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Machado-Joseph disease

Patrícia Marques, M Salgado, A Alexandre, J Leal, A Cabaça
USF Cartaxo Terra Viva, ACES Lezíria, Santarém, Portugal

Corresponding author: Dr Patrícia Marques, USF Cartaxo Terra Viva, ACES Lezíria, ARSLVT, Cartaxo, Santarém, Portugal. E-mail: msm.patricia@gmail.com

Objectives: To exhibit the main features of MJD; the framework theme and its significance in the area of Ataxia.MJD is an inherited neurodegenerative disease which occurs by autosomal dominant transmission and is highly disabling. Its prevalence in Portugal is the world's highest (3.1 / 100,000), particularly in the Ribatejo region and the Azores. The gait ataxia (abnormality) is usually the first symptom, manifesting itself from 30-40 years of age.
The diagnosis, although genetic, is based on the diversity of the clinical evaluation. Progressive motor disability, may be associated with dysarthria, dysphagia, hyperreflexia, spasticity, dystonia, tremor, Parkinsonism, parapraxis and diplopia. Other symptoms "non-motor" such as sleep disturbance, depression, chronic pain and dementia are also important in the assessment.
A study was conducted of review articles; Meta-analyzes and supporting articles to research in PubMed / MEDLINE database. The search was limited to studies published in the last 10 years, in English and Portuguese, using the MeSH terms "Machado-Joseph disease". From the articles obtained those that encompassed the defined goal were selected.
Of the 275 articles found 5 were selected: 4 review articles and one article for research support.
There is no current cure for JDM. The treatments provided are for symptomatic relief and to delay of progression of the disease.
Thus the Primary Care Services have an important role in the early detection of the most common symptoms of the disease, especially when it comes to Ataxia. When this occurs, it is advised that an imagery and genetic study be carried out. Family research, screening for other JDM cases that may benefit from symptomatic treatment is essential; and secondly, it is important for genetic counselling, since there is a 50% chance of inheriting the mutant allele.
The family doctor's role is denoted essential, because this disease leads to major functional disability, being indispensable coordination with the resources available to family, social and psychological support to these users and family.