

Progetti innovativi per le malattie rare

Registro europeo per le lipodistrofie (European lipodystrophy registry)

Ferruccio Santini et all.

Direttore UO Endocrinologia 1, Responsabile Centro Obesità e Lipodistrofie, AOU Pisana

von Schnurbein J1, Adams C2, Akinci B3, Ceccarini G4, D'Apice MR5, Gambineri A6, Hennekam RCM7, Jeru I8, Lattanzi G9, Miehle K10, Nagel G11, Novelli G12,13, Santini F4, Santos Silva E14,15,16, Savage DB2, Sbraccia P17, Schaaf J18, Sorkina E19, Tanteles G20, Vantyghem MC21, Vatier C8, Vigouroux C8, Vorona E22, Araújo-Vilar D23, Wabitsch M24.
Orphanet J Rare Dis. 2020 Jan 15;15(1):17. doi: 10.1186/s13023-020-1295-y.

per scaricare il PDF dell'articolo (in open access):

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6964101/>

Abstract

BACKGROUND

Lipodystrophy syndromes comprise a group of extremely rare and heterogeneous diseases characterized by a selective loss of adipose tissue in the absence of nutritional deprivation or catabolic state. Because of the rarity of each lipodystrophy subform, research in this area is difficult and international co-operation mandatory. Therefore, in 2016, the European Consortium of Lipodystrophies (ECLip) decided to create a registry for patients with lipodystrophy.

RESULTS

The registry was build using the information technology Open Source Registry System for Rare Diseases in the EU (OSSE), an open-source software and toolbox. Lipodystrophy specific data forms were developed based on current knowledge of typical signs and symptoms of lipodystrophy. The platform complies with the new General Data Protection Regulation (EU) 2016/679 by ensuring patient pseudonymization, informational separation of powers, secure data storage and security of communication, user authentication, person specific access to data, and recording of access granted to any data. Inclusion criteria are all patients with any form of lipodystrophy (with the exception of HIV-associated lipodystrophy). So far 246 patients from nine centres (Amsterdam, Bologna, Izmir, Leipzig, Münster, Moscow, Pisa, Santiago de Compostela, Ulm) have been recruited. With the help from the six centres on the brink of recruitment (Cambridge, Lille, Nicosia, Paris, Porto, Rome) this number is expected to double within the next one or 2 years.

CONCLUSIONS

A European registry for all patients with lipodystrophy will provide a platform for improved research in the area of lipodystrophy. All physicians from Europe and neighbouring countries caring for patients with lipodystrophy are invited to participate in the ECLip Registry.

STUDY REGISTRATION: ClinicalTrials.gov (NCT03553420).