CLINICAL PRESENTATION OF CHIARI MALFORMATION I IN CHILDREN

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PEDIATRIC NEUROSURGERY
ADVOCATE CHILDREN'S HOSPITAL, OAK LAWN CAMPUS
3.1 YEARS

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

- If nonspecific complaints are included, this becomes 7.3 years
• 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
• 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

- Cervical 15-21%
- 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, quadriplegia)

• 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 (e.g., quadriplegia)
  - Respiratory failure requiring mechanical ventilation
  - Cardiorespiratory arrest, sudden death
• Moderate
  • Focal sensorimotor deficits (mononeuropathy, e.g. 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