ERN on hereditary metabolic disorders (MetabERN)

MetabERN will develop a real-time consultation platform for clinical decision-making processes and foster translational research programmes across IMDs.

Rare inherited metabolic diseases (IMDs), of which there are more than 700, are individually rare but collectively frequent. Many metabolic diseases have severe, sometimes life-threatening, implications for patients. These conditions include disorders of all organs, can affect people of any age, and require multidisciplinary collaboration between a range of professionals.

Early diagnosis can improve outcomes but only 5 % of known IMDs are currently included in newborn screening programmes in Europe and there is a need for harmonisation of national programmes. For many of these conditions, knowledge about their natural history, the efficacy and safety of therapies, and long-term follow-up is incomplete.

MetabERN seeks to improve the lives of people affected by this highly heterogenous group of diseases by dividing them into seven main categories. It is the first pan-European and pan-metabolic network of its kind.

The network is setting up an inventory of metabolic diseases, developing patient information and training sessions, advancing collaborative diagnosis of new diseases, and establishing a long-term referral point bringing expertise to patients.

MetabERN will develop a real-time consultation platform for clinical decision-making processes and foster translational research programmes across IMDs. It will share knowledge within the network and beyond by expanding to additional regions and countries.

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