International Parkinson and Movement Disorder Society

Neurology Department

Overview Of Movement Disorders

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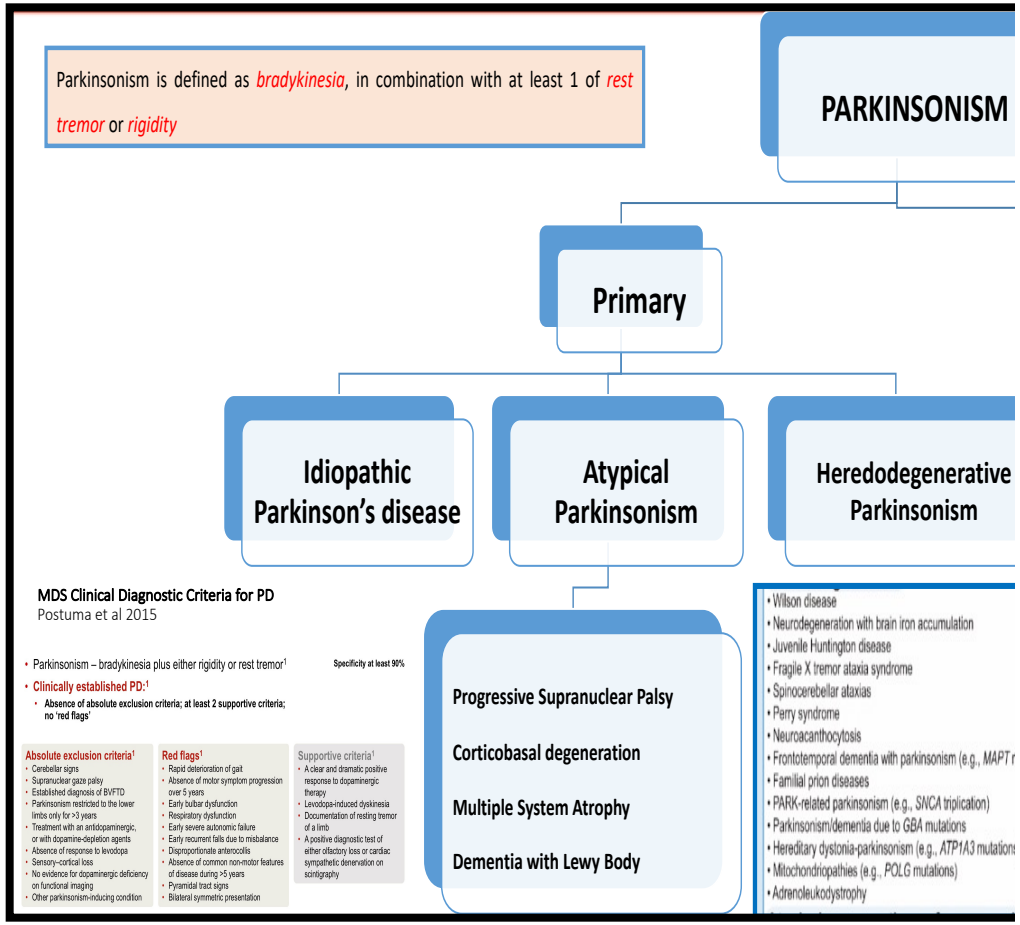
Objectives

1. Identify definitions & clinical phenotypes of MDs.
2. Recognize common types of other MDs.
3. Identify the clinical picture of Parkinson’s disease.
4. Recognize tools of treatment of PD.

What are Movement Disorders?

MOVEMENT DISORDERS

Hypokinetic MD



Hyperkinetic MD

Characteristics of Hyperkinetic MDs

CHARACTER

Dystonia: *sustained* or intermittent muscle contractions causing abnormal- often *repetitive-* movements, *postures*, or both" .

Chorea: *irregular* rapid, low amplitude, *brief* movements of extremities & face

Athetosis: involuntary writhing movements

Hemiballism: *large* amplitude involuntary movement restricted to one side of the body; usually involves proximal upper limb.

Myoclonus: sudden brief *jerk or shock-like* movements

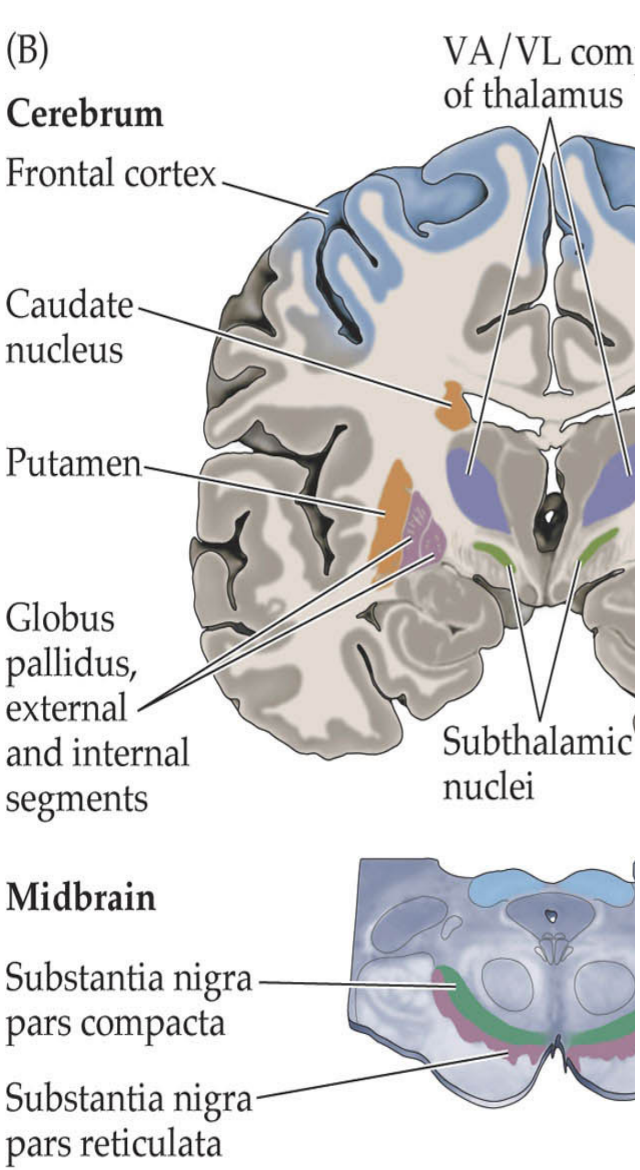
Tremor: *rhythmic oscillation* of a body part due to alternating or synchronous contractions of opposing muscles

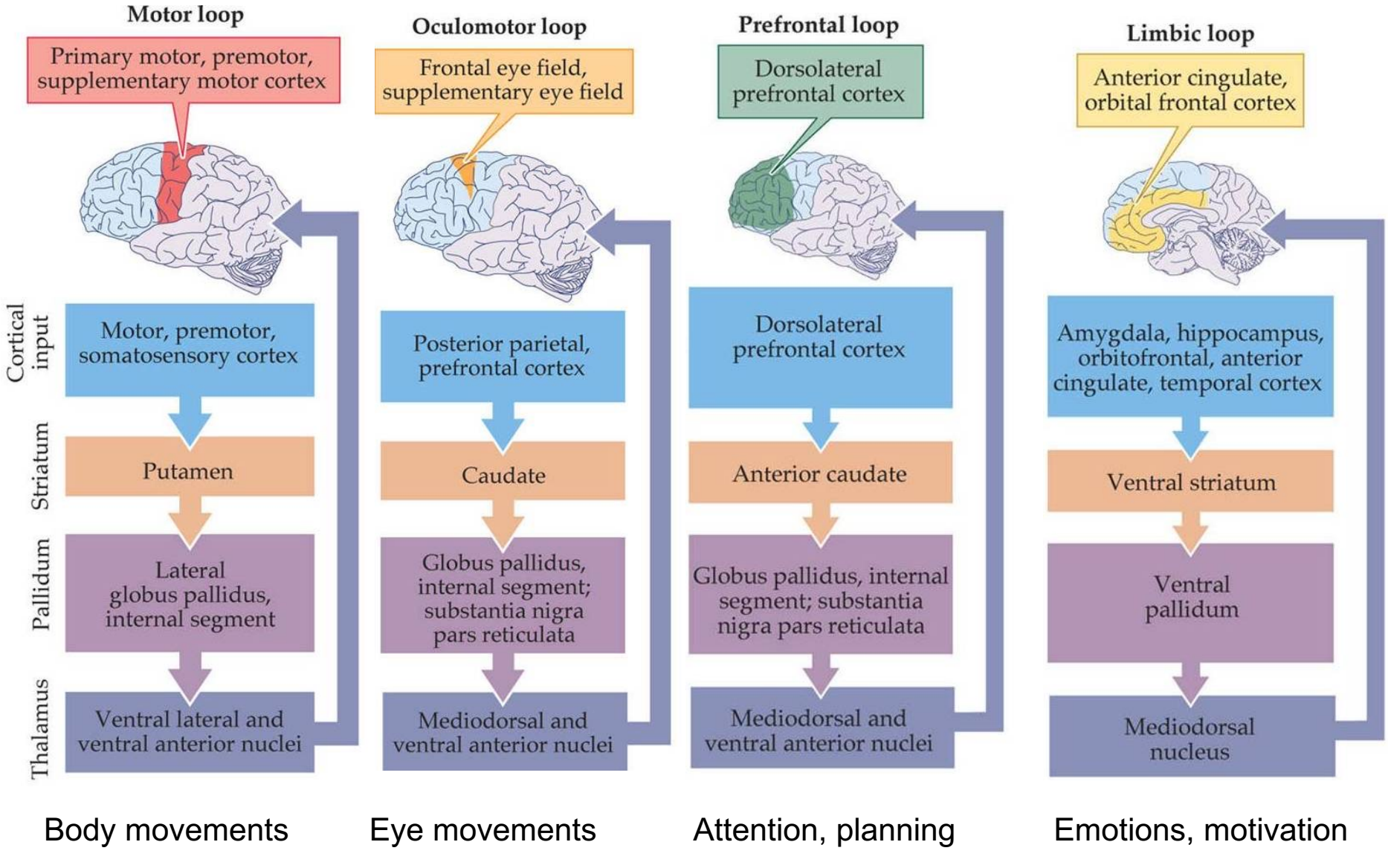
Tics: sudden, brief, purposeless, *stereotyped* simple or complex movements or vocalizations , with urge

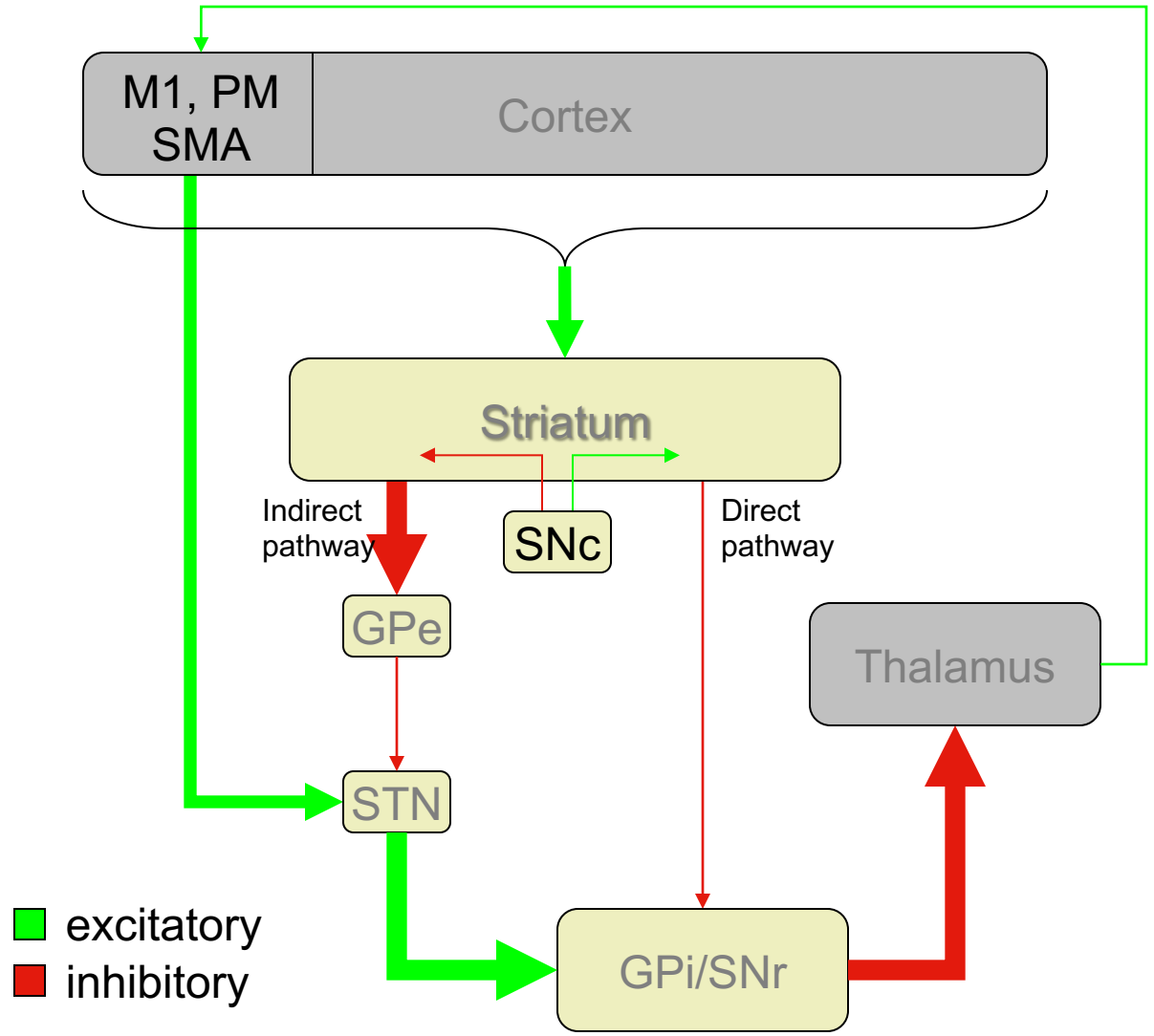
Akathisia: inner *restlessness*; often associated with external signs of restless behavior

Restless legs Syndrome: an *urge to move the legs*, usually accompanied by uncomfortable and unpleasant sensations in the legs, begin or worsen *during periods of rest or inactivity*, partially or totally *relieved by movement*, *worse in the evening*.

Parallel Organization Of Motor & Non-motor Basal Ganglia Loops

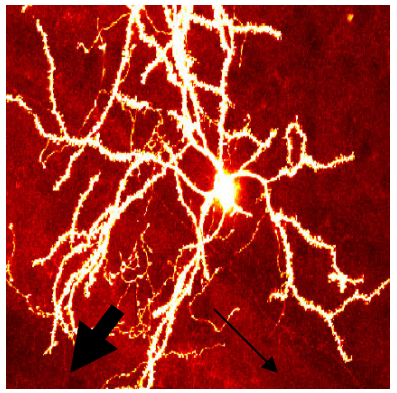






Parkinson's Disease
(Hypokinetic Movement)

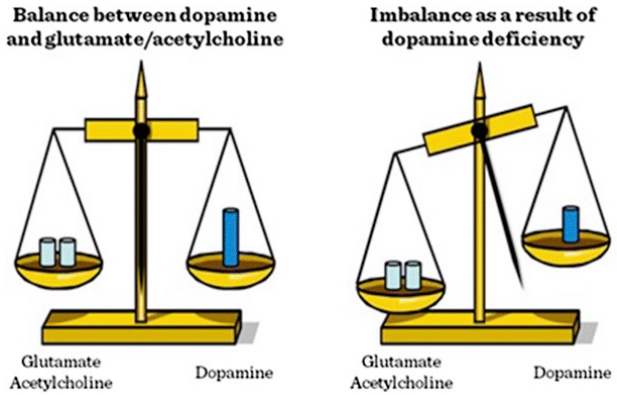
- Decreased output of SNc dopaminergic projections
  - Decrease excitation in direct pathway
  - Increase inhibition in indirect pathway
- Net effect: more inhibition of thalamus and therefore less excitatory input to motor cortex



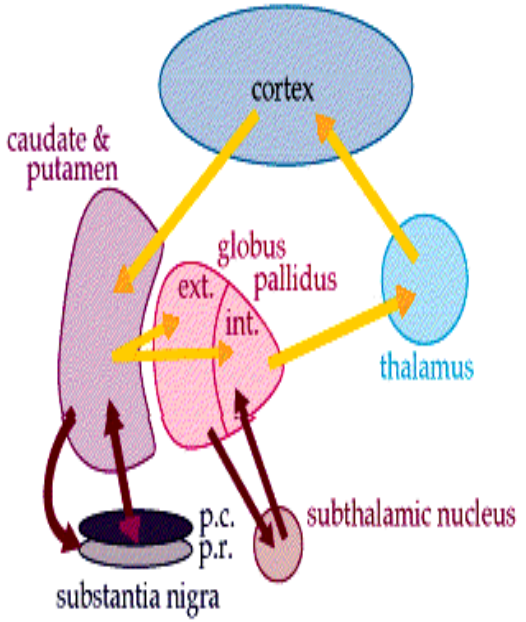
D2 (-) D1 (+)

Indirect Direct

NEUROTRANSMITTERS



- Dopamine (-): substantia nigra to corpus striatum
- ACh (+): intrastriatal putamen –caudate circuit.
- GABA (-): from corpus striatum to globus pallidus and substantia nigra. Globus pallidus to thalamus.
- Norepinephrine, serotonin, enkephalin from basal ganglia to brain stem
- Glutamate from cerebral cortex to corpus striatum , from thalamus to cerebral cortex.



Parkinsonism is defined as *bradykinesia*, in combination with at least 1 of *rest tremor* or *rigidity*

PARKINSONISM

Primary

Idiopathic Parkinson's disease

Atypical Parkinsonism

- Progressive Supranuclear Palsy
- Corticobasal degeneration
- Multiple System Atrophy
- Dementia with Lewy Body

Heredodegenerative Parkinsonism

- Wilson disease
- Neurodegeneration with brain iron accumulation
- Juvenile Huntington disease
- Fragile X tremor ataxia syndrome
- Spinocerebellar ataxias
- Perry syndrome
- Neuroacanthocytosis
- Frontotemporal dementia with parkinsonism (e.g., *MAPT* mutations)
- Familial prion diseases
- PARK-related parkinsonism (e.g., *SNCA* triplication)
- Parkinsonism/dementia due to *GBA* mutations
- Hereditary dystonia-parkinsonism (e.g., *ATP1A3* mutations)
- Mitochondriopathies (e.g., *POLG* mutations)
- Adrencleukodystrophy

Secondary

Vascular (e.g., white matter disease)
Drug-Induced (e.g., neuroleptics)
Metabolic (e.g., uremia)
Infectious (e.g., HIV, syphilis, Whipple, Lyme, prion)
Endocrine (e.g., hyperparathyroidism, hypothyroidism)
Autoimmune (e.g., Hashimoto disease, celiac disease)
Toxic (e.g., CO poisoning, manganism, MPTP)
Paraneoplastic (e.g., CRMP5 antibody)
Nutritional (e.g., vitamin B1, B12 deficiency)
Normal pressure hydrocephalus

MDS Clinical Diagnostic Criteria for PD
Postuma et al 2015

- Parkinsonism – bradykinesia plus either rigidity or rest tremor[1]
- **Clinically established PD:**[1]
  - **Absence of absolute exclusion criteria; at least 2 supportive criteria; no 'red flags'**

Specificity at least 90%

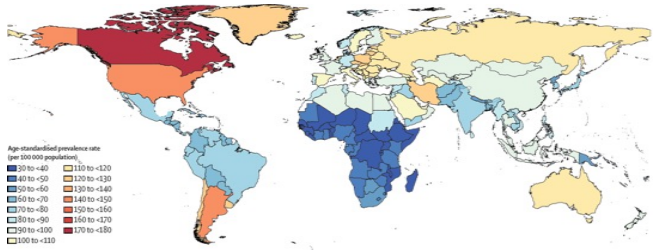
Absolute exclusion criteria[1]
- Cerebellar signs
- Supranuclear gaze palsy
- Established diagnosis of BVFTD
- Parkinsonism restricted to the lower limbs only for >3 years
- Treatment with an antidopaminergic, or with dopamine-depletion agents
- Absence of response to levodopa
- Sensory–cortical loss
- No evidence for dopaminergic deficiency on functional imaging
- Other parkinsonism-inducing condition

Red flags[1]
- Rapid deterioration of gait
- Absence of motor symptom progression over 5 years
- Early bulbar dysfunction
- Respiratory dysfunction
- Early severe autonomic failure
- Early recurrent falls due to misbalance
- Disproportionate anterocollis
- Absence of common non-motor features of disease during >5 years
- Pyramidal tract signs
- Bilateral symmetric presentation

