P-105 - PRESENTATION OF A HEREDOGRAM OF POSSIBLE MITOCHONDRIAL MYOPATHY ACCOMPANYING FOUR GENERATIONS IN A FAMILY IN AMAZONAS STATE, BRAZIL

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\textbf{INTRODUCTION:} Ernester et al. (1959) described the first mitochondrial disease on a euthyroid patient who had a long history of symptoms related to a permanent hypermetabolism state with morphological and biochemical alterations of the mitochondria (Luft's disease). \textbf{OBJECTIVE:} To describe a case report of probable mitochondrial disease in 04 generations in a family from Amazonas State in Brazil. Materials e \textbf{METHODS:} Clinical case report and heredogram survey. \textbf{RESULTS:} DLF, 41, was consulted in a medical genetics service reporting the following symptoms: Myopathy, muscle pain and exercise intolerance, Asthma, Mental Confusion, Deafness, food intolerance, Meniere's syndrome, myopathic facets and psychiatric illness. In her heredogram survey it is possible to identify several relatives in four generations on the maternal side affected by the same symptoms as hers in varying degrees. Patient reported that in the first generation there were 16 children that died before one year of age with sudden death after intense weeping and 2 surviving daughters with symptoms of myopathy; in the second generation patient reported 10 children deaths before one year old presenting the same clinical condition of the sixteen children of the first generation, one abortion occurred and 4 survivors (3 female and one male) with symptoms of myopathy. In the third generation there were 3 daughters all showing symptoms of myopathy all of them are alive in the moment; and in the fourth generation (proband) has two children of one union and another one of a second union, all of them presenting myopathy and intolerance to exercises and with IQ test above the standard considered normal. \textbf{CONCLUSION:} Based in the heredogram survey, it is possible to visualize a clear inheritance of mitochondrial disease, besides signs and symptoms compatible with mitochondrial diseases. Exoma was requested and results are being expected.