EPIDEMIOLOGY AND CLASSIFICATION OF MOVEMENT DISORDERS

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International Parkinson Disease and Movement Disorders Society (MDS)
To disseminate knowledge and promote research to advance the field of Movement Disorders

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• No fee*
• Specifically designed to enable those on a lower income to join the Society.
• All member benefits *except* the print version of the journal *Movement Disorders*.
• Apply every 3 years to renew membership.

Task Force on Africa

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MDS Outreach Education Programs

**Developing World Education Program (DWEP)**
MDS is committed to supporting quality movement disorders education in underserved areas.

**Ambassador Program**
The Ambassador Program supports travel to underserved areas for education and scientific exchange.

**Visiting Professor Program**
The Visiting Professor Program supports travel to underserved areas for education and academic exchange.

**Virtual Professor Program**
The Virtual Professor Program improves the availability of educational initiatives and training in underserved areas.

http://www.movementdisorders.org/MDS/Education.htm
Asynchronous Consultation for Movement Disorders (ACMD) Project

- Improve patient care in regions with limited access to specialists.
- Provide education on movement disorders for local physicians.
- Bring together doctors across different timezones, with different levels of access to participate.

Contact: mguttman@movementdisorders.ca
Waived dues membership

• **No fee** *

• Specifically designed to enable those on a lower income to join the Society.

• Individuals who reside in countries classified as “low-income”, “lower-middle-income” or “upper-middle-income” economies by the World Bank are eligible. *

• Benefits include all member benefits *except* the print journal *Movement Disorders*.

• Waived Dues Members need to apply every 3 years in order to renew their membership.
Learning Objectives

• To briefly describe the epidemiology of movement disorders from a global and African perspective

• To describe the current classification of movement disorders in general

• To describe the current classification of common movement disorders
Outline

- Definition of movement disorders
- Epidemiology (Global data; Africa data)
  - Parkinson disease
  - Essential tremor
- Classification
  - Broad subtypes
  - Parkinsonism
  - Dystonia
  - Tremor
  - Ataxias
What are movement disorders?

• Previously referred to as ‘extrapyramidal disorders’
  BUT…..
  – basal ganglia is not entirely extrapyramidal: intimate connections with pyramidal tract
  – other ‘extrapyramidal’ non-basal ganglia tracts exist e.g. descending vestibulospinal and reticulospinal pathways
  – some disorders of abnormal movement are not associated with basal ganglia function (e.g. ataxia, myoclonus)
  
• Preferred term is that based on clinical phenomenology rather than anatomic location
• Coined by Stanley Fahn and Lewis P. Rowland in 1968.
Definition

Neurological syndromes in which there is EITHER
an excess of movement
OR
a paucity of voluntary and automatic movements,
unrelated to weakness or spasticity
EPIDEMIOLOGY
Epidemiology: Global

• Common neurological disorders
• Availability of population-based data for all movement disorders is hampered by the broad spectrum of conditions
• Most prevalence data refer to specific movement disorders
• Hospital-based data provide insight into more disabling MDs
<table>
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<tr>
<th>Disorder</th>
<th>Prevalence rate per 100,000 population</th>
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<tr>
<td>RLS</td>
<td>9800 (age 65 – 83)</td>
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<tr>
<td>Essential tremor</td>
<td>415</td>
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<tr>
<td>Parkinson disease</td>
<td>187</td>
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<tr>
<td>Tourette syndrome</td>
<td>29 – 1052/2990</td>
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<tr>
<td>Primary torsion dystonia</td>
<td>33</td>
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<tr>
<td>Hemifacial spasm</td>
<td>7.4 – 14.5</td>
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<td>Blepharospasm</td>
<td>13.3</td>
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<tr>
<td>Hereditary ataxia</td>
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<td>Huntington disease</td>
<td>6</td>
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<tr>
<td>Wilson disease</td>
<td>3</td>
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<tr>
<td>PSP</td>
<td>2 – 6.4</td>
</tr>
<tr>
<td>MSA</td>
<td>4.4</td>
</tr>
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</table>

