CASE PRESENTATION

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Overview
• Clinical Review
• Discussion
✓ Classification
✓ Pathogenesis
✓ Epidemiology
✓ Diagnosis
✓ Treatment

Clinical Review

Chief Problem
• This case is a 4 years old girl with esotropia who is referred by her primary care physician. Her parents noted that she seems to adopt a chin down position frequently in order to see.

Clinical Review

Past Ocular History
• Esotropia

Past Medical History
• Conductive hearing loss
• Expressive language delay
• No limb anomaly, heart, and renal disease were ever noticed.
• No trauma history

Family History
• Non-contributory

Clinical Review

Review of System (02/02/2005)
• Weight: 15.2kg (25-50th)
• Height: 97.2cm (10-25th)
• HENNT: normal head shape, equal pupils, neck: supple
• Heart: regular rate and rhythm
• Musculoskeletal: no deformity.

Clinical Review

Visual Findings (03/15/2005)
• Inspection: Chin-down position
• Visual Acuity: OD: 20/50+ OS: 20/50+ (LEA)
• Sensorimotor Evaluation: abduction deficit in both eyes, -3.5 OD, -4 OS no nystagmus,
  In primary gaze: ET 25 Δ Desc and Nosc and increases to ET 40-45 Δ in both L't and R't gaze
  In down gaze: ET 35-40 Δ
  In up gaze: ET 6 Δ
• Sensory testing: attempted but patient is too young to cooperate
**Clinical Review**

- **Visual Findings:**
  - External exam: chin down head position, no obvious ptosis
  - Pupils: round and reactive with no afferent pupillary defect
  - Penlight exam: normal anterior segment
  - Intracocular pressure: within normal limits to palpitation

**Summary**

- 4 years old girl with abduction deficits in both eyes, V pattern esotropia, and chin-down head position. She has equal bilateral visual acuity which is minimal hyperopic.
- Except possible conductive hearing loss and language development delay, she is otherwise healthy with no known family history of ocular disease.

**Assessment / Diagnosis**

**Bilateral Duane Syndrome**

- An unusual congenital form of strabismus, which is characterized by horizontal eye movement limitation, and specifically by globe retraction with palpebral fissure narrowing on attempted adduction.
- Bilateral: 10-20 % in all Duane cases

**Case Discussion**

**Duane Syndrome**

Taipei 101, The highest building in the world, 580m

**Introduction**

- 1879 Hueck, the first case report
- 1905 Alexander Duane, published 54 cases
- 1974 Huber, Classification: Duane type I, II, III (clinical presentation + EMG)

**Unilateral Duane Type 1**

**Limitation of Abduction of Lt eye**

Normal abduction: narrowing of palpebral fissure and retraction of globe during adduction. Limitation of Abduction of Lt eye.

Paradoical innervation of LR shows peak innervation on adduction and defective innervation on attempted abduction. Normal electrical behavior of MR.
**Classification**

<table>
<thead>
<tr>
<th>Type I</th>
<th>Type II</th>
<th>Type III</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Sensorimotor evaluation</strong></td>
<td><strong>Sensorimotor evaluation</strong></td>
<td><strong>Sensorimotor evaluation</strong></td>
</tr>
<tr>
<td>1. Marked limitation of abduction</td>
<td>1. Marked limitation of abduction</td>
<td>1. Marked limitation of abduction</td>
</tr>
<tr>
<td>2. Inability to move the eye normally</td>
<td>2. Normal or slightly limited abduction</td>
<td>2. Increased irritation of both the LR and MR</td>
</tr>
<tr>
<td>3. Retraction of globe and narrowing of the palpebral fissure on attempted adduction</td>
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<td>3. Retraction of globe and narrowing of the palpebral fissure on attempted adduction</td>
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<tr>
<td><strong>EMG Findings</strong></td>
<td><strong>EMG Findings</strong></td>
<td><strong>EMG Findings</strong></td>
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<tr>
<td>Paradoxical innervation of the LR, with peak impulses on abduction and defective impulses on attempted adduction; normal electrical behavior of the MR</td>
<td>The LR showed peak impulses on abduction and second paradoxical on attempted abduction; normal electrical behavior of the MR</td>
<td>Normal innervation of both the LR and MR, insufficiency in primary gaze, abduction, or adduction.</td>
</tr>
<tr>
<td>A: esotropia</td>
<td>B: exotropia</td>
<td>Orthophoria</td>
</tr>
</tbody>
</table>

