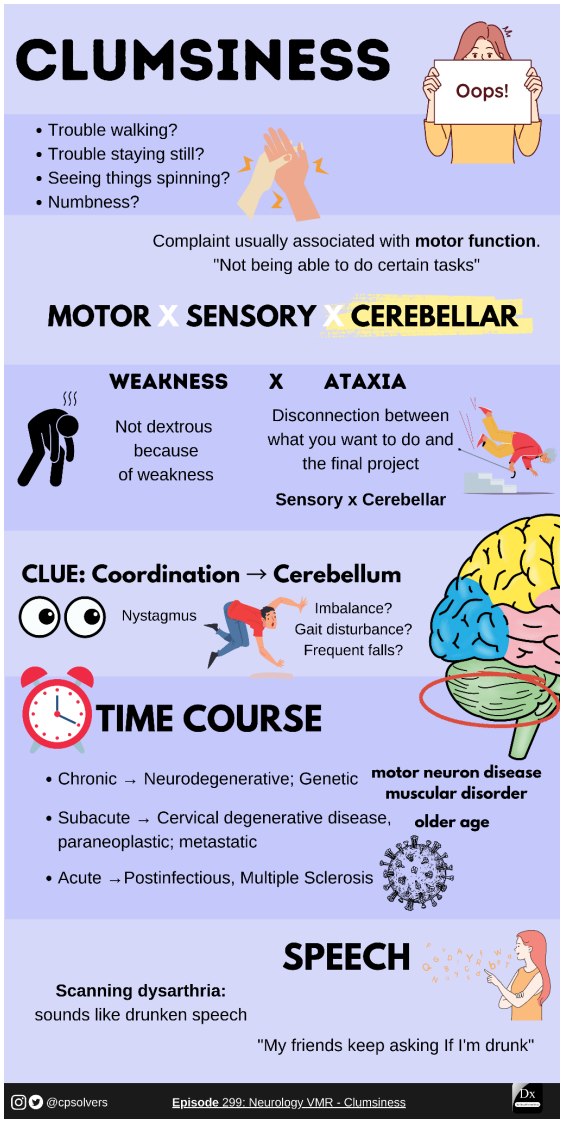
Episode 299 Recap

Authors: Gurleen Kaur and Laura Araujo

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.



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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