Clinical profile of autosomal dominant hereditary ataxia

Siti Aminah Sobana, Mochammad Faisal Afif Mochyadin, Uni Gamayani and Almira Zada

Department of Neurology Faculty of Medicine Padjadjaran University
Department of Neurology Hasan Sadikin General Hospital Bandung, Indonesia

Abstract

Hereditary ataxia is a clinically and genetically heterogenous group of disorder characterized by slow progressive incoordination of gait and associated with poor coordination of hands, speech and eye movement. It can be inherited in an autosomal dominant, autosomal reessive, x linked, or mitochondrial. This is a pedigree of a 5-generation-family with gait ataxia and in combination with symptoms such as nystagmus, tremor, sensory (numbness), and dysarthria. The affected of first generation was the mother but in the 2nd, 3rd, 4th and 5th generations the affected are males and females. The inheritance is autosomal dominant. The onset of gait ataxia is in between 30-50 years of age. The inheritance is autosomal dominant and the early symptom starts with sensory or dysarthria. There are 8 family members with gait ataxia who have Scale for Assessment and Rating of Ataxia (SARA ) between 2-28. Seven family members do not have ataxia, but have other symptoms with SARA between 0.5-2. There is anticipation within each generation. The investigation of family member no V.6.4. (male, 31 years of age) showed dysartria begins at 18 years old and gait ataxia at 30 years old. Other symptoms were nystagmus, tremor, stiffness, and numbness. Physical examination showed nistagmus, tremor, hyperreflexia, gait ataxia, and sensory disturbance. NCS study showed axonal demyelinating sensoric motoric peroneus bilateral. EMG study showed poliphasic pattern in anterior tibialis muscle. MRI showed mild dilatation of 4th ventricel, enlarge cysterna magna and with free communicating to foramen magendy (Dandy Walker Syndrome?). After 4 month physiotherapy treatment SARA score improved from 8 to 1. It need further investigation in clinical and genetic testing.

Keywords: Autosomal dominant; gait ataxia; hereditary *Author for Correspondence