Congenital Anomaly of The Upper Extremity

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Congenital Muscular Torticollis (CMT)
Etiology

exact etiology is not known
1) compartment syndrome
2) in utero crowding
3) neurogenic
4) mesenchymal cell
Clinical Finding

Head tilting : involved side
Chin rotating : the opposite side
• mass : maximum within the first 4 weeks, then gradually regressed
4-6 months : disappear mass, only tightness

7-20% combined with DDH or acetabular dysplasia
Torticollis

DDX

• Congenital anomaly (osseous type)
  Basilar impression
  Atlantooccipital anomalies
  Unilateral absence of C1
  Familial cervical dysplasia
  Atlantoaxial rotatory displacement

• Trauma

• Inflammatory

• Ophthalmic problem

• Neurogenic
  Cervical cord tumor
  Arnold Chiari syndrom
  Ocular pathology
  Proxysmal torticollis of infancy
Torticollis due to Spine deformity (Klippel-Feil syndrome)
Atlantoaxial rotatory displacement
Torticollis due to Ophthalmic problem
Treatment

• Initial treatment: conservative (effective ?, spontaneous resolution ?)
• Surgical treatment: persistent deformity after 1 years

(Op toal op timing: 1-4 years)
Treatment

1. Unipolar : mild & moderate
2. Bipolar : older age, recurred, severe case
3. Endoscopic release
4. Middle 1/3 resection
5. Complete resection : rare
Treatment

avoid damage of spinal accessory nerve, jugular vein, carotid vessel, facial nerve
Treatment

(post operative care)

cervical collar?
plaster cast?
Buckminster Brown brace?
Torticollis orthosis?
Congenital Muscular Torticollis

- preop
- postop
- Follow up

6, female

(before the age of 6 to 8 years: remodeling of facial asymmetry.)

1794787
unifocal myotomy in old age without facial asymmetry
unifocal myotomy in old age without facial asymmetry

up to 12 years without facial asymmetry: as good result as earlier operation, but not perfect.
unipolar myotomy in old age with facial asymmetry
residual facial asymmetricity after surgery in old age

established facial deformity or a limitation more than 30 degrees of motion; bad result
1, male
(1707176)
Sprengel’s Disease
(Congenital high scapula)
Sprengel’s disease

Pathology

- **Scapula**
  - high, small (smaller vertical diameter and apparently greater width)
  - Inf. angle: medial rotation
  - Sup. angle: tilted forward.

- **Clavicle**
  - tilted toward upward and laterally
Pathology

• **Omovertebra**: connection between the spinous process and scapula
  bony, cartilaginous, fibrous

• **Musculature**: **Trapezius**: most often affected
  rhomboid, levator, serratus ant: hypoplastic and fibrotic
Clinical Feature

- \( F : M = 3 : 1 \)
- Left is more often
- 3-5 cm higher in average
- Abduction is limited
  - GH joint : Normal
  - TS joint : restricted
Sprengel’s disease

Treatment

considering factors

1. severity of deformity
2. functional impairment
3. associated anomaly
4. age
Severity of deformity

- Grade 1: very mild: invisible in cloth
- Grade 2: mild: lump in undressed
  : simple excision
- Grade 3: moderate: elevated 2-5 cm
- Grade 4: superior angle near the occiput

3, 4: scapula should be lowered
high scapula due to neurogenic spasm
Case of Grade 2
Case of Grade 4
Sprengel’s disease

Functional impairment

- Omovertebra bone
  - resection of the omovertebra
- motor weakness cause by fibrosis, hypoplasia
Associated anomaly

• Klippel-Feil syndrome
• Kyphosis
• Scoliosis

: overshadow the deformity
Spina bifida
3-7 years

- < 3 : extensive and difficult
- > 7 : greater danger of brachial plexus injury
- recently, 6-9 months ? (Tachdjian)
Technique

Numerous!