Supportive criteria[1]
- A clear and dramatic positive response to dopaminergic therapy
- Levodopa-induced dyskinesia
- Documentation of resting tremor of a limb
- A positive diagnostic test of either olfactory loss or cardiac sympathetic denervation on scintigraphy

Parkinson's Disease

- PD prevalence 1 % of populations older than 65 years (Abbas et al., 2018)
- From 1990 to 2015, the number of with PD patients doubled to over 6 million, and double again to over 12 million by 2040 (Dorsey et al, 2018)
- Aging populations, increasing longevity, decreasing smoking rates, and the by-products of industrialization



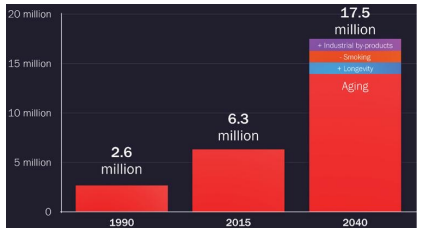


TABLE 1 *Country population, PD prevalence, and number of neurologists in surveyed African countries*

	Country	2020 Population*	Number of neurologists**	Movement disorders experts**	Specialized clinics	Number of neurologists/ million population*	Prevalence of PD per 100,000 (population based)
1	Algeria	43,851,044	600	12	–	13.68	–
2	Botswana	2,351,627	2	0	–	0.85	–
3	Burkina Faso	20,903,273	19	0	0	0.91	–
4	Burundi	11,890,784	7	0	0	0.59	–
5	Cameroon	26,545,863	33	2	0	1.25	–
6	Chad	16,425,864	4	0	0	0.24	–
7	Democratic Republic of the Congo	89,561,403	122	0	0	1.36	–
8	Djibouti	988,000	2	0	0	2.02	–
9	Egypt	102,334,404	4500	50	3	43.97	452 (≥40 years)11 557 (all ages)12 436 (all ages)13 213.15 (>40 years)14

Hamid...Shalash et al MDJ 2021

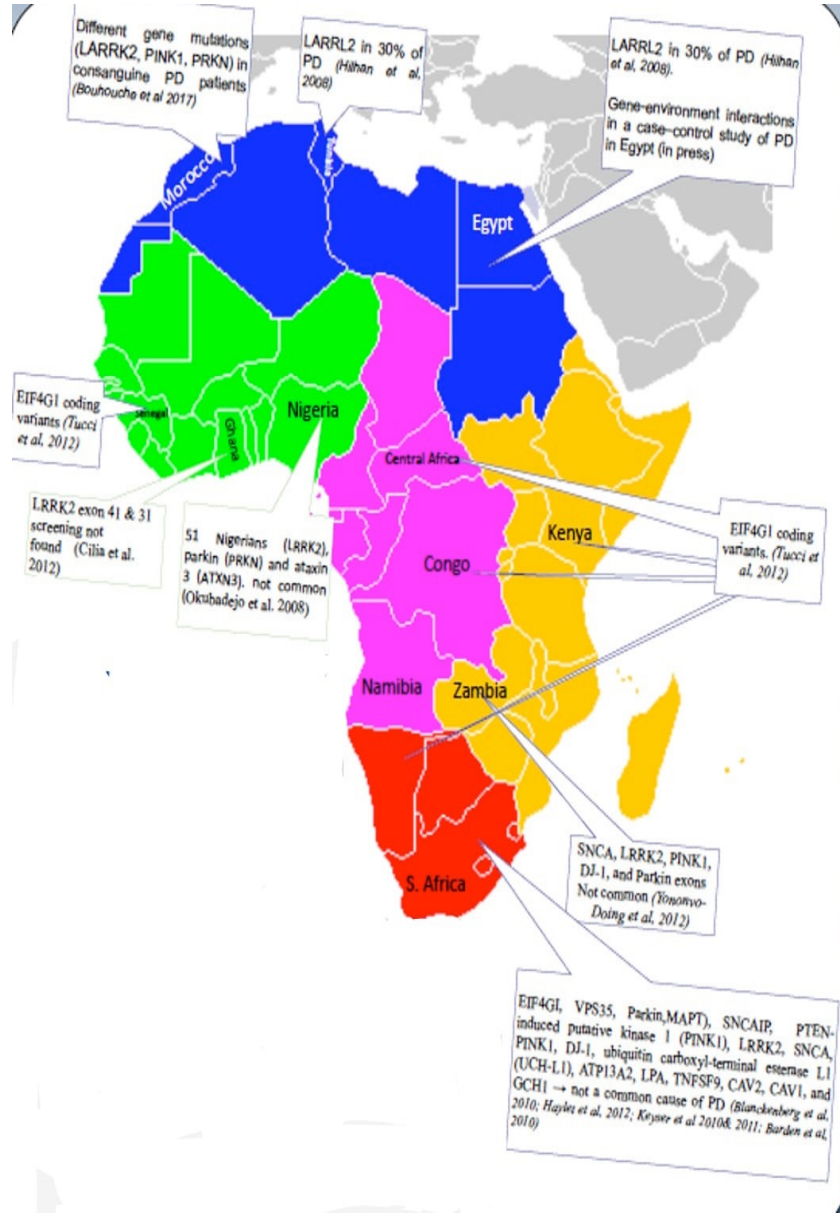
Parkinson's Disease (PD)

- **Incidence: i**ncreases dramatically with age, onset 50 - 60 years old.
- Overall incidence: 15 -25/100.000.
- 4-10% have onset before 40 years old.
  - *Young onset PD; onset before 40 or 50 years old.*
  - *Juvenile onset PD; onset before 20 years old.*
- PD is sporadic disease, rarely familial due to single gene mutations (Parkin, SNCA, LARRK).
- Susceptibility genetic loci; ↑↑ risk of PD (HLA DR, HLA DQ, SNCA, LRRK2, PARK 16), or risk (MAPT).

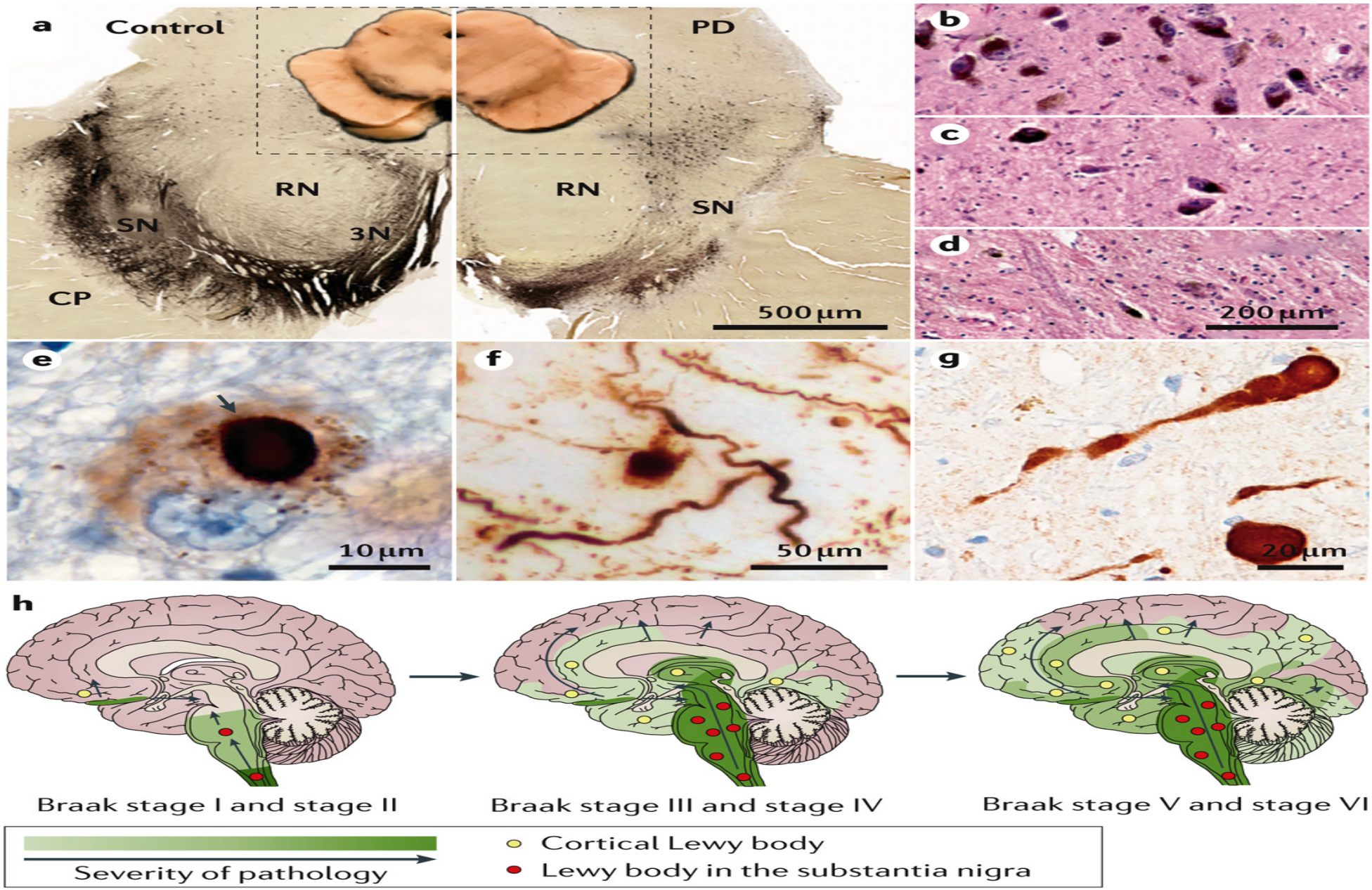
Genetic Diversity

Undiscovered genetic factors

- A genetically confirmed PD kindred from the East African region (North Tanzania) due to a homozygous *PRKN* deletion (Dekker et al, 2020)
- In Nigeria, A few genetic studies have also been conducted and did not detect pathogenic mutations in *PRKN* (parkin), *LRRK*2, and *ATXN3 (*Okubadejo et al, 2008, 2018; Oluwole et al, 2020)
- a *PTRHD1* mutation was identified in a Xhosa family with Parkinsonism and intellectual disability (Kuipers et al, 2018)
- LRRK2 in Arabic barber in Tunisia (40%), not present in Nigeria, Ghana, Tanzania, Zambia.
- Genetic studies in African populations have the potential to be of great benefit for PD research globally but have largely been unexplored.

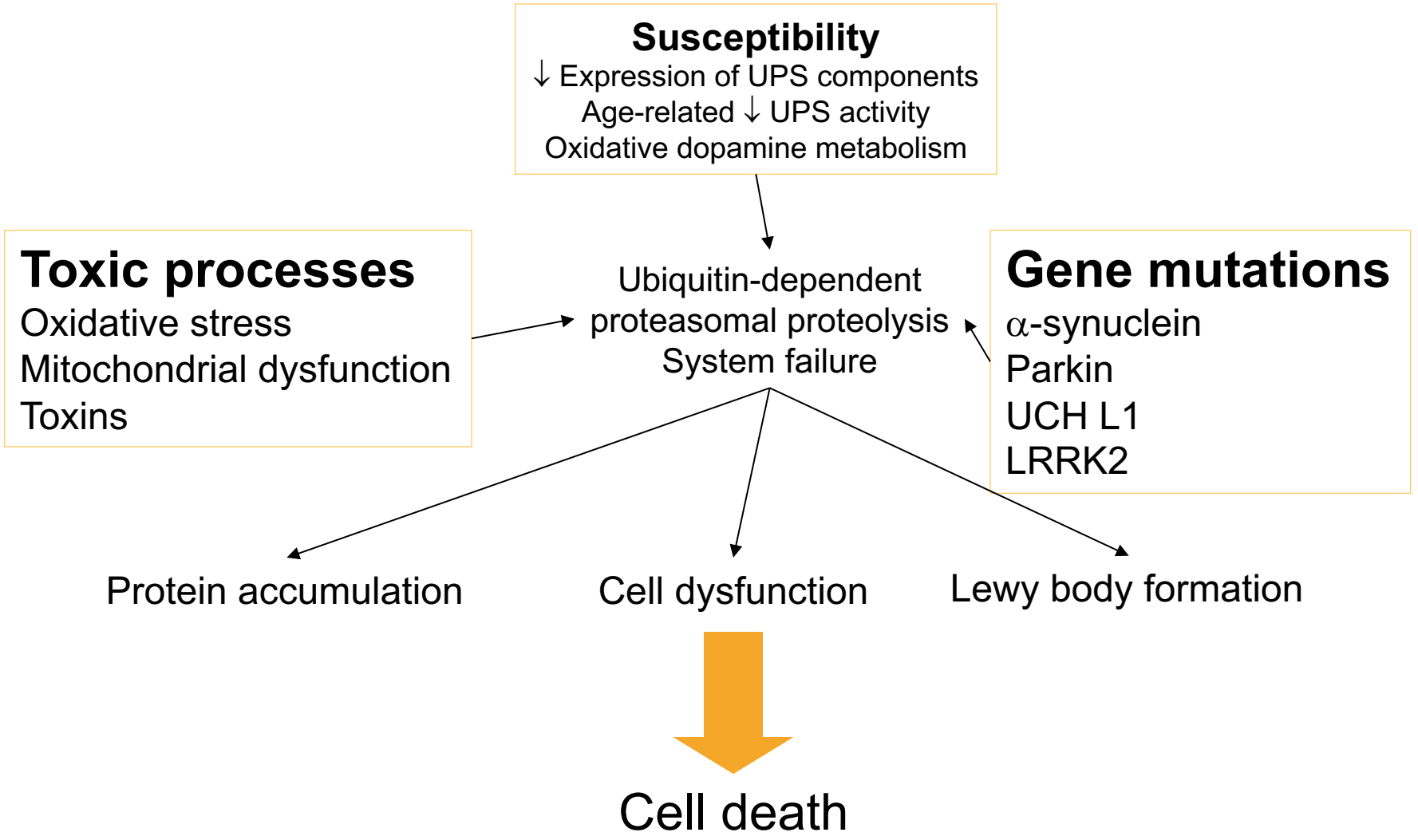


Neuropathology of PD



Poewe, W. *et al.* (2017) Parkinson disease
Nat. Rev. Dis. Primers doi:10.1038/nrdp.2017.13

PATHOGENESIS OF PARKINSON'S DISEASE



modified from McNaught & Olanow. Ann Neurol 2003;53:S73-86.

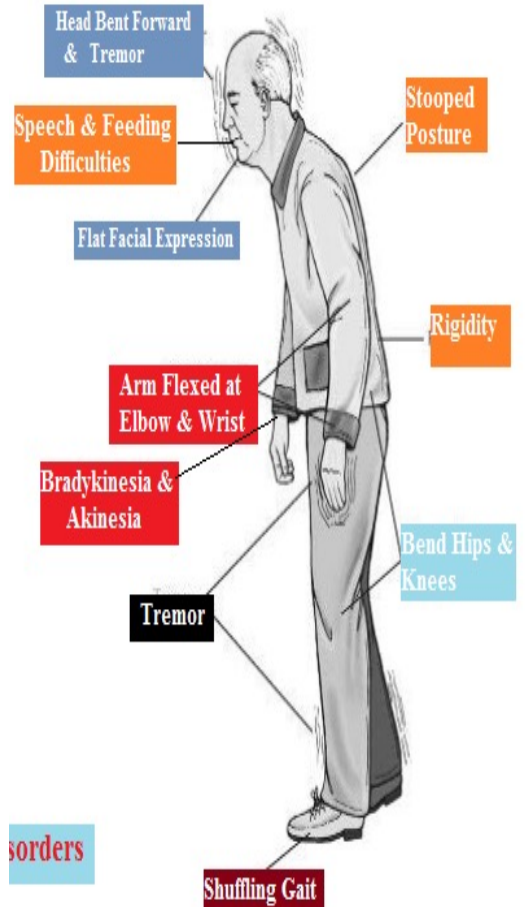
Clinical Picture

- insidious onset, progressive course.
- Start unilateral then bilateral → *Usually Asymmetric*

Bradykinesia or akinesia:

Difficulty in initiation and slowness of voluntary movements.

Decreased facial expression → mask faced, infrequent blinking. Decreased adjustment of posture.



Rigidity: 2 types:

i. *<u>Lead pipe</u>: present all throughout the movement.*

ii. *<u>Cog wheel</u>: lead pipe interrupted by tremors.*

- Proximal > distal, flexors and extensors.
- Affects muscles of limbs > neck, and trunk ⟶ flexed posture (gorilla like attitude).

PD Tremor

•Regular, rhythmic oscillatory involuntary movements.

•At rest, ± postural (reemergent tremor), distal > proximal.

Affect mainly ULs, neck, jaw and LLs.

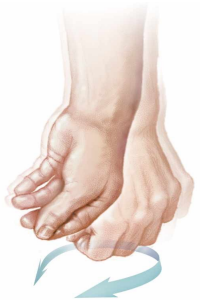
•Special characters: coarse, at rate of **4-8** c/s:

•Increased by stress, and emotions.

•Decreased by sleep and voluntary movements.

•***Pill rolling***: thumb & fingers are moving parallel & opposite to each [thumb (flexion & extension), fingers (abduction & adduction)].

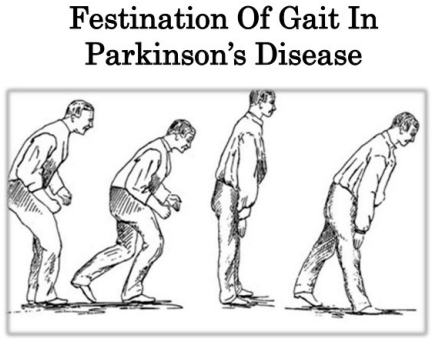
Pill rolling tremor



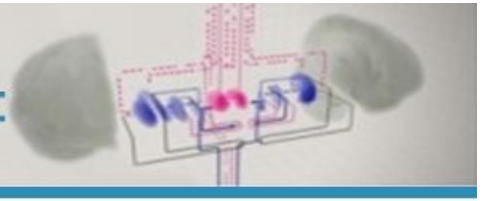
Hand tremor

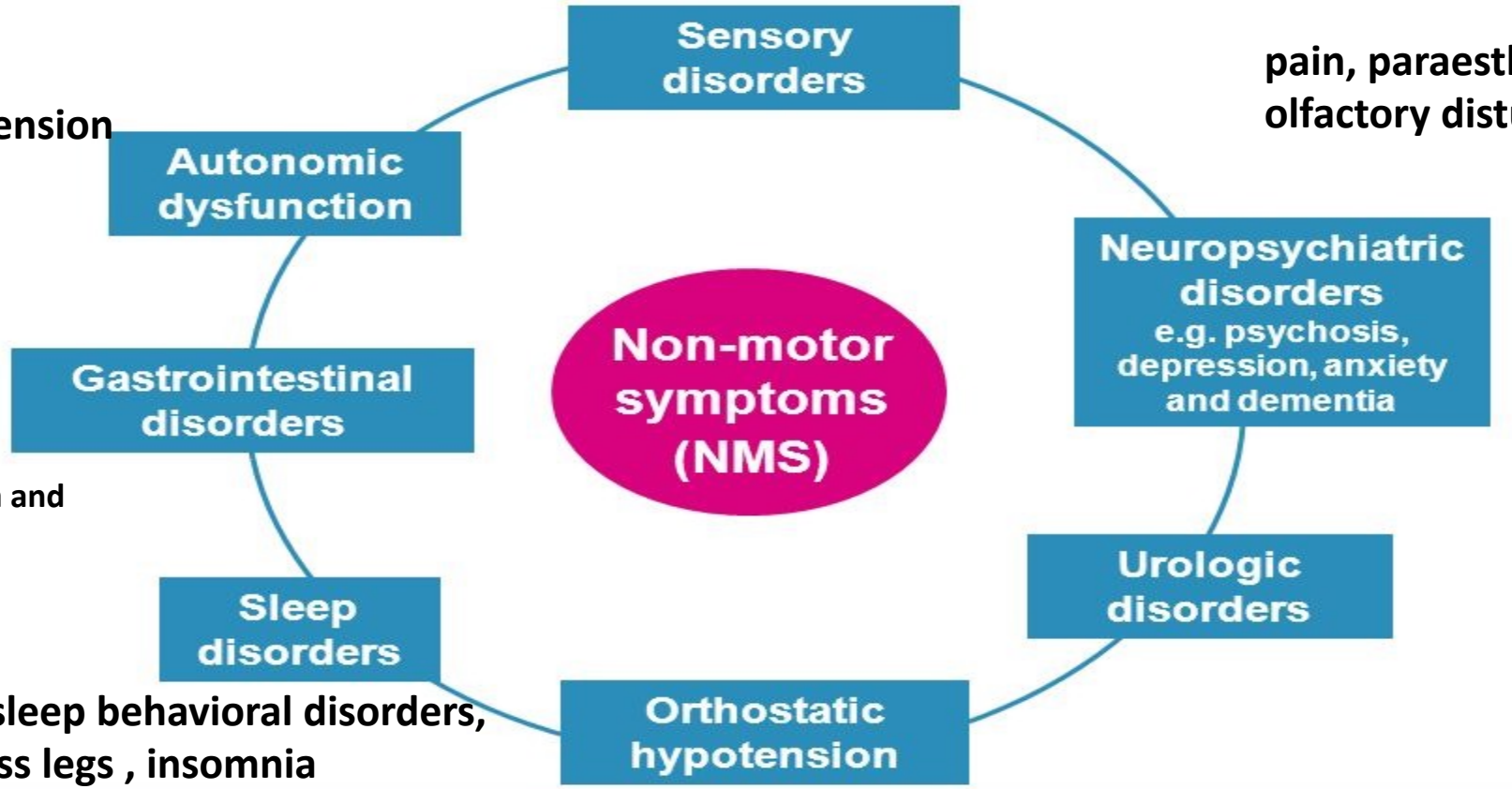
Others

- **Gait:** shuffling, short steppage, and festinant gait (running to reach the center of gravity).
- **Loss of emotional and associated movements:** *specially swinging of the arms during walking.*
- **Loss of postural reflexes;** retropulsion and propulsion**.**
- **Freezing phenomenon.**
- **The Myerson's sign, or glabellar tap sign**
- **Monotonous speech, micrographia, striatal hands.**



