

Muscular Dysgeny

A Case Study on the Muscular System
Human Anatomy and Physiology

Muscular dysgenesis or dysgeny is a lethal, recessive, genetic disease of mice that is caused by a mutation in the *mdg* gene. Skeletal muscles from dysgenic mice are paralyzed and the animals die shortly after birth.

You are a research scientist interested in finding the cause of the cellular defect associated with this genetic disease.

You surgically remove a single muscle fiber from a dysgenic mouse fetus and place it in an experimental chamber in order to study abnormalities in the control of skeletal muscle activity during dysgenesis.

1. Which of the following structures is NOT a part of the muscle fiber?

- A. transverse tubule
- B. motor end plate
- C. sarcolemma
- D. synaptic knob
- E. sarcoplasmic reticulum

2. You briefly apply a high concentration of acetylcholine (ACh) directly to the motor end plate and observe no contraction of the fiber. Since muscular dysgenesis only affects one type of cell in the body, you would suspect that the motor neurons of dysgenic mice work normally. True or False?

- A. True
- B. False

3. Arrange the events at the neuromuscular junction in the proper sequence from first to last

- 1. arrival of the action potential at the synaptic knob
- 2. generation of action potential in sarcolemma
- 3. binding of ACh to ACh receptors in the motor end plate
- 4. release of ACh into the synaptic cleft
- 5. removal of ACh from the cleft by acetylcholinesterase

- A. 1, 2, 3, 5, 4
- B. 2, 3, 1, 4, 5
- C. 1, 4, 3, 2, 5
- D. 2, 5, 1, 4, 3

4. After you apply acetylcholine to the muscle fiber, you find that an action potential is generated in the sarcolemma. This result proves that certain events or conditions occur normally within the neuromuscular junction. Which event or condition is NOT PROVED by the above result?

- A. ACh receptors are present in the membrane of the motor end plate
- B. ACh receptors bind to ACh
- C. Sodium permeability of the end plate membrane is increased
- D. Acetylcholinesterase breaks down ACh

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