: Most popular

• Woodward
  • Green

Sprengel’s disease
Treatment

extensive surgery

• normal anatomic appearance cannot be provided!
• caution must be exercised!
• expectations should be realistic!
Treatment

Green Technique

- divided at periscapular area (scapular insertion)
- traction wire (scaplua to ilium)
- Pro: definite improvement of function
- Con: difficult procedure
Treatment

Woodward Technique

- detaching the origin of the trapezius and rhomboid muscle from spinous process
- Pro: more easier (but not easy)
- Best result
Grade 4
extensive surgery

• normal anatomic appearance cannot be provided!
  • caution must be exercised!
  • expectations should be realistic!
Inferior angle
Spinous process
Atrophied Trapezius
Paravertebral muscles
Spinous process
Levator scapula
Omovertebral bone
Rhomboides
Spinous process
Preop x-ray

Sprengel’s Disease
2, Female,(1540231)

Preop clinical
Op (Woodward) at 4 Y, Postop X-ray

Follow up clinical photo
Congenital Radial Head Dislocation (CRHD)
Clinical feature

- Most common congenital anomaly of the elbow
- Ant: 47%, Post: 43%, Lateral: 10%
- Do not noted until 4-5 years of age (or school age)
12 years old girl: First visit: only clicking sound.
acquired traumatic form
(missed Monteggia, Fx of radial neck, pulled elbow)
DDX

1. bilaterality, familial Hx, trauma Hx, associated anomaly

2. Radiologic findings
   - capitulum: small, hypoplastic (primary)
   - radial head: ovoid, small (secondary)
   - radial notch of ulna: small or absence
Post-traumatic D/L
Neglected Monteggia

Congenital D/L
Inheritance

- Autosomal dominant or X-linked recessive
- Associated multiple exostosis, nail patella syndrome, antecubital pterygium syndrome
  (60% with other syndrome)
Cong.D/L.Radial Head

Treatment

- Surgical treatment is seldom necessary in childhood
- Radial head resection before skeletal maturity has been associated with several different complications

(By Chapman's Orthopedic Surgery, 2nd edition & Campbell" Operative Orthopedics, 10th edition)
Treatment

Older than Infant

Resection of the radial head

1. Progressive pain
2. Progressive LOM
3. Progressive Limitation of activity

Regardless of Age!

(common in trauma, but rare in congenital, )
Treatment

younger than 1-2 y

lengthening of the ulna
+ shortening, osteotomy of radius
+ reconstruction of the annular ligament

: preliminary procedure
Treatment

open reduction in the infancy controversy!

• recommend until 3 years?

• no evidence to justify?

(rare incidence, limited experience, small report)
04.11.4 (post op 3Y)
02.5.14: 1st op (6 months after op of the left side)
Congenital anterior dislocation of the radial head: a case with radiographic findings identical to traumatic dislocation

X-ray → Traumatic?, but No trauma hx, bilateral → Congenital?
4.5Y, boy
Immediate postop

Postop 2Y 4M
“Our study revealed that three elbows realigned successfully with open reduction of the radial head, ulnar osteotomy, and annular ligament reconstruction. The reduced radial head was well maintained and improved cosmetic deformity, and mild improvement of range of motion could be achieved.”

*J Hand Surg*(Br).2011.36;161
Congenital R-U Synostosis
Clinical feature

- mainly proximal end
- discovers early adolescence
- All most all fixed in pronation: limited supination and pronation
- bilaterality: 60%
Clinical feature

• Type I : complete synostosis
  radius is longer and arches anteriorly

• Type II : less involved with dislocation of the head
  often unilateral & other anomalies
  (absence of thumb, poly or syndactyly)
Proximal R-U synostosis with CRHD
Pathology

- initially osteochondrosis, later the osseous bridge
- Narrow & tight interosseous memb
- Abscence of brachioradialis, pronator teres, pronator quadratus, supinator
6Y, male
(1927280)
LOM on pronation & supination
Treatment

usually not necessary if patient has mild unilateral deformity or no major functional loss
Procedure to restore motion

