ATAXIA

…is NOT a foreign cab!!

GENERAL PRESENTATION

Assessing a child who presents with ataxia can be challenging, however with a clear approach and organized differential, the task becomes manageable. Children will present with their parents with the complaint of an abnormal gait and/ or tremor. The ataxic gait is often described as wide based, lurching or staggering.

Ataxia is defined as an inability to coordinate muscle activity, causing jerkiness and incoordination. It is an impairment of direction, rate and strength of voluntary movements resulting in an inability to perform precise movements as well as a loss of balance. It can also present with an intention tremor (a tremor that often worsens with intentional movement of the limbs).

Don't forget that ataxia is a symptom of underlying disease! The pathology causing ataxia can be located either within the cerebellum or its neuronal connections, or within some other body system.

ANATOMY

True ataxia is a sign of *cerebellar dysfunction*; however, loss of balance or abnormal gait can occur for several reasons; vestibular dysfunction, musculoskeletal or psychiatric abnormalities. You must use your history and physical exam to differentiate true ataxia from other causes of abnormal gait.

Anatomically, the midline cerebellum (vermis), is responsible for coordination of gait and truncal balance; whereas the cerebellar hemispheres are responsible for the coordination of appendicular or extremity motor activities. In addition, the peripheral nervous system gathers information about the body's position in space that is then relayed through the cerebellum where it is processed to maintain balance.

True ataxia can be classified into:

1. **Sensory Ataxia**: results from the loss of sensory input from the lower extremities due to diseases of peripheral nerves, dorsal roots, dorsal columns of the spinal cord or medial lemnisci.

2. **Cerebellar Ataxia**: results from a lesion or degeneration focused in the body's gait and balance center: the vermis of the cerebellum.

Characteristic	**Sensory Ataxia**	**Cerebellar Ataxia**
Location of lesion	Peripheral nerve or posterior column disease	Cerebellar vermis

Gait	Wide based gait High stepping gait	Lurching, staggering
On Exam	Can look normal while sitting Loss of peripheral position and vibration sense +ve Romberg	Unable to stay balanced sitting -worse if legs crossed, -worse with running / standing
Associated Findings	Loss of position and vibration sense extremities	Intention tremor Scanning speech Hypotonia Ocular and limb dysmetria
Speech	Normal	Often slurred
Hints	Patient often looks to feet to know their position in space	Patient looks like acute alcohol intoxication

*(1) link to additional symptoms of cerebellar disease in children

HISTORICAL INVESTIGATION

When you are faced with a child with ataxia, start with the basics: ***ABC***'s.!!! Ensure that your patient is stable and comfortable and then proceed further. In addition to a thorough history, include the following pertinent questions:

1. HPI: timing, onset, progression (congenital vs insidious vs acute), nature of ataxia, associated symptoms: fever, nausea, vomiting, lethargy, headache, head tilting?

   HINT*: signs of increased ICP: early morning headache, vomiting, change in mental status*

   a. Any previous episodes of ataxia?
   b. Onset: any recent viral infection or cough?
      HINT*: Varicella is the most common acute infectious cause of ataxia*
   c. Any medications, drugs or poisons possibly ingested?
      HINT*: Are there any visitors taking anticovulsants or antipsychotics?*
   d. Any alcohol or other illegal drugs?
   e. Any trauma?
   f. Is there the possibility of a suicide attempt?
   g. Any change in mental status?

2. PMH: Birth Hx: exposures, **TORCHES** (congenital infections including toxoplasmosis, rubella, CMV, Herpes, Syphillis), congenital anomalies (ataxia can be congenital or hereditary, but not appear until later).

