DIFFERENTIAL EXPRESSION OF BASE EXCISION REPAIR PROTEINS IN BERARDINELLI-SEIP’S SYNDROME PATIENTS FROM NORTHEAST OF BRAZIL

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Berardinelli-Seip Syndrome (BSS) or Congenital Generalized Lipodystrophy (CGL) is a rare recessive autosomal disease characterized by nearly complete absence of adipose tissue, resulting in disturbances on carbohydrates and lipids metabolism, insulin resistance, mental retard and premature aging. From 250 cases of BSS reported around the world, the Northeast of Brazil represents the higher prevalence of the syndrome with 39 cases. Since premature aging is associated with DNA damage caused by free radicals and the main DNA repair pathway involved with the removal of oxidizing DNA damage is the Base Excision Repair (BER), the purpose of this study was to analyze if the expression of the major BER enzymes are altered in BSS patients. Peripheral blood samples of 8 BSS families were collected aiming simultaneous extractions of DNA, RNA and proteins. Then, analysis of mRNA levels of APE1, PARP1 and OGG1 were conducted by real time quantitative PCR. We observed that in most BSS patients, mRNA expression levels are reduced when compared with controls. Investigation of non-synonymous polymorphisms (SNPs) that may alter the function of critical DNA repair enzymes, as APE1, PARP-1 and OGG1, will be performed. SNPs analysis will contribute to our understanding of the DNA repair role in BSS physiopathology because these enzymes also perform an important role as inflammatory regulators beyond their DNA repair function.

Key-words: Berardinelli-Seip Syndrome; Base Excision Repair (BER); APE1.