Ataxic disorders

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Introduction

- The term *ataxia is used by clinicians to denote a syndrome* of **imbalance** and **incoordination** involving gait and limbs, as well as speech; it usually indicates a disorder involving the cerebellum or its connections.

- Ataxia is a symptom, not a specific disease or diagnosis.

- Ataxia means poor coordination of movement.

Ataxia is derived from greek word

‘a’ - not

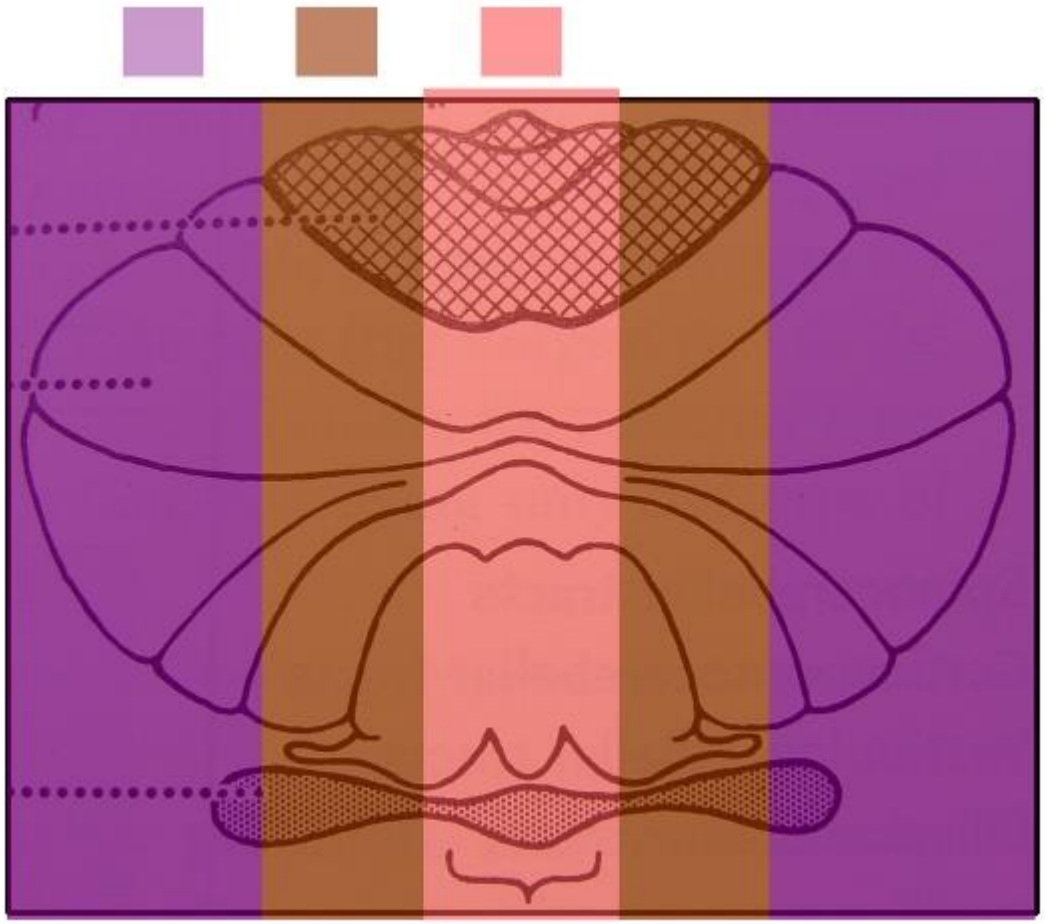
‘taxis’ - orderly

(not orderly/ not in order)

Introduction

- Ataxia can affect **coordination** of **fingers, hands, arms, speech** (dysarthria) and **eye movements** (nystagmus).