1. Resection of synostotic radius
2. Division of interosseous membrane
3. Synostotic resection + interposition of fat, fascia, silicone
4. Separation + vascularized fat-fascio graft + index procedure (osteotomy of radius)

“Not justified in textbook until now”
Treatment

The only useful surgery

osteotomy though the site of the synostosis
Indications for surgery

- Bilateral involvement
- Severe pronation (more than 60)
Complication

Compartment syndrome

- 1/3
- common in osteotomy with greater than 85 degree of correction

Cong.R-U Synostosis
Congenital synostosis of proximal R-U in KMUH (2005)

22 patients (26 elbows)

Bilateral: 5/22 (23%)

Associated anomaly: dislocation of radial head: 3

Treatment

• None had extreme position which needs osteotomy
• No serious discomfort during daily living
• Psychiatric consultation (reassurance)
Congenital Pseudoarthrosis of Clavicle
Etiology

1) Intrinsic: failure of coalescence of two separate center of membranous ossification

2) Extrinsic: compression by subclavian artery
   (근거: right side > left, left side involve시 dextrocardia 동반 되는 경우가 많다)
Clinical finding

• Painless prominence and hypermobile segment of the mid-third of clavicle
• Right side > Both side > Left side (extremely rare)
• DDX
  1) Birth injury: No callus formation, No pain, sclerotic margin
  2) Cleidocranial dysplasia: no involvement of pelvis and skull (hypoplasia of ilium and no ossifical nucleus of pubic body)
Congenital Pseudoarthrosis of Clavicle

At Birth, left side, dextrocardia
Initial dx: birth injury
Congenital Pseudoarthrosis of Clavicle

6 weeks old

Case of Birth injury

Post delivery 2 weeks
4 months old

Negative finding in bone scan
Treatment

• Optimal Op time: 3-6 Years old

• Method:
  resection of pseudoarthrosis + Iliac crest graft + plate & screw fixation

• Not essential for ensuring good shoulder motion
Congenital Clubhand
(congenital radial deficiency)
Clinical feature

- forearm bowed to the radial side with prominent distal radius
- 2/3 of the length of the opposite unaffected extremity
Clinical feature

- **Thumb**: defective or absence
- **Thenar muscle**: defective in proportion to the deficiency of the thumb
- **Flexor**: present but not well differentiated distally
- **Extensor**: deficiency at its origin
- **Neurovascular**: absence of radial A.N
Classification

- **Type 1: short distal radius:** dist. epiphysis is present but suppressed: Tx is confined to the thumb deficiency.

- **Type 2: hypoplastic radius:** dist. & prox. epiphysis is present, but growth is defective in both area; Tx would be directed toward the lengthening of the radius, but not found to be effective.
Classification

- Type 3: partial absence of the radius: supplying support of the hand
- Type 4: total absence of the radius: release of the soft tissue and provide support of the hand: most common

* TAR syndrome:
Congenital Clubhand, Type 1
Congenital Clubhand Type 2
Congenital Clubhand Type 2
Congenital Clubhand type 4

Female, 1 month,
Associated Anomaly

- rarely occur as a isolate (40% : unilateral., 77% : bilateral --associated anomaly)
- Fanconi syndrome (radial defect + severe aplastic anemia)
- TAR syndrome: thrombocytopenia + complete absence of radius
- VATER syndrome (ventricular or vertebral anomaly + T-E fistula + renal anomalies)

VACTERLs Syndrome

(Vertebral+Anus+Cardiac+Tracheal+Esophageal+Renal+Limb+Single umbilical a)
Treatment

from no treatment to aggressive correction
Treatment

main shortcoming
1) lack of wrist stability
2) impaired finger motion
3) short forearm
4) absence and defective thumb
Treatment

1) bone graft
2) fusion of the wrist
3) **Centralization:** Tx of choice
4) lengthening of the radius
5) epiphysiodesis
6) Radialization with tendon transfer
Current Treatment