Fahn S, Jankovic J. Principles and Practice of Movement Disorders, 2007: page 5
Hospital-based frequency of MD

Adapted from Fahn & Jancovic; data from 2 large MD clinics in USA 1996
Hospital-based frequency of MD: Africa

- ~5 – 15% of outpatient neurology consultations (Bower JH et al, Ethiopia, 2005; Tegueu et al, Cameroon, 2013)
- Hypokinesias – 55.4%; Hyperkinesias – 44.6% (Okubadejo et al, 2012)
- Most common – Parkinsonism/Parkinson disease
- Predominant forms: (similar to global MD clinic reports)
  - parkinsonism
  - other tremors
  - dystonia
One of the most frequent (top 10) neurological diseases encountered in primary care globally, including in Africa.

- PD frequency in specialist care: $4^{\text{th}}$ major neurological disease encountered in specialist care (46.2%) overall, 18.8% ($6^{\text{th}}$ in Africa) and 66.7% ($4^{\text{th}}$ in SE Asia).

Burden (population) of PD from 2015 to 2030

- Developing countries: 2.37 mill (39.7%) to 3.08 mill (42.5%)
- Developed countries: 3.60 mill (60.3%) to 4.16 mill (57.5%)
Proportion (%) of PD in emerging and developed countries: 2015 to 2030.
PD epidemiology ii

- Affects ~ 1% population 65 – 85 yrs; 4.3% >85 yrs

- Prevalence rates vary widely: methodological, geographic, genetic, environmental differences

- In metanalysis, steady increased prevalence with age

- Age-standardized DALY rates rank (Global Burden of Disease Study 2015) for all neurologic dx (14 categories)
  - Global: 11th
  - North, central SSA, western SSA: 11th
  - Eastern SSA: 12th
  - Southern SSA: 10th
Global prevalence of PD

Parkinson’s Disease in Africa: A Systematic Review of Epidemiologic and Genetic Studies

Njideka U. Okubadejo, MD, James H. Bower, MD, Walter A. Rocca, MD, MPH, and Demetrius M. Maraganore, MD

Movement Disorders. 2006;21(12):2150 - 2156

FIG. 1. Distribution of epidemiologic and genetic studies of Parkinson’s disease in Africa by country. The number of published reports for each type of study (clinical series, prevalence, incidence, or genetic) is reported in parentheses. Red, East Africa; Blue, West Africa; Green, North Africa; Violet, South Africa. [Color figure can be viewed in the online issue, which is available at www.interscience.wiley.com.]
PD epidemiology iii: incidence

- Based on most recent systematic review and metanalysis (Hirsch L, et al. Neuroepidemiology 2016;46:292 – 300)

- Overall incidence (females >40): 37.55 per 100,000 person-years

- Overall incidence (males > 40): 61.21 per 100,000 person-years

- Increased incidence with age in both sexes
Descriptive study of General Neurology Clinic based PD cases: 1996 – 2006

124 parkinsonism – 98 (79.0% PD) and 26 (21.0% others)

Mean time from onset to diagnosis: **24.6 months** (median 12 mo)

Severity at presentation (Hoehn and Yahr stage 2)
PD: environmental risk factors

- Variable evidence for specific risk factors
- Rural living, agricultural occupation, well water consumption
- Pesticides – paraquat, rotenone, organochlorines, organophosphates, permethrin, benomyl
- Solvents
- Polychlorinated biphenyls
- Mild to moderate head injury
- Toxins: MPTP
- Behavioural risk factors (*reduced risk*): caffeine and cigarette smoking
PD: genetic factors

• Monogenic causes of PD/parkinsonism
  – Classic PD: PARK-SNCA, PARK-LRRK2, PARK-VPS35 (all autosomal dominant)
  – Early onset PD: PARK-parkin, PARK-PINK1, PARK-DJ1 (all autosomal recessive)
  – Parkinsonism: PARK-ATP13A2, PARK-FBX07, PARK-DNAJC6, PARK-SYNJ1

