# PREVALENCE OF C161 T AND PRO ALA POLYMORPHISMS IN PPAR? IN PATIENTS WITH BERARDINELLI SYNDROME

Baracho, MFP; Santos, MGN; Rodrigues, AC; Fajardo, CM; Vasconcelos, RC; Hirata, MH; Brandão-Neto, J.

Universidade Federal Do Rio Grande Do Norte

Berardinelli syndrome or Congenital Generalized Lipodystrophy (CGL) is a rare recessive autosomic disease characterized by the almost total absence of subcutaneous adipose tissue, hypertriglyceridemia and insulin-resistant diabetes. Peroxisome proliferator-activated receptor gamma (PPARy) is a nuclear receptor involved in metabolic control, playing an important part in insulin sensibility and in adipocyte differentiation. The aim of the present study was to investigate the presence of polymorphisms of the PPARy gene in 18 patients with Berardinelli syndrome. DNA was extracted from peripheral blood and amplified using the polymerase chain reaction (PCR) technique and digested with BbsI and BstuI enzymes to detect C161T and PRO12ALA polymorphisms, respectively, using the restriction fragment length polymorphism (RFLP) technique. Polyacrylamide gel electrophoresis detected the presence of polymorphisms for the pro12ala genotype in only 3 (16.7%) white female patients with consanguine parents. Low polymorphism positivity was considered insufficient to justify the intense metabolic alterations observed in all the patients; this suggests the need to investigate other polymorphisms.

**Supported by**: DNACENTER-RN, LABMOL/USP-SP.
**Key words**: lipodystrophy, PPARy, polymorphisms.