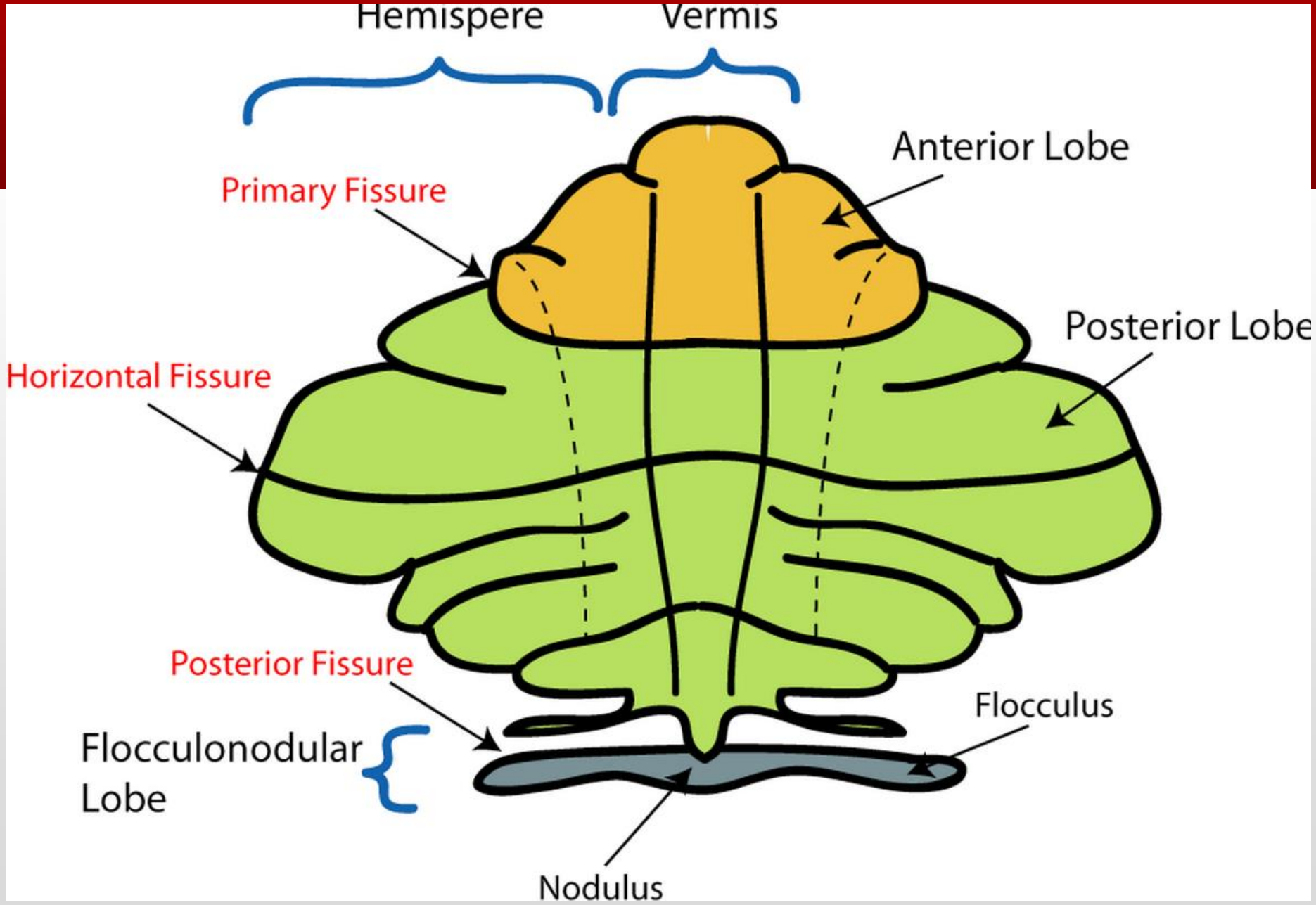
- Ataxia can also result from **disturbances of sensory input to the cerebellum**, especially proprioceptive input and also involvement of vestibular system.

