An approach to hyperkinetic movement disorders:

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Phenomenological Classification of Movement Disorders

• Movement Disorders are classified broadly into two main groups:

**HYPOKINETIC DISORDERS**: too little movement
bradykinesia (slowness of movements)
(Parkinson’s Disease and other akinetic rigid syndromes)

**HYPERKINETIC DISORDERS**: too much movement
dyskinesias- (different types of involuntary movements)
The Tricky World of Hyperkinetic Disorders

- Five main types:
  - Tremor
  - Tics
  - Chorea
  - Myoclonus
  - Dystonia

3 types of jerks
History and examination in a movement disorder case

Three points to stress in history:

• **Birth history** - anoxia, peri-natal problems, milestones (delayed onset movement disorders)

• **Family history** - positive, negative and “absent” family history (many movement disorders conditions are inherited)

• **Drug and toxin history**: neuroleptics, antiepileptics, illegal substances, toxins (many movement disorders are drug related)
Special points in examination of a movement disorder case

• Cognition/speech
• **Eye movements**- saccades and pursuit
  - Vertical gaze palsy and slow saccades in Progressive supranuclear palsy in a parkinsonian patient
  - Difficulty initiating saccades in Huntington’s disease
• **Gait**: Arm swing, stride-length, freezing, postural reflexes
• **Bradykinesia**: Repeated finger tapping- decrement and fatiguing, foot tapping
History and examination in a movement disorder case

• Level 1: **Phenomenology**- What is the main category of movement disorder?- Hypokineti
  vs Hyperkinetic – (could be mixed- but make up your mind to wear one pair of spectacles )

• Level 2: **Distribution** of movement disorder, associated signs or features including history (age
  etc.) to help consider etiology (DEFINE THE SYNDROME!)

• Level 3: **Investigations** keeping in mind history and signs and the syndrome to arrive at diagnosis
Step 1 - hypokinetic;
Step 2 - decreased left arm swing and slow asymmetric parkinsonian syndrome- no other signs;
Step 3 – he is young for PD – investigate for ‘young onset parkinsonian syndrome’s - found to have parkin gene mutations
Hyperkinetic Disorders

- Five main types:
  - Tremor
  - Tics
  - Chorea
  - Myoclonus
  - Dystonia

Decide which group does the patient best fit
Tremor

• **Definition**: Rhythmic oscillation of a body part.
Consensus Statement on the Classification of Tremors, From the Task Force on Tremor of the International Parkinson and Movement Disorder Society

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Tremor: - 3 aspects to pay attention to

- **Phenomenologically** (when does it appear?)
  - *Rest*: occurs when affected body part is at rest
  - *Postural*: occurs when arms are outstretched
  - *Kinetic*: occurs during movement of body part
    (intention tremor: exacerbation of kinetic tremor
     towards the end of a goal directed movement)
  - *Task or position specific*: on certain tasks e.g.
    writing, orthostatic- on standing
Tremor - 2nd aspect

Anatomic distribution (which body parts are predominantly affected):

• Limb- arms / legs; unilateral or bilateral; symmetric or asymmetric
• Head
• Tongue
• Trunk
3rd Aspect: Pay attention particularly for any associated features particularly look for these 4 associated signs if present

- Dystonia
- Cerebellar
- Parkinsonism- look for bradykinesia
- Reflexes- & sensory
Why do we pay attention to when the tremor occurs

**Resting tremor:**
- Parkinson’s disease and other parkinsonian disorders, dystonic tremor, one component of rubral tremor, severe ET

**Postural:**
- Essential tremor, Physiological
- PD, Dystonic tremor etc.

**Kinetic:**
- Cerebellar disorders
Why do we pay attention to the associated features

- The concept of “Isolated” versus “Combined” tremor
Axis 1: clinical features

- historical features
  - age at onset
  - temporal onset and evolution
  - past medical history
  - family history
  - alcohol and drug sensitivity

- tremor characteristics
  - body distribution
  - activation conditions
  - tremor frequency

- associated signs
  - signs of systemic illness
  - neurologic signs
  - soft signs

