

A 4-year-old boy is brought to the office by his parents for difficulty walking. The child started walking at age 17 months. He often falls while walking and has a tendency to walk on his toes. His maternal uncle had similar symptoms and died in his teens. Examination shows weakened hip muscles, thigh atrophy, bilateral calf enlargement, and positive Gower sign. What is the best test to confirm this patient's diagnosis?

- ☐ A. Electromyography
- ☐ B. Genetic testing
- ☐ C. Muscle biopsy
- ☐ D. Nerve conduction studies
- ☐ E. Serum aldolase levels
- ☐ F. Serum creatine kinase levels

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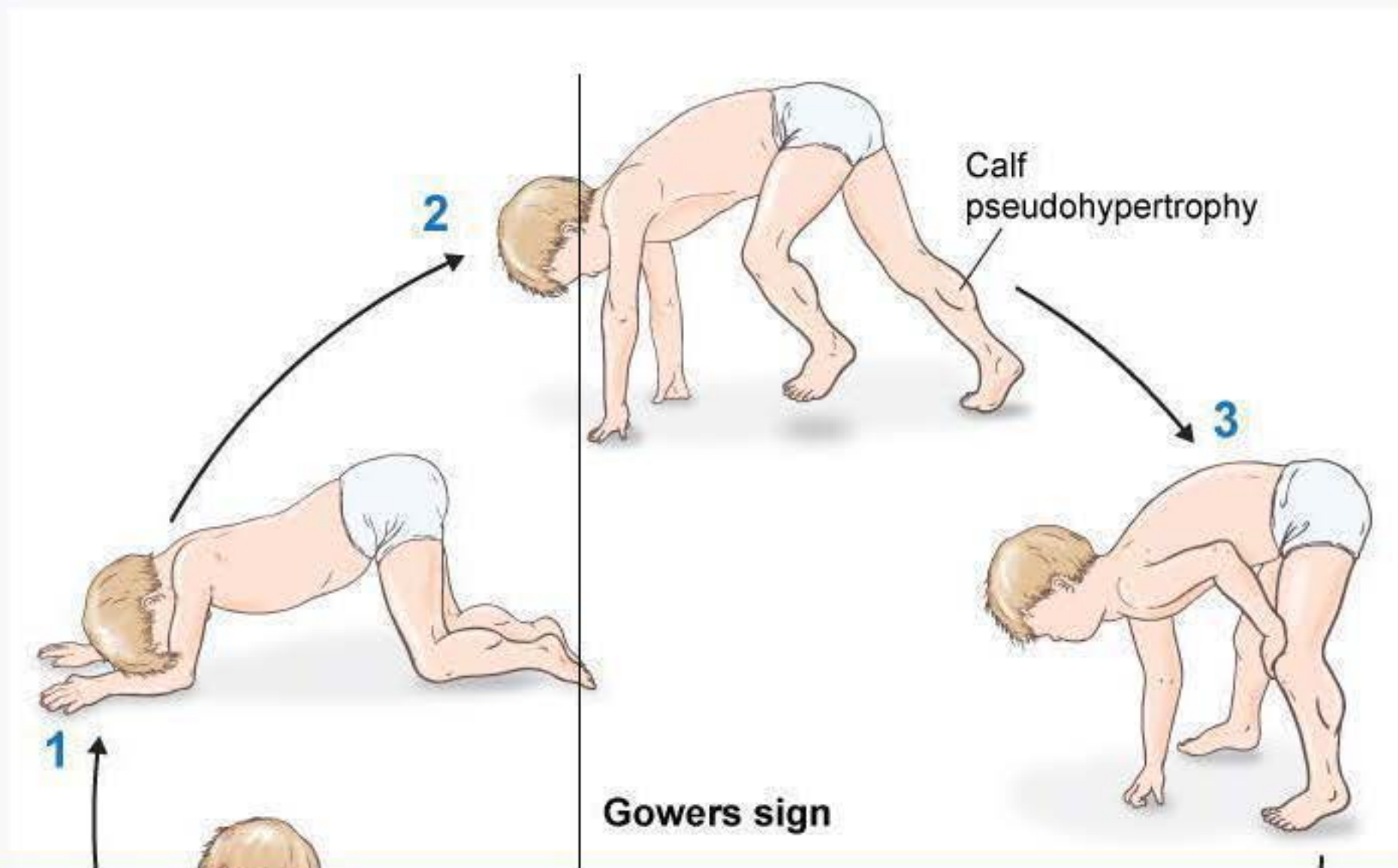
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- ☐ A. Electromyography [1%]
- ☒ B. Genetic testing [40%]
- ☐ C. Muscle biopsy [57%]
- ☐ D. Nerve conduction studies [0%]
- ☐ E. Serum aldolase levels [0%]
- ☐ F. Serum creatine kinase levels [2%]