• Genetic risk variants (GWAS): > 26 genetic risk variants consistently associated
ESSENTIAL TREMOR

• Most common tremor
• Worldwide prevalence: 4.9 – 39.2 per 1000
• Highest prevalence >60: 13.0 – 50.5 per 1000

Prevalence in Lagos: 23.8 per 1000 (age-adjusted to WHO New world population)
• Consistent increase in prevalence with advancing age
CLASSIFICATION
Broad subtypes of MD

• Hypokinesias

• Hyperkinesias
Hypokinesias

1. Parkinsonism
(Akinesia/bradykinesia)
2. Apraxia
3. Cataplexy and drop attacks
4. Freezing
5. Rigidity
6. Blocking (holding) tics
7. Catatonia, psychomotor depression, and obsessional slowness
8. Hesitant gaits
9. Hypothyroid slowness
10. Stiff muscles
<table>
<thead>
<tr>
<th>Hyperkinesias</th>
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<tbody>
<tr>
<td>1. Tremors</td>
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<tr>
<td>2. Dystonia</td>
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<tr>
<td>3. Chorea/Athetosis/Ballism</td>
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<tr>
<td>4. Ataxia/dysmetria</td>
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<tr>
<td>5. Myoclonus</td>
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<tr>
<td>6. Hemifacial spasm</td>
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<tr>
<td>7. Tics</td>
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<tr>
<td>8. Hyperekplexia</td>
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<tr>
<td>9. RBD</td>
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<tr>
<td>10. Myokymia and synkinesis</td>
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<tr>
<td>11. Restless legs</td>
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<tr>
<td>12. Moving toes/fingers</td>
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<tr>
<td>13. Paroxysmal dyskinesia</td>
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<td>14. Abdominal dyskinesias</td>
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<tr>
<td>15. Akathisia</td>
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<tr>
<td>16. Hypnogenic dyskinesias</td>
</tr>
<tr>
<td>17. Jumpy stumps</td>
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<tr>
<td>18. Myorhythmia</td>
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<td>19. Stereotypy</td>
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</tbody>
</table>
PARKINSONISM

- Blank facial expression
- Slow, monotonous, slurred speech
- Forward tilt to posture
- Reduced arm swinging
- Rigidity and tremor of extremities and head
- Short, shuffling gait
Parkinsonism: classification

I. Primary (Parkinson disease, idiopathic PK)

II. Atypical parkinsonism: MSA, PSP, CBD, parkinsonism with dementia

III. Heredodegenerative disorders

I. Secondary: infections, toxins, drugs, tumor, trauma, vascular, metabolic
Venn diagram showing the relationships between Parkinson Disease, Secondary Parkinsonisms, Hereditary Parkinsonisms, and Atypical Parkinsonisms.

**Secondary Parkinsonisms**
- Vascular
- Infectious
- Drug/toxins
- Metabolic
- Tumor/trauma
- Normal pressure hydrocephalus

**Hereditary Parkinsonisms**
- PARK gene/loci
- Spinocerebellar ataxias
- Huntington disease
- Lubag disease
- Wilson disease
- Neuronal brain iron accumulation disorders

**Atypical Parkinsonisms**
- Progressive supranuclear palsy
- Corticobasal degeneration
- Multiple system atrophy
- Dementia with Lewy bodies
Classification of PD (motor subtypes)

i. Tremor dominant

ii. Postural impairment and gait disturbance (PIGD)
Multiple system atrophy (subtypes)

i. MSA - cerebellar

ii. MSA – parkinsonian
Progressive supranuclear palsy subtypes/phenotypes

- Classic PSP (Richardson disease)
- PSP – parkinsonism
- PSP – pure akinesia with freezing of gait
- PSP – corticobasal syndrome
- PSP – behavioural variant of frontotemporal dementia
- PSP – cerebellar ataxia
- PSP – primary lateral sclerosis
Corticobasal degeneration (CBD) subtypes/phenotypes