- additional laboratory tests
  - electrophysiological tests
  - structural imaging
  - receptor imaging
  - serum and tissue biomarkers
FIG. 1. (A) Axis 1 classification of tremor is based on clinical features from the patient's medical history and physical examination. Additional tests are sometimes useful. (B) Axis 2 classification is etiology. A syndrome in Axis 1 may have multiple etiologies, and a particular etiology may produce multiple syndromes.
The concept of “isolated” and “combined” tremor

In addition to characterizing tremor, the physical exam is devoted to the identification of associated or concomitant signs that may aid in clinical diagnosis. We propose two broad categories of tremor in Axis 1: *isolated tremor* in which tremor is the only abnormal sign and *combined tremor* in which other abnormal signs are present. Combined tremor may occur with other *neurological signs* (e.g., dystonic postures, rigidity, bradykinesia, or myoclonus) or with relevant *systemic signs* (e.g., Kayser-Fleischer ring, hepatosplenomegaly, or exophthalmos).
FIG. 3. Axis 1 tremor syndromes. Tremor syndromes are listed in this figure according to the predominant presenting symptoms.
Essential tremor
1) isolated tremor syndrome of bilateral upper limb action tremor
2) at least 3 years’ duration
3) with or without tremor in other locations (e.g., head, voice, or lower limbs)
4) absence of other neurological signs, such as dystonia, ataxia, or parkinsonism.

It is important to emphasize that the definition of ET in Axis 1 allows for the existence of multiple etiologies for this common syndrome. Patients frequently have a family history, and small doses of alcohol may improve the tremor. However, these clinical features are not consistent enough to be included in the definition of ET. It was discussed to include onset of tremor in the upper limbs as a further criterion, but there are no convincing data that support this criterion. Some studies have included patients with neurological signs of uncertain relationship to tremor (i.e., “soft neurological signs”), such as mild memory impairment, impaired tandem gait, and subtle body posturing that could be dystonic. There is no consensus on which of these additional signs are acceptable within the definition.

Essential tremor plus: Tremor with the characteristics of ET and additional neurological signs of uncertain significance such as impaired tandem gait, questionable dystonic posturing, memory impairment, or other mild neurologic signs of unknown significance that do not suffice to make an additional syndrome classification or diagnosis. ET with tremor at rest should be classified here.

The ET plus syndrome does not include other clearly defined syndromes like dystonic tremor and task-specific tremor.

Exclusion criteria for ET and ET plus
• Isolated focal tremors (voice, head)
• Orthostatic tremor with a frequency >12 Hz
• Task- and position-specific tremors
• Sudden onset and step-wise deterioration
ET is a “syndrome” due to different etiologies.

- There is genetic heterogeneity
- Many cases appear to be sporadic.
- It is an early phenotype of hereditary dystonia (eg, ANO3), hereditary ataxia (eg, SCA12), and Parkinson disease.

Deuschl et al. *Mov Disord* 2015; 30: 1327-34
Stamelou et al. *Mov Disord* 2014; 29: 928-934
Examples of Axis 1 classification

Isolated tremor syndromes

- Essential tremor
- Task-specific tremor
- Primary orthostatic tremor

Combined tremor syndromes

- Dystonic tremor
- Rest tremor
- Bradykinesia
- Rigidity
- Tremor with ataxia
Rest Tremor

• Typical feature of Parkinson’s disease, and sometimes in other parkinsonian syndromes.
• Parkinson’s disease: classically a “pill-rolling” rest tremor, frequency 3-6 Hz.
• Variable, distractable
• May appear on actions like walking or distraction
• Resting tremors can be seen in other conditions
Conditions with Rest tremor

- PD - classic pill rolling
- Atypical parkinsonism - (MSA more than PSP or CBD, but generally not typical pill rolling)
- Neuroleptic drug induced
- Dystonia / SWEDDS
- Rubral tremor
- Fragile X
- Neuropathic tremor
PD resting tremor examples

Arm tremor

Legs

Jaw
DAT SPECT scan - normal
Patients With Adult-Onset Dystonic Tremor Resembling Parkinsonian Tremor Have Scans Without Evidence of Dopaminergic Deficit (SWEDDs)

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Mov Disord 2007
Recognise particular tremor syndromes
Rubral tremor
Palatal tremor syndromes - again check for isolated or combined

Progressive ataxia and palatal tremor - PAPT
Orthostatic tremor syndromes
Diagnostic criteria of OT

- A subjective feeling of unsteadiness during stance, which increases while standing, relieved by walking and sitting down;
- Presence of 13-18 Hz tremor on surface EMG recordings. All leg, trunk and arm muscles can show this tremor that is typically absent during tonic activation, while the patient is sitting or lying down.