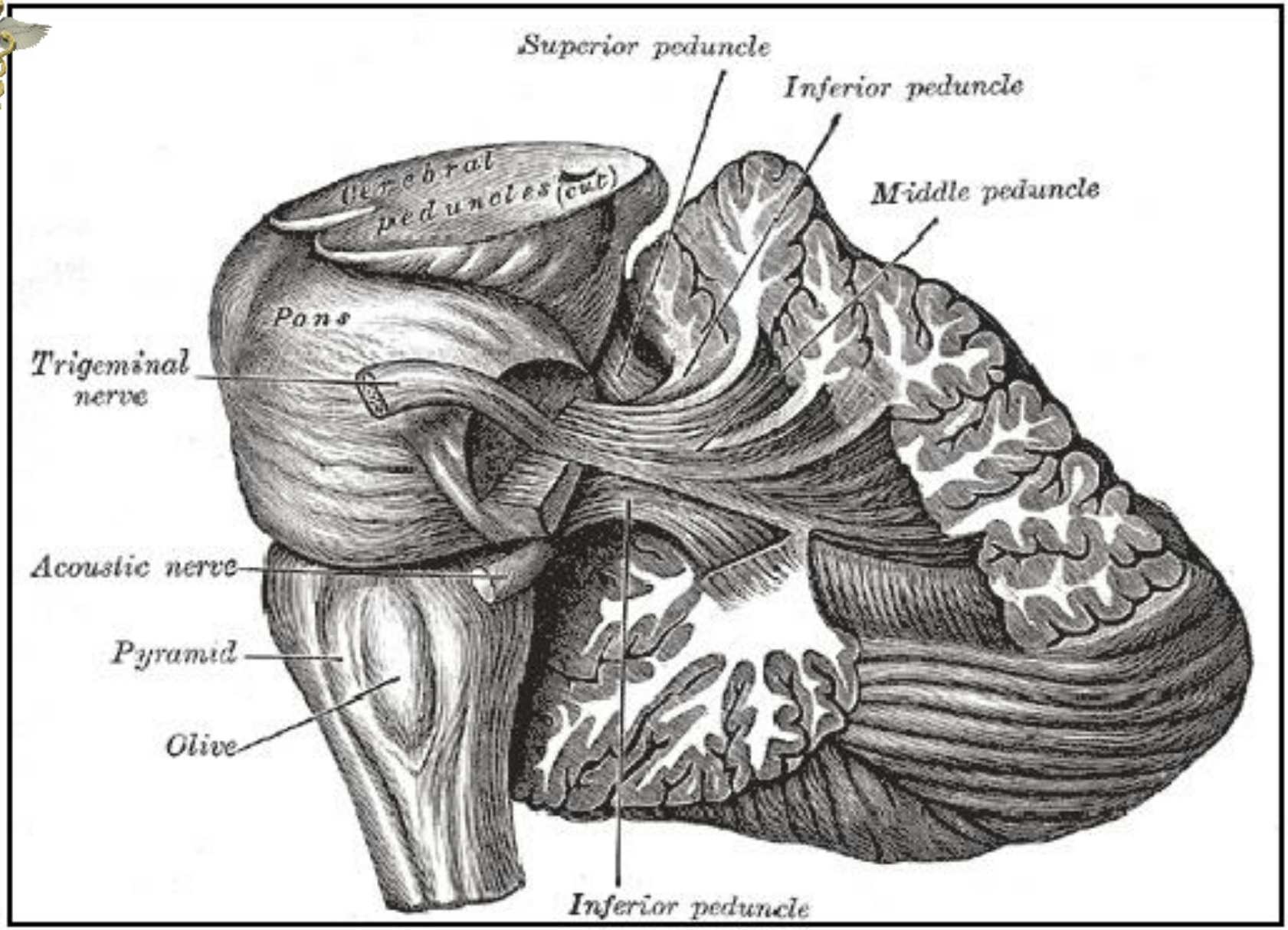


NEO CEREBELLUM

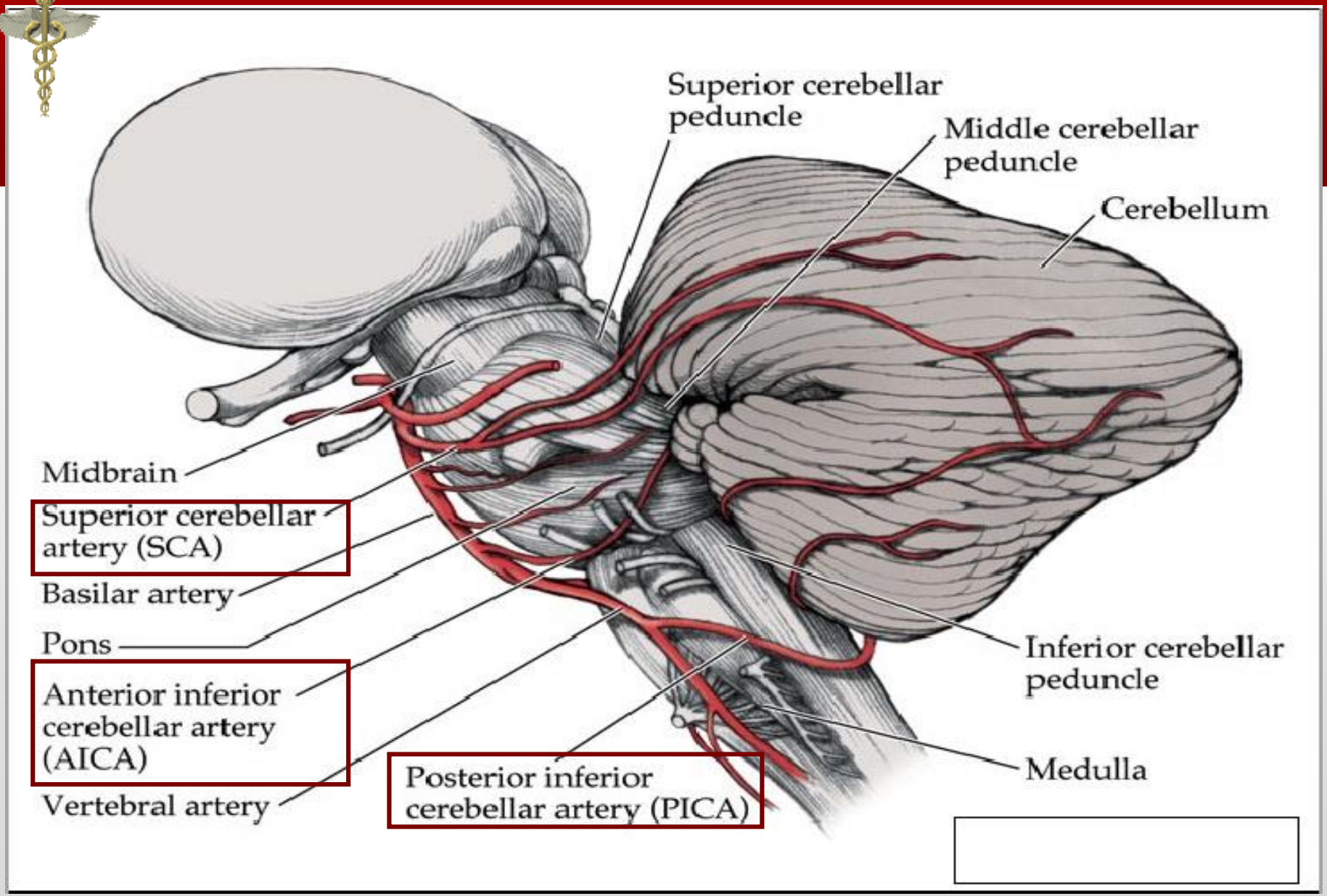
SPINO CEREBELLUM

FLOCCULONODULAR LOBE


Hemispere
Vermis
Anterior Lobe
Primary Fissure
Posterior Lobe
Horizontal Fissure
Posterior Fissure
Flocculus
Flocculonodular Lobe
Nodulus


Superior peduncle
Inferior peduncle
Middle peduncle
Cerebral peduncles (cut)
Pons
Trigeminal nerve
Acoustic nerve
Pyramid
Olive
Inferior peduncle

ARCHICEREBELLUM/ FLOCCULO NODULAR/ VESTIBULO CERELLUM	PALLEOCEREBELLUM/ SPINOCEREBELLUM/ VERMIS & PAR VERMIAN REGION	NEOCEREBELLUM/ CELEBELLAR HEMISPHERES/ PONTO CEREBELLUM
Eye movements, gross balance and orientation	Posture, Muscle tone, Axial muscle control, Locomotion	Coordinating movements, Fine motor control
Inferior cerebellar pudencle	Inferior/ middle/ superior pudencle	Middle/ superior pudencle


Superior cerebellar peduncle
Middle cerebellar peduncle
Cerebellum
Midbrain
Superior cerebellar artery (SCA)
Basilar artery
Pons
Anterior inferior cerebellar artery (AICA)
Vertebral artery
Posterior inferior cerebellar artery (PICA)
Inferior cerebellar peduncle
Medulla

ATAXIA

*"errors in the **RATE, RANGE, FORCE & DIRECTION** of movement"*

- GAIT ATAXIA
- TRUNCAL ATAXIA
- LIMB ATAXIA

CLASSIC FEATURES

Dyssynergia: results in jerky *decomposed* movements

- **Dysmetria**: inaccuracy in reaching target due to premature arrest of movement (hypometria) or overshoot the target (hypermetria)
- -**Dysdiadochokinesis**: irregularities of force, speed, and rhythm

Other features

Hypotonia: decrease in resistance to passive movement of muscles related to *depression of gamma motor neuron activity* (usually seen transiently in acute phase of cerebellar lesions), pendullar knee jerk.

