FRIEDREICH'S ATAXIA: CLINICAL MANAGEMENT AND PRESENTATION OF CASES

Dionyssiotis Yannis^{1,2}, Danopoulou Anna², Xanthopoulou Elpida², Siafaka Ioanna³, Kokolaki Maria³, Vadalouka Athina³

¹ Hospice for the Disabled, ASYLON ANIATON, Kypseli, Athens, Greece

² Physical Medicine and Rehabilitation Department, European Interbalkan Medical Center, Thessaloniki, Greece

³ Hellenic Society of Pain Management and Palliative Care (PARH.SY.A.), Athens, Greece

Introduction:

Hereditary ataxias are a genetically heterogeneous group of diseases characterized by gait ataxia, loss of balance, poor coordination, oculomotor dysfunction and ataxic dysarthria. Symptoms occur slowly over time with progressive deterioration. The majority of hereditary ataxias with an early onset are inherited in an autosomal recessive manner.

Purpose:

The aim of the study is to review Friedreich's ataxia complications related to pain and present cases.

Method:

We present two cases (brother and sister) with Friedreich's ataxia (FDRA) that are chronically hosted since 15 years. Both are wheel chair bound, and need assistance in all activities of daily living due to incomplete tetraplegia (quadriplegia). Nystagmus, ataxic dysarthria with slow and slurred speech, dysphagia, hearing impairment, common manifestations of the disease, is also present. During the neurologic examination deep tendon reflexes are found to be diminished to absent, mainly on lower limbs, and muscle strength is pathologically reduced, while dystonia and spasticity are present in upper limbs. The patients also suffer from diabetes mellitus and cardiomyopathy.

Results:

The patients were provided with nursing care, adequate nutrition, bladder catheterizations in order to prevent complications i.e. pressure ulcers and a tailored rehabilitation program was organized to maintain their motor function and reduce pain. Pain was assessed with visual analogue scale (VAS) and spasticity using Ashworth scale.

Discussion-Conclusion:

Hypertrophic cardiomyopathy and myocardial fibrosis, lead to heart failure. Our patients are treated with cardioselective beta blocker metoprolol. Figure presents ECG of the female patient. Increased R wave amplitude and inverted T waves are typical findings.

Increase of spasticity *is the key component* in order to intervene early in complications in these patients. Although there is no known cure that can alter the natural course of this chronic, degenerative disease, symptomatic treatment can help the patients maintain the maximum possible function and quality of life.

Current recommendations insist that treatment for diabetes should be initiated early and insulin is preferred over oral antidiabetics. Our patients are treated with metformin, pioglitazone, glimepiride without achieving good glycemic control and thus we are considering conversion to insulin. Both patients have indwelling urinary catheters due to neurogenic bladder dysfunction (overactive detrusor muscle), due to inability to perform intermittent self-catheterization. They are treated with Citalopram for depression, which is common in patients with FDRA. Patients are provided with nursing care and adequate nutrition. Frequent repositioning and mobilization are necessary for the prevention of pressure ulcers. Although there is no disease modifying agent symptomatic treatment can help the patients to maintain the maximum possible function and quality of life.