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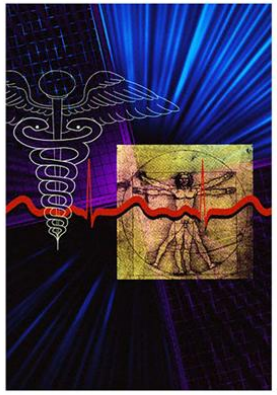

info@virtualmeded.com

Question of the Week

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Case Study

- Two boys with mental disability are found to have mutations in a gene on the X chromosome that has no homology with globin genes.
- Both are also noted to have deficiency of alpha-globin synthesis with alpha thalassemia.
- Which of the following is the best explanation for their phenotype?

A. The mutation disrupted an enhancer for an alpha-globin pseudogene
B. The mutation disrupted an X-encoded transcription factor that regulates the alpha-globin loci
C. There is a second mutation that disrupts an enhancer near the alpha -globin gene
D. There is a DNA rearrangement that joins the mutated X chromosome gene with an alpha -globin gene
E. There is a second mutation that disrupts the promoter of an alpha -globin gene

Answer

- The correct answer is B.

Explanation

- The boys have an X-linked recessive condition called alpha-thalassemia/mental retardation or ATR-X syndrome (309510).
- The X-encoded gene has an unknown function in brain as well as being a factor that regulates alpha-globin gene transcription. In order to affect all four alpha -globin genes, the X-encoded gene must produce a trans-acting factor; second mutations altering enhancers or promoters would be cis-acting and affect only one alpha-globin gene.
- Pseudogenes are functionless gene copies, so altered expression would not influence alpha globin chain synthesis

The end

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