**Pathogenesis**

- **Embryogenesis**
  - Sporadic DRS (90%), 10-20 times risk for having other congenital malformations (skeletal, auricular, ocular, neural), similar as thalidomide embryopathy -- teratogenic event during 4 to 8 gestational age

- **Hereditary Factors**
  - Familial DRS (10%)
  - AD, isolated or associated with Okihiro syndrome, Holt-Oram syndrome, Rubinstein-Taybi syndrome
  - AR, isolated or associated
  - Polygenic, associated with Wildervanck syndrome

- **Pathogenesis: Genetic View**
  - **Familial Study**
    - Identified chromosome: 2q31, 8q13, 22q11, 4q27,
  - **Identified genes** (2004)
    - SALL4 (20q13): human sal-like gene
    - Mutation identified in Duane-radial ray syndrome (DRRS, Okihiro syndrome), suggest SALL4 plays a critical role in abducens motoneuron development.
### Pathogenesis, in conclusion

**Sporadic:** Teratogenic event during the 4th to 8th weeks of gestational age.

**Familial:** Genetic background, mostly AD, polygenic or incomplete penetrant with variable expression.

**Mechanical Innervational Anomalies**

- Parasympathetic innervation (parasympathetic denervation syndrome)
- Narrowing of palpebral fissure
- Abnormal high or low insertions of MR
- Anomalous innervation of the vertical muscles
- Cocontraction

**Associated Congenital Anomaly**

- Sporadic Duane Syndrome, 30-50%
- Embryogenesis

<table>
<thead>
<tr>
<th>Structure and/or anomaly</th>
<th>Gestational age (wk)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cranial nerves and nuclei (III, IV, VI)</td>
<td>5-8</td>
</tr>
<tr>
<td>Extraocular muscle innervation</td>
<td>6-6</td>
</tr>
<tr>
<td>Pinna and external auditory canal</td>
<td>6-8</td>
</tr>
<tr>
<td>Auditory ossicles and semicircular canals</td>
<td>6-9</td>
</tr>
<tr>
<td>Palate</td>
<td>7-10</td>
</tr>
<tr>
<td>Oral fissure</td>
<td>7-8</td>
</tr>
<tr>
<td>Vertebrae</td>
<td>4-8</td>
</tr>
<tr>
<td>Upper extremity</td>
<td>3-7</td>
</tr>
<tr>
<td>Enteric neural crest migration</td>
<td>7-12</td>
</tr>
<tr>
<td>Epibulbar dermoids</td>
<td>4-5</td>
</tr>
<tr>
<td>Retinal agenesis</td>
<td>5-6</td>
</tr>
<tr>
<td>Ventricular septal defect</td>
<td>6</td>
</tr>
</tbody>
</table>

**Associated Ocular Congenital Anomaly**

**Frequent Reported**

- Nystagmus
- Epibulbar dermoid
- Anisocoria
- Coloboma
- Ptosis
- Crocodile tears

**Less Frequent Reported**

- Optic nerve hypoplasia
- Cataract
- Anisocoria
- Marcus-Gunn jaw-wrinkling syndrome

### Epidemiology

- Incidence: 1-4% of all strabismus patients
- Sexual distribution: female 60%
- Unilateral: Bilateral = 5:1
- Laterality: in unilateral case: Lt: Rt = 3:1
- Type 1 > Type 3 ≥ Type 2
- 30-50% associated with other congenital anomaly

### Associated Ocular Congenital Anomaly

- **Klippel-Feil Syndrome:** Congenital brachiocephalic, fused cervical vertebrae
- **Goldenhar Syndrome:** Half facial anomaly, partially formed or totally absence of ear, missing eye or normal eye

### Associated non-ocular anomaly

- **Klippel-Feil Syndrome:** Congenital brachiocephalic, fused cervical vertebrae
- **Goldenhar Syndrome:** Half facial anomaly, partially formed or totally absence of ear, missing eye or normal eye
**Diagnosis**

**Modes of testing**

- Observation is the only requirement in making the diagnosis
- Forced Duction or Force Generation Test
- Saccadic Velocity Testing
- Electromyography
- Hess-Lancaster Screen

**Typical findings:**

1. Defects in Abduction and Adduction
2. Esotropia and Exotropia
3. Upshoots and Downshoots
4. Aaaa V patterns
5. Exodeviation and globe retraction
6. Torsional cases
7. Bilateral Duane cases
8. Amblyopia and reduced stereovisuality

**Differential Diagnosis**

- **Abducens Nerve Palsy**
  1. Diplopia (+)
  2. Esotropia without vertical deviation and global retraction

