Overview of Facial Dystonia

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- I do not intend to discuss an unapproved/investigative use of a commercial product/device in my presentation.
Outline

• Define dystonia
• Types of dystonia
  • Eyelid myokymia
  • Benign essential blepharospasm
  • Orofacial dystonia
    – Meige
    – Brueghel
  • Hemifacial spasm
  • Apraxia of lid opening
  • Aberrant regeneration of facial nerve
What is dystonia?

• Involuntary muscle contractions
  • Sustained contraction vs intermittent contraction (tonic vs clonic)
  • → abnormal postures or repetitive movements

• Can affect 1 muscle, groups of muscles, or muscles throughout the body

• Thought to be caused by damage or abnormalities in basal ganglia

• Can be idiopathic, genetic, or acquired
Facial muscle anatomy

https://clinicalgate.com/eyelid-anatomy-and-function/

https://crystal-touch.nl/our-facial-muscles-and-their-functions/
Temporal branch
Zygomatic branch
Buccal branch
Mandibular branch
Cervical branch
Motor root of facial nerve
Types of facial dystonia

- Eyelid myokymia
- Blepharospasm
- Orofacial dystonia
  - Meige
  - Brueghel
- Hemifacial spasm
- Apraxia of eyelid opening
- Aberrant regeneration of facial nerve
Eyelid myokymia

- Fascicular contractions within orbicularis muscle
  - Twitching, flickering, rippling movements
- Usually affects one side
- Associated with stress, fatigue, caffeine
  - “Medical student’s disease”
- Treatment: avoid triggers, usually self-resolves
- Very rarely is the first manifestation of BEB or HFS
Benign Essential Blepharospasm

- Involuntary episodic closure of eyelids
  - Contractions of eyelid protractors
  - Tonic – eye is closed for prolonged time
  - Clonic – multiple contractions
- Ranges from mildly increased blink rate to forceful contractions
- Eyelid involvement ONLY
- BOTH sides affected
Blepharospasm

- Population
  - Ages 40-60 years
  - Female > male (2-4:1)
  - Prevalence of 1.6-30/100,000

- 2,000 cases diagnosed annually in the US
Blepharospasm

- Can be isolated or caused by other conditions
- Cause: defective circuit

https://www.hindawi.com/journals/prm/2021/6664736/
https://en.wikipedia.org/wiki/Basal_ganglia
Blepharospasm

- Treatment:
  - Artificial tears, lid scrubs, tinted lenses
  - Botulinum toxin injections
  - Consider oral medications
  - Consider excision of eyelid muscles (myectomy)
Meige Syndrome

- Blepharospasm + oromandibular dystonia (lower facial spasm)
- BOTH sides affected

https://bjo.bmj.com/content/81/6/439
Meige Syndrome

- **Population**
  - 30-70 years old, mean age 55 years old
  - Female > males (2:1)
  - 5/100,000

- **Can start as blepharospasm and then spread to involve lower face**
  - Spread is more common early in disease
Meige syndrome

- Cause: dopaminergic and cholinergic hyperactivity
  - Decreased functioning of inhibitory neurons
  - Abnormal sensorimotor processing
  - Abnormal control of CN nuclei in brainstem by basal ganglia
- Can be primary or secondary
Meige Syndrome

- **Treatment:**
  - “Sensory tricks”
  - 1\(^{st}\) line systemic medication
  - 2\(^{nd}\) line botulinum toxin
    - Reserved for blepharospasm component
  - Deep brain stimulation
Brueghel Syndrome

- Jaw opening dystonia
- +/- eyelid involvement

https://jnnp.bmj.com/content/jnnp/39/12/1204.full.pdf
https://n.neurology.org/content/46/6/1767
Hemifacial Spasm

- Involuntary upper and lower facial contractions
- Usually affects one side only
- Population
  - Average age 50-60 years old
  - Women > men (2:1)
  - 8-15/100,000 in US

https://www.swedish.org/services/neuroscience-institute/our-services/cerebrovascular-center/conditions-we-treat/hemifacial-spasm
Hemifacial Spasm

• Causes
  • Compression of facial nerve
    – Most common = abnormal artery compressing at exit point
  • Idiopathic
Hemifacial Spasm

• **Treatment:**
  - Botulinum toxin injections
  - Neurovascular decompression surgery (Janetta procedure)

https://danishbhatti.com/review-of-hemifacial-spasm-differentials/
Blepharospasm  Orofacial spasm  Hemifacial spasm

Apraxia of Eyelid Opening

- Inability to open eyes voluntarily after closing eyelids, in absence of squeezing
  - BOTH sides affected
- Commonly associated with blepharospasm but can be isolated
- Cause: poorly understood
- Treatment: botulinum toxin
  - Also: myectomy, eyelid crutches, ptosis surgery, frontalis sling
Apraxia of Eyelid Opening

Apraxia of lid opening in blepharospasm

D R Jordan, R L Anderson, K B Digre

Abstract

Apraxia of lid opening is a nonparalytic motor abnormality characterized by difficulty in initiating the act of lid elevation. It has been reported with extrapyramidal disorders, including Parkinson's disease, Huntington's chorea, progressive supranuclear palsy, and Shy-Drager syndrome. We found seven cases (7%) of functionally disabling apraxia of lid opening in 100 consecutive blepharospasm patients studied. It is important for physicians treating blepharospasm to be aware of the association between these two visually debilitating disorders.
Aberrant regeneration

• Involuntary facial movements accompanying voluntary movements

• Cause: nerve regeneration after CN VII palsy resulting in abnormal connections between peripheral branches of facial nerve

• Types
  • Ocular-oral synkinesis
  • Crocodile tears
  • Hypertonicity
<table>
<thead>
<tr>
<th></th>
<th>Blepharospasm</th>
<th>Orofacial spasm</th>
<th>Hemifacial spasm</th>
<th>Apraxia of eyelid opening</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Affected sites</strong></td>
<td>Eyelids</td>
<td>Eyelids Lower face</td>
<td>Eyelids Lower face</td>
<td>Eyelids</td>
</tr>
<tr>
<td><strong>Laterality</strong></td>
<td>Both sides</td>
<td>Both sides</td>
<td>One side</td>
<td>Both sides</td>
</tr>
<tr>
<td><strong>Cause</strong></td>
<td>Unknown</td>
<td>Unknown</td>
<td>Compression of CN VII(vessel) Can be idiopathic</td>
<td>Unknown</td>
</tr>
<tr>
<td><strong>Treatment</strong></td>
<td>Botox Myectomy</td>
<td>Oral medications Botox</td>
<td>Botox Decompression of CN VII</td>
<td>Botox Myectomy Ptosis surgery Frontalis sling</td>
</tr>
</tbody>
</table>
References

WillsEye Hospital

www.willseyeonline.org