Serum Transthyretin Levels in Brazilian Patients With Familial Amyloid Polyneuropathy.

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Familial amyloid polyneuropathy (FAP) is an autosomal dominant polyneuropathy of adult onset which used to lead to death within 10 years on average after the first symptoms. Serum transthyretin (TTR) levels are reduced in familial amyloidotic polyneuropathy (FAP) in others populations. A single study of patients with senile systemic amyloidosis (SSA) in Sweden found that those individuals also had a significantly lower mean serum TTR concentration. It is noteworthy that there are no reports of levels of TTR in the serum of patients with FAP in Brazil. The objective of this work is to assess the TTR serum levels of patients with FAP and compare these levels to those of healthy patients and to correlate these levels with the clinical development of the disease. We compared the serum TTR levels, as determined by ELISA method. Our data shows that FAP patients have low levels of TTR in serum compared to controls. Patients in early stage of disease (phase 1) present high serum TTR levels than those in more advanced stages (phase 2), which indicates a correlation of TTR serum levels and the progression of disease. Although patients who develop the disease later (50-60 years) had even lower serum levels of TTR than that presented by patients who develop the disease early (30-40 years) for the same disease stage, which may indicate that the depletion of TTR in serum is more exacerbated in late onset of disease. These data suggest that TTR serum levels can be used as a marker for progression of FAP disease in Brazilian patients.

Key words: Amyloidosis, Transthyretin, FAP.