

Dr Marianne Becker, Gwennaëlle Crohin, Rita Da Rocha Oliveira

Rare diseases (RDs) are complex, heterogeneous, and frequently disabling conditions that affect around 300 million people worldwide. Approximately 75% of these diseases affect children, many of whom experience prolonged and challenging diagnostic journeys with sequential referrals to numerous specialists and frequently invasive diagnostic procedures. This diagnostic odyssey can have serious consequences for the physical and mental health of these children, and for their families' psychosocial and financial wellbeing. According to the 2022 EURORDIS Rare Barometer survey, 60% of the 76 respondents from Luxembourg reported having consulted more than 5 healthcare professionals to receive a diagnosis, 47% declared lacking psychological support, and 64% had insufficient coordination of their diagnostic pathway.

Given the lack of coordinated care during the diagnostic journey, there is an urgent need to develop an integrated diagnostic pathway for paediatric patients. This presentation will describe a qualitative research study protocol, which aims to design such an integrated diagnostic pathway for paediatric patients with a suspected rare disease. Within the scope of this research, we will conduct semi-structured interviews with patients and caregivers, and focus groups (FGs) with healthcare professionals, to collect data on the diagnostic experiences and unmet needs of patients and caregivers, as well as on the perspectives of healthcare professionals. We aim to identify barriers, facilitators and potential areas for improvement during the diagnostic process, and ultimately develop an integrated diagnostic pathway with the main focus on patients' unmet needs and preferences.

To optimize ecological validity, the semi-structured interview and FG guidelines were co-designed with an Advisory Committee of different stakeholders including paediatrician(s), clinical geneticist(s), patient representative (s), and researcher(s).