



# HOX and PAX Genes

Virtual MedEd

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# Question

- Mutations involving homeobox genes (HOX genes) are most likely to be associated with which of the following?
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- A. Abnormal limbs and phocomelia
- B. Arachnodactyly and dissecting aortic aneurysm
- C. Congenital goiter and hypothyroidism
- D. Congenital pigment abnormalities and deafness
- E. Synpolydactyly and brachydactyly

# Answer

- The correct answer is E.

# Explanation

- Abnormalities of genes that control normal morphogenesis during embryonic development are associated with the development of congenital malformations.
- Two important types of genes associated with morphogenesis include homeobox (HOX) genes and paired box (PAX) genes.
- HOX genes act in temporal or spatial combinations and play an important role in the patterning of limbs, vertebrae, and craniofacial structures.
- Mutations of HOX genes may cause extra digits (synpolydactyly) or short digits (brachydactyly).
- It is interesting to note that vitamin A is an upstream regulator of HOX genes, and the use of retinoic acid during pregnancy may produce congenital abnormalities.

- Mutations of certain PAX genes are associated with malformations.
- Mutations of PAX-3 are seen in Waardenburg syndrome (congenital pigment abnormalities and deafness), while PAX-6 mutations may be seen with aniridia, and PAX-2 may be involved with Wilms syndrome.
- In contrast, abnormal limbs and phocomelia (underdeveloped limbs that are short stumps without fingers or toes) can result from thalidomide use, while arachnodactyly and dissecting aortic aneurysm in adults are seen in Marfan's syndrome.
- Congenital goiter or hypothyroidism can result from iodide deficiency.

The end

[info@virtualmeded.com](mailto:info@virtualmeded.com)