3. Family History: sickle cell, metabolic disease, epilepsy, migraines or Friedrich's ataxia.

PHYSICAL EXAM

You must complete a full physical exam including vital signs and growth parameters. Be sure to also include the following observations or exam:

General: awake, alert, acute distress, dysmorphic features
Skin: scars, crusted varicella lesions
HEENT: fluid in middle ear, cervical adenopathy, papilledema, hemorrhages
Chest: chest rales, arrhythmias, murmurs
Abdomen: hepatosplenomagaly, masses
MSK: decreased strength
Neuro: full neuro exam: cranial nerves, tone, power, reflexes, sensation (pain, vibration, light touch, temperature and proprioception),focal neurological findings (ex) sudden onset of hemiparesis in a non speaking child may look like abnormal gait)

Assessment of Ataxia (Testing Cerebellar Function):

- Assess patient for **truncal control** by having the child sit with the buttocks resting on the edge of a firm surface. You need to see if the child can keep balanced in this position without support from their extremities.
- Then ask the child to walk several steps with **natural gait**, then assess **tandem gait** (walk heel toe). You can also ask the child to walk several times around an object (ie. chair). A child with organic cerebellar disease may fall or lean towards the side of the lesion.
- Assess **tremor** by asking the patient to hold their hands out in front of them.
- Assess **dysmetria** (error in estimating the amplitude of movement) by asking them to touch their nose with their index finger and then touch your finger. Challenge them by moving your finger to different locations. Look for an inability to connect with your finger or their nose, (for example, if they poke themselves in they eye they likely have dysmetria!!)
- Assess **dysdiadochokinesia** (the inability to switch on and off antagonizing muscle groups) by rapid alternating movements of the hand. Do this by asking to child to tap the palm of one hand with the palm of the other and then quickly turn over the hand to tap the palm

with the dorsum of the other hand (pronation and supination as quickly as possible).

- **Romberg test**: ask the child to keep their feet shoulder width apart with arms extended in front and assess their balance. Then ask the child to close their eyes and maintain balance.

Where is the lesion if balance is lost with:

-eyes closed? (With the eyes closed, the child must depend on proprioception from their lower limbs to maintain balance. A deficit in proprioception (peripheral nerve or posterior column) will result in loss of balance with eyes closed)

-eyes open?; (a deficit in the cerebellum will result in difficulty standing whether the eyes are open or not).

HINTS FOR NEURO EXAM IN KIDS:

1. Kids greater than 4-6 years and of normal intelligence will be able to participate in a screening neuro exam.
2. Have the child stand in front of you and demonstrate the desired motor acts.
3. Be clear and concise with what you want them to do.
4. Be ready to support them if you are doing balance tests.
5. Younger kids (<4 years), you must rely heavily on your observational skills and history from the parents. Be sure to assess tone and reflexes.

DON'T FORGET::

- *loss of balance with **closed eyes** = **THINK PERIPHERAL DISEASE** (peripheral proprioception nerves)*
- *loss of balance with **open eyes** = **THINK CENTRAL DISEASE** (cerebellum)*

DIFFERENTIAL DIAGNOSIS

Another clue to the cause of ataxia, is in its acuity of onset.

ACUTE ATAXIAS:

1. Drug Ingestion (ie anticonvulsant)
2. Post-Infectious Immune – Acute Cerebellar Ataxia and Guillain Barre Syndrome
3. Brainstem encephalitis
4. Trauma
5. Acute hemorrhage in brain tumor
6. Cerebral hemorrhage – vascular disease
7. Acute labyrinthitis
8. Brain Tumours (may be insidious onset)
   a. Cerebellar Astrocytoma
   b. Cerebellar hemangioblastoma (Von Hippel Lindau disease)

c. Ependymoma
d. Medulloblastoma
e. Supratentorial Tumors
f. Neuroblastoma

INSIDIOUS OR INTERMITTENT ATAXIA

1. Brain Tumors (as above)
2. Benign Parxoysmal Vertigo
3. Basilar Migraine
4. Hereditary Ataxia
    a. Autosomal recessive inheritance
        i. Ataxia telangiectasia
        ii. Friedrich's Ataxia
        iii. Hartnup's disease
        iv. Hypobetalipoproteinemia
        v. Refsum's disease
        vi. Wilson's disease
5. Congenital Malfomations
    a. Basilar impression
    b. Cerebellar aplasia
    c. Dandy Walker Malformation
    d. Vermal apalsia
    e. Chiari malformation
6. Metabolic/Inborn Errors of Metabolism
    a. Maple syrup urine disease
    b. Ornithine transcarbamylase deficiency

PROCEDURES FOR INVESTIGATION

HINT: One approach to working through investigations is to think in this pattern:
1. Blood 2. Other body fluids 3. Imaging and 4. Consults.