Dysarthria: often *scanning* type with irregularities in tone, with words broken into syllables; often slow with occasional rapid portions ("explosive speech")

Other features

Ocular Motor Abnormalities:

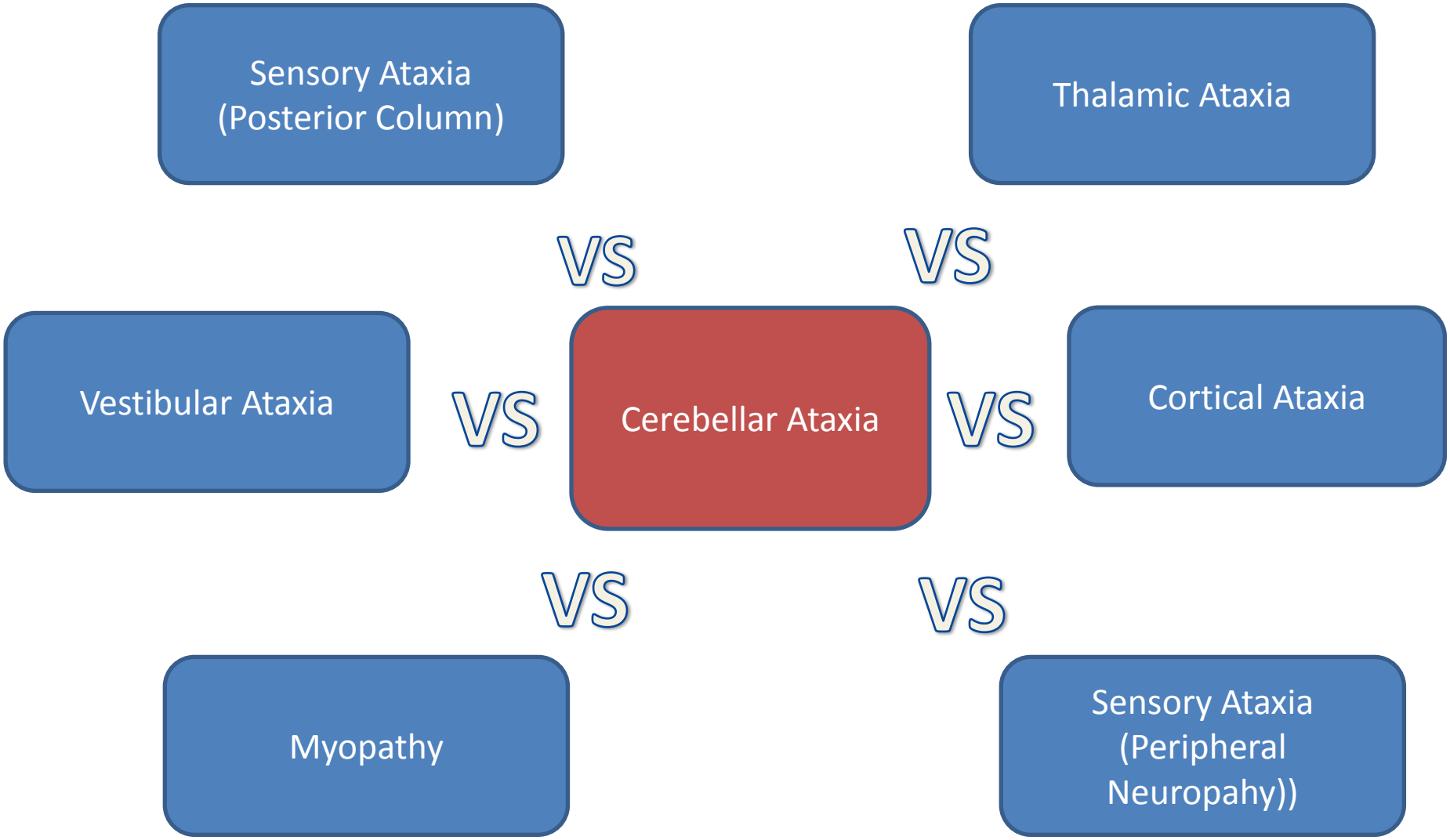
- usually if vestibular connections or flocculonodular lobe affected

