AN INTERESTING CASE OF QUADRIPARESIS IN THE YOUNG

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Abstract: We present here an interesting case of motor neuron disease, pure lower motor neuron type, spinal muscular atrophy in a 21-year old male who presented with weakness of all 4 limbs since 1 year of age. Patient had progressive wasting and weakness with fasciculation of muscles. On examination higher mental function was normal. There was no cranial nerve, sensory, cerebellar, autonomic involvement. All blood investigation was within normal limits. MRI brain was normal. NCS, EMG, muscle biopsy done.

Keyword: quadriparesis, spinal muscular atrophy, motor neuron disease, minipolymyoclonus

A 21-year old male came with complaints of weakness of all 4 limbs since 1 year of age and cough with expectoration - 1 week. Initially patient had weakness of both lower limbs which then progressed to weakness of both upper limbs and involved neck and trunk muscles also. There was both proximal and distal muscle weakness of both upper and lower limbs with twitching of muscles in tongue and hand muscles. No history suggestive of cranial nerve, sensory, cerebellar involvement. No similar illness in past PERSONAL HISTORY: mixed diet taker. Unmarried. FAMILY HISTORY: not born of consang. marriage. 3 sibling elder died in uterus. Younger sibling died after birth reason not known on examination patient was conscious, oriented. afibrile. not anemic. No cyanosis/ jaundice/ clubbing/ pedal edema/ lymphadenopathy. No neurocutaneous marker. Thyroid examination-normal HEIGHT NECK RATIO-13(normal). Kyphoscoliosis present with convexity to right and disappears on bending forward. Wasting of muscles in upper and lower limb present with minipolymyoclonus. Fasciculation in tongue and hand muscles were present. Contracture present in knee. VITAL SIGNS: pulse 75/min. bp 120/70mmhb in supine position. resp. rate 20/min. single breath count-30 CNS EXAMINATION: HIGHER MENTAL FUNCTION-normal. CRANIAL NERVE EXAMINATION-NORMAL BULK OF MUSCLE-severe wasting of both upper and lower limb with wasting of trunk muscle also, fasciculation of hand and tongue present. MINIPOLYMYOCOLONUS present TONE: hypotonia of all 4 limbs.

POWER: power 3 in upper limbs except hand muscles 2.power 3 in lower limbs except 2 in ankle

REFLEXES: superficial reflex present. Deep tendon reflex not elicitable. SENSORY SYSTEM: all sensation intact. EXAMINATION OF CEREBELLUM AND AUTONOMIC NERVOUS SYSTEM-normal. Coordination could not be tested because of weakness of muscle. BLADDER AND BOWEL: intact. EXAMINATION OF SPINE AND CRANIUM-normal. OTHER SYSTEM EXAMINATION: CVS- s1 s2 heard. no murmur RS-nvbs heard. P/A soft.
ELECTROMYOGRAPHY REPORT
SKELETAL MUSCLE BIOPSY shows only fibroadipose tissue suggestive of spinal muscular atrophy PROBABLY TYPE2

DISCUSSION SPINAL MUSCULAR ATROPHY
d/t degeneration of ant.horn cells and in some types bulbar neuron.autosomal recessive with homozygous deletion on smn on chromosome 5. 5 types. sma type 1 fetal movt.towards end of preg. or first few months of life severe hypotonia, a weak cry and respiratory distress severe head lag, frog leg position sensory normal muscle stretch reflex absent bell shape chest deformity fasciculations of tongue present .atypical infantile sma-smard-resp.distress.lactic acidosis,cardiomyopathy . sma type 2-chronic sma.most common.delayed motor mile stones.lower limb weakness>upper limb weakness.minipolymyoclonus highly suggestive of diagnosis.mild and slow progression.kyphoscoliosis present sma type-3.before 3 years-type 3a.after 3 years-type 3 b.waddling gait with protrubent abdomen.patient use gowers maneuver sma type4-mostly affects adults. quadriceps weakness is a prominent feature.ad adult onset sma-finkel type sma

SMA VARIANTS:
Ø bulbar hmn1 or bvilsØ bulbar hmn2 or faziolonde disease Ø kennedy diseaseØ cmtØ davidenkow syndromeØ rukuyan spinal muscular atrophy

en.wikipedia.org/wiki/Spinal muscular atrophy
www.smafoundation.org/bradleys neurology.
harrisons principles of internal medicine.