

Specific Objectives COST ACTION BM1105:

GNRH deficiency: Elucidation of the neuroendocrine control of reproduction

Specific objectives that will be addressed by the Working Groups over the four years of the project include:

- Reach out to and include clinicians, physician-scientists, genetic counselors, and basic researchers from all European countries (deliverable: Europe-wide network of experts)
- Recruit collectively a cohort of at least 1,000 well-phenotyped patients with GnRH deficiency, and create a common web-accessible database for storing de-identified genetic and phenotypic data on patients (deliverable: database)
- Facilitate the joint investigation of specific patients/families with unique phenotypes identified through the database (deliverable: joint clinical and scientific research projects; publications)
- Facilitate access to and shared use of cutting-edge platforms for genetic research (deliverable: genetic studies, publications)
- Identify 1 or more gene(s) contributing to the disease in each patient, and/or at least 10 novel disease genes (deliverable: genes, publications)
- Characterise the role of each newly identified gene in animal models and cell-based systems (deliverable: genes, publications)
- Fully describe the phenotypic spectrum of GnRH deficiency (including its response to hormonal treatment) and to delineate the underlying genotype-phenotype correlations (deliverable: publications)
- Create genetic counseling guidelines for GnRH-deficient patients and their family members based on the emerging disease architecture (deliverable: genetic counseling guidelines, endorsement by medical society, publication)
- Connect adult and pediatric endocrinologists to optimize therapeutic approach for patients in transition between childhood and adulthood, and to share clinical experience/protocols to identify the influence of hormonal regimens on clinical endpoints such as quality of life and fertility (deliverables: clinical treatment guidelines, endorsement by medical society, publication)
- Recruit and train 100 or more young investigators in reproductive research (deliverables: Training School participation and full-time research/fellowship positions)
- Facilitate joint applications for national and European funding (deliverable: research-supporting grants)
- Reach out to patients, their families, and patient advocacy groups to provide support, education and expert clinical care (deliverable: online, telephone and in-person interaction capabilities at the national level as well as across the network)