30th EUROPEAN CONGRESS ON OBESITY
17-20 May 2023 | The Convention Centre Dublin | Dublin, Ireland

DIVERSE GENOTYPES AND PHENOTYPES OF A LARGE COHORT OF BRAZILIAN PATIENTS WITH FAMILIAL PARTIAL LIPODYSTROPHY (FPLD)

C. VALERIO[1], L. VIOLA[1], N. GUIDORIZZI[2], V. VERAS[3], V. FERNANDES[3], G. LIMA[3], A. FLOR[3], R. MUNIZ[1], M. FREITAS[2], F. PAULA[2], L. ZAJDENVERG[4], J. R. DANTAS[4], A. MATOS[1], R. JÚNIOR[3]

1Institute of Diabetes and Endocrinology of Rio de Janeiro (IEDE), Department of Metabolism - Rio de Janeiro, Brazil.
2 Clinical Hospital of Ribeirão Preto, University of São Paulo (USP) Medical School - Ribeirão Preto, Brazil.
3 Brazilian Group for the Study of Inherited and Acquired Lipodystrophies (BRAZLIPO), Brazil.
4 Federal University of Rio de Janeiro (UFRJ), Internal Medicine Department - Nutrology and Diabetes Session. Rio de Janeiro,Brazil

INTRODUCTION

Familial partial lipodystrophy (FPLD) is a rare disorder that results in partial loss of subcutaneous fat. This condition is characterized by the accumulation of dysfunctional adipose tissue in ectopic depots, mainly in the visceral compartment and organs such as the muscle, liver, pancreas, and epicardial tissue. As a result, FPLD patients often develop insulin resistance and associated complications such as type 2 diabetes mellitus (T2D), hypertriglyceridemia, polycystic ovary syndrome (PCOS), pancreatitis, metabolic fatty liver, and cardiovascular disease.

There are few data on the clinical and molecular prevalence of FPLD in the Brazilian population.

AIM

To assess clinical, metabolic, and genetic characteristics of FPLD type 2 to 6 in the Brazilian Population.

RESULTS

Ninety-three patients from 37 Brazilian families with genetically confirmed FPLD diagnosis, registered in the Brazilian Group for the Study of Inherited and Acquired Lipodystrophies (BRAZLIPO) database, were included in this study.

These patients had a mean age of 44.7 years ± 14.9, and the majority of them were women (80.6%). Most of the mutations found were in the LMNA gene (87%), followed by the PPARG gene (8.6%), PLIN1 gene (3.2%), and MFN2 gene (1.1%).

Type 2 Diabetes mellitus (T2DM) was diagnosed in 59.1% of the patients, mainly affecting females (64%). Glycemic control was predominantly poor as 51.8% of the patients with T2DM had an A1c ≥ 8.0%.

Severe hypertriglyceridemia (≥ 500 mg/dL) and pancreatitis were found in 35.9% and 9.7% of patients, respectively. Metabolic-associated fatty liver (MAFL) and established cardiovascular disease were found in 57% and 11.8% of the patients, respectively.

COMPLICATIONS	PRESENT
T2DM	58.7% (54/92)
T2DM w/ A1c ≥ 8.5%	51.8% (28/54)
TG ≥ 500	35.9% (33/92)
Pancreatitis	9.7% (09/93)
Cardiovascular disease	11.8% (11/93)
MAFLD	57% (53/93)

METHOD

This is a multicenter cross-sectional study where we assessed the genetic, clinical characteristics, metabolic abnormalities, and end-organ complications of patients followed in four Brazilian reference centers for lipodystrophies. Clinical diagnosis of FPLD was based on documented fat loss in selected areas (core diagnosis) plus a screening of metabolic abnormalities. For genetic confirmation of the FPLD, panel sequencing of lipodystrophy candidate genes was performed. The main results are presented as mean ± SD and relative and absolute proportions.

CONCLUSIONS

For the first time, we describe a large cohort of Brazilian patients with FPLD, where Dunnigan Type was found to be the most prevalent. These Brazilian patients with FPLD have multisystem complications, with a high prevalence of diabetes mellitus (T2DM), hypertriglyceridemia, metabolic fatty liver (MAFL), and cardiovascular disease. Efforts in registering the clinical, metabolic, and genetic characteristics of FLPD can lead to optimal management and care of patients

STUDY CENTERS

FMRP-USP RIBEIRÃO PRETO
IEDE
UNIVERSIDADE FEDERAL DO CEARÁ
UFRJ UNIVERSIDADE FEDERAL DO RIO DE JANEIRO

REFERENCES

1. Brown RJ, Araujo-Vilar D, Cheung PT, et al. The diagnosis and management of lipodystrophy syndromes: A multi-society practice guideline. Journal of Clinical Endocrinology and Metabolism. 2016;101(12):4500-4511. 2. Valerio CM, Zajdenverg L, de Oliveira JEP, Mory PB, Moyses RS, Godoy-Matos AF. Body composition study by dual-energy x-ray absorptiometry in familial partial lipodystrophy: Finding new tools for an objective evaluation. Diabetol Metab Syndr. 2012;4(1). 3. Foss-Freitas MC, Akinci B, Luo Y, Stratton A, Oral EA. Diagnostic strategies and clinical management of lipodystrophy. Expert Rev Endocrinol Metab. 2020;15(2):95-114. 4. Montenegro RM, Costa-Riquetto AD, Fernandes VO, et al. Homozygous and heterozygous nuclear lamin a p. R582C mutation: Different lipodystrophic phenotypes in the same kindred. Front Endocrinol (Lausanne). 2018;9(AUG):1-9

CONTACT INFORMATION

Corresponding author:
dracynthiavalerio@gmail.com