Consensus statement of the MDS (Deuschl et al, Mov Disord 1998)
Chorea

• **Definition**: Irregular, brief, purposeless movements that flit from one body part to another
Particular things to differentiate chorea from tics/myoclonus

• Movements are unpredictable (Tics are stereotyped- even if multiple)

• Look out for the “quasi –purposive” action within the chorea- adjusting the spectacles, adjusting their belt, picking up the trouser, etc- this is not present in myoclonus or tics

• Always take the shoes and socks off and look for chorea in the toes- tics and myoclonus are very uncommon in feet.
Chorea

- Many causes: Acquired and inherited
  - Drugs/ Oral contraceptives
  - Basal ganglia lesions
  - Sydenham’s chorea
  - Antiphospholipid antibody syndrome
  - Huntington’s disease/ HD like diseases
  - Neuroacanthocytosis
Chorea

• 5 main considerations with regard forming the syndrome
  - Age of onset
  - Type of onset e.g. acute/subacute/ or chronic and worsening
  - Distribution of chorea
  - Other clinical features
  - Family history/drug history
Age of onset is crucial
Movement disorder onset age – since birth

Benign Hereditary Chorea - TTFI (thyroid transcription factor 1 gene) mutation
Particular distribution and type of onset is crucial
Making syndromes- additional features are crucial

- Chorea with dementia:
  HD, Prion, C9ORF72, others
- Chorea with eye movement abnormality:
  HD, ataxia telangiectasia,
- Chorea with peripheral neuropathy
  Neuroacanthocytosis, etc
- Chorea with ataxia
  NA, DRPLA, SCA’s
Tics

• Brief, repetitive and stereotyped movements or vocalisations.

• Tics are usually suppressible for a short period of time, but at the expense of mounting inner tension.

• Very common: 3-4% of the population are affected at some time in their lives, almost always starting in childhood.
Tics

• **Motor:**
  - eye blinking
  - head jerks
  - arm/leg jerks
  - complex sequence

• **Vocal:**
  - sniffing
  - grunting
  - snorting

When pointed out if they perceive the movements patients will often say its happening in response to a particular sensation of discomfort they feel in that body part.
Motor tics: Tics are a caricature of normal movements
DSM-IV TIC DISORDERS
(typical tic disorders begin before age 18)

Motor and/or phonic tics occurring many times a day, nearly every day for ≥ 4 weeks, but < 12 months

TRANSENT TIC DISORDER

• Motor or phonic tics occurring many times a day, nearly every day, or intermittently for ≥ 1 year
• Tic-free interval < 3 months

CHRONIC TIC DISORDER

• Motor and phonic tics occurring many times a day, nearly every day, or intermittently for ≥ 1 year
• Tic-free interval < 3 months

TOURETTE’S SYNDROME
Tics and coprolalia and copropraxia
TOURETTE ‘SPECTRUM’

- ADHD
- Obsessive-compulsive disorder
- Anxiety disorders
- Depression
- Echophenomena
- Coprophenomena
- Paliphenomena
- Self-injurious behaviour
- Simple tics
- Complex tics
Late onset vocalisation (or tics) think secondary causes
Secondary causes

• Excitotoxic drugs- amphetamines, cocaine, ecstasy etc.
• Degenerative conditions- Neuroacanthocytosis, HD, DRPLA etc.
• Fragile X
• New associations of later onset tic disorders- Idiopathic cervical/segmental dystonia with tics; Paroxysmal Kinesigenic Dyskinesia with tics
Myoclonus

• **Definition:** Brief shock-like jerks.