Non-motor symptoms of Parkinson's disease:
Patient burden





Postural hypotension sialorrhea

pain, paraesthesia, olfactory disturbance

dribbling of saliva, dysphagia and choking, reflux, constipation

REM sleep behavioral disorders, restless legs , insomnia

Adler CH. Mov Disord 2005;20(Suppl 11):S23-9 .

frontiers in Neurology

ORIGINAL RESEARCH
published: 24 May 2018
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Non-Motor Symptoms as Predictors of Quality of Life in Egyptian Patients With Parkinson's Disease: A Cross-Sectional Study Using a Culturally Adapted 39-Item Parkinson's Disease Questionnaire

Ali S. Shalash[1], Eman Hamid[1], Hanan Hani Elrassas[2], Ahmed Safwat Bedair[1], Abdelrahman Ibrahim Abushouk[3], Mohamed Khamis[1], Mostafa Hashim[2], Nahed Salah-Eldin Ahmed[1], Samia Ashour[1] and Mahmoud Elbalkimy[1]*

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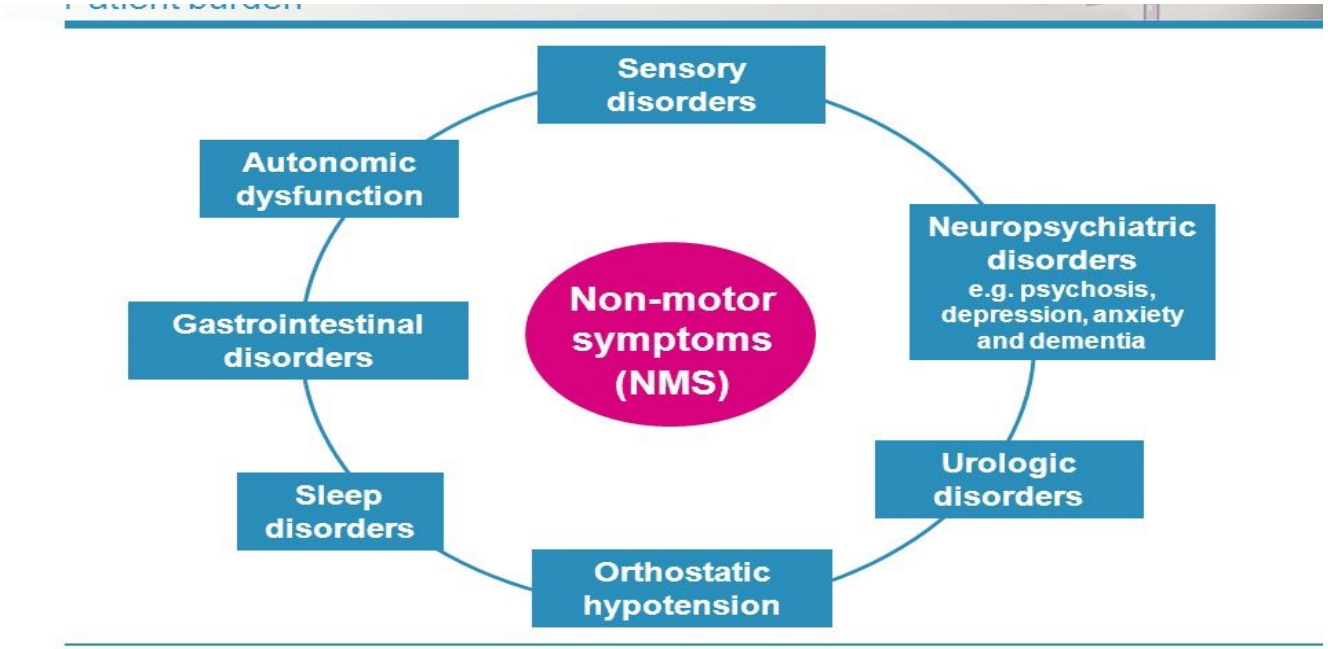

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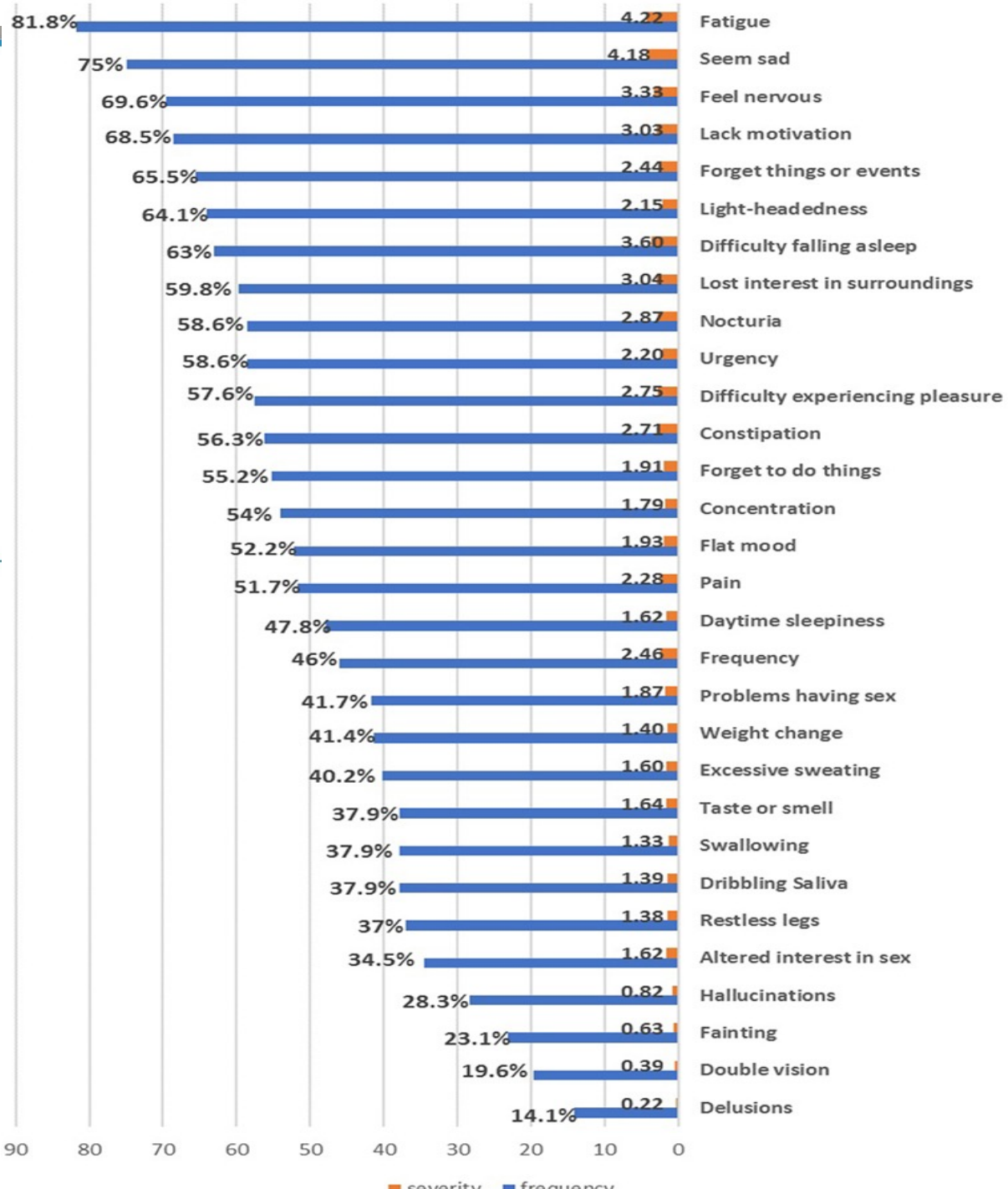
Shalash et al Front Neurol 2018



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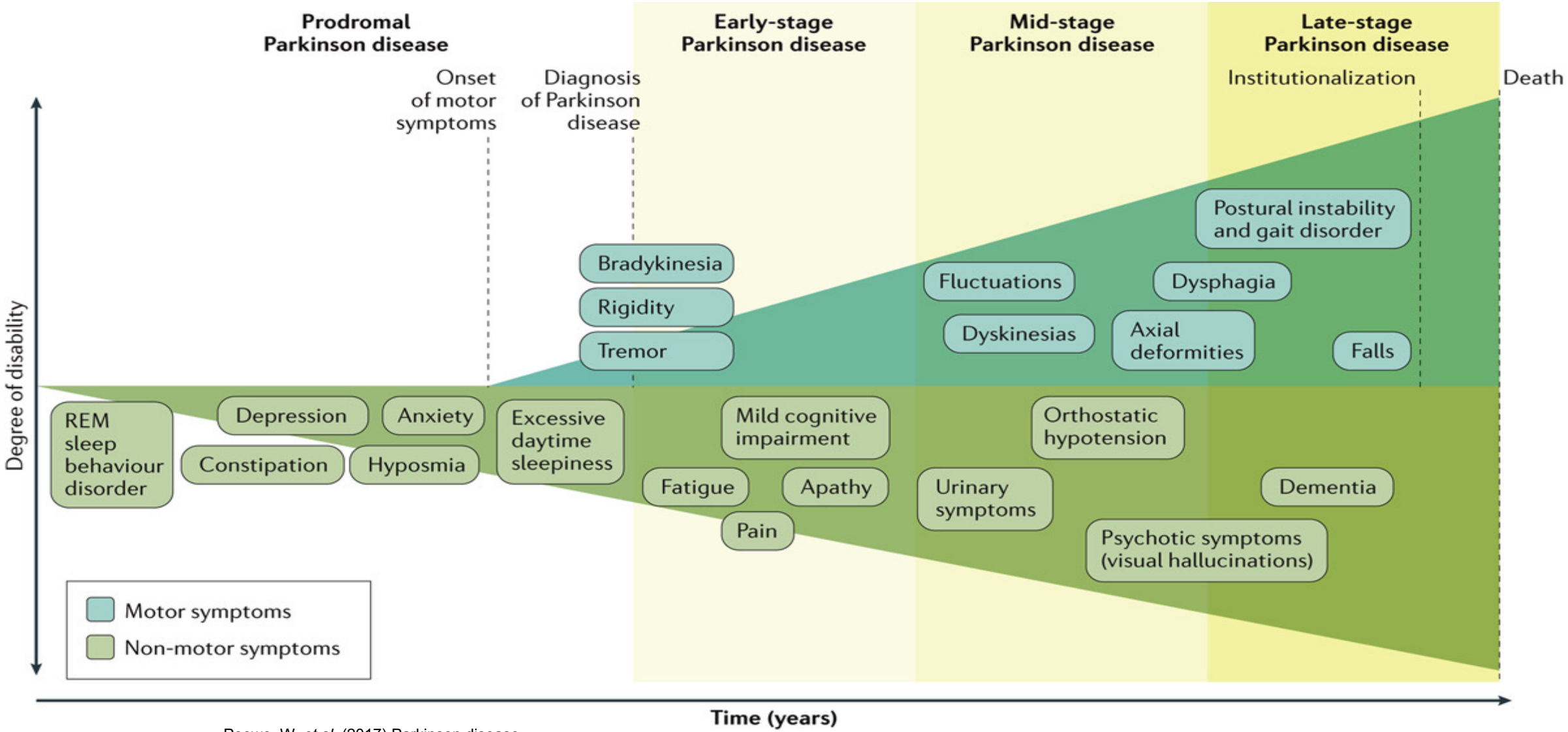
Clinical symptoms associated with PD progression



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Nature Reviews | Disease Primers

Differential Diagnosis of PD:

1. **Essential tremors:** AD, kinetic, and/or postural, absent at rest and increase with movement.

2. **Atypical Parkinsonian (Parkinsonism Plus) Syndromes** *include:*

- **Progressive supranuclear palsy (PSP):** characterized by symmetrical akinetic rigid syndrome, early falling, supranuclear gaze palsy, dysarthria, dysphagia, and pyramidal dysfunction.
- **Multiple system atrophy (MSA):** characterized by variable presentations of parkinsonism, cerebellar and pyramidal signs, and autonomic dysfunction.
- **Corticobasal degeneration (CBD):** Asymmetrical rigidity & bradykinesia, dystonia, myoclonus, Progressive aphasia, progress to dementia, Cortical sensory loss.
- **Dementia with Lewy bodies (DLB).**

Differential Diagnosis of PD:

3) **Vascular parkinsonism**:

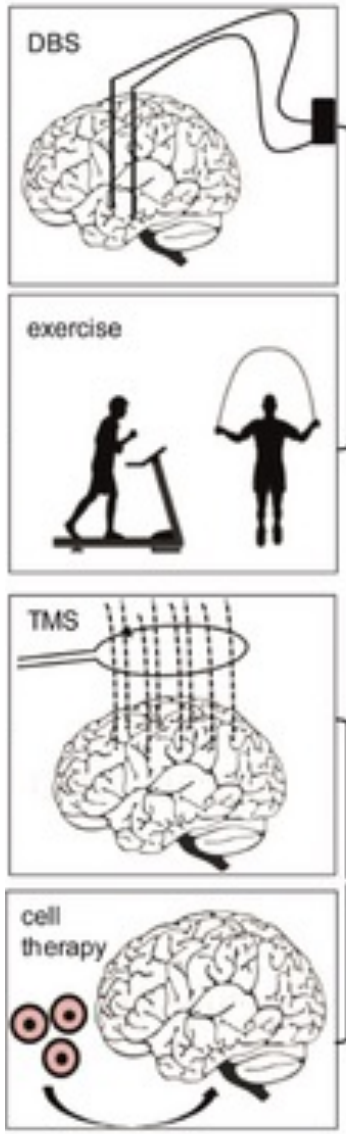
- Old age, risk factors of stroke, gradual or stepwise onset.
- Rigidity is predominant, pyramidal signs.
- Early and severe gait disturbance with falling.
- Mainly affecting lower limbs, so called lower body parkinsonism.
- Early cognitive impairment.
- Poor response to levodopa.
- Abnormal MRI brain; subcortical infarcts.

4) **Drugs and toxins (CO) induced parkinsonism**.

5) **Wilson's disease** (young onset).

TREATMENT OF PARKINSON'S DISEASE

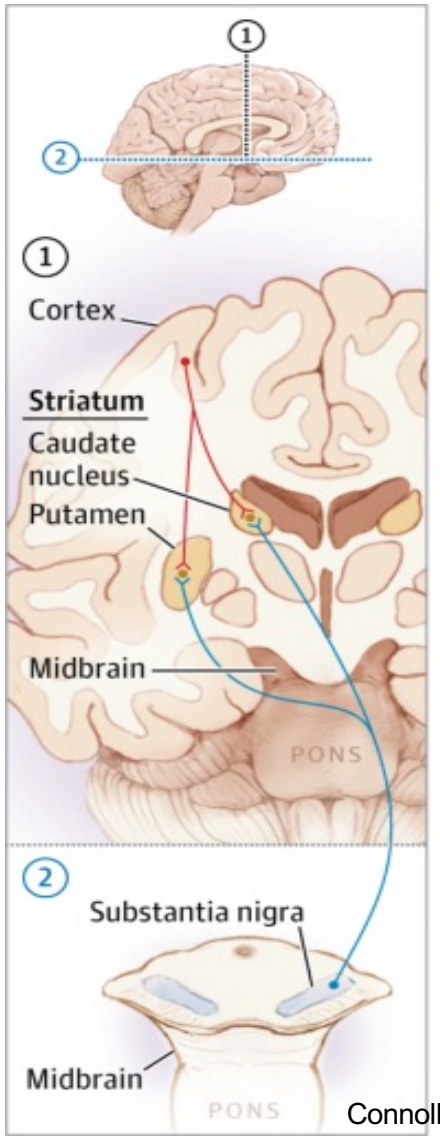
- **Medical**
  - Dopaminergic agents
  - Anticholinergics
  - MAO-B inhibitors
  - Therapies of NMSs
  - Others
- **Surgical**
  - Ablative
  - Advanced therapies; DBS, Duodopa, apomorphine infusion.
  - Restorative
- **Physical therapies**
- **Others:** botulinum toxin, TMS

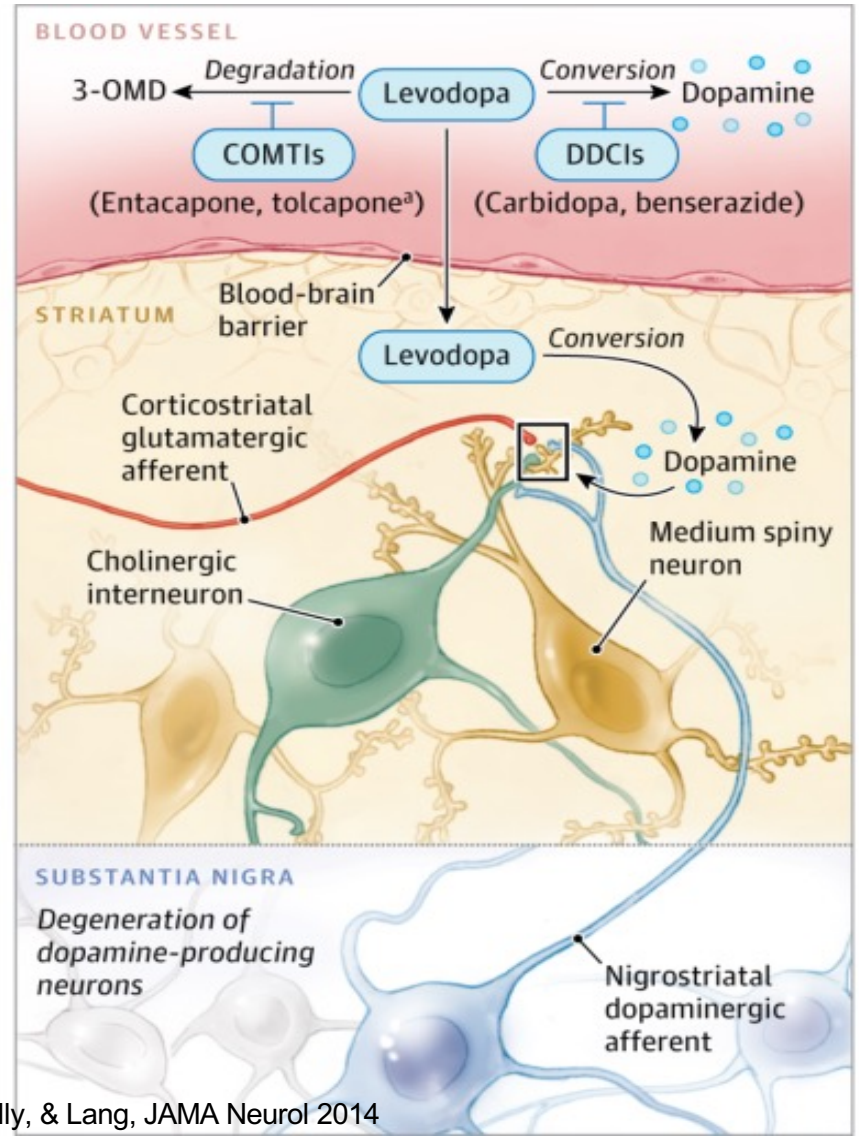


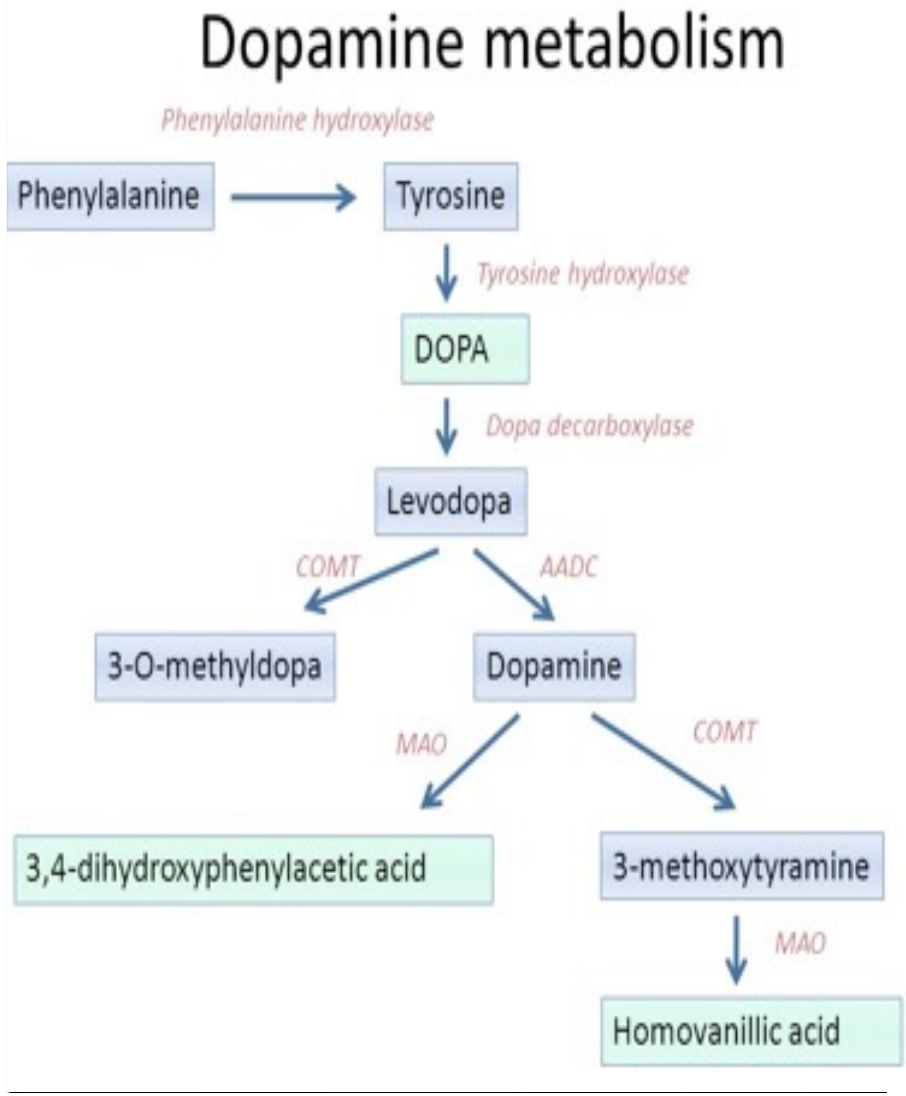
Pharmacological Treatment of PD

- Levodopa
- Dopamine Agonists
- Monoamine Oxidase-B Inhibitors
- *Catechol-O-methyltransferase* (COMT) *inhibitors*
- *Others:*
  - *Anticholinergics*
  - *Amantadine*
  - *Clozapine*
  - *Istradephyline;* Adenosine A2A receptor antagonist

