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RARE DISEASES AND UNDIAGNOSED DISORDER

Clarifying the molecular cause in a large number of patients with a rare disease. Everyone living with a rare disease should be able to receive high quality services, treatment and support.

Most of the genetic diseases known are rare disorders. The impact of these genetic disorders on public health is relevant since they are individually rare but collectively frequent, affecting lives of approximately 25 million people in Europe about 50% of these patients remain without a molecular diagnosis.



The University of Bologna have traditionally been interested in understanding the molecular causes leading to different genomic and genetic disorders. This commitment resulted also in the **active participation in several European Reference Networks (ERN)**, aimed at facilitate discussion on complex or rare diseases that require highly specialised treatment: [ENDO ERN](#), [ERN LUNG](#), [ERN ITHACA](#), [ERN SKIN](#), [ERN BOND](#) and [ERN EURACAN](#), through the St. Orsola-Malpighi University Hospital and the Rizzoli Orthopaedic Institute.

Several research group have exploited the use of **NGS approaches** to identify causative mutations in patients with a range of different rare Mendelian phenotypes, including syndromic and non-syndromic neurodevelopmental disorders. A list of the main diseases studied includes: intellectual disabilities, neurodegenerative disorders (such as Hereditary spastic paraplegias), Rare Skeletal Diseases (Osteochondrodysplasias), focal epilepsies and epileptic encephalopathies, rare pediatric dermatology diseases, rare endocrine conditions such as Turner Syndrome and lipodystrophies, myelodysplastic syndrome, hereditary optic neuropathies and several neuromuscular disorders among others.

HIGHLIGHTS

Different research groups have established an **extensive network of collaborations with clinical and research centers across major research strengths**, such as NGS sequencing of germ-line and somatic DNA from patients with genetic diseases and the implementation of methods for statistical assessment of disease-gene associations of rare variants for rare Mendelian/complex disorders (trio analysis, gene collapsing analysis).

Gene-editing tools for functional assessment of the genetic variants identified through NGS high-throughput data is also a major area of excellence of the University of Bologna.

The University of Bologna participates to a number of national and international consortia for the analysis of rare variants in the general population (*Network for Italian Genomes* - [NIG](#)) and in diseases ([Epi25](#): a collaborative project aimed to exome sequence as many as 25,000 patients with epilepsy and the *Epilepsy Genome Initiative* - [EGI](#)).