

ERN on congenital malformations and rare intellectual disability (ERN ITHACA)

This ERN brings together experts in rare congenital malformations and rare intellectual disability disorders. Congenital malformations affect one in 40 babies. For more common malformations, such as cleft lip, there are well-established care networks. For rarer conditions, expertise is scattered across the EU. Many malformations occur together as part of 'syndromes' associated with abnormal growth, development or social adaptation. Over **8 000 syndromes** have been described, and most occur at a frequency of **less than 1 in 2 000**.

Chromosome disorders are one of the commonest causes of malformations and intellectual disability. New tests, such as exome and genome sequencing, have improved the prospects of diagnosis but are not routinely available in more than 50% of highly specialised centres.

Expanding access to this technology is a key goal of ERN ITHACA. The network is also developing telehealth initiatives with virtual



multidisciplinary teams across EU centres, and will use virtual online clinics to improve access to diagnostics without requiring patients to travel.

ERN ITHACA will network parents and patients to develop best practice and initiate guideline development where required. It will establish criteria for patient registry data, advance training for health professionals and facilitate research. The network will work with existing networks in the field and with ERNs

with whom there are complementary interests, while keeping patients at the centre of its activities.

NETWORK COORDINATOR

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