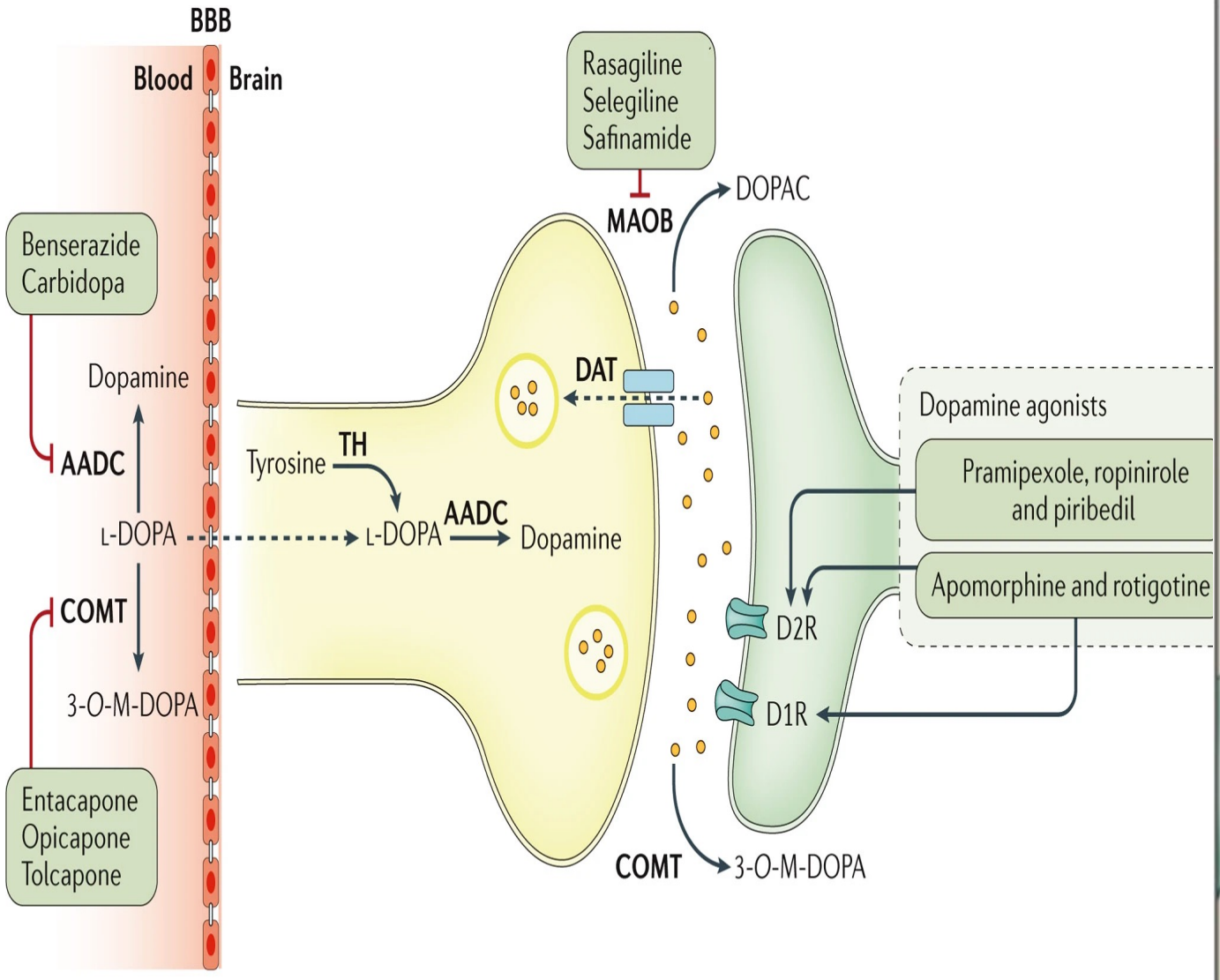
Pharmacological Treatment of PD



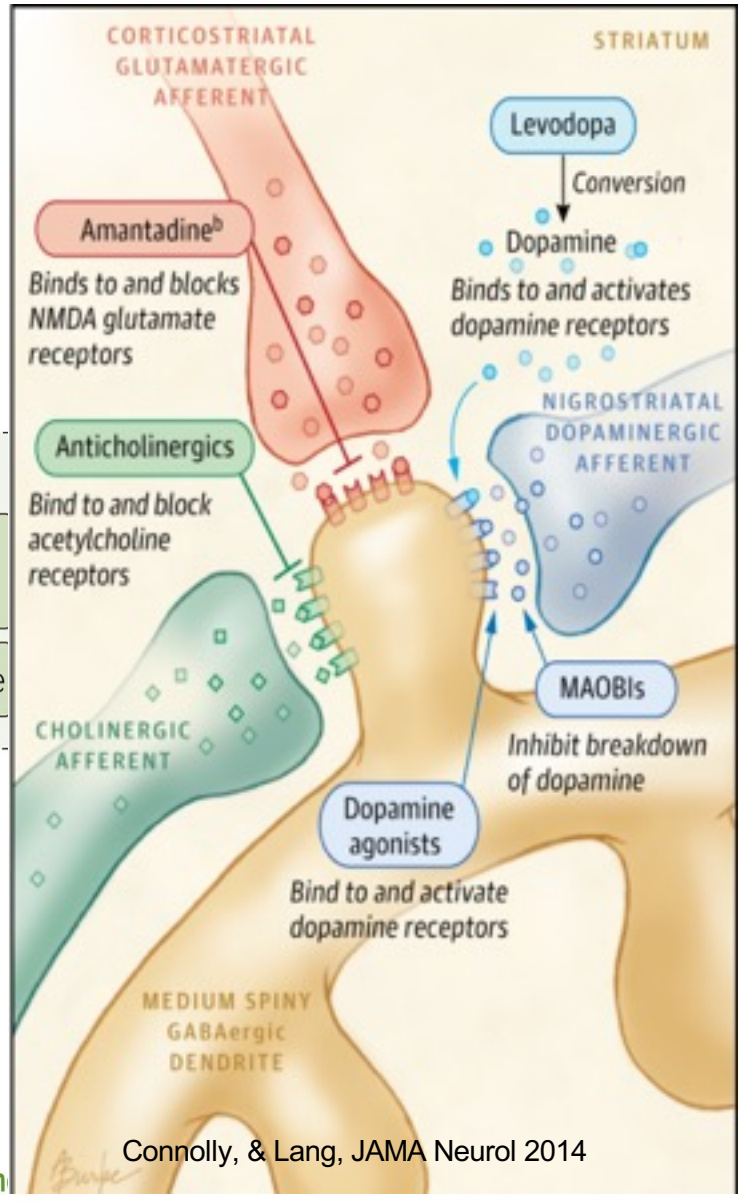




Connolly, & Lang, JAMA Neurol 2014



Powe et al 2017



Connolly, & Lang, JAMA Neurol 2014

Treatment of Non-Motor Manifestations:

1. ***Dementia***: rivastigmine, donepezil, memantine.
2. ***Depression/ Anxiety***: antidepressants (SSRI, SNRI).
3. ***Psychosis***: assess medication, clozapine (efficacious) or quetiapine, pimavanserin.
4. ***Impulse control disorders***: reduce dopamine agonists, clozapine, and quetiapine, donepezil.
5. ***Drooling***: anticholinergics, botulinum toxin.
6. ***Postural hypotension***: increasing salt intake, changing position slowly, wearing elastic stockings and avoiding aggravating factors, midodrine, fludrocortisones.
7. ***Constipation***: laxative, macrogol.
8. ***Fatigue***: methylphenidate.

Applying Advanced Therapies, Egypt Experience

PD DBS insured in Egypt June 2021

Pre DBS

Post DBS

Ali Shalash, Zeiad Fayad , Ain Shams Team

RESEARCH ARTICLE

Availability of Therapies and Services for Parkinson's Disease in Africa: A Continent-Wide Survey

MDJ, 2021

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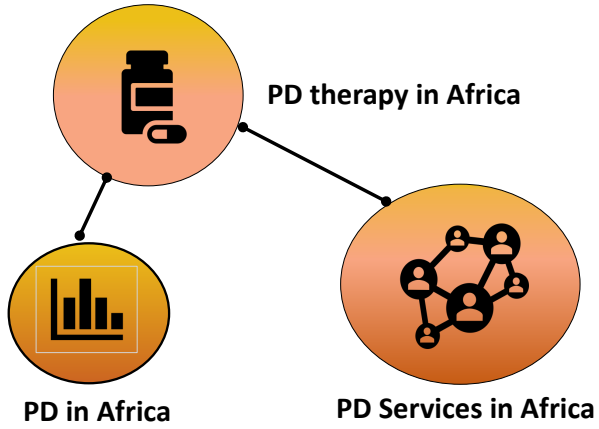
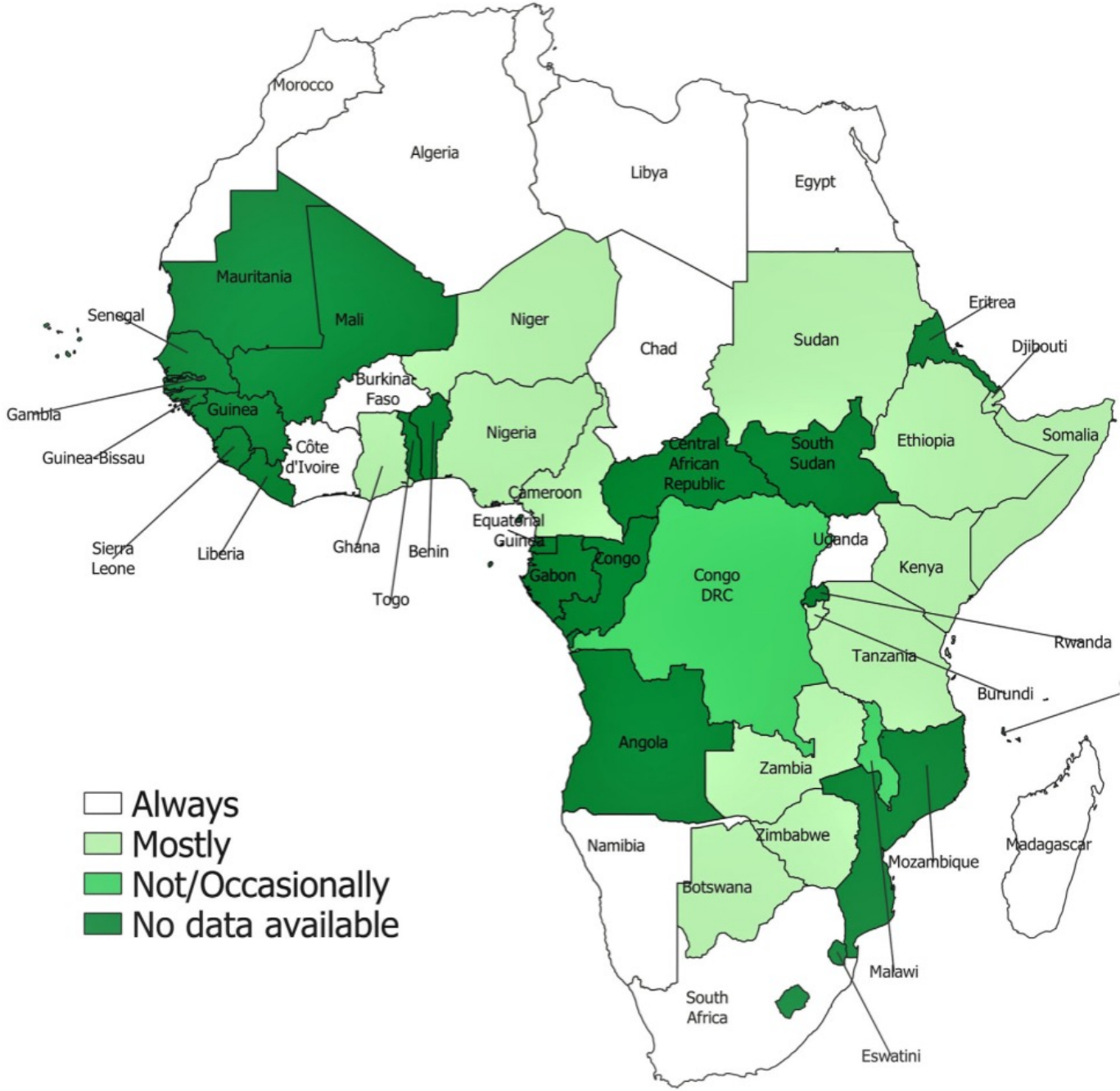
- **28 countries (of 43 contacted countries).**
- **51.9% of the 54 countries within Africa.**
- **84.7% of the total continent population.**



Availability of Levodopa in Africa

Levodopa preparation was

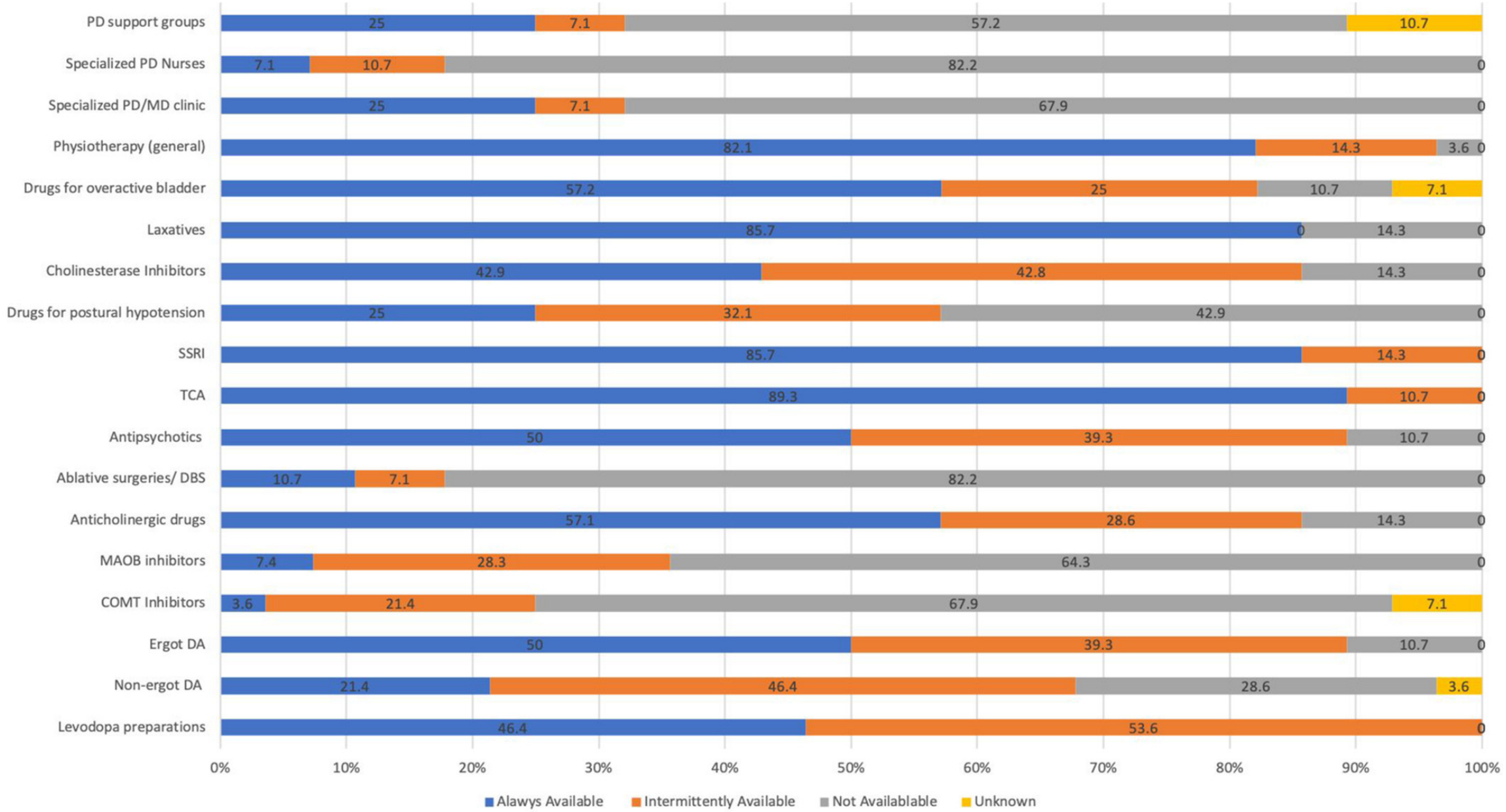
- Always available in 13 countries (46.4%),
- Mostly available in 13 countries (46.4%)
- Occasionally/ sometimes available in 2 countries



f Africa indicating countries and availability of levodopa (as always, mostly, not/occasionally, or data not available).

