

A 15-year-old Caucasian male is brought to the office by his mother for the evaluation of a six-month history of unstable gait and speech difficulty which are getting worse over time. His past medical history is insignificant. He is not taking any medications, and denies smoking or alcohol consumption. His blood pressure is 120/70 mmHg and pulse is 80/min. Musculoskeletal examination showed scoliosis and feet deformity with 'hammer toes.' The neurologic examination showed dysarthria, dysmetria, nystagmus, and absence of deep plantar reflexes on lower extremities. What is the most common cause of death in this patient population?

- ☐ A. Cardiomyopathy
- ☐ B. Renal failure
- ☐ C. Diabetes-related complications
- ☐ D. Malignancy
- ☐ E. Septic shock

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- ☒ A. **Cardiomyopathy** [74%]
- ☐ B. Renal failure [11%]
- ☐ C. Diabetes-related complications [2%]
- ☐ D. Malignancy [8%]
- ☐ E. Septic shock [4%]

[Proceed to Next Item](#)**Explanation:**

User Id:

This clinical scenario is typical for Friedreich ataxia, which is the most common type of spinocerebellar ataxias. It is an autosomal recessive disorder, and its symptoms usually begin before 22 years of age. Neurological manifestations (e.g., gait ataxia, frequent falling, dysarthria) result from degeneration of the spinal tracts (spinocerebellar tracts, posterior columns, pyramidal tract). Non-neurological manifestations may also occur, and these include concentric hypertrophic cardiomyopathy, diabetes, and skeletal deformities (e.g., scoliosis and 'hammer toes'). The median survival after disease onset rarely exceeds 20 years. The most common causes of death are cardiomyopathy and respiratory complications. Cardiomyopathy develops in up to 90% of the patients.

(Choice B) Renal failure is not characteristic of Friedreich ataxia.

(Choice C) Diabetes develops in 10-20% of the patients with Friedreich ataxia, but it is a rare cause of death.

(Choice D) Friedreich ataxia does not predispose to cancer.

(Choice E) Septic shock may develop due to respiratory complications, but it is not as common as cardiac complications.

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Educational Objective:

Friedreich ataxia is the most common type of spinocerebellar ataxias. Remember the combination of neurologic (ataxia, dysarthria), skeletal (scoliosis, feet deformities) and cardiac (concentric hypertrophic cardiomyopathy) manifestations of the disease. The most common causes of death are cardiomyopathy and respiratory complications.

*very high-yield topic for USMLE!

Time Spent: N/A

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