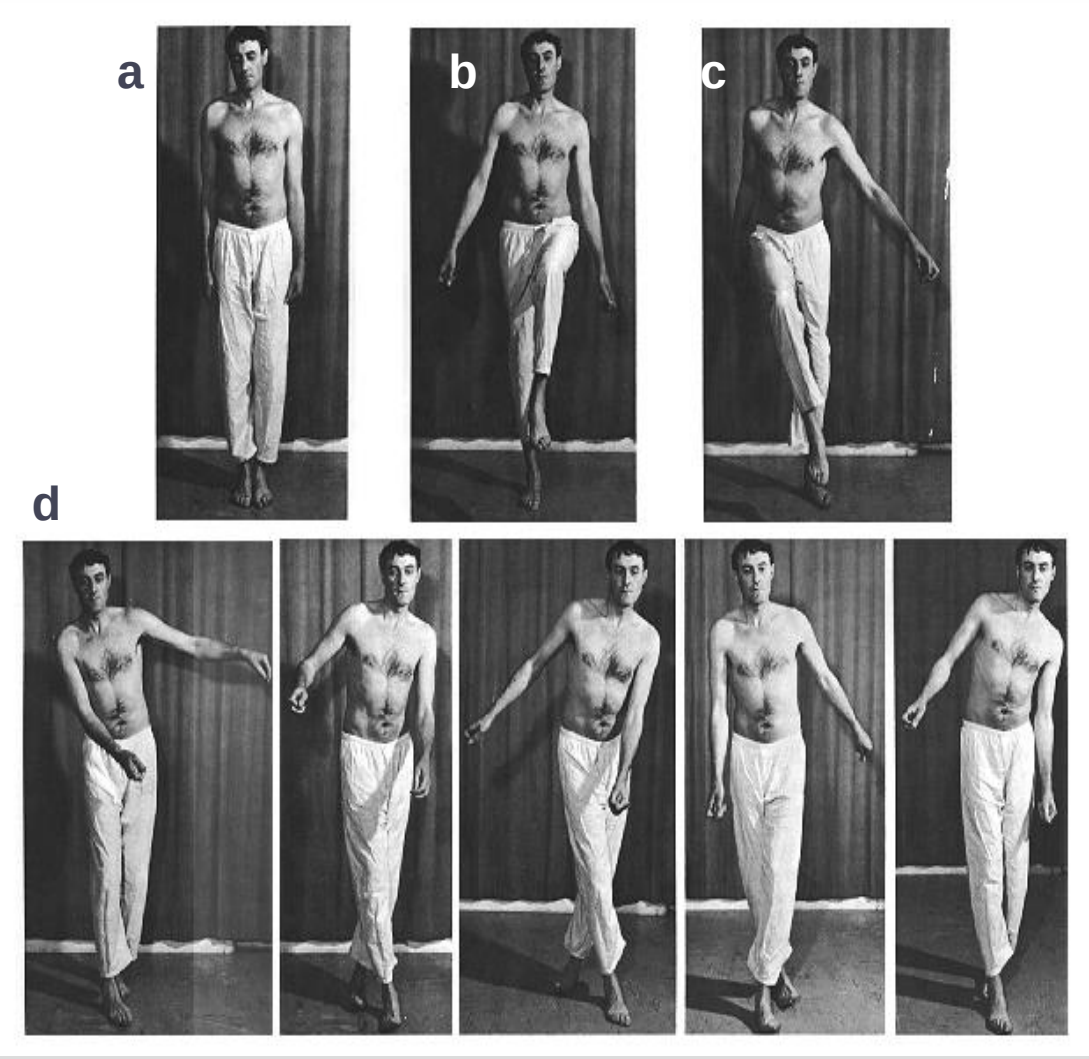
- pursuit movements *no longer smooth*, but saccadic

- may over- or under-shoot target with attempts at fixation (ocular dysmetria)

- **Writing abnormalities**

Positional projectile vomiting (posterior fossa lesions)


Sensory Ataxia (Posterior Column)
Thalamic Ataxia
VS
VS
Vestibular Ataxia
VS
Cerebellar Ataxia
VS
Cortical Ataxia
VS
VS
Myopathy
Sensory Ataxia (Peripheral Neuropahy))



Cerebellar Ataxia

Ataxic gait and position:
Left cerebellar tumor

a. Sways to the right in standing position

b. Steady on the right leg

c. Unsteady on the left leg

d. Ataxic gait

SENSORY ATAXIA

"Disturbances in the sensory input to the cerebellum"

- Tests of proprioception- Joint sense, passive movement.

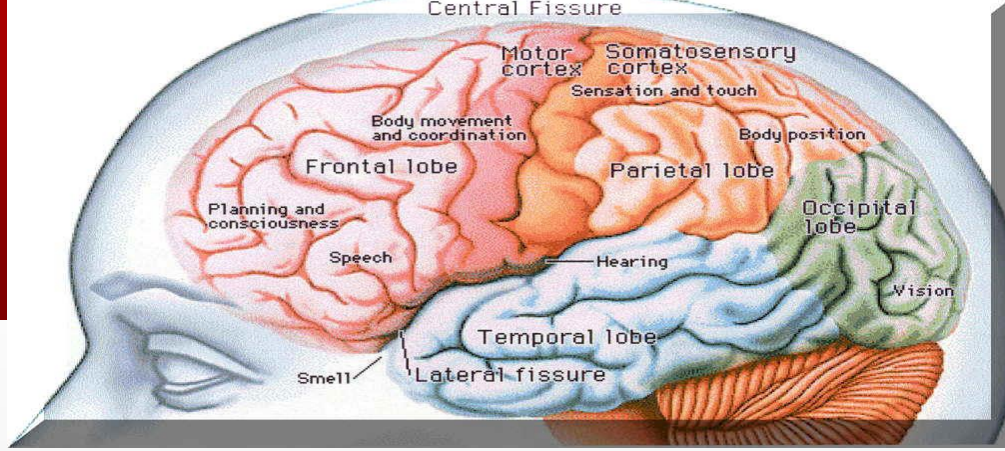
"The corrective effects of the Visual system"

- Classical Sensory Ataxic Gait
- Romberg's sign
- Loss of tendon reflexes
- Features of Peripheral neuropathy

TABLE 43.2 Associated Findings Helpful in Distinguishing Sensory from Cerebellar Ataxia

Sensory Ataxia	Cerebellar Ataxia
Sensory loss, especially for joint position and vibration	Nystagmus, ocular dysmetria, and other eye movement abnormalities
Steppage gait	Reeling, ataxic gait
Decreased reflexes	Other signs of cerebellar disease (dyssynergia, dysmetria, dysdiadochokinesia, hypotonia, rebound, impaired check response)

Cortical Ataxias



- FRONTAL LOBE ATAXIA refers to disturbed coordination due to dysfunction of the **contralateral frontal lobe**;

-Results from disease involving the frontopontocerebellar fibers en route to synapse in the pontine nuclei.

