


Episode 299 Recap

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This week, the [CPSolvers](#) featured an episode from [Neurology VMR](#). [Maria](#) presented the case of a 72-year-old woman with a twenty year history of progressive clumsiness and difficulty walking, now requiring a wheelchair. Over the past five years, it had progressed to clumsiness of her arms/hands and difficulty speaking. She had a family history significant for multiple family members requiring wheelchairs. Neurological exam was notable for slow speech with unusual separation of syllables, reduced muscle bulk, decreased light touch and pinprick sensation in lower extremities to the mid-shins, difficulty with finger-to-nose testing, and dysdiadochokinesia. Genetic testing revealed 39 repeats of ATXN7 gene leading to the final diagnosis of spinocerebellar ataxia type 7.

CLUMSINESS




- Trouble walking?
- Trouble staying still?
- Seeing things spinning?
- Numbness?

Complaint usually associated with **motor function**.
"Not being able to do certain tasks"

MOTOR X SENSORY X CEREBELLAR


WEAKNESS X ATAXIA




Not dextrous because of weakness

Disconnection between what you want to do and the final project


Sensory x Cerebellar




CLUE: Coordination → Cerebellum




Nystagmus




Imbalance?
Gait disturbance?
Frequent falls?




TIME COURSE



- Chronic → Neurodegenerative; Genetic **motor neuron disease** **muscular disorder**
- Subacute → Cervical degenerative disease, **older age** paraneoplastic; metastatic
- Acute → Postinfectious, Multiple Sclerosis






SPEECH



Scanning dysarthria:
sounds like drunken speech

"My friends keep asking if I'm drunk"

 @cpsolvers  Episode 299: Neurology VMR - Clumsiness  Dx

Teaching points

[Spinocerebellar ataxia](#)

- **What?** → autosomal dominantly inherited diseases, >40 genetically distinct subtypes
 - **Pathophysiology?** → dynamic repeat expansion mutations or non-repeat mutations; ataxia from damage to cerebellum and other regions of nervous system, including spinal cord.
 - **Clinical presentation?** → progressive loss of balance and coordination accompanied by slurred speech; onset of ataxia often occurs in mid-adulthood; as ataxia progresses, coordination of extremities deteriorates leading to loss of fine motor skills
 - **Other associated findings?** → speech and swallowing problems; may have oculomotor abnormalities on exam due to cerebellar dysfunction
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Train your brain

Test your recall by answering our weekly quiz question [here](#).

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