

A 6-year-old female with Down syndrome is brought to the physician for behavioral changes. Over the past few weeks, she has begun refusing to do her usual activities. Her parents also report that she seems dizzy and state that she has developed urinary incontinence. On examination, she is hypotonic but hyperreflexic with a positive Babinski reflex. Her gait is ataxic. Which of the following is the most likely diagnosis?

- ☐ A. Alzheimer disease
- ☐ B. Atlantoaxial instability
- ☐ C. Hypothyroidism
- ☐ D. Mental retardation
- ☐ E. Spinal cord infarction



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- ☐ A. Alzheimer disease [5%]
- ☒ B. **Atlantoaxial instability** [61%]
- ☐ C. Hypothyroidism [7%]
- ☐ D. Mental retardation [3%]
- ☐ E. Spinal cord infarction [24%]

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### Explanation:

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Atlantoaxial instability is a malformation seen in 10-15% of patients with Down syndrome, and most commonly occurs due to excessive laxity in the posterior transverse ligament, which causes increased mobility between the atlas (C1) and the axis (C2). Fortunately, only 1-2% of Down syndrome patients with atlantoaxial instability are symptomatic. Symptoms usually progress over several weeks and result from compression of the spinal cord. Presenting symptoms include behavioral changes, torticollis, urinary incontinence, and vertebrobasilar symptoms such as dizziness, vertigo, and diplopia. On examination, upper motor neuron symptoms such as leg spasticity, hyperreflexia, a positive Babinski sign, and clonus are often present. Patients with Down syndrome are normally hypotonic, and they may remain hypotonic or have increased tone with symptomatic atlantoaxial instability.

Atlantoaxial instability is suspected on physical examination and diagnosed with lateral radiographs of the cervical spine in flexion, extension, and in a neutral position. Open mouth radiographs can also be helpful in visualizing the odontoid. Treatment consists of surgical fusion of the first cervical vertebrae (C1) to the second (C2).

**(Choice A)** Although there is a higher prevalence of Alzheimer disease among individuals with Down syndrome, it usually presents around ages 40-50.

**(Choice C)** The risk of hypothyroidism is increased in individuals with Down syndrome. These patients present with decreased growth velocity, weight gain, cold intolerance, fatigue, constipation, or declining school performance. Neurologic symptoms can include



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**(Choice A)** Although there is a higher prevalence of Alzheimer disease among individuals with Down syndrome, it usually presents around ages 40-50.

**(Choice C)** The risk of hypothyroidism is increased in individuals with Down syndrome. These patients present with decreased growth velocity, weight gain, cold intolerance, fatigue, constipation, or declining school performance. Neurologic symptoms can include delayed reflexes, but upper motor neuron signs should not be present.

**(Choice D)** Mental retardation is seen in individuals with Down syndrome and can vary in its severity. However, progressive neurologic symptoms are not seen with mental retardation alone.

**(Choice E)** Spinal cord infarction presents with the acute onset of severe pain, weakness, and paresthesias. This condition is rare and more commonly seen in adults, and there is no increased risk in patients with Down syndrome.

**Educational objective:**

Atlantoaxial instability should be suspected in any patient with Down syndrome who presents with upper motor neuron findings.

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