- Hyperreflexia,
- Increased tone.

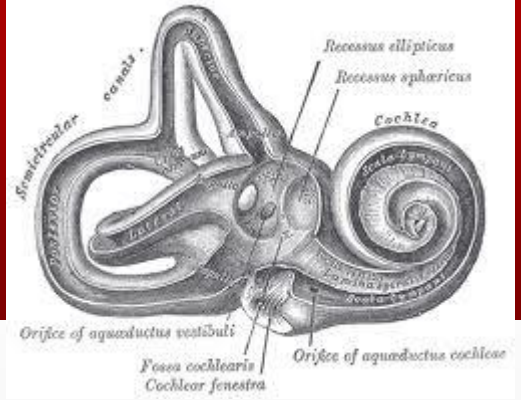
Table 24-2 Features of Cerebellar Ataxia, Sensory Ataxia, and Frontal Gait Disorders

	Cerebellar Ataxia	Sensory Ataxia	Frontal Gait
Base of support	Wide-based	Narrow base, looks down	Wide-based
Velocity	Variable	Slow	Very slow
Stride	Irregular, lurching	Regular with path deviation	Short, shuffling
Romberg	+/–	Unsteady, falls	+/–
Heel → shin	Abnormal	+/–	Normal
Initiation	Normal	Normal	Hesitant
Turns	Unsteady	+/–	Hesitant, multistep
Postural instability	+	+++	++++
			Poor postural synergies getting up from a chai
Falls	Late event	Frequent	Frequent

Muscle weakness

- Simple “tests of muscle power” can help detect muscle weakness in various muscle groups

Labrynthine Disorders



- Ataxia associated with vestibular nerve or labyrinthine disease.
- It results in a disorder of gait associated with a significant degree of **dizziness, light-headedness, or the perception of movement** .

BEWARE OF EXTREMELY ANXIOUS PATIENTS!!! (PSYCOGENIC)

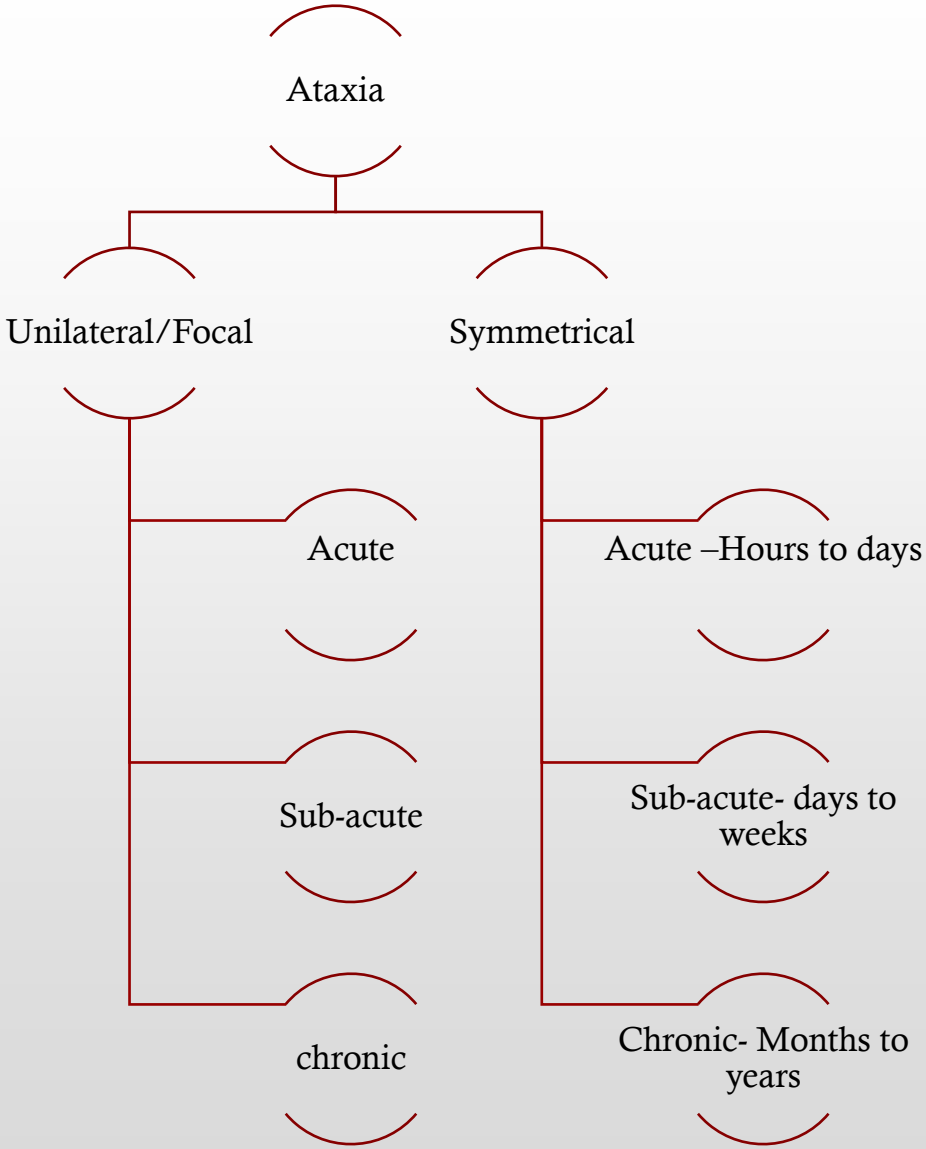
THE "FOUR" QUESTIONS????

- Mode of ONSET ?
- PROGRESSION ?