1. initial treatment at birth: serial casting

   definite surgery : 6 months- 1 years

   1) proper preoperative splinting, stretching and tendon transfer

   2) osteotomy of the ulna : bowing >30

   3) thumb reconstruction
2. 1-3 years old who first present
: external fixator to stretch tight radial structure
(casting will fail in this age group)
Congenital ulnar clubhand (Ulnar dysplasia)
Type 1: Hypoplasia of the ulna + hand malformation
Type 2: partial loss of ulna + anlage + stable elbow, radius bowing
Arthrogryposis
Clinical feature

not specific disorder but a symptom complex associated neurogenic and myopathic

1) multiple symmetric joint contracture
2) atrophied extremities
3) marked limitation of motion
Clinical feature

More severe distally than proximally
Most upper extremity is affected
Clinical feature

Deformity of upper extremity

- shoulder: adducted and Int. rotated
- elbow: fixed in extension: (most limiting deformity)
- forearm: pronated
- wrist: wrist flexion with ulnar deviation
- fingers: gathered together, thumb in palm
Arthrogryposis

Clinical Feature

- Internal rotation & adduction
- Fixed in extension
- Pronated
- Flexion with ulna deviation

Gather together
Thumb in palm

(1824618)
Arthrogryposis
Ultimate goal of Treatment

- Independent upper extremity function for allow for activity of daily living
  1) bring to the hand to mouth for hygiene and feeding
  2) can be use for pushing up to a sitting position
  3) using an ambulatory aid, such as a crutch
Arthrogryposis

Treatment

PT, casting, splinting

Goal: passive elbow flexion by 2 y (6 months) of age

- obtained
  - No active motion until 5 Y (4Y)
    - Triceps lengthening & capsulotomy
  - Tendon transfer (Ticeps to Bicep)
- Not obtained
Treatment

- Timing of treatment: neonatal period
- ROM exercise and stretching followed prolonged splint
- normal intelligence & sensate skin
- marked adaptability and surgery is often unnecessary
Treatment

surgical release

(no response to adequate trial of 6 months to 1 year)
Treatment

Elbow

Dynamic or static splint: until 40-50 flex.
Older 2 years: elastic harness
No passive motion: post. capsulotomy & lengthening of triceps
Good hand and wrist function: Steindler flexoplasty
Treatment

shoulder

• active and passive stretching
• **External rotation osteotomy** at proximal 1/3 (if it can not approximate 90 degree ext. rotation)
• shoulder fusion is not necessary
Treatment

Wrist

- volar capsulotomy & tendon transfer (FCR & FCU to the dorsum of wrist)
- rigid type: prox. row carppectomy with tendon transfer

Finger

- casting & splinting
- surgery seldom improve the function
Congenital Transverse failure of forearm
Congenital Transverse Failure of Forearm (congenital amputation)
Congenital Transverse Failure of Forearm with Congenital Radial head
Subclavian artery supply disruption?
The most common level is proximal forearm
Unilateral
Remarkably few functional deficit
Treatment

**BE or AE type**

* 6 months
  - Prosthetic team
  - Passive hand or mitt
  - When able to sit independently

* 2 to 3 years
  - Active terminal device (TD)

**More proximal or distal failure**

- No prosthesis in Infancy
- Body powered (cable-operated)
- Myoelectric prosthesis

* Krukenberg forearm: rarely indicated
Congenital Elbow Synostosis
Clinical Features

- Humeroradial > Ulnotrochlea
- No forearm motion
  (in position of 60-90 flexion ?)
- Associated upper limb malformation
  (ulnar clubhand, phocomelia)
Treatment

Dysfunctional position: corrective derotational osteotomy

- Excision & motion restoration procedure
- Continuous passive motion device

Not improved results
Humeroradial synostosis
+ Ulnotrachlear synostosis
+ Ulnar clubhand
+ Hand: agenesis of 5th metacarpal bone
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