Many causes –

- Physiological,
- Fragment of epilepsy
- Metabolic encephalopathies/ Hypoxia
- Progressive myoclonic ataxia/epilepsy
- SSPE/CJD/other encephalitides
Symptomatic myoclonus

- Storage disease
- Progressive Myoclonic ataxia (PMA) causes
- Infectious or postinfectious- CJD, SSPE
- Hypoxic
- Metabolic
- Toxic and drug induced
- Malabsorption- coeliac disease
- Paraneoplastic
### Helpful Generalisations

<table>
<thead>
<tr>
<th>Clinical</th>
<th>Cortical</th>
<th>Basal ganglia</th>
<th>Brainstem</th>
<th>Spinal</th>
</tr>
</thead>
<tbody>
<tr>
<td>Focal (EPC) Multifocal</td>
<td>Neck &amp; arms Dystonic posture Alcohol response</td>
<td>Generalised Prox &gt; distal ++ reflex (esp auditory)</td>
<td>Axial (sparing face + distal) Segmental (often rhythmic) If reflex rarely sound</td>
<td></td>
</tr>
<tr>
<td>Distal &gt; prox +/- epilepsy +/- reflex ++ action</td>
<td>Epsilon-sarcoglycan (DYT 11) Wilson's SSPE</td>
<td>Brainstem pathology GLRA1 mutation (hered hyper-ekplexia)</td>
<td>Denervation Spinal lesions H. zoster/HIV</td>
<td></td>
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</tbody>
</table>

**NB.** Peripheral myoclonus rare. Psychogenic common.
Cortical myoclonus: PMA

Myoclonus dominates with underlying ataxia

Multifocal eg cortical tremor, coeliac disease, AMRF syndrome, some mitochondrial encephalopathies, Unverricht Lundborg, drugs

May not be a correlate on routine EEG

Action jerks, max distally, reflex to touch
Basal ganglia myoclonus: look out for the dystonia and characteristic features as part of the syndrome

- Epsilon-sarcoglycan gene mutations
- Early onset, benign course
- Alcohol benefit
- Arrhythmic jerks face & arms, co-existent dystonia, OCD
- No stimulus sensitivity
- EMG: long duration jerks + dystonia
- Other causes: Wilson’s, BG lesion, etc
Phenomenology and Classification of Dystonia: A Consensus Update

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ABSTRACT: This report describes the consensus outcome of an international panel consisting of investigators with years of experience in this field that reviewed the definition and classification of dystonia. Agreement was obtained based on a consensus development methodology during 3 in-person meetings and manuscript review by mail. Dystonia is defined as a movement disorder characterized by sustained or intermittent muscle contractions causing abnormal, often repetitive, movements, postures, or both. Dystonic movements are typically patterned and twisting, and may be tremulous. Dystonia is often initiated or worsened by voluntary action and associated with overflow muscle activation. Dystonia is classified along 2 axes: clinical characteristics, including age at onset, body distribution, temporal pattern and associated features (additional movement disorders or neurological features); and etiology, which includes nervous system pathology and inheritance. The clinical characteristics fall into several specific dystonia syndromes that help to guide diagnosis and treatment. We provide here a new general definition of dystonia and propose a new classification. We encourage clinicians and researchers to use these innovative definition and classification and test them in the clinical setting on a variety of patients with dystonia. © 2013 Movement Disorder Society

Key Words: dystonia; classification; definition
Dystonia

- Involuntary muscle spasms leading to abnormal posturing of limbs and writhing movements (athetosis) - tremor is part of dystonia
- **Isolated dystonia**: without any structural damage often inherited (called primary or idiopathic in the old classification)
- **Combined dystonia**: Due to variety of environmental or heredodegenerative causes dystonia combined with other signs (referred to as secondary symptomatic dystonia in old classification)
- **Paroxysmal dystonia**: brief episodes of dystonia/dyskinesia
1. **Age at onset**
   - Infancy (birth to 2 years)
   - Childhood (3–12 years)
   - Adolescence (13–20 years)
   - Early adulthood (21–40 years)
   - Late adulthood (>40 years)

2. **Body distribution**
   - Focal (one site, i.e. torticollis)
   - Segmental (contiguous body parts)
   - Generalised
Axis I. Clinical Characteristics

3. Temporal Pattern
   - Persistent
   - Action-specific
   - Diurnal fluctuations
   - Paroxysmal

4. Associated features
   - Isolated dystonia
   - Combined dystonia (myoclonus, parkinsonism, etc.)
   - Occurrence of other neurological or systemic manifestations
Isolated dystonia:
Two common phenotypes depending on age of onset

