CLINICAL PRESENTATION OF CHIARI MALFORMATION I IN CHILDREN

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PEDIATRIC NEUROSURGERY

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3.1 YEARS

- Average duration of symptoms: 3.1 years (range 1month -20 years)

- If nonspecific complaints are included, this becomes 7.3 years

3-4 %

- The frequency of radiographic findings consistent with Chiari I among children undergoing brain or spine imaging for any indication

SYMPTOMS GENERALLY EXHIBIT THE FOLLOWING CHARACTERISTICS

- Clinical presentation varies with respect to age
- Younger patients tend to
  - present sooner, with
  - shorter symptom duration than adults

- Compression on brain stem and cervicomedullary junction
- Abnormal CSF flow dynamics
  - Syringomyelia
  - Scoliosis

15-37 %

- Discovered incidentally within asymptomatic patients or patients evaluated for nonspecific symptoms like headache or dizziness

CLINICAL PRESENTATION: 1. HEADACHES

- 27-70% of children with symptomatic CM1.
- In adolescents/adults:
  - Occipitocervical location
  - Provoked or intensified by Valsalva-type maneuvers:
    - Sneezing, coughing
    - Laughing, screaming
    - Defecation
    - Running, repetitive jumping

CLINICAL PRESENTATION: 2. SCOLIOSIS

- Demonstrates strong association with syringomyelia
- Most (but not all) pediatric CM1 patients with scoliosis have underlying syringomyelia
- Not all CM1 patients with syringomyelia have scoliosis

- Scoliosis associated with Chiari is associated with
  - Unusual curvature types
  - Unusual locations

CLINICAL PRESENTATION: 3. SYRINGOMYELIA

- Cervical15-21%
- Cervicothoracic 12-25%
- Thoracic 15-16%
- Lumbar 3-4%
- Holocord 39-44%

CLINICAL PRESENTATION (LESS COMMON): 4. BRAINSTEM OR UPPER CERVICAL SPINAL CORD COMPRESSION)

- Medulla: RESPIRATORY FUNCTION → sleep apnea
- Brainstem or upper C spine: Sensorimotor deficits (hemiparesis, upper extremity paraparesis, quadriparesis)
- Lower cranial nerves: (up to 10% of pediatric population):
    - Dysphagia
    - Absent gag reflex
    - Dysarthria
    - Vocal cord dysfunction
    - Abnormal extraocular motility

NEONATAL PERIOD – INFANCY (UP TO 3 YEARS)

- LIMITATIONS OF COMMUNICATION
- Generalized irritability
- Crying spells w/ behavioral patterns suggesting neck discomfort (reaching for the head or neck)
- Concerning signs:
  - Opisthotonus
  - Frequent neck extension/arching
  - Apneic episodes

- **Medullary compression with lower cranial nerve dysfunction**: ***oropharyngeal dysfunction*** – one of the most common presenting symptoms 0-2 yo
  - Dysphagia, choking, or aspiration
  - Poor feeding, failure to thrive
  - Gastroesophageal reflux
  - Persistent cough
  - Snoring or episodic sleep apnea
  - Stridor
  - Recurrent respiratory infections

- Syringomyelia: sensorimotor deficits – less frequent compared with other groups

- In this (as well as other age groups) there does not seem to exist a correlation between the extent of tonsillar herniation and the presence or absence of syringomyelia

TODDLERS (3-5 YEARS)

- As they develop improved ability to communicate and localize their pain, they may be able to verbalize more effectively complaints
- May report headache or discomfort in the upper neck
- 40-57 % complain of occipital headache (prompted by Valsalva)

- Frequently syringomyelia and/or scoliosis. May report
  - Back or shoulder pain
  - Paresthesias
  - Gait disturbance and/or
  - clumsiness
- PE:
  - Cosmetic irregularity along the spine
  - Subtle sensorimotor deficits
  - Hyperreflexia

CHILDHOOD AND ADOLESCENCE (5 YEARS AND OLDER)

- Most commonly: occipital headache and/or neck pain, often induced by Valsalva-type maneuvers (straining for a bowel movement, laughing, coughing, sneezing etc) and of short duration
- To warrant surgical consideration, these headaches should be severe enough to impact activities of daily living (ie missing school) or quality of life.
- They may exhibit oropharyngeal dysfunction

- Scoliosis, typically associated with syringomyelia (19-76% of pts) → may lead to back or shoulder pain in this group
- Classic symptoms suggesting syringomyelia include:
  - Upper extremity weakness – prominently affecting intrinsic muscles of the hand
  - Pain and temperature sensory loss (anterolateral spinothalamic tract) in a cape-like distribution
  - Preservation of light touch sensation and proprioception (dorsal columns)
  - Absence of superficial abdominal reflexes ipsilateral to the convexity of scoliosis

- Vocal cord dysfunction, hoarseness, dysarthria
- Absent/reduced gag reflex
- Extraocular motor deficits (eg esotropia)
- Sensory loss

CLINICAL CONDITIONS ASSOCIATED WITH CM1

- Several craniovertebral junction abnormalities like basilar invagination → CM1 up to 33-38%
- Most commonly (but still rare) associated conditions:
  - Hydrocephalus (8-10%)
  - NF 1 (up to 5%)
  - Growth hormone deficiency (4%)
  - Sprengel deformity (abnormal elevation of scapula)
  - Pierre Robin syndrome

RARE PRESENTATIONS OF CM1 IN CHILDREN

- More obscure presentations based on their acuity, rapid progression:
  - Dysphagia
  - Hemiparesis
  - Respiratory distress
  - Gait dysfunction
  - Anisocoria

PRESENTATION ACCORDING TO SEVERITY

- Severe:
  - Syncopal episodes, drop attacks
  - Acute spinal cord injury after trauma (eg quadriplegia)
  - Respiratory failure requiring mechanical ventilation
  - Cardiorespiratory arrest, sudden death

- Moderate
  - Focal sensorimotor deficits (mononeuropathy, eg plantar flexion weakness)
  - Urinary incontinence
  - Torticollis
  - Trigeminal or glossopharyngeal neuralgia
  - Sensorimotor hearing loss

- Mild:
  - Nystagmus (typically downbeating)
  - Chronic hiccoughs
  - Chronic cough
  - Cerebellar or cerebellovestibular dysfunction (eg vertigo)

REVIEW OF SYMPTOMS

- There are common symptoms with different severity and combinations in each patient
- Other conditions can coexist: syringomyelia, bone abnormalities in CC junction, tethered cord, genetic disorders
- The degree of tonsillar herniation does not correlate with severity of symptoms
- Not everything can be attributed to Chiari
- There is no evidence that seizures are correlated with Chiari

PAIN

- Headache
  - SUBOCCIPITAL
  - INTENSE PRESSURE AGGRAVATED BY VALSALVA MANEUVER
  - There might be some relation of CM1 and migraines
- Neck ; Upper back pain
  - Muscles hurt, their function is affected and become to act antagonistically

EYE AND EAR

- Nystagmus
- Strabismus – cranial nerves

- Cerebellar compression → tinnitus, balance abnormalities,

SLEEP APNEA

- Headaches might be associated with sleep apnea in advanced cases
- 60% of children were found to have some degree of sleep apnea

- Chiari can be underdiagnosed, missed, or overdiagnosed
- The accurate diagnosis/management is based on a multitude of factors
  - Symptoms
    - Duration and evolution over time
    - Current presentation
  - Neurological examination
  - Imaging