The Telethon Undiagnosed Diseases Program (UDP) pursues the goal of providing a diagnosis to pediatric patients with a genetic disease without a name. This task will be accomplished through collaboration among three Italian clinical centers in Rome, Monza and Naples, coordinated by the Telethon Institute of Genetics and Medicine (Tigem) in Pozzuoli – Naples, where the genetic analysis will take place.

Patients will access the program through their referring physician, who will fill in an online form on the dedicated Telethon UDP website. The requests will be evaluated by the Program’s clinical teams; where the absence of a diagnosis is confirmed, the patients will be examined in one of the three clinical centers. This medical examination might allow identifying a diagnosis; otherwise the patients will undergo whole exome sequencing (WES) through a “family trio” analysis. The outcome will be fed back to the patients via the direct involvement of their referring physicians.

The program will address 350-400 cases and aims at identifying the causative variants in at least 25-35% of isolated cases and in at least 40% of those with hereditary features, resolving a total of 100-120 cases. To the goal of identifying “second cases”, the Telethon UDP team will collaborate with international programs and will use internationally adopted tools and platforms, such as Phenotips, Phenome Central and MatchMakerExchange.

Not only will these results bring a diagnosis to patients and their families, but also they will set the basis for investigating their diseases to the aim of finding treatments and cures.

Workflow of Telethon Undiagnosed Diseases Program

- Web-based
  - Insert requests
  - Referring Physician
  - PATIENT

- Clinical Evaluation
  - by three UDP clinical centers
  - DIAGNOSIS

- Genetic analysis
  - by WES of family trios