**Young onset:** (below 28 yrs)
lower limb onset,
spreads,
tends to generalise;
cranial-cervical
less affected/spared
often familial: DYT1 gene +ve

Prevalence: 3/100,000

**Adult onset:**
affects upper body;
focal or segmental;
cranio-cervical most common
(F>M)
mostly sporadic
Non-DYT-1
Prevalence: 8, 33, 58*, and even 732**/100,000
Two main forms of isolated (primary) dystonia depending on age of onset
Generalised DYT1 gene dystonia

Adult onset focal
Dystonia phenomenology: Tremor in dystonia
Ask or pick up the sensory-geste which indicates this is dystonia
Fixed head tilt following neck injury

**Fixed postures are unlikely to be true dystonia**

<table>
<thead>
<tr>
<th>TABLE 2. List of pseudodystonias (imitators of dystonia)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Dystonic (tonic) tics</td>
</tr>
<tr>
<td>Head tilt (vestibulopathy, trochlear nerve palsy)</td>
</tr>
<tr>
<td>Bent spine, camptocormia, scoliosis</td>
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<tr>
<td>Atlanto axial and shoulder subluxation</td>
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<td>Arnold-Chiari malformation</td>
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<tr>
<td>Soft tissue neck mass</td>
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<tr>
<td>Congenital muscular torticollis</td>
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<tr>
<td>Congenital Klippel-Feil syndrome</td>
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<tr>
<td>Satoyoshi syndrome</td>
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<tr>
<td>Dupuytren’s contractures</td>
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<td>Trigger digits</td>
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<tr>
<td>Neuromuscular causes (Isaacs syndrome, etc.)</td>
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<td>Spasms (hypocalcemia, hypomagnesemia, alkalosis)</td>
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<tr>
<td>Orthopedic and rheumatological causes</td>
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<td>Sandifer syndrome</td>
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<td>Deafferentiation (pseudoathetosis)</td>
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Assessment of Patients With Isolated or Combined Dystonia: An Update on Dystonia Syndromes

Victor S. C. Fung, PhD, FRACP,1 H. A. Jinnah, MD, PhD,2 Kailash Bhatia, MD, FRCP,3 and Marie Vidailhet, MD, PhD4

ABSTRACT: The clinical evaluation of a patient with dystonia is a stepwise process, beginning with classification of the phenomenology of the movement disorder(s), then formulation of the dystonia syndrome, which, in turn, leads to a targeted etiological differential diagnosis. In recent years, there have been significant advances in our understanding of the etiological basis of dystonia, aided especially by discoveries in imaging and genetics. In this review, we provide an update on the assessment of a patient with dystonia, including the phenomenology of dystonia and highlighting how to integrate clinical, imaging, blood, and neurophysiological investigations in order to formulate a dystonia syndrome. Evolving or emerging dystonia syndromes are reviewed, and potential etiologies of these as well as established dystonia syndromes listed to guide diagnostic testing. © 2013 Movement Disorder Society

