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Genomics, proteomics and *in silico* analysis of spontaneous abortions

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C tudies showed that about 20% of all recognized clinical pregnancies end in a spontaneous abortion mainly in the first semester. $oldsymbol{\mathcal{O}}$ Risk factors associated with the occurrence of a sudden miscarriage have been established and genetic factors are the most prevalent; as a problem that affects innumerable couples, it is important to increase the quality of prognosis and diagnosis. Genomics, bioinformatics and proteomics techniques can be used as powerful tools to provide an integrated molecular analysis of genotypic and phenotypic factors potentially related to sudden miscarriage. A genomic phase of investigation was based on the study of intronic, exonic and untranslated regions of certain genes using PCR amplification followed by sequencing and in silico analysis. The genes of interest are involved in angiogenesis and apoptosis processes. The proteomic phase was divided into three stages. First, protein extraction using the SDS-PAGE technique was optimized. Second, proteins were separated using the SDS-PAGE×IEF technique. Finally, spots extracted from the electrophoresis gels were identified by a MALDI-TOF/MS technique. Some of spontaneous abortions samples had a normal karyotype, wherein the most prevalent alterations found were single nucleotide polymorphisms (SNPs). Using the software Human Splice Finder, it was estimated that 75% and 23% of these differences were in intronic and exonic regions respectively. Considering the exonic regions, 54% of the amino acid substitutions encoded by SNPs could lead to an alteration in the function of some protein domains and or cause damage to some protein structures according to ProtFun 2.2 and PolyPhen-2 software predictions. In the MALDI-TOF/MS analysis, it was possible to identify 23 different proteins related to glycolysis, regulation of the cell cycle, transcription and angiogenic mechanisms and stress responses. HSP 70 and 90 were also identified. From these results we can hypothesize the presence of an undetected bacterial infection. Anti-HSPs produced in excess may have affected fetal growth as the balance between HSPs and their respective antibodies is essential for healthy embryonic development.

Biography

Agata Carvalho was an MSc student of University of Trás-os-Montes and Alto Douro in collaboration with Hospitalar Centre of Trás-os-Montes and Alto Douro. She has a degree in Genetics and Biotechnology and an MSc in Molecular Genetics, both from the University of Trás-os-Montes and Alto Douro. She published 1 paper in a European conference proceeding with scientific refereeing and also presented oral and panel communications in other conferences.

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