HISTORY

- Focal /Symmetric involvement ?
- Localisation of the cerebellar lesion ?

EXAMINATION


Ataxia
Unilateral/Focal
Symmetrical
Acute
Sub-acute
chronic
Acute –Hours to days
Sub-acute- days to weeks
Chronic- Months to years

ACUTE ONSET ATAXIA

- *INTOXICATION:* alcohol(Vermian Atrophy), lithium , phenytoin(should be avoided in seizure with ataxia) , barbiturates

- *POST INFECTIOUS:* Acute Viral Cerebellitis(CSF supportive of acute Viral infection), Varicella zoster virus.

- *VASCULAR:* Infarction (AICA, PICA syndromes), Haemorrhage, Subdural hematoma (Focal and ipsilateral cerebellar signs)

SUB ACUTE ATAXIA

- *INTOXICATION:* Mercury(parasthesiass, restricted visual defects), Solvents, Glue
- *NUTRITIONAL:* B1 and B12 deficiency
- *INFECTION:* HIV
- *DEMYELINATING:* Multiple Sclerosis
- *NEOPLASTIC:* Glioma, Metastases

CHRONIC ATAXIA

- *AUTOIMMUNE CAUSES :* Paraneoplastic syndromes, Gluten hypersensitivity, Anti GAD(Glutamic acid decarboxylase) antibodies.
- *HYPOTHYROIDISM*
- *INFECTIONS:* Syphilis (Tabes Dorasalis)
- *CONGENITAL LESIONS:* Arnold-Chiari and Dandy Walker Syndromes
- *INHERITED ATAXIAS:* AD,AR,XR,XD,Mitochondrial

PROGRESSION

- Progressive
- Static
- Intermittent symptoms
- Reversible Ataxias

PROGRESSIVE ATAXIA

CLASSIFICATIONS OF GREENFIELD AND OF HARDING

into three main groups:

- (1) **SPINOCEREBELLAR ATAXIAS**, with unmistakable involvement of the spinal cord (Romberg sign, sensory loss, tendon reflexes, Babinski signs);
- (2) **PURE CEREBELLAR ATAXIAS**, with no other associated neurologic disorders; and
- (3)**COMPLICATED CEREBELLAR ATAXIAS**, with a variety of pyramidal, extrapyramidal, retinal, optic nerve, oculomotor, auditory, peripheral nerve, and cerebrocortical accompaniments including what is now referred to as multiple system atrophy

STATIC ATAXIAS

- Vascular causes

REVERSIBLE ATAXIAS

- Infectious causes – post viral
- Thyroid(hypothyroidism)
- Drugs
- Toxins

INTERMITTENT SYMPTOMS

- Episodic Ataxias (Inherited etiology)

FOCAL / SYMMETRIC ATAXIAS

- Cerebellar symptoms on same side of lesion, or
- Bilateral symptoms

FOCAL ATAXIAS

Vascular causes, Multiple Sclerosis, Cerebellar abscess, cerebellar glioma, PML (HIV), Congenital causes.

SYMMETRIC ATAXIAS

Intoxication, Nutritional, Post inhectious, Hypothyroid, Autoimmune causes

LOCATION OF LESION

CEREBELLAR HEMISPHERIC SYNDROME	Ipsilateral head & Body cerebellar signs	Infarct, Neoplasm, Abscess, Demyelination
ROSTRAL VERMIS SYNDROME	gait and Trunk Ataxia	Alcoholism, B1 deficiency
CAUDAL VERMIS SYNDROME	Disequilibrium, Trunk ataxia	Medullobalstomas
PANCEREBELLAR SYNDROME	Bilateral signs	Toxins, metabolic, Infections, Autoimmune, Inherited
CEREBELLAR PEDUNCLES	Dramatic cerebellar symptoms	

PICA
(Lateral medullary-Wallenberg Syndrome)

Dysfunction	Effects
Vestibular nucleus	Vestibular system: vertigo, diplopia, nystagmus, vomiting
Inferior cerebellar peduncle	Ipsilateral cerebellar signs, including ataxia
Central tegmental tract	Palatal myoclonus
Lateral spinothalamic tract	Contralateral deficits in pain and temperature sensation from body
Spinal trigeminal nucleus	Ipsilateral loss of touch pain and temperature sensation from face
Nucleus ambiguus (which affects vagus X and glossopharyngeal nerves IX)	Dysphagia, hoarseness, diminished gag reflex
Descending sympathetic fibers	Ipsilateral Horner syndrome

AICA
(Lateral Inferior Pontine Syndrome)

- Vestibular N.→ i/l vertigo, nystagmus
- Cochear n. → i/l deafness
- 7th Cranial Nerve→ i/l facial palsy
- Cerebellum → i/l Ataxia
- 5th cranial nerve→ i/l hemisensory loss of face
- Spinothalamic Tract→ C/L hemisensory loss

i/l ipsilateral
C/L contralateral.

THE NEXT STEP ...RULE OUT

Acquired ataxias

↓

Inherited ataxias

↓

SPORADIC or IDIOPATHIC ATAXIAS

ACQUIRED ATAXIAS

- First rule out the **Structural causes** (MRI Brain/ CT head)

-CVJ (Cranio vertibro Junctional) anomalies

-Posterior fossa tumors

-Demyelinating diseases

-Hypoxic encephalopathies

-Vascular causes- infarct, haemorrhage

INHERITED ATAXIAS

- AD
- AR
- MITOCHONDRIAL DISTURBANCES
- X LINKED RECESSIVE
- X LINKED DOMINANT

AUTOSOMAL DOMINANT

- SPINO CEREBELLAR ATAXIAS (Types1→31)-previously olivopontocerebellar atrophies
- DentatoRubroPallidoLuysian Atrophy
- EPISODIC ATAXIAS (Types 1→7)

SCA(spino cerebellar ataxia) SALIENT FEATURES

- 3-5th decade of life ONSET, loss of ambulation over 10-15 yrs. from onset
- Differs from each SCA→responsible for various ages of presentation and variable phenotypic expression
- CAG polyglutamate repeat expansion in most of them.