Hamid.....Shalash MDJ 2021

Availability of Therapies and Services for Parkinson's Disease in Africa: A Continent-Wide Survey



Movement Disorders, First published: 03 June 2021, DOI: (10.1002/mds.28669)

Hamid....Shalash et al MDJ 2021

Mucuna pruriens in Parkinson disease

A double-blind, randomized, controlled, crossover study

OPEN

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ABSTRACT

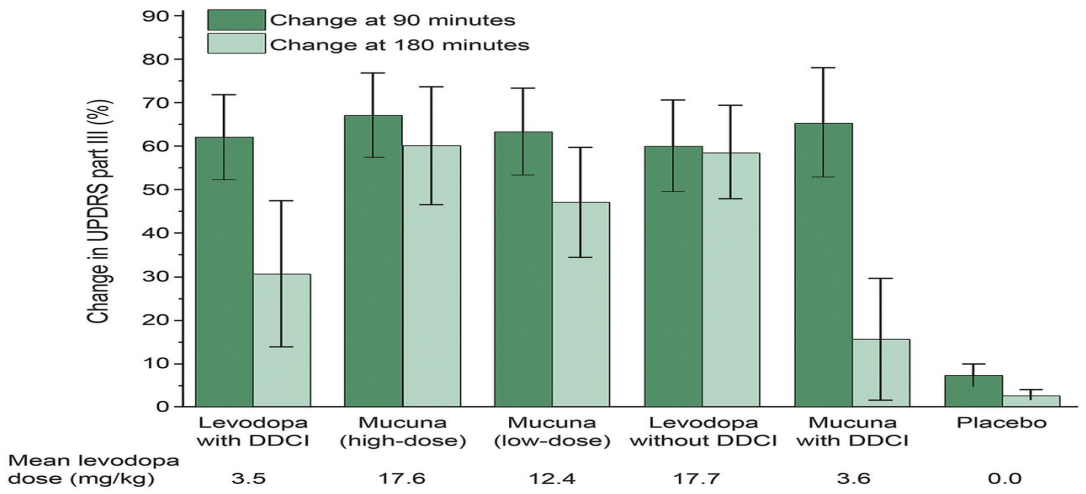
Objective: To investigate whether *Mucuna pruriens* (MP), a levodopa-containing leguminous plant growing in all tropical areas worldwide, may be used as alternative source of levodopa for indigent individuals with Parkinson disease (PD) who cannot afford long-term therapy with marketed levodopa preparations.

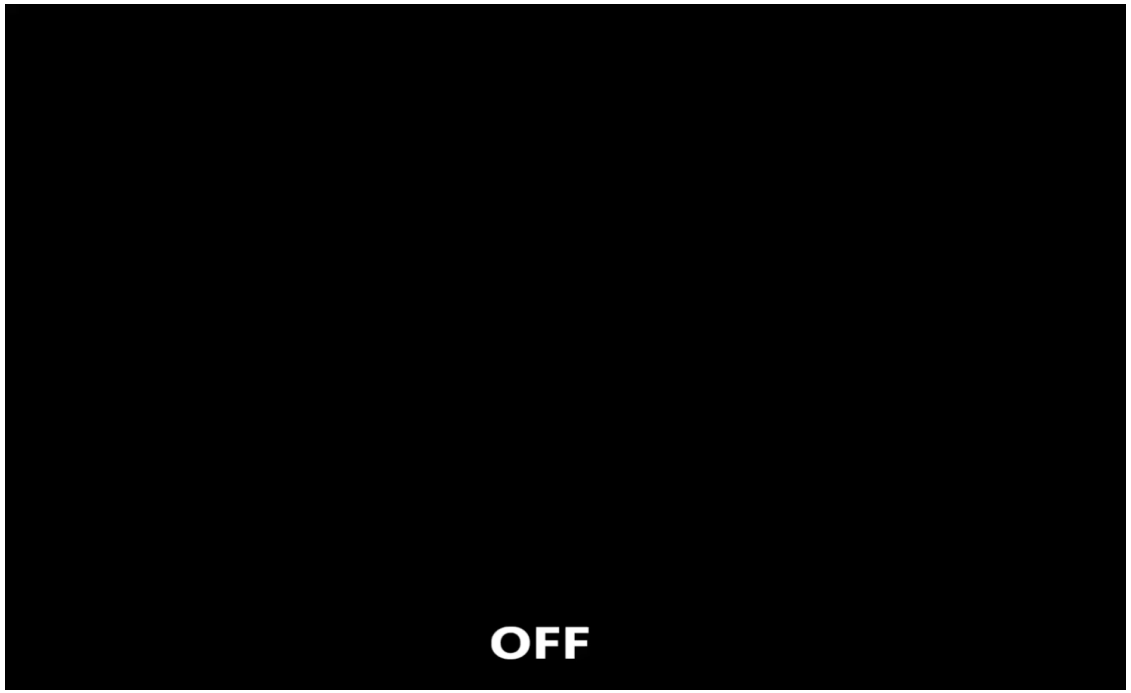
Methods: We investigated efficacy and safety of single-dose intake of MP powder from roasted seeds obtained without any pharmacologic processing. Eighteen patients with advanced PD received the following treatments, whose sequence was randomized: (1) dispersible levodopa at 3.5 mg/kg combined with the dopa-decarboxylase inhibitor benserazide (LD+DDCI; the reference treatment); (2) high-dose MP (MP-Hd; 17.5 mg/kg); (3) low-dose MP (MP-Ld; 12.5 mg/kg); (4) pharmaceutical preparation of LD without DDCI (LD−DDCI; 17.5 mg/kg); (5) MP plus benserazide (MP+DDCI; 3.5 mg/kg); (6) placebo. Efficacy outcomes were the change in motor response at 90 and 180 minutes and the duration of on state. Safety meas-

Roberto Cilia et al. Neurology 2017;89:432-438

- Mucuna Pruriens is a leguminous plant whose seed contain Levodopa without Dopa-Decarboxylase Inhibitor
- Available in all tropical areas worldwide

MP-Ld showed similar motor response with fewer dyskinesias and AEs, while MP-Hd induced greater motor improvement at 90 and 180 minutes, longer ON duration, and fewer dyskinesias. MP-Hd induced less AEs than LD+DDCI and LD−DDCI.





In courtesy of Dr Roberto Cilia (in press)

JAMA Neurology | Special Communication

Six Action Steps to Address Global Disparities in Parkinson Disease
A World Health Organization Priority

Nicoline Schiess, MD, MPH; Rodrigo Cataldi, PhD; Michael S. Okun, MD; Natasha Fothergill-Misbah, PhD; E. Ray Dorsey, MD; Bastiaan R. Bloem, MD, PhD; Maria Barretto, PhD; Roongroj Bhidayasiri, MD; Richard Brown, PhD; Lorraine Chishimba, MD; Neerja Chowdhary, MD; Max Coslov, MPhil; Esther Cubo, MD, PhD; Alessandro Di Rocco, MD; Rachel Dolhun, MD; Christopher Dowrick, MSc, MD; Victor S. C. Fung, MBBS, PhD; Oscar S. Gershanik, MD; Larry Gifford, BS; Joyce Gordon, BS; Hanan Khalil, PhD; Andrea A. Kühn, MD; Sara Lew, BA; Shen-Yang Lim, MBBS, MD; Maria M. Marano, MSc; Jacquie Micallef, BSW; Jolynne Mokaya, DPhil; Emile Moukheiber, MD; Lynda Nwabuobi, MD; Njideka Okubadejo, MBChB; Pramod Kumar Pal, MBBS, MD, DM; Hiral Shah, MD; Ali Shalash, MD; Todd Sherer, PhD; Bernadette Siddiqui, MA; Ted Thompson, JD; Andreas Ullrich, MD, MPH; Richard Walker, MD; Tarun Dua, MD

Advocacy & Awareness

- Increase awareness, patients' supportive groups, increase education & training, fight discrimination, contacting stakeholders

Prevention & Risk Reduction

- An increased risk has been reported among those with exposure to pesticides.
- Amphetamine or methamphetamine, lack of physical activity, heavy metals, air pollution, traumatic brain injury, and industrial solvents, such as trichloroethylene (TCE)
- Avoid exposure to pesticides, protective tools, physical activities, caffeine.

Diagnosis, Treatment, and Care

- Strengthening Health and Social Systems and Building Capacity. Education & training, tele-education & telemedicine,
- Ensuring the Availability of Essential Drugs, Diagnostics, and Interdisciplinary Therapies.

Caregiver Support

- Provision of a timely diagnosis; effective communication and education about caregiver roles, medications, and adverse effects; and rehabilitation and palliative care strategies, including governmental entitlements and discussions of decision-making capacity. Social workers, patient support groups, and community-based support.

Research

- investigate cultural and population differences of variable risk factors, genetics, and phenomenology.

Schiess et al, JAMA Neurology 2022

Characteristics of Hyperkinetic MDs

Dystonia: *sustained* or intermittent muscle contractions causing abnormal- often *repetitive-* movements, *postures*, or both" .

Chorea: *irregular* rapid, low amplitude, *brief* movements of extremities & face

Athetosis: involuntary writhing movements

Hemiballism: *large* amplitude involuntary movement restricted to one side of the body; usually involves proximal upper limb.

Myoclonus: sudden brief *jerk or shock-like* movements

Tremor: *rhythmic oscillation* of a body part due to alternating or synchronous contractions of opposing muscles

Tics: sudden, brief, purposeless, *stereotyped* simple or complex movements or vocalizations , with urge

Akathisia: inner *restlessness*; often associated with external signs of restless behavior

Observe MD During Examination

- Rhythmic vs. arrhythmic
- Sustained vs. nonsustained
- Paroxysmal vs. Nonparoxysmal
- Slow vs. fast
- Amplitude
- At rest vs. action
- Patterned vs. non-patterned
- Combination of varieties of movements
- Supressibility

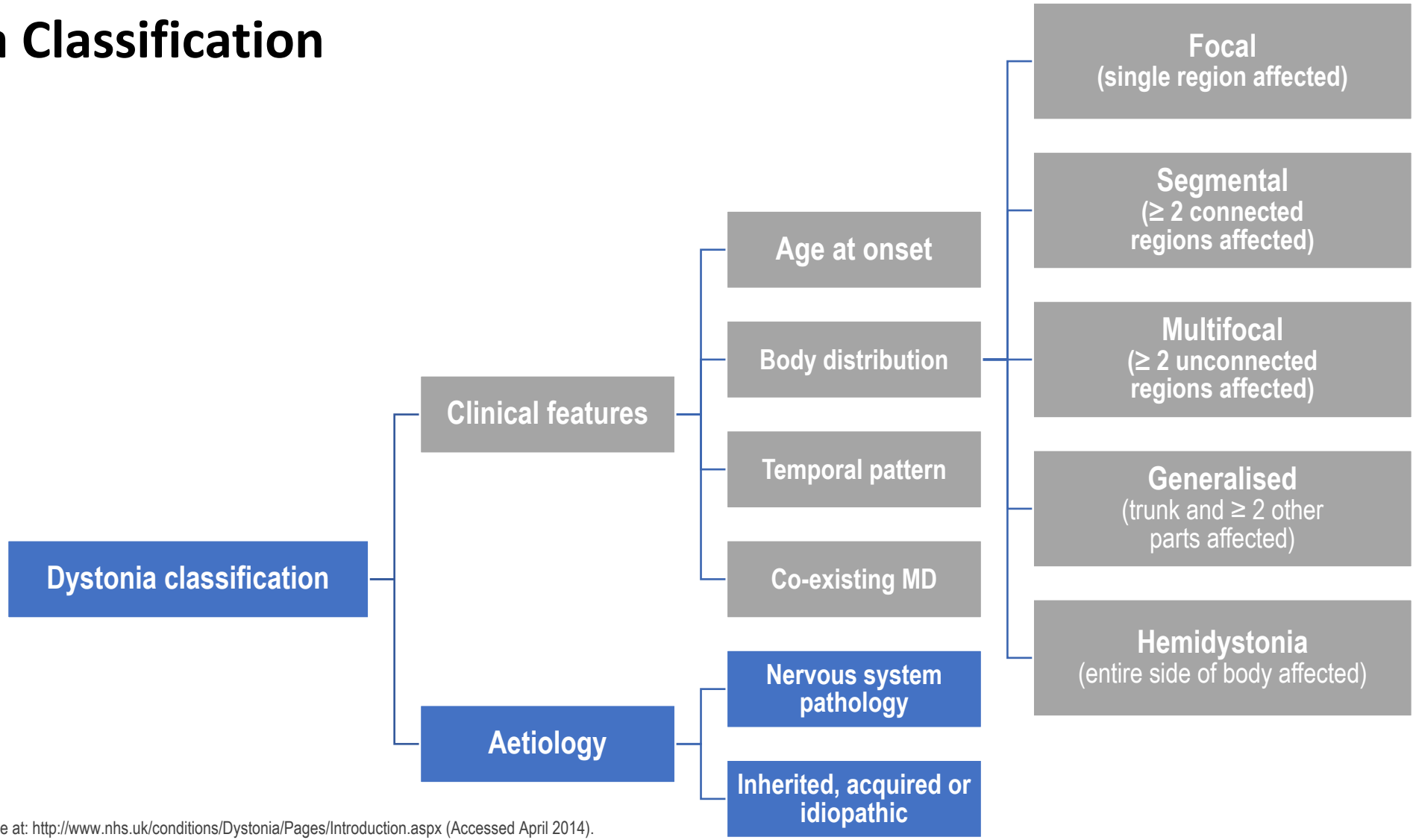
- Observe any involuntary movements during history and their distribution; speech and vocalizations

DYSTONIA

"It is a movement disorder characterized by sustained or intermittent muscle contractions causing abnormal, often repetitive, movements, postures, or both”

- It’s typically patterned, twisting, and may be tremulous.
- It is often initiated or worsened by voluntary action and associated with overflow muscle activation.
- ± Alleviating maneuvers (sensory tricks or gestes antagonistes).
- ± Task-specificity: selective activation by specific tasks (e.g. writing, playing music).

Dystonia Classification

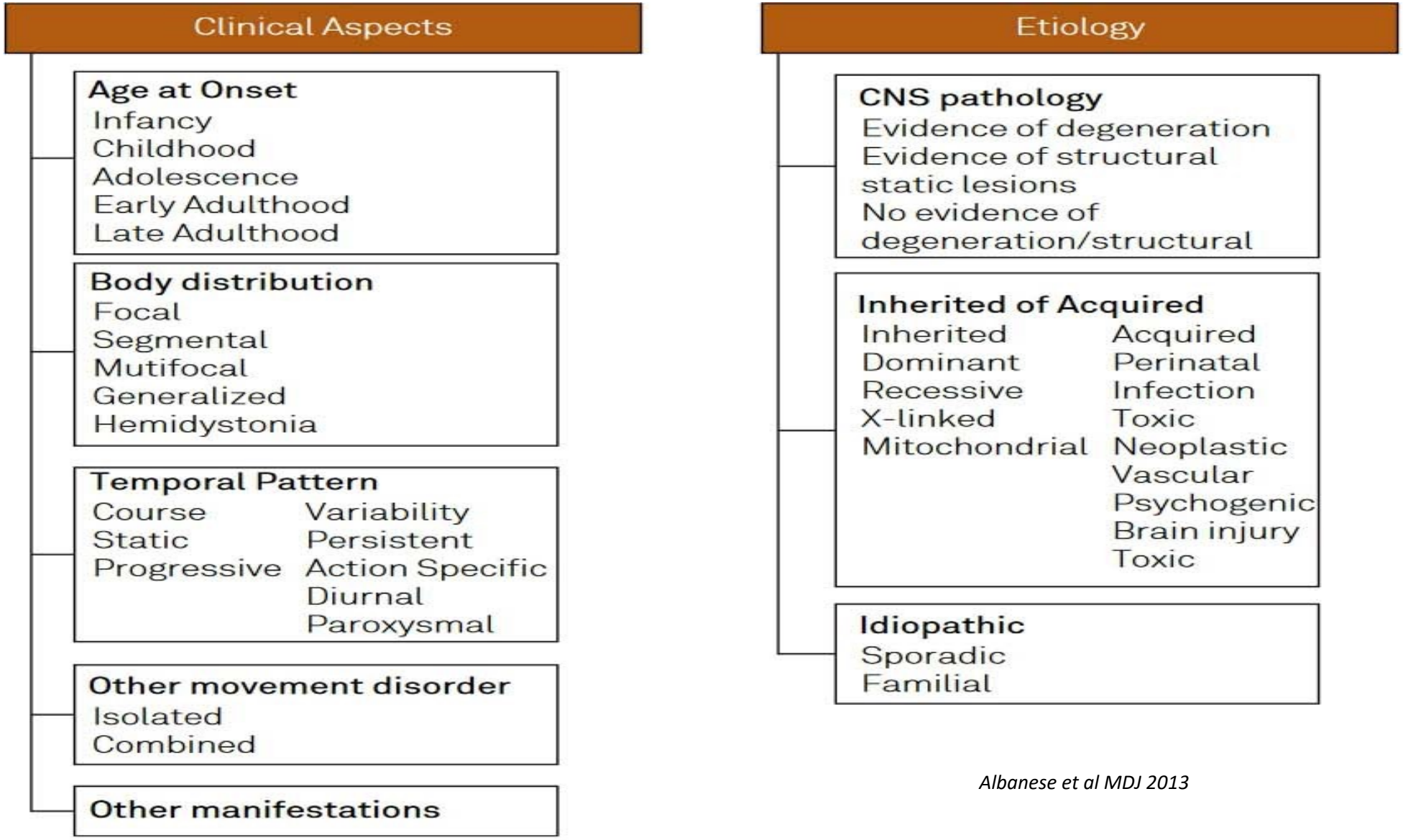


MD, movement disorders.

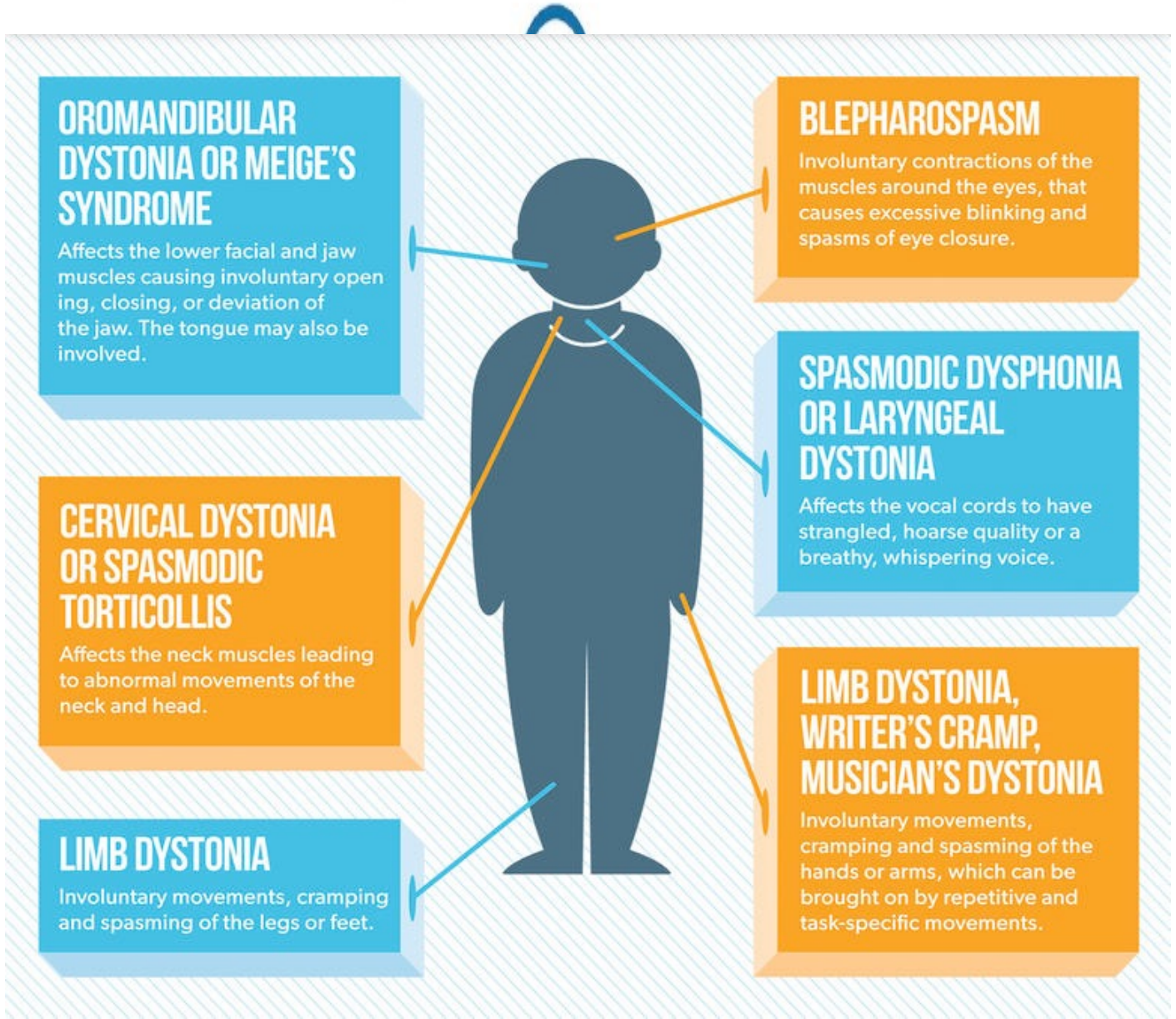
1. NHS choices. Dystonia. Available at: http://www.nhs.uk/conditions/Dystonia/Pages/Introduction.aspx (Accessed April 2014).
2. Skogseid. Acta Neurol Scand Suppl 2014;198:13–9.

