Poster P.01.1

A NATION-WIDE ITALIAN REGISTRY FOR PATIENTS WITH MUSCULAR DYSTROPHIES AND MYOPATHIES

D"Amico A.[1], Comi G.[3], Tupler R.[4], Bruno C.*[2]

[1]BAMBINO GESU' HOSPITAL ~ ROMA ~ Italy, [2]G.GASLINI INSTITUTE ~ GEONVA ~ Italy, [3]Fondazione I.R.C.C.S. Ca' Granda Ospedale Maggiore Policlinico ~ MILANO ~ Italy, [4]Università degli Studi di Modena e Reggio Emilia ~ MODENA ~ Italy

Muscular dystrophies and myopathies represent heterogeneous group of rare conditions, with different clinical and molecular features.
In light of the extraordinary progress in understanding on the pathogenesis and the molecular bases and to the incoming therapeutic approaches for several muscle disorders, patient registries are becoming essential.
We propose to build-up a registry on Muscular dystrophies and myopathies that will be part of the Telethon Italian NMD registry. This registry will be developed on a IT platform which has a user-friendly web interface allowing both direct patients' and clinicians' participation. The registry will be focused on Congenital Muscular Dystrophies (CMDs), Congenital Myopathies (CMs), Limb girdle muscular dystrophies (LGMDs) and Facioscapulohomeral dystrophy (FSHD), and will involve the 4 clinical networks centers already established for these diseases through previously funded Telethon projects. All the researchers involved have gained expertise working on specific projects. This registry, by guaranteeing the collection of accurate clinical data according to the best clinical practices, will be useful to i) provide epidemiological data, ii) better describe natural history, iii) obtain a better genotype-phenotype characterization, iv) identify new phenotypes, and v) improve management of specific disorders
We believe that this registry may facilitate feasibility studies, trial design and rapid identification of the most suitable patients to be included in clinical trials.

Le distrofie muscolari e le miopatie congenite rappresentano un gruppo eterogeneo di malattie rare che si presentano con caratteristiche cliniche e molecolari differenti. Alla luce degli enormi progressi compiuti nella comprensione della patogenesi e delle basi molecolari di queste patologie e in vista degli approcci terapeutici emergenti per molte di queste patologie, riteniamo che la costituzione di registri di malattia sia molto importante.
Con questo progetto proponiamo la costituzione di un registro di Distrofie muscolari e Miopatie che sia integrato nel registro Telethon Italiano sulle malattie neuromuscolari [vedi www.registronmd.it].
Questo registro sarà sviluppato su una piattaforma elettronica di semplice utilità con un interfaccia che permetta la partecipazione e l'inserimento di dati da parte dei pazienti e dei ricercatori.
Il registro sarà sviluppato su Distrofie Muscolari Congenite (CMDs), Miopatie Congenite (CMs), Distrofie Muscolari dei Cingoli (LGMDs) e sulla distrofia Facio ScapoloOmerale (FSHD e coinvolgerà le 4 reti cliniche che, grazie al supporto del Telethon, si sono costituite negli ultimi anni.
Tutti I ricercatori coinvolti nei network hanno acquisito una importante esperienza su queste patologie, lavorando su specifici progetti di ricerca.
Questo registro, garantendo la raccolta accurata di dati, conforme alle linee giuda di buona pratica clinica sarà molto utile per: i) fornire dati epidemiologici su queste patologie, ii) descrivere in modo accurato la storia naturale, iii) definire meglio le correlazioni genotipo-fenotipo, iv) identificare nuovi fenotipi clinici e v) migliorare la gestione clinica delle diverse forme di patologia, Riteniamo che questi obiettivi sono la base per favorire lo sviluppo di studi clinici, contribuire al disegno di trials clinici e identificare al meglio pazienti idonei alla partecipazione a questi studi.

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Distrofie Muscolari e Miopatie

Coordinator: Adele D'Amico
Partners: Claudio Bruno, Giacomo Comi, Rossella Tupler
Duration (N. Years): 2
Starting year: 2019-2020

Telethon Project (nr):

GSP18002

Disease Name:

CMD, LGMD, FSHD, CM

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congenital muscular dystrophies, limb girdle muscular dysotrphies, FSHD