Country: Italy

Organization: National Center for Rare Diseases

Program/Project: The National Center for Rare Diseases of the National Institute of Health (Istituto Superiore di Sanità - ISS) (www.iss.it/cnmr) promotes and develops experimental research and public health actions, as well as to provide technical expertise and information on rare diseases and orphan drugs, for the prevention, treatment and surveillance of these diseases. A project dedicated to Undiagnosed Rare Diseases (URD) has been funded within the 2015 by the Italian Ministry of Foreign Affairs and International Cooperation. The pilote phase of the project consists of a Coordinating Centre (ISS) and 6 clinical participating centres: Tor Vergata University, Rome; L'Aquila University; Mario Negri Institute, Milan; Turin University; Ferrara University; Rare Diseases Centre, Udine. Expected results of the project include: to establish a National database containing information regarding URD patients using common standards and terminology for URD classification; to gather URD patients with similar phenotypes to facilitate match making; to make diagnoses of rare diseases that have not been previously diagnosed; to perform genetic molecular analysis in selected cases. Deep sequencing (WES/WGS) of relevant family members and trios is provided in selected patients.

ISS is IRDiRC Member Institution

President of ISS: Prof Gualtiero Ricciardi

Director of National Center for Rare Diseases: Dr Domenica Taruscio (M.D.)

Website of the URD project: http://www.udnpitaly.com

Organization: Telethon Foundation

Program/Project: Telethon Foundation is a non-profit organisation recognised by the "Ministry of the University and Scientific and Technological Research" (Ministero dell'Università e della Ricerca Scientifica and Tecnologica)". Its mission is to foster research leading to cures for rare genetic diseases. Telethon started up an "Undiagnosed Diseases" programme with the goal of providing a diagnosis to pediatric patients with severe and complex genetic disease without a name. Standardized clinical evaluation is provided by experts of three clinical centers in Rome Milan and

Naples. Prioritized cases are studied at the Telethon Institute of Genetics and Medicine (TIGEM) by high coverage whole exome sequencing (WES) of family trios/quartets. Data are analyzed using a shared pipeline at TIGEM (Pozzuoli) and Ospedale Pediatrico Bambino Gesù (Rome).

Telethon Foundation is IRDiRC Member Institution

Chief Scientific Officer: Dr. Lucia Monaco

Coordinator of the Telethon "Undiagnosed Diseases" Program: Prof Vincenzo Nigro (TIGEM)

Website of Telethon Undiagnosed Diseases project: http://www.telethon.it/node/49281