ERN on congenital malformations and rare neurodevelopmental disabilities (ERN ITHACA)

ERN ITHACA (Intellectual disability, TeleHealth, Autism and Congenital Anomalies) echoes the diagnostic 'odyssey' experienced by so many patients with developmental anomalies. The network brings together more than 70 clinical genetics department across EU academic hospitals, including experts in rare neurodevelopmental disorders (NDDs) - mainly intellectual disability (ID) and autism spectrum disorder (ASD) - as well as rare multiple congenital anomalies.

ERN ITHACA covers the clinical and biological/genetic diagnosis of these developmental anomalies, the coordination of multidisciplinary care and treatment, and prenatal diagnosis and foetal pathology.

Rare developmental anomalies affect many children and adults - for example, approximately two percent of newborns will be affected by ID and at least one percent by ASD (with or without ID). Roughly half of patients with ID, and more than one in ten with ASD, have a monogenic or a chromosomal disorder. Congenital malformations affect one in 40 babies, often as part of complex syndromes which also display NDDs. More than 5,000 rare syndromes have been described.

ERN ITHACA unites medical experts and ePAG representatives, providing collaborative support for clinical research, developing best practice consensus and guidelines, and improving the early diagnosis, care and cure of patients. The network has also established the International Library of Intellectual disability and Anomalies of Development (ILIAD) patient registry.

The network develops telemedicine and tele-expertise to facilitate collegial discussions between referring doctors and researchers across the EU, and produces training and e-learning tools for health professionals, lay persons and ePAGs.

NETWORK COORDINATOR

Professor Alain Verloes
Université de Paris & Assistance Publique-Hôpitaux de Paris, Hôpital Universitaire Robert-Debré, Paris, France