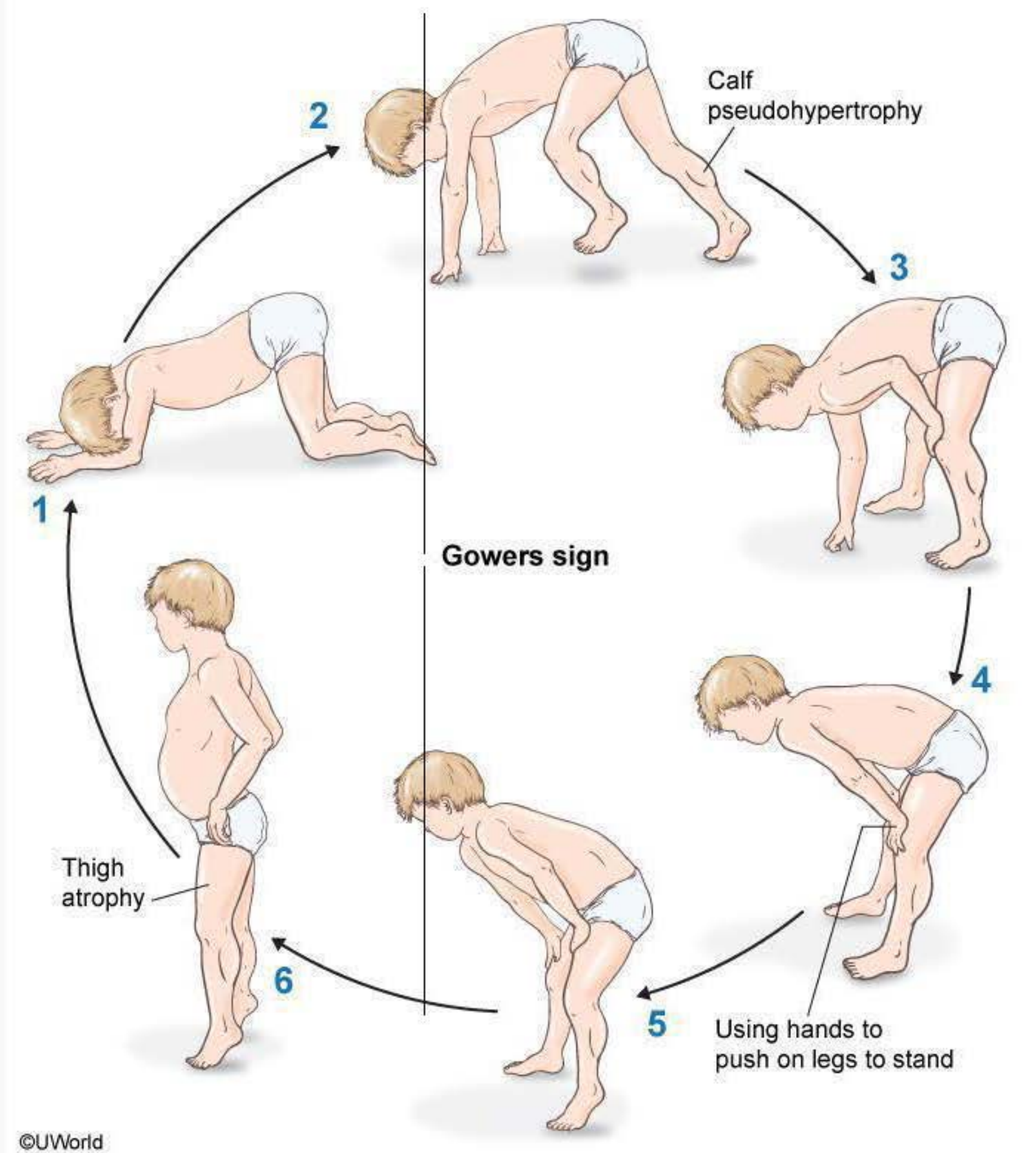
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Explanation:

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The patient most likely has Duchenne muscular dystrophy (DMD), the most common muscular dystrophy in children. The myopathy typically presents at age 2-5 with bilateral calf pseudohypertrophy and Gower sign, as seen in this patient. Similar





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The patient most likely has Duchenne muscular dystrophy (DMD), the most common muscular dystrophy in children. The myopathy typically presents at age 2-5 with **bilateral calf pseudohypertrophy** and **Gower sign**, as seen in this patient. Similar symptoms in the maternal uncle are consistent with an **X-linked recessive** transmission. The gold standard for diagnosis is genetic testing, which would show deletion of the **dystrophin gene** on Xp21.

**(Choices A and D)** In DMD, electromyography demonstrates a myopathic pattern with normal nerve conduction velocities. However, these tests are supportive and not confirmatory.

**(Choice C)** Muscle biopsy would show fibrosis and fatty infiltration and can support the diagnosis in this patient. Immunohistochemistry staining of muscle tissue would show absent dystrophin.

**(Choices E and F)** Serum creatine kinase and aldolase levels are elevated as early as infancy even before clinical manifestations. These elevations reflect muscle damage and release of these enzymes in the serum. As the disease progresses and more muscle is replaced by fat and fibrosis, these levels eventually drop. These tests are typically used for screening, as elevations can be seen in other myopathies.

#### Educational objective:

Serum creatine phosphokinase and aldolase levels are elevated in screening for muscular dystrophies. Fibrosis and fatty infiltration on calf muscle biopsy support the diagnosis. Genetic studies are the gold standard for confirmation.

#### References:

1. [Muscle disease.](#)
2. [Entries in the Leiden Duchenne muscular dystrophy mutation database: an overview of mutation types and paradoxical cases that confirm the reading-frame rule.](#)