- Corticobasal syndrome (classic CBD)
- CBS – frontal behavioural variant
- CBS – posterior cortical atrophy syndrome
- CBS – progressive non-fluent /agrammatic aphasia
- CBS – PSP syndrome
DYSTONIA
Dystonia – Axis I: Clinical characteristics

• **Age at onset**
  - Infancy (birth – 2 years)
  - Childhood (3 – 12 years)
  - Adolescence (13 – 20 years)
  - Early adulthood (21 – 40 yrs)
  - Late adulthood (> 40 years)

• **Body distribution**
  - Focal
  - Segmental
  - Multifocal
  - Generalized (with or without leg involvement)
  - Hemidystonia

• **Temporal pattern**
  - Disease course: Static or Progressive
  - Variability: persistent, action-specific, diurnal, paroxysmal

• **Coexistence of other movement disorders**
  - Isolated dystonia (±tremor)
  - Combined (with another MD e.g. myoclonus, park)

• **Other neurological manifestations (complex)**

• **Other systemic features**

Movement Disorders, Vol. 28, No. 7, 2013
Dystonia – Axis II: Aetiology

• **Nervous system pathology**
  – Evidence of degeneration
  – Evidence of structural (often static) lesion
  – No evidence of degeneration or structural lesion

• **Inherited or acquired**
  – Inherited: AD, AR, X-linked recessive, Mitochondrial
  – Acquired: Perinatal brain injury, infection, drug, toxic, vascular, neoplastic, brain injury, psychogenic
  – Idiopathic: Sporadic, Familial
TREMOR
Tremor: classification by phenomenology

A) Rest tremor

B) Action tremor

   i.  *Postural tremor*

      i.  Position-independent postural tremor
      ii. Position-dependent postural tremor

   ii.  *Kinetic tremor*

      a) Simple (non – goal directed)
      b) Target-directed (intention)

   iii.  *Task-specific kinetic tremor*

   iv.  *Isometric tremor*
## Tremor: syndromic classification

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<table>
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<tbody>
<tr>
<td>a)</td>
<td>Physiologic tremor</td>
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<tr>
<td>b)</td>
<td>Enhanced physiologic tremor</td>
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<tr>
<td>c)</td>
<td>Essential tremor</td>
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<tr>
<td>d)</td>
<td>Dystonic tremor</td>
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<tr>
<td>e)</td>
<td>Parkinsonian tremor</td>
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<td>f)</td>
<td>Cerebellar tremor</td>
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<td>g)</td>
<td>Holmes’ tremor</td>
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<td>h)</td>
<td>Palatal tremor</td>
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<tr>
<td>i)</td>
<td>Drug/Toxin-induced</td>
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<td>j)</td>
<td>Tremor in periph. neuropathy</td>
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<td>k)</td>
<td>Psychogenic tremor</td>
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<tr>
<td>l)</td>
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ATAXIAS
Classification

• Age of onset

• Time course: *acute, subacute, chronic, episodic*

• Anatomic involvement

• Distribution: *focal or generalized/symmetrical*

• Cause: acquired or inherited or sporadic
Hereditary ataxias

• Autosomal dominant: *Spinocerebellar ataxias (SCAs), DRPLA, episodic ataxias 1 & 2*

• Autosomal recessive: e.g. *Friedreich ataxia, Ataxia telangiectasia*

• X-linked

• Mitochondrial e.g. *MERRF, MELAS*
Systematic Approach to Diagnosis of Movement Disorders

- Presence of one or more movement disorders?
  - Identify all subtypes of movement disorders
    - Define the dominant movement disorder
    - Identify associated neurological features
    - Identify associated non-neurological features
      - Clinically based syndrome
        - Diagnostic work-up
          - Diagnosis
          - Diagnosis

References


• World Health Organization. Neurological disorders: public health challenges


