



POSNA

The Core Curriculum

Autosomal dominant disorders

Objectives

1. Define autosomal dominance
2. Describe the method of transmission of autosomal disorders, and the distinguishing features of a pedigree of an autosomal dominant disorder
3. List 3 relatively common autosomal disorders seen in orthopaedic practice

Discussion point

1. What are common physical features of autosomal disorders?

Discussion

Autosomal dominant disorders are relatively commonly seen in orthopaedic practice. Affected individuals are heterozygous, with one normal and one mutated gene. Since the mutated gene determines phenotypic expression, the disorder is characterized as dominant. Homozygosity is generally fatal. Variable expressivity is characteristic of autosomal dominant disorders. Transmission is characterized by one half of offspring of affected individuals being affected, regardless of sex. Examples of autosomal dominant disorders are achondroplasia, pseudoachondroplasia, the multiple epiphyseal dysplasias, chondrodysplasias, osteogenesis imperfecta, Marfan syndrome, polydactyly, hereditary motor sensory neuropathies I and II (Charcot-Marie-Tooth disease), myotonic dystrophy, and neurofibromatosis. A common feature of autosomal dominant disorders is that they are structural disorders. While they may impair function and have some reduction in life expectancy, survival to at least reproductive age is the rule.

References

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