UNICAS PMP22: ÚNICAS. Implementation of a Pediatric Network for Personalised Medicine in rare pediatric diseases. A Pilot Project.

Description

The ultimate goal of Pediatric Precision Medicine is to stratify children affected by diseases to improve diagnosis and medical treatments. Translational investigators are bringing to bear ever greater amounts of heterogeneous clinical data and scientific information to create classification strategies that enable the matching of intervention to underlying mechanisms of disease in subgroups of patients.

Most pediatric genetic diseases such as neurodevelopmental disorders, inherited metabolic diseases, neuromuscular disorders, epilepsies and other ultra-rare monogenic conditions are important causes of infantile death and disability in Spain and worldwide.

The main objectives of this project are the 1- Establishment of a Pediatric Research Network on Rare Diseases for Precision Medicine in Spain; 2- Application of Electronic Medical Record standards by mean of the introduction of a minimum clinical data set in four disease groups in order to obtain a set of common standards for clinical data that we are planning to include in the ERNs, CSUR and CIBERs ; 3- Implementation of common tools among pediatric units and the deployment of Use Cases for IMPaCT-Data and IMPaCT-Genomics; 4- Application of automated tools for analyzing and sharing the genomic information of children with rare diseases and the interaction with IMPaCT -Genomics.

The project will deploy a federated data platform following the guidelines, documentation and demonstrators developed by IMPaCT-Data. This pilot project is intended to give the initial kick-off of the Network, to structure its minimum bases taking advantage of existing phenotypic and genomic data in 300 patients and carry out genomic studies and structured phenotypic analysis as a pilot study in 200 further patients with four groups of pathologies. Once the Network is established, funding both in clinical care as well as research at a Regional, National, European and International levels will be applied for.

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