Dystonia

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Introduction

• Syndrome of sustained muscle contraction-
  Repetitive movements or postures

• Due to co-contraction of antagonistic muscles
Types of Dystonia

- Focal dystonia ➔ one body part
- Segmental dystonia ➔ two or more body adjacent body part
Classification of Dystonia

Primary Dystonia
- Primary Dystonia (childhood onset generalized primary dystonia)
- Sporadic Dystonia (Adult onset primary focal dystonia)
- Dopa responsive Dystonia
- Heredodegenerative dystonia
  - Wilsons disease (AR)
  - Huntington's disease (AD)
  - SCAs (AD)
  - Lubag (X linked dystonia parkinsonism)
  - Rapid Onset dystonia parkinsonism
  - NBIA (PKAN) (AR)
  - Neuroacanthosis
- Degenerative syndromes
  - MSA
  - PSP
  - CBD

Secondary Dystonia
- Perinatal trauma/hypoxia
- Stroke
- Focal brain lesions (Putamen)
- Drug induce dystonia's
  - Acute Drug induced dystonia
  - Tardive Dystonia
  - Tardive dyskinesia
Evaluation

• Adequate history
  • Age at onset
  • Course of illness
  • History of psychiatric illness
  • Neuroleptic drug use

• Physical examination
  • Focus and identify the abnormal movement i.e. pattern recognition- challenging even to experienced neurologist
Investigations

• Exclude Wilson's disease
  – Serum Cu
  – 24hr urinary copper
  – Slit lamp examination for KF rings
• Check DYT1 gene <25 years
• Other genetic tests
  • HD
  • SCAs
• MRI of Brian
• Fresh blood film for acanthocytes
Acanthocytes

An abnormal red blood cell that has thorny projections of protoplasm
Clinical manifestations of Adult onset focal and Segmental Dystonia

• Blepharospasms / Hemifacial Spasms
• Oromandibular dystonia
• Lingual dystonia
• Meige syndrome (Blepharospasms + oromandibular dystonias)
• Cervical Dystonia
• Spasmodic dysphonia
• Task specific dystonias (writers cramps)
PRIMARY
DYSTONIA (Oppenheim's Dystonia)

DYT1 gene
Affects 1/3000
Ashkenazi Jews
AD with low penetrance

• Childhood onset
• Starts from the foot and has a variable spread to segmental or generalized
• cranio cervical usually spared
• Action induced dystonia

• DNA testing is available—usefulness in prenatal or presymptomatic diagnosis

• Treatment
  • Trial of dopamine in absence of DNA
  • High dose anticholinergic
    • Baclofen
  • Benzodiazepines
  • Dopa depleting drugs
Dopa- responsive dystonia (DRD) - Segawa’s disease

- AD with incomplete penetrance
- Mutations in GTP hydroxylase gene (DYT 5)
- AR forms exist where the mutations are found in Tyrosine Hydroxylase gene

- Girls > Boys
- Lower limb onset – action induced generalized dystonia
- **Diurnal variation of symptoms - Almost normal in the morning and deteriorate throughout day**
- Mild parkinsonism
- Para paresis in some presentation
- Some cases similar to CP

- L-DOPA up to 275 mg tid
  - No Dopa dose fluctuations
Pantothenate kinase- associated neurodegeneration (PKAN)

Neurodegeneration with Brian Iron accumulation
Formerly *Halloverdin Spatz* syndrome
  - Dystonia
  - Optic atrophy
  - Dementia
  - Retinitis pigmentosa
  - Parkinsonism
  - Death

- AR condition/ childhood onset
- Atypical forms occur in adulthood but no EOT sign on MRI
- PANK2 gene mutations- pantothenate kinase an important enzyme in Vit B5(pantothenate) phosphorylation
- HARP(Hypobetalipoprotienemia, acanthocytosis, retinitis pigmentosa and pallidal degeneration) syndrome linked to PANK2 gene mutations
Adult Onset focal and Segmental Dystonia
Task Specific Dystonia

Writers Cramps 1

Flexion Type
Task specific Dystonia

Writer's cramps 2
Extension Type
Sporadic Dystonia

Adult onset

Cervical Dystonia
Sporadic Dystonia
Adult onset 2
Meigs Syndrome
Sporadic Dystonia
Adult onset 3
Hemifacial Spasms
Acute Drug Induced Dystonia

Metoclopramide

Occurs 1-3 days of use of drugs
Abnormal postures, neck dystonia, tongue or jaw postures

Treat with IV/IMCogentin 2mg or Benadryl 50mg
**Tardive Dystonia**

- Truncal dystonia is most typical form
- Focal and segmental dystonia—bleparospasms, cervical dystonia and oromandibular occur and extremities can be involved
- M>F (younger age)
- Seen after **chronic** use of dopamine blocking agents (median duration is 5.1 years)

**Treatment**

- Gradually withdraw the neuroleptic
- Substitute atypical antipsychotics
- Serpasil
- Valproate
- Clonazepam
- Baclofen
- Diazepam
- Vitamin E
- BoNT

**PISA syndrome**
Drug Induced
Tardive Dystonia (Amodiaquine chronic use)
Tardive Dyskinesia

• Stereotyped movements often involving the facial and oral muscles manifest as tongue protrusion, chewing, lip smacking, grimacing
• Trunk and extremities can be involved

• Seen after chronic use of dopamine blocking agents (6 weeks)

• Treatment
  – Gradually withdraw the neuroleptic
  – Serpasil
  – Valproate
  – Clonazepam
  – Baclofen
  – Diazepam
  – Vitamin E
• **Post Traumatic Dystonia** *(Children or adolescence surviving head injury)*
  
  – Hemidystonia
  – Refractory to medical treatment
  – BoNT effective
  – DBS/Thalamotomy
• **Paroxysmal Kinesiogenic Dystonia (DYT 11)**
  - Childhood onset
  - Episodic dystonia induced by rapid movement to an unexpected stimulus, spells decrease in adulthood
  - Last <1 minute occurring >100 times a day
  - Responds well to anticonvulsants

• **Paroxysmal Non-Kinesiogenic Dystonia (DYT 8)**
  - Infancy onset
  - Episodic, last longer (10 mins) and less frequent
  - Episodes precipitated by Stress, Caffeine and ethanol
  - Does not respond to anticonvulsants – benzodiazepines, neuroleptics, anticholinergic
Management of Dystonia

• Consider trial of Dopa in DRD - <40 years, child or adolescent for DRD

• Anticholinergic drugs
  • Benzhexol up to 80mg/day
  • Use other if not helpful
    – Tetrabenazine
    – Pimozide
    – Sulpiride

• BoNT- Focal dystonias

• Thalamic DBS may be an option
BoNT

• For most patients BoNT is the agent of first choice for Dystonia
• For early onset dystonias(<20) drug therapy can be tried
  • Dopa for 3/12-non response rules out DRD
  • Trihexyphenidyl
  • Benzodiazepines
    – Clonazepam
• Physiotherapy
BoNT

• Several preparations available
  – Botox (Type A)
  – Dysport (Type A)
  – Xeomin (Type A)
  – Neurobloc (Type B)

  – Wide therapeutic range
  – EMG guided techniques.
Syringes and needles
• Reconstitute with 1ml N/S for Deep IM injections
• Reconstitute with 2.5ml N/S for facial injections
Conclusion

• Pattern recognition is the key
• Establish age at onset, family history drugs and psychiatric history
• *It is challenging even to experienced neurologist*
• Use Dopamine in all childhood onset dystonias for up to 3 months if DNA not available- DRD
Thank you