AUTOSOMAL RECESSIVE ATAXIAS

- FRIEDREICH'S ATAXIA
- ATAXIA TELANGIECTASIA
- ATAXIA WITH ISOLATED VIT.E DEFICIENCY
- ABETALIPOPROTEINEMIA
- ENZYME DEFICIENCIES (Maple Syrup urine disease, Urea cycle defects, Sialidosis, Adrenoleucodystrophy,Organic aciduria, Pyruvate dehydogenase def.)

FRIEDREICH'S ATAXIA

- **Friedreich's ataxia** is an autosomal recessive inherited disease that causes progressive damage to the nervous system.
- Unstable expansion of GAA repeats→FRATAXIN protein→iron accumulation in mitochondria→neuronal injury.
- May present as classical or associated with vit E deficiency.
- Progressive staggering gait, frequent falling and titubation.
- May be associated with progressive scoliosis,foot deformity, cardiomegaly, conduction defects.
- *NATURAL HISTORY:*

-onset <25 yrs. At ADOLESCENCE

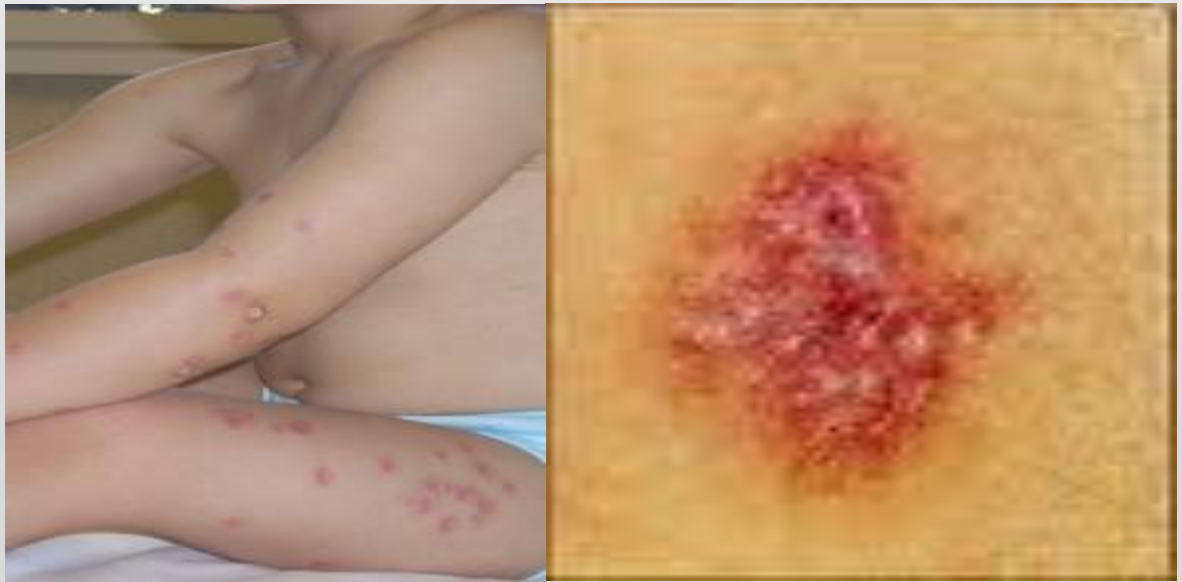
-loss of ambulation 15 yrs. Since onset

-Death usualy due to cardiac complications.

Median age of death 35 years.

ATAXIA TELANGIECTASIA

- Present in 1st decade.
- OCULOMOTOR APRAXIA , TELANGIECATSIAS IN EYES, SKIN,deficits in cerebellar function and nystagmus



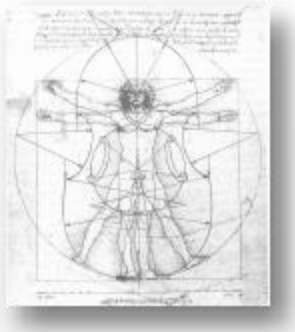
SPORADIC or IDIOPATHIC ATAXIAS

- Unknown genetic defects after ruling out acquired causes
- Old age of onset
- Presents with Dysautonomia –Orthostatic hypotension, erectile dysfunction, Urinary incontinence

Investigations

MRI →Brain and Upper cervical cord
CT Head
Vit. E, B12 levels
Total cholesterol levels, Thyroid hormones
NCV and EMG studies (to rule out other systems' involvement)
Toxicology screen (includes phenytoin levels)
Serology screen (for autoantibodies)
CSF analysis
Genetic Analyses (GAA, CGG, CAG repeat analyses)

ALGORITHM



Pt. with progressive ataxia

IMAGING
(MRI,CT)

PEDIGREE
CHART

AD

SCA1,2
MJD
SCA6,7
SCA10,12
DRPLA
SCA17

AR

Singleton patient

ACQUIRE
D CAUSES

FA
AT
AVED(ataxia with isolated vit E def)
ABETALIPOPROTEINEMIA

SUMMARY

- RULE OUT “ATAXIA MIMICKERS”
- CONFIRM PREDOMINANT CEREBELLAR INVOLVEMENT WITH RESPECTIVE TESTS
- ANSWER THE “FOUR” QUESTIONS

(Onset, progression, Symmetry, Localisation of lesion)

- RULE OUT ACQUIRED CAUSES
- GENETIC ANALYSES


THANK
YOU!!