Old Dystonia Classification

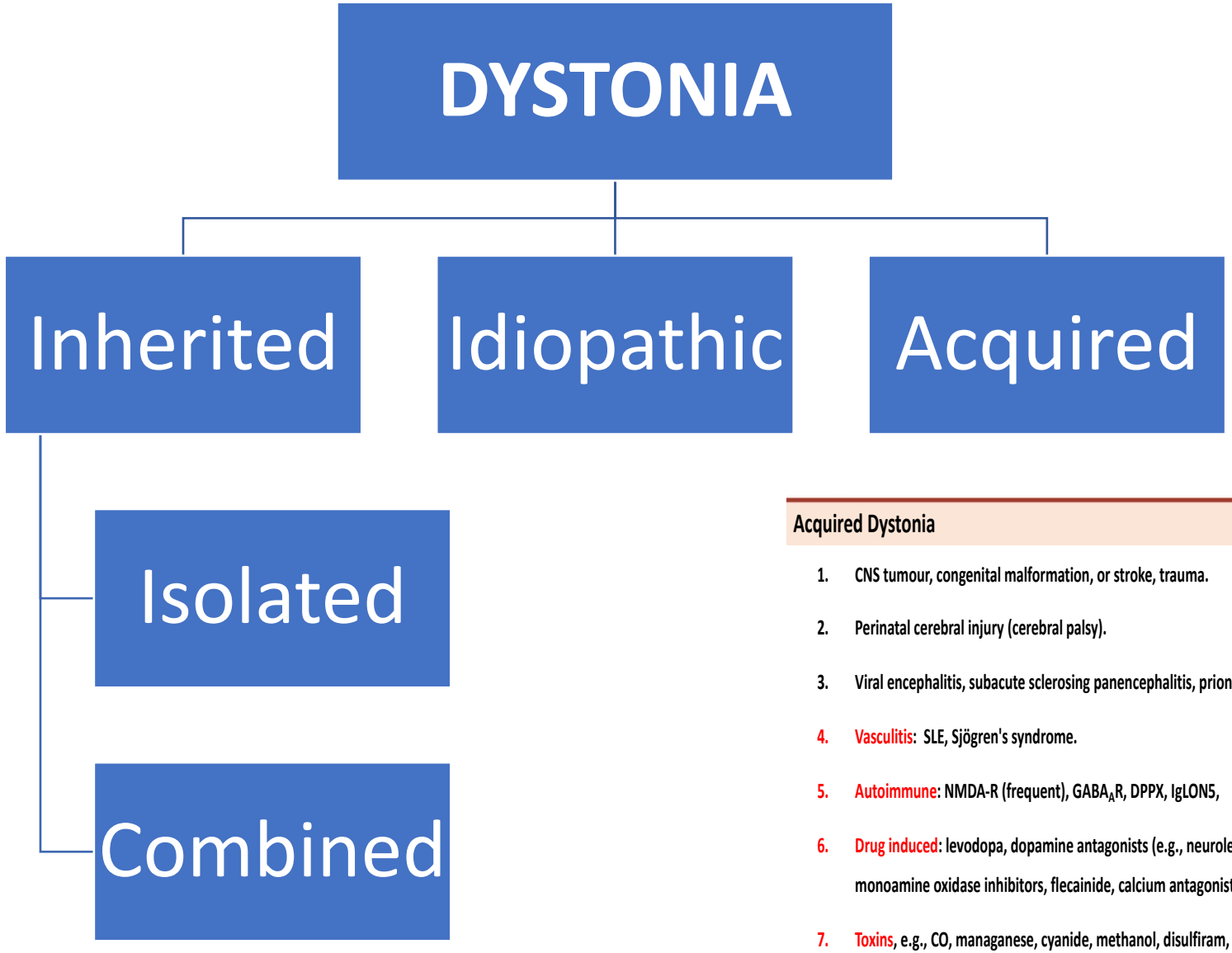
Recent Dystonia Classification



Albanese et al MDJ 2013


OROMANDIBULAR DYSTONIA OR MEIGE'S SYNDROME
Affects the lower facial and jaw muscles causing involuntary open ing, closing, or deviation of the jaw. The tongue may also be involved.
BLEPHAROSPASM
Involuntary contractions of the muscles around the eyes, that causes excessive blinking and spasms of eye closure.
SPASMODIC DYSPHONIA OR LARYNGEAL DYSTONIA
Affects the vocal cords to have strangled, hoarse quality or a breathy, whispering voice.
CERVICAL DYSTONIA OR SPASMODIC TORTICOLLIS
Affects the neck muscles leading to abnormal movements of the neck and head.
LIMB DYSTONIA, WRITER'S CRAMP, MUSICIAN'S DYSTONIA
Involuntary movements, cramping and spasming of the hands or arms, which can be brought on by repetitive and task-specific movements.
LIMB DYSTONIA
Involuntary movements, cramping and spasming of the legs or feet.


Torticollis
(neck turning)
Laterocollis
(head tilt)
Anterocollis
(neck flexion)
Retrocollis
(neck extension)



Acquired Dystonia

1. CNS tumour, congenital malformation, or stroke, trauma.
2. Perinatal cerebral injury (cerebral palsy).
3. Viral encephalitis, subacute sclerosing panencephalitis, prion disease, tuberculosis
4. Vasculitis: SLE, Sjögren's syndrome.
5. Autoimmune: NMDA-R (frequent), $GABA_AR$, DPPX, IgLON5,
6. Drug induced: levodopa, dopamine antagonists (e.g., neuroleptics, prochlorperazine, metoclopramide), SSRI, buspirone, cocaine, monoamine oxidase inhibitors, flecainide, calcium antagonists.
7. Toxins, e.g., CO, managanese, cyanide, methanol, disulfiram, carbondisulphide, and methanol.
8. Metabolic: hypoparathyroidism
9. Paraneoplastic syndromes
10. Functional

INHERITED ISOLATED DYSTONIAS

Gene (previous DYT symbol[a])	Inheritance	Age at onset	Prevalent site at onset	Distribution	Body parts involved	Additional signs
TOR1A (DYT1)	Autosomal dominant *Clinical penetrance of only 30-40%* *The GAG deletion in TOR1A*	First to third decade	Lower limbs much more likely than upper limbs	Mostly generalized	• Lower limbs[b] • Upper limbs • Trunk	None
THAP1 (DYT6)	Autosomal dominant (autosomal recessive in rare cases)	Second to third decade (ranging from first to seventh decade)	Neck and upper limbs Older-onset cervical or craniocervical dystonia with likely involvement of the larynx	Focal, segmental and generalized	• Neck[b] • Upper limbs[b] • Orofacial areas • Larynx • Lower limbs	None
GNAL (DYT25)	Autosomal dominant (autosomal recessive in rare cases)	Fourth decade (ranging from first to seventh decade)	Neck	Mostly focal or segmental and occasionally generalized	• Neck[b] • Orofacial areas • Larynx • Upper limbs • Lower limbs	None
ANO3 (DYT24)	Autosomal dominant	Fourth to fifth decade (ranging from first to fifth decade)	Neck and larynx	Segmental	• Neck[b] • Upper limbs • Orofacial areas • Larynx	None

COMBINED DYSTONIA

		GCH1	DYT5a	DYT-GCH1	Dopa-responsive	AD, AR
Combined	**Dystonia + Parkinsonism**	TH	DYT5b	DYT-TH	Dopa-responsive	AR
		SPR	Not assigned	DYT-SPR	Dopa-responsive, cognitive impairment	AR
		TAF1 [1]	DYT3	DYT-TAF1	Neurodegeneration	XL
		PRKRA	DYT16	DYT-PRKRA	Dystonia w/mild parkinsonism	AR
		ATP1A3	DYT12	DYT-ATP1A3	Rapid-onset	AD
	Dystonia + Myoclonus	SGCE	DYT11	DYT-SGCE	Psychiatric disease	AD
	Paroxysmal Dystonia + Other Dyskinesia	PNKD [2]	DYT8	PxMD-PNKD	Paroxysmal nonkinesigenic dyskinesia	AD
		PRRT2	DYT10	PxMD-PRRT2	Paroxysmal kinesigenic dyskinesia	AD
		SLC2A1	DYT18	PxMD-SLC2A1	Paroxysmal exertion-induced dyskinesia	AD
		ECHS1	Not assigned	PxMD-ECHS1	Paroxysmal exertion-induced dyskinesia	AR

c.207C>G mutation in sepiapterin reductase causes autosomal dominant dopa-responsive dystonia

OPEN

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ABSTRACT

Objective: To elucidate the genetic cause of an Egyptian family with dopa-responsive dystonia (DRD), a childhood-onset dystonia, responding therapeutically to levodopa, which is caused by mutations in various genes.

Methods: Rare variants in all coding exons of *GCH1* [illegible] sequencing was applied for 1 unaffected and 2 [illegible] functional consequences of detected genetic vari[illegible] determined by high-performance liquid chromatog[illegible]

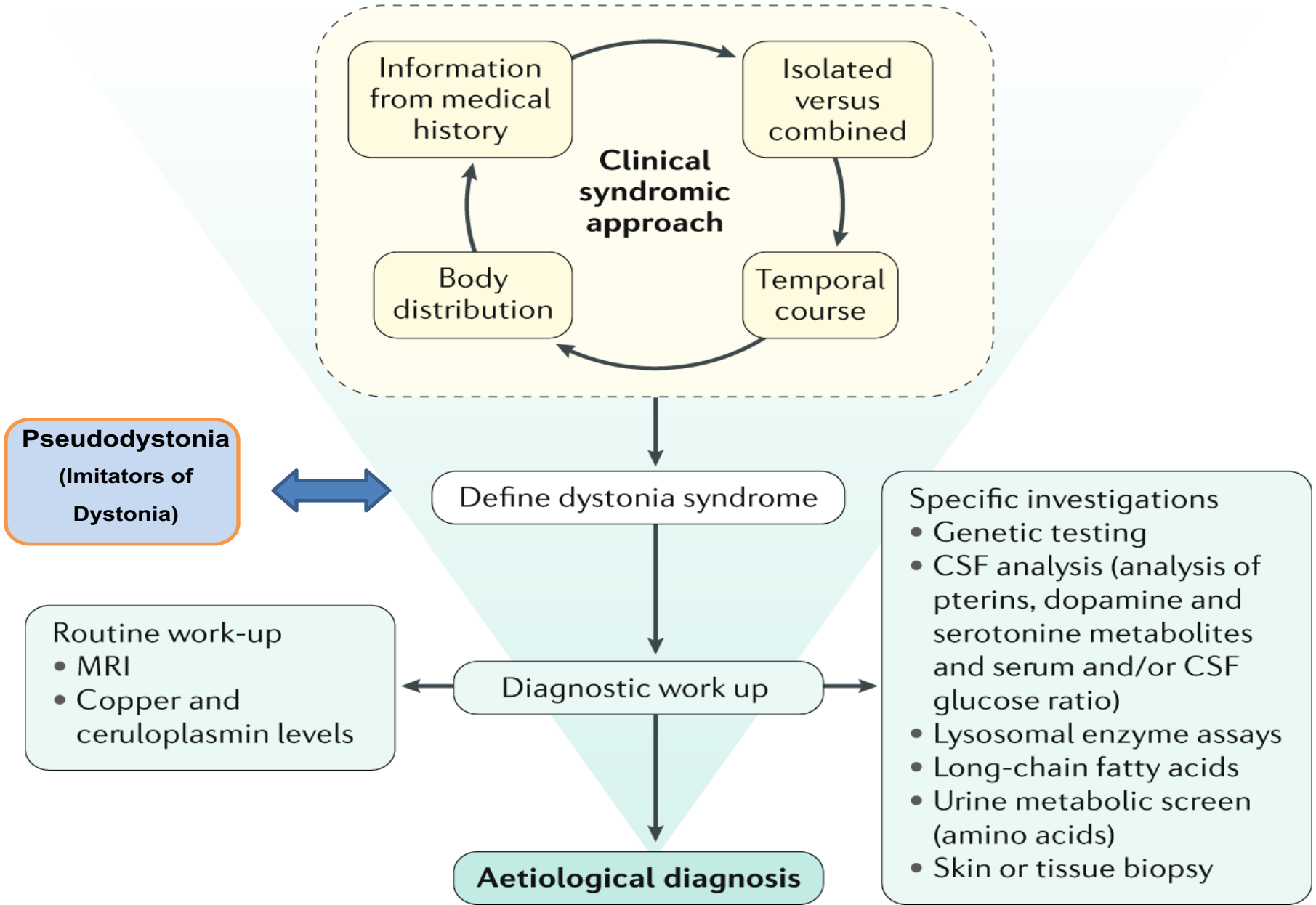
Results: A heterozygous rare nonsynonymous vari[illegible] c.207C>G, p.Asp69Glu) was found in all affecte[illegible] sepiapterin were above the standard of normal co[illegible] ing functional biochemical consequences of the mu[illegible] the tetrahydrobiopterin pathway, required for levod[illegible] variant in dihydrofolate reductase (*DHFR*, rs709[illegible] significantly stronger associated with the biochemical abnormality and the clinical disease state as opposed to 1 variant only.

Conclusions: The rare *SPR* mutation can cause autosomal dominant DRD with incomplete penetrance. The common *DHFR* variant might have synergistic effects on production of tetrahydrobiopterin and levodopa, thereby increasing penetrance. *Neurol Genet* 2017;3:e197; doi: 10.1212/NXG.0000000000000197

Paroxysmal Dyskinesias

	Paroxysmal kinesigenic choreoathetosis (PKD)	Paroxysmal Non kinesigenic choreoathetosis (PNKD)	Paroxysmal exercise-induced dystonia (PED)
Duration	Very brief	0.5-1 hr	2 min – 2 hrs
Triggering factors	Sudden movements	Alcohol, coffee, tobacco, emotions, fatigue, hunger	Prolonged or sustained exercise
Age of onset	7-15 yrs	Infancy - childhood	2 – 30 yrs
Treatment	carbamazepine	Benzodiazepine Anticonvulsants Acetazolamide	Gabapentin L-dopa
Gene	PRRT2 (Chr 16p11)	MR1 (Chr 2q35), KCNMA1 (10q22)	SLC2A1 (1p34.2)

Diagnostic Approach for Dystonia *(Balint et al, 2018)*



Management of Dystonia

Pharmacological Therapies

ABCD

Anticholinergics

Baclofen

Clonazepam

Dopamine-related medications

Botulinum Toxin Injection

Surgical Interventions

DBS

Ablative Surgeries

Dorsal Rhizotomy

Termsarasab et al.Medical treatment of dystonia. *J Clin Mov Disord* 2016

International Journal of Neuroscience

Outcome of Pallidal Stimulation of Idiopathic Generalized Dystonia with Predominant Mobile Truncal Dystonia: Cases Report

Ali S Shalash, Zeiad Y Fayed, Eman Hamid, Hisham Radwan, Mohamed A Nada, Mohammed Eid & Walid A Abdel Ghany

Table 1 Demographic and clinical data of patients with truncal dystonia underwent pallidal stimulation

	Patient 1	Patient 2	Patient 3
Gender	female	female	male
Age at surgery (years)	46 (2017)	20 (2014)	15 (2018)
Age of disease onset (years)	45	10	25
Duration (at surgery) (years)	1	10	12
Clinical presentation	dystonia started at right shoulder and arm, followed by trunk (on action), and generalized gradually, with severe mobile (tremulous) truncal dystonia; camp-tocormia, lateral tilt, twisting	Dystonia started at right upper limb on action, that generalized gradually with mobile (tremulous) truncal dystonia; camptocormia, lateral tilt	Dystonia started at left lower limb, mobile (tremulous) generalized, truncal dystonia; camptocormia, lateral tilt.
Pre-BFMDRS Total/trunk	57.5/16	66/16	84/16
Post-BFMDRS Total/trunk	3/2	5/1	16/2
Percentage of Improvement Total/Trunk	94.78/87.5%	92.4/93.75%	80.95/87.5%
Dystonia Disability Scale-Preop	20	25	24
Dystonia Disability Scale-Post-op	1	4	12
Dystonia Disability Scale-%improvement	95%	84%	50%
Last follow-up (years)	3	6	1.5
Current (Best) DBS setting			
Right	(C+2-) 3.8v/90us/130 hz	(C+0-) 2.7v/60us/130 hz	(C+2-) 3.8v/120us/130hz
Left	(C+10-) 3.9v/90us/130hz	(C+8-) 3.2v/60us/130hz	(C+10-) 3.8v/120us/130hz

CHOREA

- Irregular rapid, low amplitude, brief movements of extremities & face.
- Semi purposeful or apparently purposeful.
- Severe cases → obvious movement of the hand, feet, face.
- ± Facial grimacing, eye brow movement and respiratory noises.
- Increase by stress and disappear during sleep.
- ± Athetosis (slower, distal, writhing and sinuous).

- Ballismus: high amplitude movement of a limb in a flinging or flailing motion, including the most proximal segments

- Motor impersistence: difficulty sustaining ongoing movement e.g. inability to maintain forced eye closure, or protrude the tongue for long periods.

Special Signs

1. Tongue sign: patient cannot keep his tongue protruded outside mouth.
2. Boat hands or scaphoid hands
3. Pronator signs.
4. Dancing gait.

Causes of Choreic Syndromes

GENETIC CHOREAS

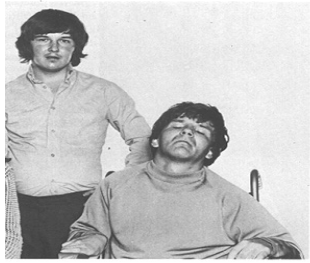
1. Huntington's disease (AD).
2. Huntington's disease-like 2 (AD).
3. Dentatorubropallidoluysian atrophy (DRPLA)
4. Neuroacanthocytosis
5. Ataxia teleangiectasia
6. Benign hereditary chorea (AD)
7. Spinocerebellar ataxia (types 2, 3, or 17)
8. Paroxysmal kinesigenic choreoathetosis

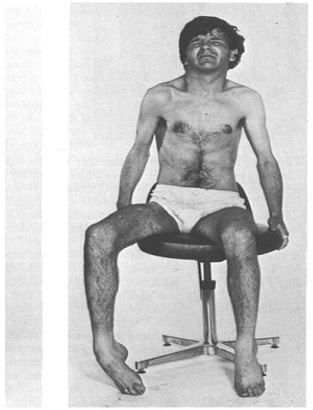
AQUIRED CHOREAS (non-genetic)

1. **Structural basal-ganglia lesions:** stroke
2. **Parainfectious & autoimmune disorders:** Sydenham's chorea, SLE.
3. **Infectious chorea**
4. **Metabolic or toxic encephalopathies**
5. **Drug induced**
6. **Functional forms.**

Huntington's Disease

- AD causing choreic and mental changes.
- Caused by unstable trinucleotide CAG repeat expansion in chromosome 4p, HTT gene, which encodes a protein (Huntingtin) widely expressed in neuronal and other tissues.
- Age: 30 – 55 years around 40 years.
- < 20 years → juvenile HD (Westphal variant) 5 %.
- >60 years → elderly onset disease 25%.
- Gradual, progressive, 2/3 starts by motor symptom (chorea).