- **Moebius Syndrome**
  Congenital diplopia, sixth and seventh n. palsies
  1. Unilateral or bilateral limitation of abduction, adduction and convergence
  2. Mask-like face
  3. Abs. Abnormalities of the limb, chest, and tongue

- **Congenital Oculomotor Apraxia**
  - Janky headnods
  - Gradual improvement with time

**Treatment Goals of Treatment**

Reducing the face turn, tropia and enlarge the binocular diplopia-free field, increase the rotational movements of the affected eye

- **Positive indications of surgery:**
  1. noticeable horizontal ocular deviation
  2. abnormal head position

- **Relative indications:**
  1. marked retraction of the globe on attempting abduction
  2. ophthalmologically unacceptable downshoot and/or upshoot
  3. relative contraindication:
     - Disrupt binocular development in young age

**Horizontal Surgery**

<table>
<thead>
<tr>
<th>Condition</th>
<th>Procedure</th>
<th>Indication</th>
<th>Expected result</th>
</tr>
</thead>
<tbody>
<tr>
<td>MRs with esotropia</td>
<td>Bilateral MR recession</td>
<td>Example = 15 s</td>
<td>Reduced abnormal head turn 75%</td>
</tr>
<tr>
<td></td>
<td></td>
<td>1. Reduced abnormal head turn 75%</td>
<td></td>
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<td></td>
<td></td>
<td>2. Correct alignment</td>
<td></td>
</tr>
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<td></td>
<td></td>
<td>3. Rarely overcorrected</td>
<td></td>
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<tr>
<td></td>
<td></td>
<td>4. No improvement of abduction beyond the primary position</td>
<td></td>
</tr>
<tr>
<td>MRs with exotropia</td>
<td>Bilateral MR recession</td>
<td>Example = 20 s</td>
<td>Reduced abnormal head turn 75%</td>
</tr>
<tr>
<td></td>
<td></td>
<td>1. Reduced the long-term risk of the development of contraction of the ipsilateral medial rectus muscle by reducing its tonic innervation (Hering Law)</td>
<td></td>
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<tr>
<td></td>
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<td>2. Overcorrection, markedly reduced saccadic velocities in abduction</td>
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</table>

**Vertical Transposition**

**Transposition of the superior and inferior rectus muscles toward the lateral rectus muscle. (partial: longitudinal splitting superior and inferior rectus muscles in half)**

<table>
<thead>
<tr>
<th>Procedure</th>
<th>Advantage</th>
<th>Disadvantage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Complete vertical transposition</td>
<td>1. Increase abduction</td>
<td>1. Sacrificed some degree of abduction</td>
</tr>
<tr>
<td></td>
<td>2. Enlarge the diplopia-free field to 60º</td>
<td>2. Usually full abduction was not obtained</td>
</tr>
<tr>
<td></td>
<td>4. Poor eye alignment</td>
<td>4. Anterior segment ischemia</td>
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<tr>
<td></td>
<td>5. Anterior segment ischemia</td>
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</tr>
</tbody>
</table>
**Posterior fixation Suture & Y-Splitting Procedure**

- Augmentation, combined with MR recession
- Posterior fixation suture (Faden procedure)
  - effectively prevent the slippage of the muscle belly over the globe.
  - Alternative procedure to treat upshoots and downshoots.
- Y-splitting procedure
  - Results in a broad lateral rectus insertion that stabilizes its position.

**Prognosis**

- Not very good
- Major complications (second operation)
  - Undercorrection
  - Overcorrection
  - New vertical deviation
  - Ant. segment ischemia

So, Let it be ~~~

**Summary**

- Clinical presentation: limitation of horizontal movement, palpebral fissure narrowing and global retraction while adduction.
- Epidemiology: Female, Lt eye, unilateral, sporadic
- Associated congenital anomalies: oculocutaneous, neural, auricular, musculoskeletal, renal.
- Pathogenesis: paradoxical innervation and mechanical, CNS anomalies
- Genetic: ch2q, 4q, 8q, 20q (SALL4), 22q.
- Diagnosis: clinical presentation
- Treatment: MR recession or VRT with augmentation

**References**

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**Pathogenesis**

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**Management**
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Orchid Island, Taiwan.
The rich decorated canoes of Yami people.
THANKS FOR YOUR ATTENTION

Beautiful coasts, Taiwan

Mountains and Rivers

Tradition

Kaohsiung Port

Education
National Taiwan University