AN INTERESTING CASE OF QUADRIPARESIS IN THE YOUNG

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Abstract : A 14 YEAR OLD GIRL ADMITTED WITH HEARING LOSS AND VISUAL DIMINUTION SHE ALSO HAD WEAKNESS AND WASTING OF BOTH UPPER AND LOWER LIMBS MUSCLES. ROUTINE INVESTIGATION DONE. EMG, NCS DONE. WE PRESENT HERE A RARE CASE OF MADRAS MOTOR NEURON DISEASE VARIANT WITH PRIMARY OPTIC ATROPHY AND SENSORINEURAL HEARING LOSS.

Keyword : quadriparesis, motoneuron disease, madras motor neuron disease, madras motor neuron disease variant.

WEAKNESS OF TRUNK MUSCLE

A 14 year old female came with complaints of hearing loss -7 years, defective vision- 6 years weakness of all 4 limbs- 2 years. patient has hearing loss which is progressive, bilateral and not associated with ear discharge, tinnitus or ear ache. patient has progressive loss of vision, which is painless and not improving with pinhole test. Initially patient had weakness of both lower limbs which then progressed to weakness of both upper limbs and involved neck 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 h/o unsteadiness in walking. no h/o sensory involvement family history patient not born of consanguineous marriage. she has 1 sibling who is normal. on examination patient was conscious, oriented, afebrile, thin built, moderately nourished. no pallor, no cyanosis/jaundice/ clubbing/pedal edema/lymphadenopathy. kyphoscoliosis is present with convexity to right side. No thyromegaly. height neck ratio 13. wasting of muscles in upper and lower limb present.no neurocutaneous marker. fasciculation of tongue muscle present.

WEIGHTING OF HAND MUSCLES

VITAL SIGNS PR-75/min, BP- 120/70mmhg .RR- 20/min. SINGLE BREATH COUNT-30.

HIGHER MENTAL FUNCTION-normal
CRANIAL NERVE: 2nd nerve- visual acuity reduced both eyes field of vision is reduced in both eyes. fudus- optic atrophy present in both eyes. 8th nerve- weber test - lateralized to right ear. rinnies test=ac>bc in both ears 9th and 10th- gag reflex defective. left>right 12th- wasting and weakness of tongue muscles. fasciculation of tongue muscles present other cranial nerves normal

MOTOR SYSTEM
BULK OF MUSCLE- reduced on both upper and lower limb TONE: hypotonia of all limbs. POWER: 3 in upper limbs except hand muscles which is 2. power 3 in lower limbs except in ankle which is 2
REFLEX: ALL SUPERFICIAL REFLEX-present, bilateral plantar flexor. ALL DEEP TENDON REFLEX-absent
SENSORY SYSTEM- all modalities of sensation intact.
CEREBELLM: normal
CRANIUM: normal
SPINE- KYPHOSCOLIOSIS PRESENT
NO MENINGEAL SIGNS
NO INVOLUNTARY MOVEMENTS
ANS- normal
OTHER SYSTEM EXAMINATION:
CVS- s1 s2 heard. no murmur
RS-nvbs heard.
P/A soft.

LAB INVESTIGATIONS; COMPLETE BLOOD COUNT - NORMAL
BLOOD SUGAR,UREA,CREATININE were within normal limits.
CPK - normal
CHEST XRAY , ECG - normal
MRI BRAIN was normal
EMG showed delayed compound motor unit action potential with
mild reduction in amplitude with prolonged distal latency.

OPHTHALMIC EVALUATION- primary optic
atrophy right>left

ENT EVALUATION- bilateral severe sensorineural hearing loss.
OGTT-normal study.

AUDIOMETRY
IN VolVEMENT OF LMN DISTRIBUTION OF MUSCLE WASTING
AND WEAKNESS
SENSORINEURAL DEAFNESS
PRIMARY OPTIC ATROPHY
INVoLvement of MULTIPLE CRANIAL NERVES , MADRAS
MOTOR
NEURON DISEASE VARIANT IS DIAGNOSED.
DIAGNOSIS: MADRAS MOTOR NEURON DISEASE VARIANT

DISCUSSION
FIRST DISCOVERED BY MEENAKSHI
SUNDARAM, JAGANNATHAN AND RAMAMOORTHI IN 1970S.
SUB GROUP OF MND FIRST DESCRIBED IN MADRAS IN
YOUNGER AGE GROUPAGE OF ONSET 10-30 YEARS

PREDOMINANTLY AFFECTS MALES
MADRAS MOTOR NEURON DISEASE IS COMMON IN
YOUNG PEOPLE.
ETIOLOGY IS PROPOSED TO BE INFLAMMATORY.
BENIGN COURSE ABSENCE OF FAMILY HISTORY
GRADUAL ASYMMETRIC INVOLVEMENT OF ALL 4
LIMBS LATER MANIFESTING AS CLASSIC
ALS WEAKNESS OF FASCIAL AND BULBAR MUSCLES
IN 60%

MOST STRIKING IS SENSORINEURAL DEAFNESS
BIOCHEMICAL PARAMETER- IMPAIRED OGTT
REDUCED SERUM CITRATE
INCREASED SERUM PYRUVATE DUE TO ALTERED
CARBOHYDRATE METABOLISM
FAZIO LONDE DISEASE-AR,EARLY AGE AT
ONSET,RARITY OF PYRAMIDAL SIGNS, NORMAL
HEARING,RAPIDLY PROGRESSIVE FATAL COURSE
SPORADIC ALS-OTHER FEATURES LIKE
CHOREA,CEREBELLAR ATAXIA, ABSENCE OF
DEAFNESS, LATE INVOLVEMENT OF BULBAR NUCLEI

DIFFERENTIAL DIAGNOSIS
1)BROWN VIALETTA VAN LAERE SYNDROME
2)JUVENILE ONSET ALS
3)SPINAL MUSCULAR ATROPHY
4)FAZIO LONDE DISEASE
5)POSTPOLIO PROGRESSIVE MUSCULAR ATROPHY

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