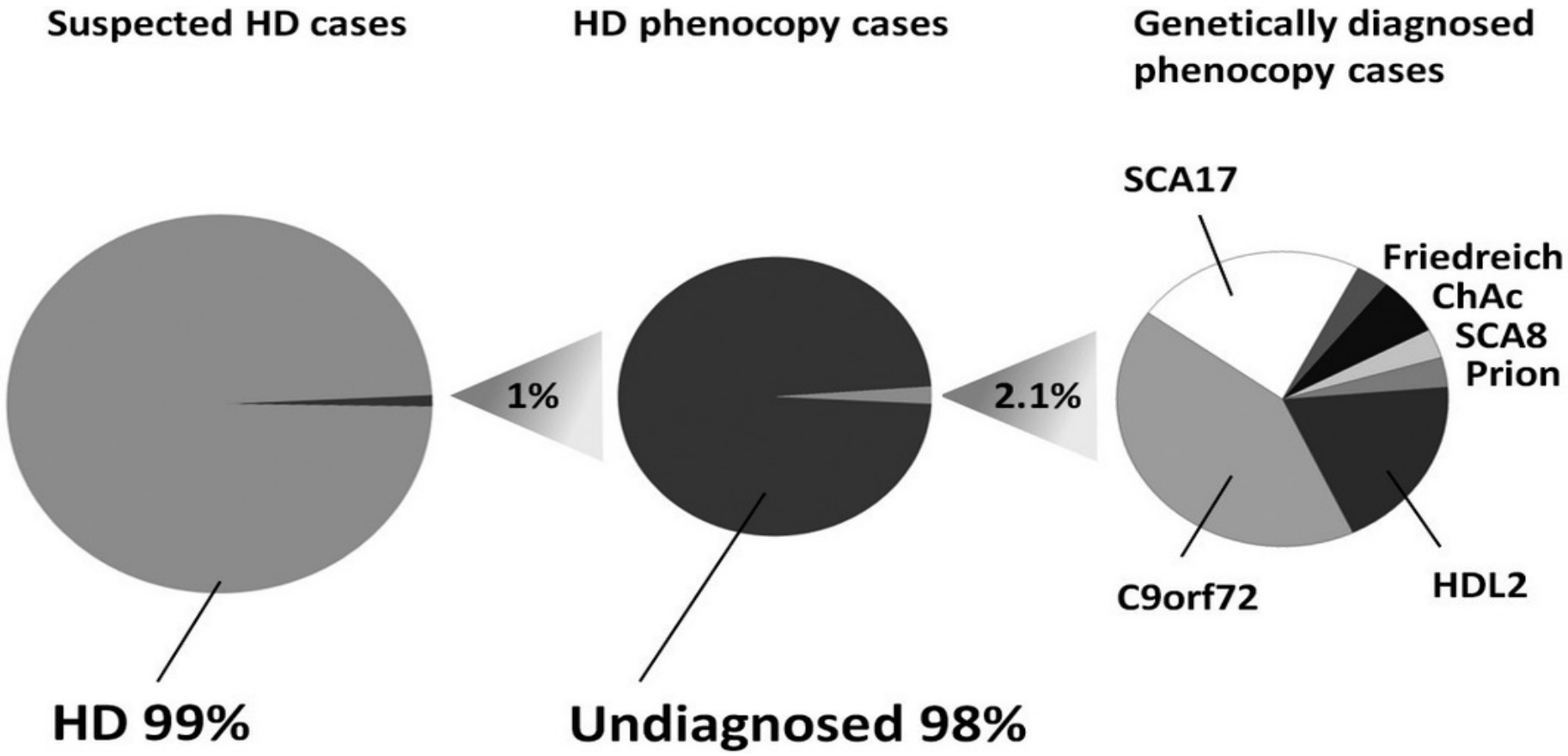




- **Motor**
  - Chorea
  - Parkinsonism
  - Dystonia
  - Myoclonus
  - Motor impersistence
  - Gait disorder

- **Psychiatric**
  - Personality
  - Affective; depression
  - Obsessive compulsive
  - Psychosis (rare)
- **Cognitive**
  - Executive dysfunction
  - Dementia

Huntington Disease Phenocopies



Schneider & Bird, Mov Disord Clin Pract. 2016

RESEARCH ARTICLE

AMERICAN JOURNAL OF
medical genetics
Neuropsychiatric Genetics

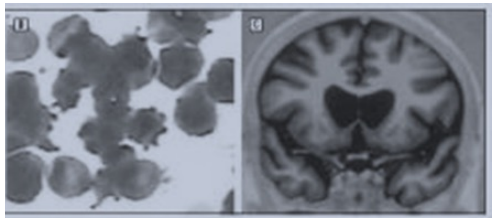
Junctophilin 3 (*JPH3*) Expansion Mutations Causing Huntington Disease Like 2 (HDL2) are Common in South African Patients with African Ancestry and a Huntington Disease Phenotype

Amanda Krause,[1,2]* Claire Mitchell,[1] Fahmida Essop,[1,2] Susan Tager,[3,4] James Temlett,[3,5] Giovanni Stevanin,[6,7] Christopher Ross,[8] Dobrila Rudnicki,[9] and Russell Margolis[10]

origin of the *JPH3* mutation. In a sample of unrelated South African individuals referred for diagnostic HD testing, 62% (106/171) of white patients compared to only 36% (47/130) of black patients had an expansion in *HTT*. However, 15% (20/130) of black South African patients and no white patients (0/171) had an expansion in *JPH3*, confirming the diagnosis of Huntington disease like 2 (HDL2). Individuals with HDL2 share many clinical features with individuals with HD and are clinically indistinguishable in many cases, although the average age of onset and diagnosis in HDL2 is 5 years later than HD and individual clinical features may be more prominent. HDL2 mutations contribute significantly to the HD phenotype in South Africans with African ancestry. *JPH3* haplotype studies in 31 families, mainly from South Africa and North America, provide evidence for a founder mutation and support a common African origin for all HDL2 patients. Molecular testing in individuals with an HD phenotype and African ancestry should include testing routinely for *JPH3* mutations.

Workup of Chorea

1. Routine blood work, Thyroid studies, ESR, ANA, antiphospholipid antibodies, ASO titers…
2. Test for thyroid function, renal and liver function, electrolytes, erythrocyte sedimentation rate, antinuclear antibodies, anti double-stranded DNA antibodies, ↑↑ CPK, anticardiolipin antibodies, and lupus anticoagulant.
3. Test for acanthocytes in peripheral fresh blood film. perform three assays.
4. Perform brain MRI.
5. Genetic test: for Huntington disease. If the latter genetic test is negative, consider spinocerebellar ataxia type 17 and C9orf72 in white individuals, and Huntington disease-like syndrome type 2 in subjects with black African ancestry.



Treatment of Chorea

- Treat underlying acquired cause: autoimmune, WD.
- No protective treatment
- **Symptomatic treatment of chorea:**
    1. Dopamine receptor blockade
        - Typical neuroleptics—caution!
        - “Atypical” neuroleptics: tiapride, olanzapine, and risperidone Presynaptic dopamine depletion: Tetrabenazine 25-100 mg/day, deutetrabenazine (HD), valbenazine (tardive dyskinesia)
    2. Glutamate antagonism: Amantadine
    3. GABA-ergic: Valproic acid.
- Treat associated symptoms: psychiatric, seizures
- Botulinum toxin, DBS for certain cases

Tic Disorders

Definition: Rapid, non-rhythmic, stereotyped involuntary movements usually affecting the face, head, or UL.

- More semi purposeful, which may be:
- Simple or complex, motor or vocal.
- Acute, subacute or chronic.
- Brief or sustained "dystonic tics."
- **Motor tics:**
- *Simple motor tics:* blinking, head jerking, shrugging shoulder, grimacing.
- *Complex motor tics;* picking at the body or object, gestures, rubbing or manipulative movements.
- **Vocal tics:** simple (noises, cough, sniffs) or complex (words, phrases).
- Waxing and waning, transient remissions.
- Persist during sleep (all stages)

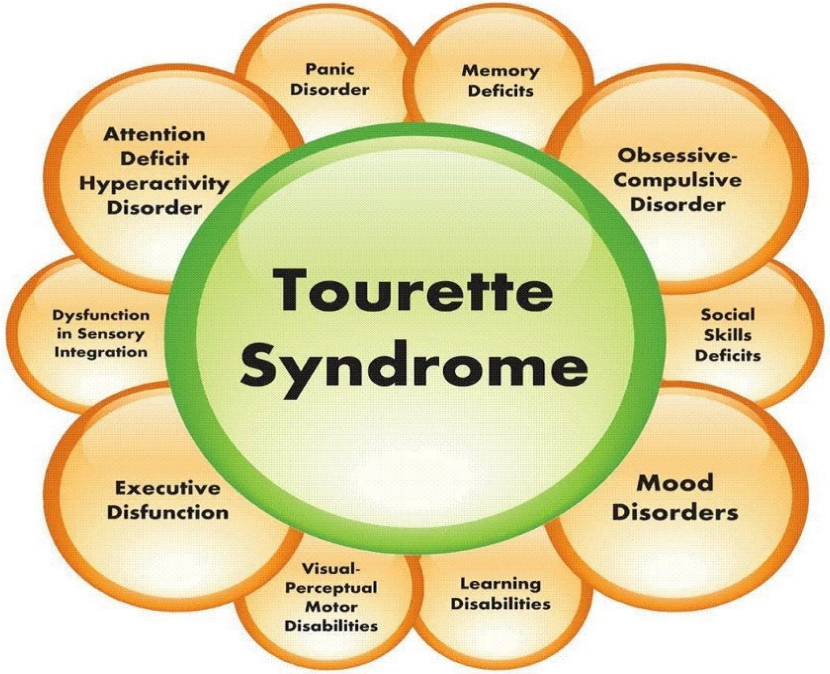
Causes of Tics

Primary	Secondary
Transient tic disorder (< 1 year)	a) HD, choreo-acanthocytosis
Chronic motor tic disorder .	b) Drugs;
Chronic vocal tic disorder	
Tourette's syndrome	c) Encephalitis, CJD, PANDAS
	d) PD, PSP.
	e) Rett's syndrome.
	f) Focal BG lesion.

Tourette's Syndrome

DSM-IV Diagnostic Criteria

- Both multiple **motor** and one or more **vocal** tics have been present at some time during the illness, although not necessarily concurrently.
- The tics occur many times a day (usually in bouts) nearly every day or intermittently throughout a period of more than 1 year, and during this period there was never a tic-free period of more than 3 consecutive months.
- The onset is before age 18 years.
- The disturbance is not due to the direct physiological effects of a substance (e.g., stimulants) or a general medical condition (e.g., Huntington's disease or postviral encephalitis).

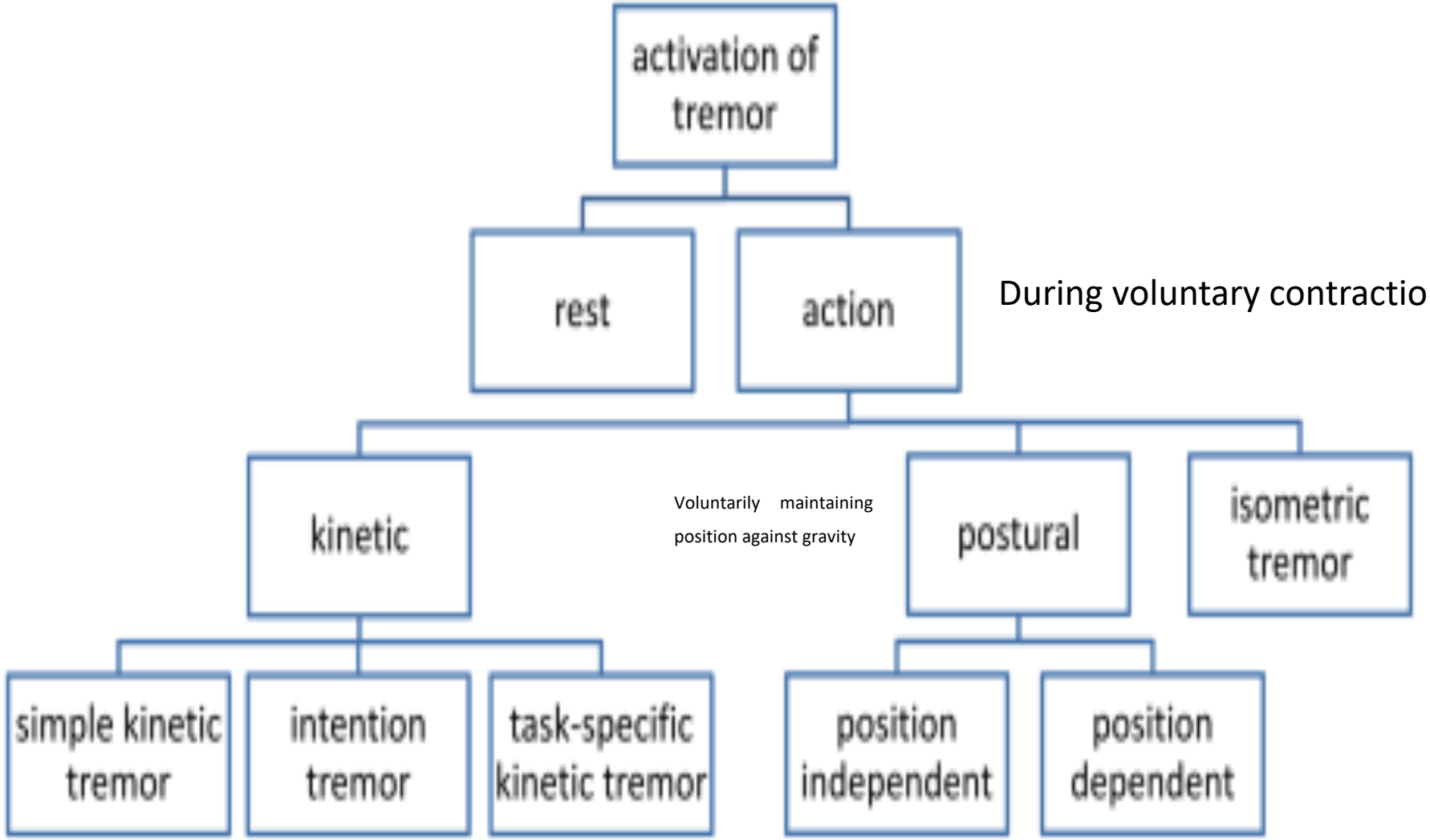


Treatment of Tic Disorders

1. Education; Patient, family, and school
2. Counseling for family and patient
3. Relaxation therapy
4. Supportive therapy
5. Habit Reversal Therapy
6. Pharmacological: α-2 agonists (Clonidine), neuroleptics
7. Treatment of comorbidities.

Consensus Statement on the classification of

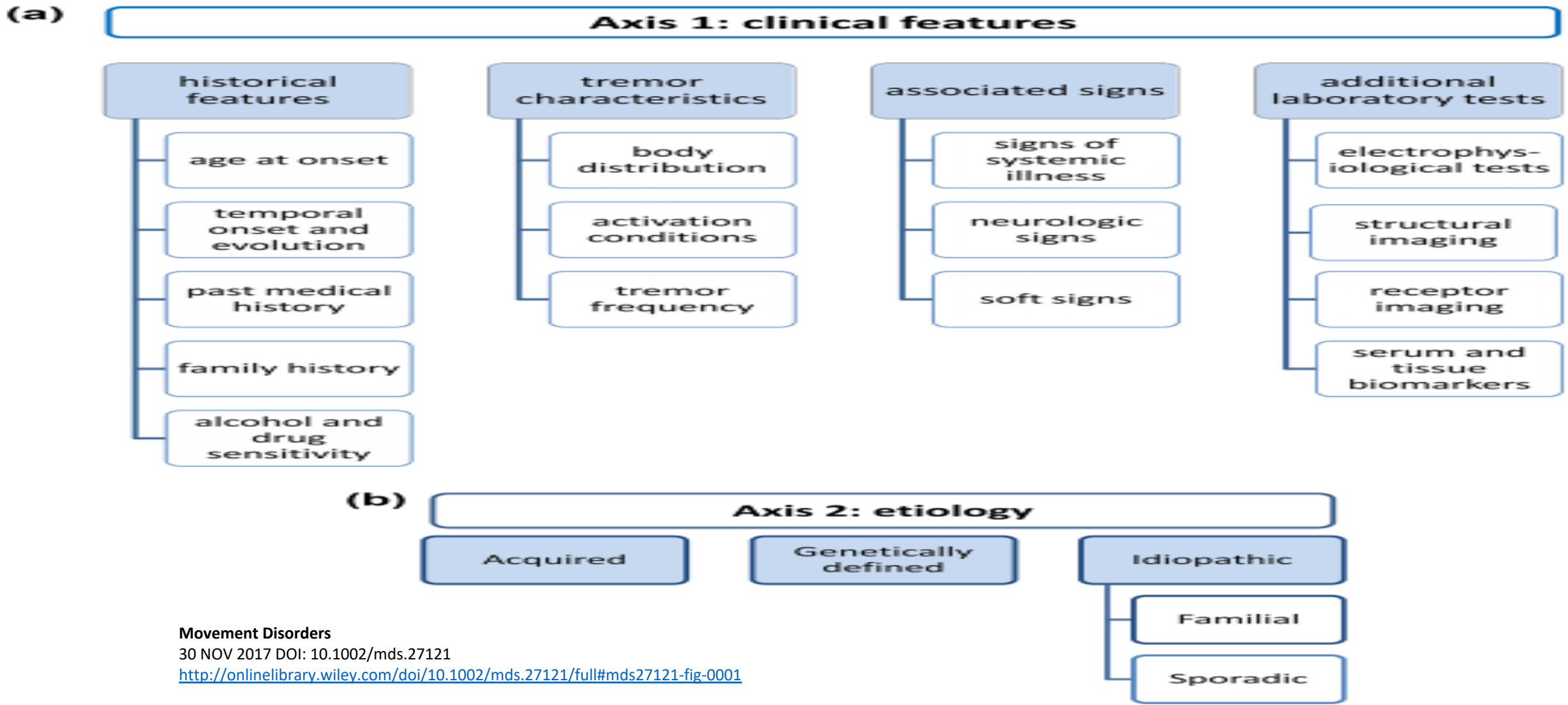
tremors, ***the task force on tremor of the MDS***



During voluntary contraction of muscle

Movement Disorders
30 NOV 2017 DOI: 10.1002/mds.27121
http://onlinelibrary.wiley.com/doi/10.1002/mds.27121/full#mds27121-fig-0002

Consensus Statement on the classification of tremors, *the task force on tremor of the MDS*



Movement Disorders
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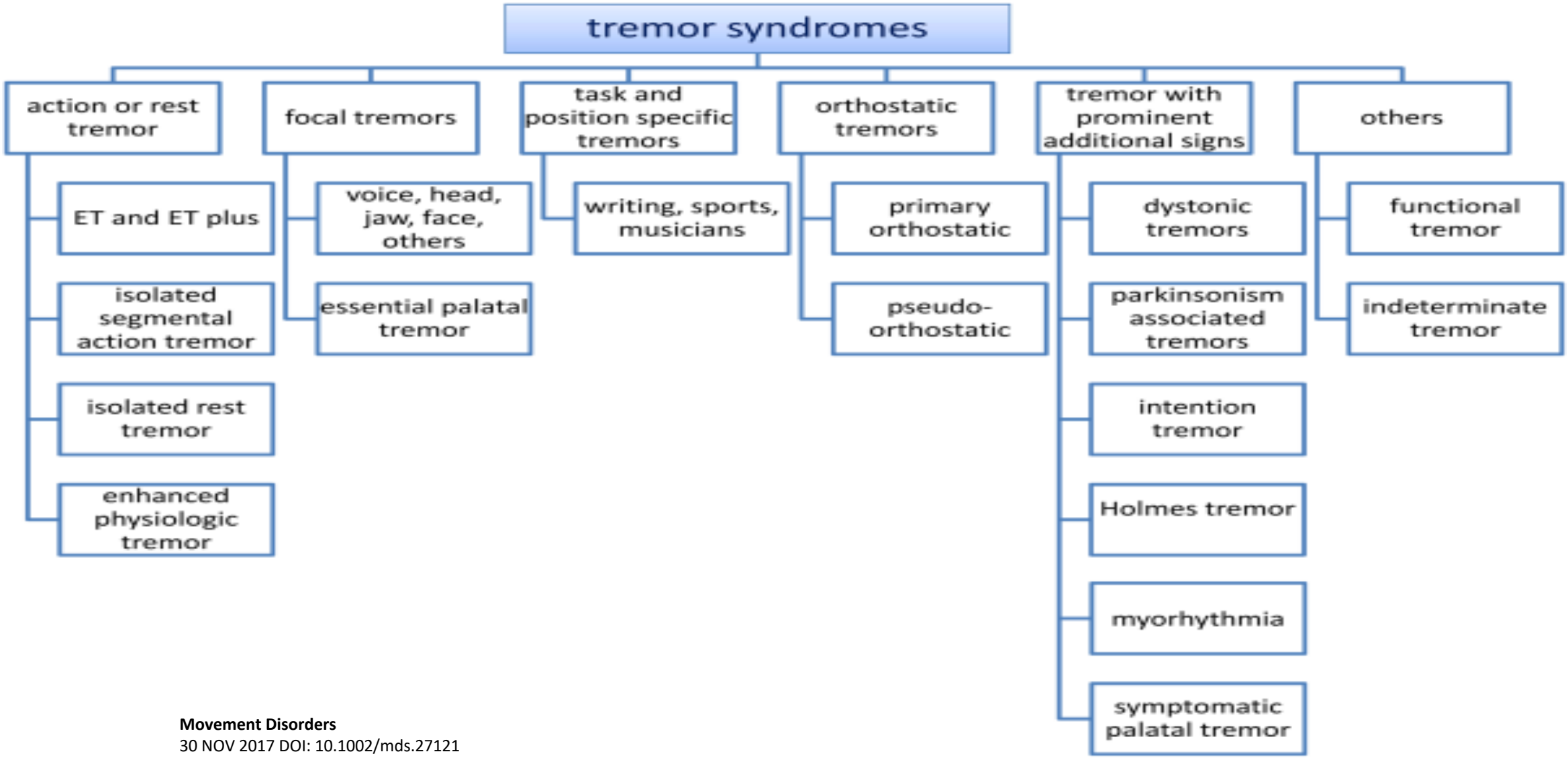
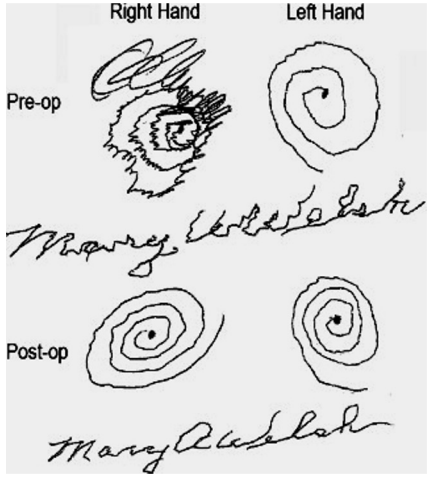


Movement Disorders
30 NOV 2017 DOI: 10.1002/mds.27121
http://onlinelibrary.wiley.com/doi/10.1002/mds.27121/full#mds27121-fig-0003

Essential Tremor (ET):

- Sporadic or familial AD, insidious, and progressive.
- Bimodal onset; late adolescence and older adulthood.
- Mixture of kinetic and (>) postural tremor, 4 -12 hz.
- Bilateral roughly symmetric or mild asymmetric.
- Start in UL (95%), head, face, voice….
- ± mild intentional component, ± mild gait ataxia.
- No other neurological deficits (except Froment's sign).
- Isolated focal or task specific (vs. dystonic).



