terns of muscle weakness and preservation of deep tendon reflexes did not differ between patients with SMA type 2 and 3.

doi:10.1016/j.nmd.2011.06.900

P3.7

Trained health professionals of an intensive care unit of a public hospital dedicated to spinal atrophy patients: Increase of life expectancy and quality of life

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Spinal Muscular Atrophy (SMA) is a genetically heterogeneous group of neuromuscular disorders. The most frequent form is a severe infantile type, the onset of symptoms being before 6 months of life, with a life expectancy of only 2 years. We report the initiative of a public hospital staff to dedicate an intensive care unit (ICU) to SMA patients. We reviewed the clinical data of 10 patients admitted to the ICU of a public hospital located nearly 50 km from Sao Paulo City, Brazil. Patients age, first symptoms, onset of symptoms, lab exams and family history were analyzed. Ten patients were found to be admitted at an intensive care unit of a hospital near Sao Paulo with the clinical diagnosis of spinal muscular atrophy: six females. Their ages varied from 4 to 11 years (average 6.5). First symptoms were hypotonia in 7 out of 10, two were found less active, not moving their limbs as usual and one had difficulties sucking. Age of onset ranged from 1 to 9 months (average 4.6). The neurological examination showed tongue fasciculations in three of them. All are tetraplegic. Fifty per cent had no copies of SMN1, 20% had exons deletions of the SMN1 gene, 10% had no copies of SMN2 and 20% had non conclusive molecular analysis. We found three cases in our sample with a history of consanguinity. Our data stress the importance of creating medical centers with professionals dedicated to neuromuscular cronic diseases requiring hospital admission. Life expectancy was highly increased as well as quality of life. The children were very well adapted to the condition of living in a hospital, with a quality of life much better than the one they would have in their houses.

doi:10.1016/j.nmd.2011.06.901

P3.8

Cardiac involvement in patients with spinal muscular atrophies

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The spinal muscular atrophies (SMAs) comprise a group of disorders characterized by progressive weakness of the lower motor neurons. Several types of SMAs have been described based on age onset of clinical features: acute infantile (SMA type I), chronic infantile (type II), chronic juvenile (SMA type), and adult onset (SMA type IV) forms. The incidence is about 1:10,000 live births with a carrier frequency of 1:50. The mortality and/or morbidity rates of SMAs are inversely correlated with the age at onset. A X-linked recessive adult form of bulbospinal muscular atrophy (BSMA), has also been described, characterized by bulbar and lower motor neuron weakness. SMAs are believed to only affect skeletal muscles; however, new data on SMA mice suggest they may also impact the heart. Aim of the study was to evaluate the records of 37 type II/ III SMA and 6 BSMA patients, aged 6–65 years. All patients had a standard ECG and 25/37 a routine echocardiography. The parameters analysed were the following: Heart rate (HR), PQ interval, PQ segment, Cardiomyopathic Index ventricular and supraventricular ectopic beats, pauses $\geqslant$2,5, ventricle diameters, wall and septum thickness, ejection fraction, fiber shortening. The results show that HR was within the normal limits (85 $\pm$ 15b/m) in all patients; the Cardiomyopathic Index was higher than the normal values (2,6–4,2) in 10% of ARSMA and 16, 6% of XLSMA. Left ventricular systolic function was within the normal limits in all SMA patients, both autosomal recessive and X-linked. A dilation of the left ventricle – without systolic dysfunction – was observed in only two patients with ARSMA, aged respectively 65 and 63 years; however they were hypertensive and/or affected by coronary artery disease. Data here reported contribute to reassure patients and their clinicians that type II/III SMAs and BSMA do not present heart dysfunction.

doi:10.1016/j.nmd.2011.06.902

P3.9

Late-onset lower motor neuronopathy (LOSMoN) - clinical and genetic evaluation of two new families

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Autosomal dominant adult onset spinal muscular atrophy is a multifarious entity with only a few disease-causing genes identified so far. We have recently described two large Finnish families with a spinal lower motor neuronopathy beginning with widespread painful symptoms of cramps-fasciculations after age 30 and followed by slowly progressive weakness of muscles. EMG studies showed chronic neurogenic changes in all limb muscles. Slightly reduced sensory amplitudes without slowing of nerve conduction velocities was observed in some but not all patients. Muscle biopsy findings were neurogenic with fiber type grouping and grouped atrophic fibers combined with some secondary 'myopathic' features such as increase of internal nuclei. We identified two new families from the same geographic region in Finland including one interesting case with an earlier disease onset at about age 15, based on typical symptoms and electrophysiological studies. The most helpful diagnostic finding is the presence of visible fasciculations throughout the body at an early stage when EMG studies may be otherwise normal. Over 90% of patients had cramps and fasciculations, absent patellar and achilles reflexes, mildly to moderately elevated CK values and MRI studies consistently showing posterior lower leg involvement predominating in medial gastrocnemius. Faciobulbar muscles were spared. Disease course was fairly benign and none of the patients was confined to the wheelchair. Although, the most disabled patients needed/required mobility scooters outdoors because of muscle fatiguability and poor balance. The phenotype, with except for the young onset individual, is well in line with the disease in the previously reported families. Molecular genetic data are compatible with the previously suggested linkage on chromosome 22 as identified by a common core haplotype of some 3.5 Mb. Fine mapping of the linked region to narrow down the region of interest further is ongoing.

doi:10.1016/j.nmd.2011.06.903