. In this way, detailed data about the mutations, the enzymes involved, the synthesis of enzyme rate, the level of accumulated substances in cells, and other aspects determining the child's phenotype will be obtained. The medical data will be also obtained from the doctor attending the child from the Counselling Centre for Rare genetical diseases. The information about endocrinological and cardiological status, pulmonary problems, physiological functioning and more will be gathered. This will provide a holistic and interdisciplinary view of the child and family functioning that will be assumed in the case study.

The author of the project assumes to recognize the challenges and difficulties accompanying people with RGD and their families to open discussions in the world of science, and in order to be able to comprehensively support their development and contribute to increased quality of life in the future.