Key Words: diagnosis; phenomenology; etiology; differential diagnosis; secondary dystonia
<table>
<thead>
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</tr>
</thead>
<tbody>
<tr>
<td>Isolated dystonia syndromes that are red flags for the subsequent development of a combined dystonia syndrome or neurodegenerative disease</td>
</tr>
<tr>
<td>Cranial dystonia in young adults and children</td>
</tr>
<tr>
<td>Adult-onset lower limb dystonia</td>
</tr>
<tr>
<td>Adult-onset nontask-specific limb dystonia</td>
</tr>
<tr>
<td>Truncal dystonia</td>
</tr>
<tr>
<td>Adult-onset generalized dystonia</td>
</tr>
<tr>
<td>Hemidystonia</td>
</tr>
<tr>
<td>Combined dystonia</td>
</tr>
<tr>
<td>Dystonia with or without parkinsonism of infantile or childhood onset</td>
</tr>
<tr>
<td>Dystonia with or without parkinsonism of adolescent and young adult onset</td>
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<tr>
<td>Dystonia and parkinsonism in older adults</td>
</tr>
<tr>
<td>Dystonia with spasticity (with or without parkinsonism)</td>
</tr>
<tr>
<td>Dystonia with cerebellar ataxia</td>
</tr>
<tr>
<td>Dystonia with myoclonus</td>
</tr>
<tr>
<td>Dystonia as part of paroxysmal dyskinesia</td>
</tr>
<tr>
<td>Dystonia with chorea</td>
</tr>
<tr>
<td>Dystonia with tics</td>
</tr>
<tr>
<td>Dystonia with other neurological involvement</td>
</tr>
<tr>
<td>Dystonia with deafness</td>
</tr>
<tr>
<td>Dystonia with ophthalmological abnormalities</td>
</tr>
<tr>
<td>Dystonia with peripheral neuropathy</td>
</tr>
<tr>
<td>Dystonia with progressive dementia (see progressive dystonia with normal brain MRI)</td>
</tr>
<tr>
<td>Dystonia with systemic disease</td>
</tr>
<tr>
<td>Dystonia with endocrine abnormalities</td>
</tr>
<tr>
<td>Dystonia with hematological abnormalities</td>
</tr>
<tr>
<td>Dystonia with solid organ involvement</td>
</tr>
<tr>
<td>Syndromes according to brain imaging</td>
</tr>
<tr>
<td>Dystonia with MRI evidence of neuronal brain iron accumulation</td>
</tr>
<tr>
<td>Dystonia with basal ganglia lesions</td>
</tr>
<tr>
<td>Dystonia with leukoencephalopathy</td>
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<tr>
<td>Dystonia with basal ganglia calcification</td>
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<tr>
<td>Progressive dystonia with normal brain MRI or generalized atrophy</td>
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</table>

*See Supporting Tables 1–18 for a list of etiologies of this syndrome. MRI, magnetic resonance imaging.
Isolated young limb onset generalised dystonia
Onset age 55 years- Isolated craniocervical dystonia
Adult-onset foot dystonia
Its about Pattern Recognition
Young onset limb onset generalised dystonia......BUT

Loss of balance, speech affected, spasticity...hence combined dystonia suggestive of a symptomatic dystonia- (glutaric acidemia type 1)
Adult onset oro-mandibular mouth opening  primary dystonia
17 year old /F: 2 year history of mouth opening spasms
Clues Suggesting Symptomatic/ heredodegenerative dystonia etiologies: usually combined dystonia

- Unusual pattern for age of onset
- **Rapid progression or early bulbar/ pronounced oromandibular involvement**
- Fixed rather than mobile spasms
- Hemidystonia
- Sensorineural Deafness
- **Parkinsonism**
- Abnormalities of eye movements, cognitive impairment, optic atrophy or RP, postural loss, pyramidal signs

**Syndromic associations guide investigations needed**

**Brain imaging is most informative**
Further Pattern recognition

- Recognise characteristic patterns or conditions causing particular distribution of dystonia
Schneider et al, Severe Tongue protrusion dystonia Neurology 2006
Prominent oro-mandibular dystonia

- Neuroleptic drugs
- Acanthocytosis
- Nbia’s (Neurodegeneration with brain iron accumulation disorders)
- Wilson’s
- Lesch Nyhan
- Etc.
Dystonic opisthotonus

Dystonic Opisthotonus: A “Red Flag” for Neurodegeneration With Brain Iron Accumulation Syndromes?

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11-2006
Other simple facts and points in a movement disorder patient

- Hemidystonia/Hemicchorea always rule out contralateral structural cause (basal ganglia lesion etc)
- Remember to exclude treatable causes:
  - DRD
  - Wilson’s
Dopa Responsive dystonia

• An inherited condition characterised by early onset dystonia and parkinsonism.
• Responds very well to small doses of levodopa, and response lasts for life.
• Many people with DRD are misdiagnosed as having other conditions e.g cerebral palsy.
• Therefore, levodopa should be considered in all patients with dystonia, particularly those with young onset or with family history.
Some other characteristic hyperkinetic MD
Hand washing
Stereotypies - Rett’s syndrome
Mirror movements
Stereotypy-like movements in NMDA encephalitis
Test cases

• Level 1: **Phenomenology** - What is the main category of movement disorder?

• Level 2: **Distribution** of movement disorder, associated signs or features including history (age etc.) to help consider etiology

• Level 3: **Investigations** keeping in mind history and signs to arrive at diagnosis
Test case:
Neuroleptic induced dystonia
Final point- make your syndrome – don’t jump straight to etiology

• Thank you for your attention