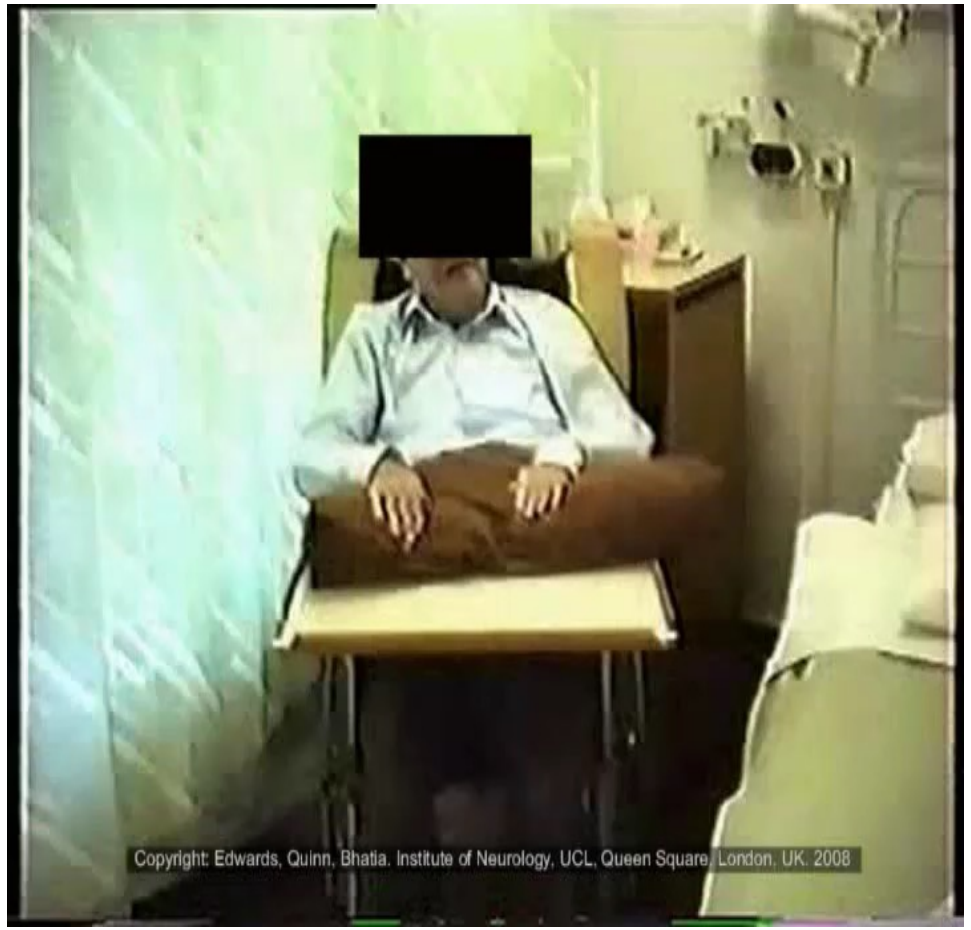
Treatment of ET

1. *Propranolol* (up to 320 mg/day), and other B blockers.
2. *Primidone* (up to 250 mg three times daily).
3. *Gabapentin* (1200 to 3600 mg/day), and *topiramate.*
4. Mirtazapine, alprazolam, Phenobarbital, Clonazepam
5. *Botulinum toxin* for task-specific, severe isolated head, or dystonic tremors.
6. Wearing wrist weights while eating, drinking from a heavier mug, using a fat pen rather than a thin one, and using heavier utensils
7. *Surgical*: Vim thalamotomy or Vim DBS.

MYOCLONUS

- **Classification of Myoclonus:** The essential feature is the sudden, brief, and **shock-like** movement.

Clinical Presentation						
Distribution	1.	Focal	1.	Segmental		
	1.	Multifocal	1.	Generalized		
Relation to activity:						
	a)	Spontaneous	a)	Action	a)	Reflex
Pattern	I.	Rhythmic	I.	Irregular		
	I.	Repetitive or oscillatory				
Etiology	1.	physiological	1.	Essential		
	1.	Epileptic	1.	Symptomatic		
	1.	Psychogenic				
Neurophysiologic origin						
	1)	Cortical	1)	Subcortical		
	1)	Spinal	1)	Peripheral nerve or root		



- Positive Myoclonus: muscle contractions.
- Negative Myoclonus; interruptions of tonic muscle activity = asterixis= flapping tremor.
- **Treatement:** piracetam, levetiracetam, valproic acid and clonazepam

Causes of Myoclonus

Generalised Myoclonus:

a) **Essential myoclonus**: Non-progressive condition in which myoclonus is only or most important neurological symptom and sign.

b) **Progressive myoclonic encephalopathies (PME):**

▪ *Myoclonus (with or without seizures) is part of a progressive encephalopathy.*

e.g., mitochondrial encephalomyopathy (esp. MERFF); Creutzfeldt–Jacob disease, Alzheimer's disease; and *Metabolic myoclonus* (e.g. uraemia, hepatic failure,

a) **Static myoclonic encephalopathies**: obvious myoclonus occurs after some acute and now static cerebral insult, e.g. postanoxic action myoclonus (Lance–Adams syndrome).

b) **Myoclonic epilepsies:** epilepsy is the main problem, but myoclonus is present.

Focal Myoclonus: *myoclonus is restricted to one small discrete part of the body.* → Spinal myoclonus, Hemifacial spasm.

Wilson's Disease

Onset: the ages of 5 and 35 years (mean 13 years)

Hepatic
18-84%

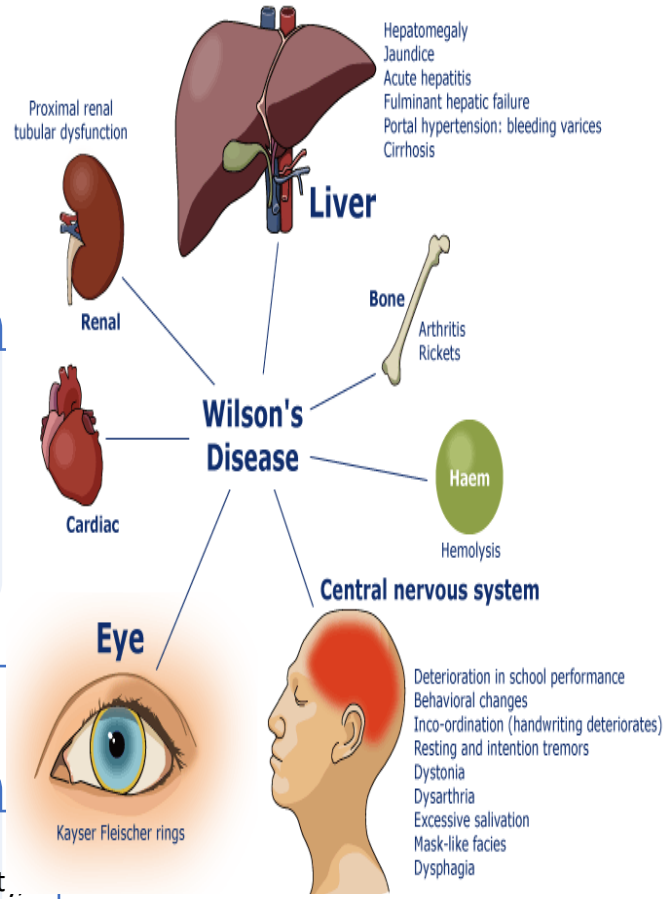
Asymptomatic
Acute hepatitis
Chronic hepatitis
Cirrhosis

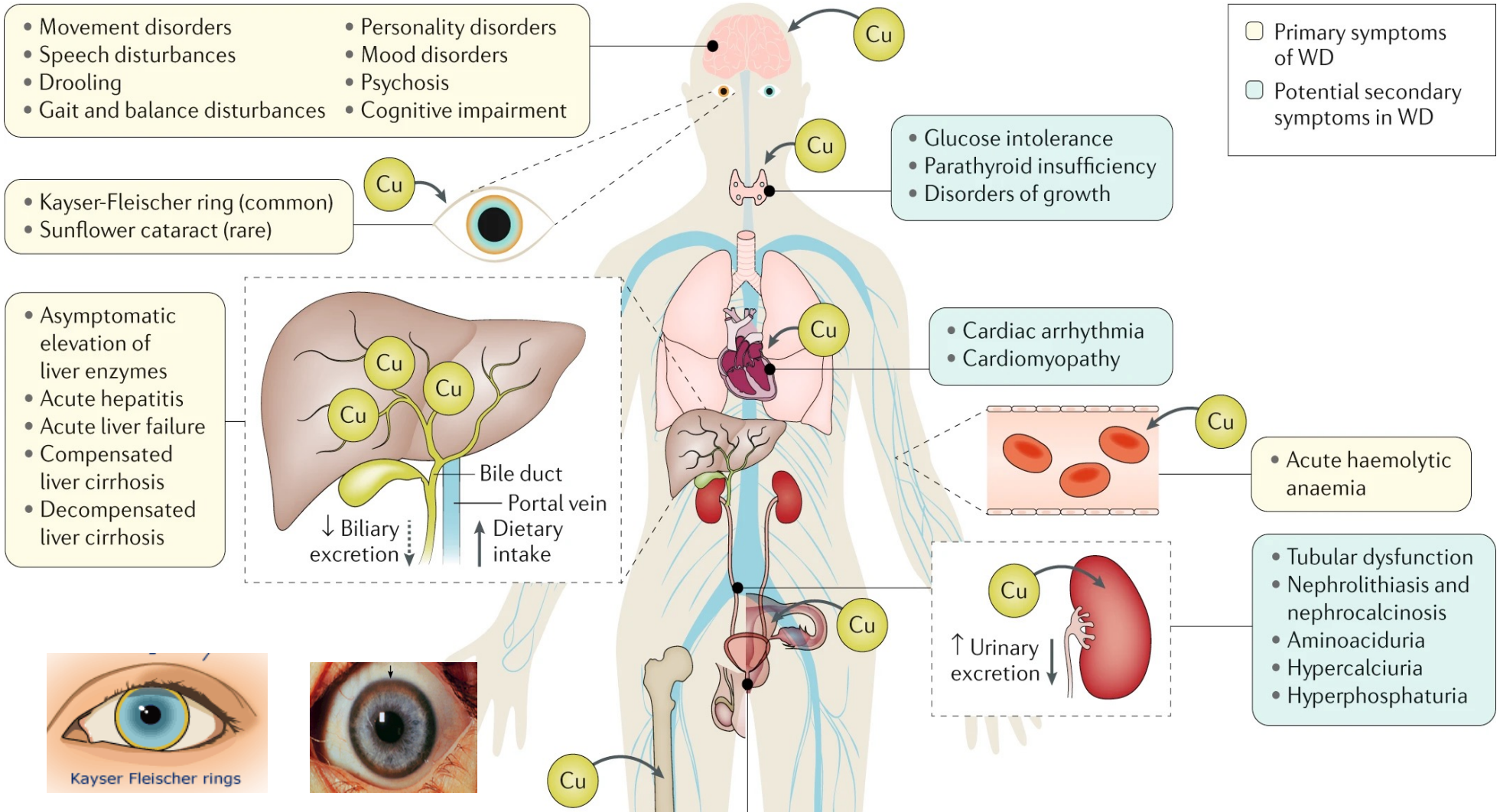
Neurological
18-73%
AOO: 20-30 yrs

Dysarthria, dystonia, tremor, parkinsonism, chorea, or myoclonus, ataxia

Psychiatric
10-100%

Emotional lability, hypersexuality, impulse control disorders, psychosis, depression, or attempted suicide- Drop in scholastic grades or work performance





Członkowska et al, Nature Reviews 2018

Prominent Extensor Truncal Dystonia in Egyptian Patients With Wilson's Disease

TABLE 1. Clinical and MRI brain characteristics in patients with Wilson's disease with and without dystonia[a]

Characteristic	Mean ± SD [Range]/No. of Patients (%)			P^b	Significance
	Patients With WD: Total	WD With Dystonia	WD Without Dystonia		
No. of patients	22 (100)	14 (63.6)	8 (36.4)		
Age, y	19.9 ± 6.3 [11–38]	20.8 ± 6.98 [11–38]	18.4 ± 4.95 [13–25]	0.358	NS
Sex: Male/female	14/8	9/5	5/3	0.642	NS
No. with positive family history	13 (59.1)	8 (57.1)	5 (62.5)	0.546	NS
Duration of illness, y	7.4 ± 7.2 [0.5–30]	8.3 ± 8.03 [0.5–30]	6.1 ± 5.97 [0.5–16]	0.489	NS
Age of onset, y	12.5 ± 4.1 [8–25]	12.5 ± 4.74 [8–25]	12.6 ± 3.33 [8–14]	0.943	NS
Neurological presentation	9 (40.9)	5 (35.7)	4 (50)	0.239	NS
Parkinsonism	13 (59.1)	9 (40.9)	4 (50)	0.662	NS
KFR	17 (77.3)	12 (85.7)	5 (62.5)	0.309	NS
Dysarthria severity	1.727 ± 1.316	2.288 ± 1.204	0.750 ± 0.886	0.007	S
Walking impairment	1.409 ± 1.141	1.857 ± 1.167	0.625 ± 0.517	0.006	S
BFMDRS score		36.9 ± 29.94			
No. with extensor truncal dystonia	11 (50)	11 (87.6)			
Score		8.9 ± 3.73			
MRI brain abnormality					
Bilateral lentiform lesions	19 (59.1)	12 (85.7)	7 (87.5)	0.709	NS
Bilateral lentiform and caudate lesions	15 (68.2)	10 (71.4)	5 (62.5)	0.510	NS
Bilateral GP lesions	10 (45.5)	7 (50)	3 (37.5)	0.454	NS
Brainstem lesions	6 (27.3)	5 (35.7)	1 (12.5)	0.255	NS
Bilateral thalamic lesions	5 (22.7)	3 (21.4)	2 (25)	0.620	NS
Treatment					
Compliant patients	9 (40.9)	5 (35.7)	4 (50)	0.616	NS
D-penicillamine, mg	403.41 ± 308.44; n = 16	330.36 ± 257.61; n = 10	531.25 ± 364.43; n = 6	0.146	NS
Zinc sulfate, mg	100 ± 76.38; n = 16	108.93 ± 88.04; n = 10	84.38 ± 51.65; n = 6	0.419	NS

WD gene, ATP7B

Shalash et al Mov Dis 2014

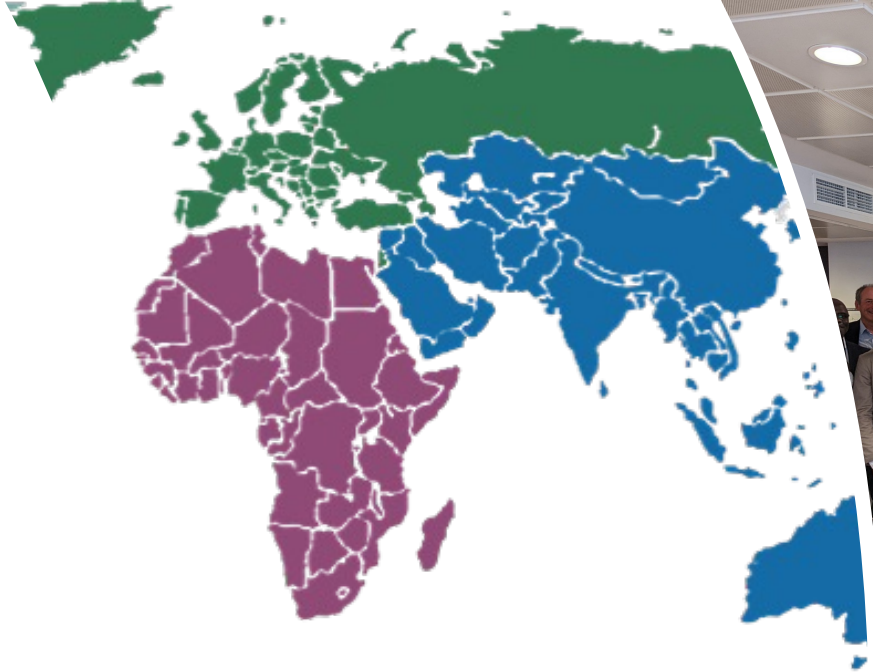
CHALLENGES

1. Awareness, early diagnosis, advocacy.
2. Lack of basic medications (unavailable and unaffordable)
3. Lack of different medications for motor, LD-induced complications, non-motor
4. Dealing with old medications
5. Shortage of Neurologists, Nurses, neurosurgeons,…..
6. Lack of trainig and education
7. Unavailability of functional surgeries and advanced therapies
8. Barriers of Telemedicine
9. Need for comprehensive care for PD patients
10. Limited resources for research

RECOMMENDATIONS

1. Ealry diagnosis: awarness, screening, acess to health care
2. Contact stakholders, provision of affordable drugs.
3. Promote training of neurologists, practiciners, and alied health professionals.
4. Improve knolwedge of available tools.
5. Facilitate access for PD treatment.
6. Use of available and afordable alternatives.
7. Non-pharmacological interventions.
8. PD supportive groups.
9. Overcome telemedicine barriers.
10. Comprehensive care for PD patients.

The International Parkinson and Movement Disorder Society (MDS)



- Task Force on Africa
- African Steering Committee (Created in 2017)
- African MDS education committee 2019
- MDS Africa Section 2021

Membership Benefits

Learn more about all MDS member benefits at: **www.movementdisorders.org/benefits**

Delegates from the African Section can apply for a FREE No-Fee Membership with MDS

Video Library

Over 2,000+ searchable videos

MDS Journals

MDS peer-reviewed Journals

Moving Along

Quarterly newsletter

Member Directory

Online-only directory

Reduced Registration Fees

Discounted rates for live courses and MDS Congresses

E-Learning

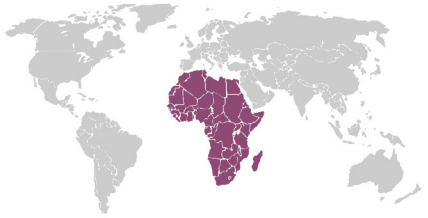
Coffee Break CME, Journal CME, Fundamentals Course Series, Interactive Courses and more

Voting Rights

Regular and Waived Dues Members are eligible to vote in MDS leadership elections

Rating Scales

MDS-UPDRS & UDysRS Online Training Programs



MDS-AS Online Regional Course

International Parkinson and Movement Disorder Society African Section

Upcoming MDS-Africa Education

MDS-AS African Multicenter Grand Rounds 5-part Series
Session 4 (Functional Movement Disorders): November 11 , 2022 at 16:00 EET

IN-PERSON MDS-Africa School for Young Neurologists
Tunis, Tunisia | December 1-3, 2022

Applications close September 2022

Registration is FREE for MDS Members. Non-members from the African region can apply for No-Fee Membership and register at no cost.

2020 MDS-AS Education

- 1 – In person MDS-AS Education Course
- 2 - MDS-AS Regional Online Courses
- 4 - Outreach Programs
- 1 – Developing World Education Programs (DWEP)
- 2 – MDS Supported/Endorsed Meetings

MDS-AS Online Regional Course

International Parkinson and Movement Disorder Society African Section

MDS Outreach Programs

"Does Your Program Need…"

- MDS Endorsement (use of MDS logo)?
- MDS Financial Support for your local Event?
- MDS Expert Recommendations / Honoraria Support?
- Technology Support through access to MDS Zoom Webinar Account?

Contact the MDS Secretariat to learn more at education@movementdisorders.org

Ain Shams Movement Disorders Group

AIN SHAMS UNIVERSITY
كلية الطب
Neurology Department

- Care
- Education
- Research

Thank You!


Enkosi
Ngiyabonga
شكراً
Amesege'nallo'
Zikomo Kwambiri
Siyabonga kakulu
Thank You
Asante sana
Ndatenda
Murakoze
N'itumezi
Masvita
Kea leboha
Zikomo
International Parkinson and
Movement Disorder Society