TITLE: Clinical outcomes after Preimplantation genetic diagnosis of Familial amyloid polyneuropathy (Corino de Andrade disease).

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ABSTRACT

Familial amyloid polyneuropathy, first described in 1952 on Northern Portugal, presently named Transthyretin-related hereditary amyloidosis (V30M), is a chronic fatal hereditary autosomal dominant neurodegenerative disorder. The progressive sensory, autonomic and motor neuropathy eventually leads to cachexia and/or cardiovascular collapse and result in death 10-20 years after the onset of symptoms. It is caused by a single nucleotide mutation in the Transthyretin gene.

Disease prevention is crucial, as most individuals are heterozygous and no toxic amyloid deposition occurs until adulthood, with patients diagnosed at reproductive ages. Current treatments are mainly directed towards symptoms relieve, but other have been developed to halt the progression of the disease. These include liver transplantation, Tafamidis and gene therapy. However, the disease can only be eradicated by the use of PGT-M. This is the first large report of PGT-M applied to patients with the V30M variant. It showed that patients have a high likelihood of achieving a live birth per treatment cycle (48%) and that the disease itself, transplantation or Tafamidis treatments seem to have no negative impact. The high rate of live birth obtained should represent a strong stimulus for patients to use PGT-M in order to definitively avoid the transmission of the disease.

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