**Assessment of Respiratory Function in Friedreich’s Ataxia**

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**INTRODUCTION:**

Hereditary ataxias constitute an extensive group of clinically and genetically heterogeneous neurodegenerative diseases. The ataxias are divided into autosomal dominant, autosomal recessive, X-linked, mitochondrial ataxias, congenital and sporadic ataxias.

Friedreich’s ataxia (FRDA) is the most common cause of autosomal recessive ataxia worldwide. It was first described by Nikolaus Friedreich in 1863. FRDA is caused by expanded guanine-adenine-adenine (GAA) triplet repeats in the first intron of the frataxin gene (FXN), resulting in reduction of messenger RNA and protein levels of frataxin in different tissues. So, Friedreich ataxia (FRDA) is a rare condition that cause nervous system damage and movement problems, including muscle weakness and impaired coordination (ataxia). Heart problems, vision problems, spine problems, and diabetes can occur, too. Whitin 10 to 20 years of the first symptoms, an individual with FRDA generally requires a wheelchair. Friedreich ataxia, develop disorders of the sleep, respiratory complications and dysphagia. Aspiration pneumonia is one of the main causes of death in these patients. There are few studies on the assessment of pulmonary function in ataxias, as well as what is the form of prevention for aspiration pneumonia and ventilatory failure. The purpose of this study is, therefore, to evaluate the respiratory function in Friedreich ataxia.

**OBJECTIVES:** To evaluate the lung function of individuals diagnosed with Friedreich's ataxia through respiratory function tests and phrenic nerve conduction when compared to the healthy controls. Furthermore, to correlate respiratory assessment data with demographic data (age, duration of the disease), anthropometric data (weight, height, BMI, cervical and abdominal circumference) and data from the SARA and ICARS scales on the Friedreich's group**.**

**METHODS:** A total of 16 individuals (15 adults and 1 children) with FRDA and 20 healthy controls were recruited.

The patients and controls underwent, assessment spirometry, Inspiratory and expiratory pression test, sniff test and Peak cough flow test. We too, correlated tests with ICARS and SARA scale, respectively.

**RESULTS:** Thirty-five (15/42.9% Friedreich's group and 20/57.1 healthy controls), with an age mean (standard deviation) of 37.1 (11.0) were studied. There were no differences in sex or age between the groups, but Friedreich's group had significantly lower weight, height, and BMI (p=0.04, p<0.01 and p<0.01, respectively). Friedreich's group had significantly lower values of respiratory parameters (forced vital capacity, forced expiratory volume in 1 s, PEF, oxygen saturation, respiratory and cardiac frequencies, maximal inspiratory effort in the supine and seated positions, and sniff nasal inspiratory pressure, all parameters with p<0.01), when compared to the control group. There are no differences among the mean and maximum phrenic amplitude’s (p=0.42 and p=0.37, respectively) between groups. There was a significant negative correlation observed between age and forced vital capacity (rho = -0.86; p<0.01), age and maximal inspiratory effort (rho = -0.69; p<0.01), and age and Tiffeneau index (rho = 0.71; p = 0.01). A moderately significant correlation (rho=-0.68; p=0.02) was observed between forced expiratory volume in 1 second and cervical circumference based on anthropometric data. Forced vital capacity (rho=-0.67; p=0.02), maximal inspiratory effort (rho=-0.73; p=0.01), maximal inspiratory effort in seated position (rho=-0.68; p=0.02), and maximal inspiratory effort in supine position (rho=-0.73; p=0.01) all showed a negative, moderate correlation with the SARA scale. Additionally, ICARS scale and forced vital capacity (rho=-0.61; p=0.04), maximal inspiratory effort (rho=-0.75; p<0.01), maximal inspiratory effort in seated position (rho=-0.82; p<0.01), and maximal inspiratory effort in supine position (rho=-0.83; p<0.01) were found to significant negatively moderately correlation.

**DISCUSSION**: We demonstrate that patients with Friedreich’s ataxia have many respiratory difficulties comparing with healthy control. This has not been observed in follow-up studies of this patients. Limitation of the study was patient’s number. We report that the respiratory assessment is significantly important in patients with FRDA. Our study shows significant dysfunction in all the outcome measures used, ICARS and SARA scores are significatively reduced when compared with healthy controls (p < 0.001).

**CONCLUSION:** More studies are necessary to identify respiratory complications in Friedreich ataxia and these findings could be useful when approaching progressive neurological disorders.