1. <u>Bloodwork</u>:
    - CBC and differential (infection)
    - Electrolytes with bicarbonate (imbalances),
    - Monospot
    - Toxicology screen (for anticonvulsants, hypnotics and phenothiazines, for alcohol and drugs of abuse; for heavy metals)
    - Consider metabolic screen and IgA (for ataxia telangiectasia)
2. <u>Urine</u>: toxicology screen
3. <u>Lumbar Puncture</u>: if differential includes meningitis and fundi are normal (ie) don't cause a tonsillar herniation of the brain due to increased ICP); look for CSF protein if demyelinating disease is suspected.
4. <u>Imaging</u>: Consider CT/ MRI if there are focal neurological findings.
5. <u>Consults</u>: Neurology.

CONCLUSION:

In summary, ataxia is the inability to coordinate muscle activity and can be challenging to diagnose in children. A thorough history and physical exam in conjunction with various investigations are key to understanding the underlying etiology. Finally, remember you must know and be able to rule out the life threatening causes of ataxia.

SUPPLEMENTARY INFORMATION

Classification of most common causes of ataxia by age classification. (2)

REFERENCES:

Behrman, R. E. and Klliegman, R. M. Nelson Essential of Pediatrics, 4th ed. WB Saunders Company, 2002.

DeAngelis, C. Ataxia. Pediatrics in Review, 16 (3): 114-115, 1995.

Fishman, M. A. Pediatric Neurology. Chpt 17: Ataxia: (Allan Percy): 267-273. Toronto: Grune & Stratton, 1986.

Goldbloom, Richard. Pediatric Clinical Skills. San Francisco, Churchilll Livingstone: 1997.

Swaiman, Kenneth. Pediatric Neurology: Principles and Practice. 2nd ed. Vol: 1; Chpt 19: 261-269: Toronto, Mosby: 1994.

LINKS

(1)
Classic Findings of cerebellar Dysfunction
Hypotonia
Nystagmus
Staggering gait
Titubation

Other symptoms and signs of cerebellar dysfunction
Action tremor
Asthenia
Ataxia
Decompensation of movements
Dysdiadochokinesis
Dysmetria
Dyssynergia
Hypotonia
Impaired rebound

Pendular DTRs

Exam and arms and hands
Finger to nose
Finger to finger to nose
Rapid alternating movements
Rebound
Tone evaluation

Exam of gait
Foot tapping
Heel to shin maneuver
Heel and toe waling
Hopping in one place
Romberg
Routine walk and run
Tandem walk
Walk in circle

(2)
Commonest causes of ataxia by age groups:

<1 year	**1-5 years**	**5-10 years**	**>10 years**
Congenital Malformations	Drug ingestion	Drug ingestion	Friedriech's Ataxia
Mild arrested hydrocephalus	Acute cerebellar ataxia	Acute cerebellar ataxia	Miller Fisher syndrome
Cerebral Palsy	Myoclonic Encephalopathy and neuroblastoma	Brain Tumors	Cerebellar Hemorrhage
Marinesco-Sjogren Syndrome	Inborn errors of metabolism	Wilson's disease	Multiple Sclerosis
	Brain Tumors	Adrenaleukodystrophy	Oligopontocerebellar degeneration
	Ataxia Telangiectasia	Hereditary ataxia	Hereditary ataxia
	Refsum's disease		