



# POSNA

## The Core Curriculum

### **Autosomal recessive disorders**

#### **Objectives**

1. Define autosomal recessive
2. Describe the characteristics of a pedigree of an autosomal recessive disorder
3. List 2 examples of autosomal recessive disorders

#### **Discussion point**

1. If autosomal dominant disorders tend to be structural problems, how would autosomal recessive disorders be characterized?

#### **Discussion**

Patients with autosomal recessive disorders are homozygous for the affected gene. Both parents, therefore must be heterozygous, or carriers, if there is no phenotypic evidence of the disorder. The risk of being affected with an autosomal recessive disorder with 2 heterozygous parents is 1:4. Many autosomal recessive disorders produce inborn errors of metabolism, resulting from an enzymatic deficiency. Lysosomal storage diseases, which include the mucopolysaccharidoses and Gaucher's disease are autosomal recessive disorders. Limb-girdle muscular dystrophy and Frederich's ataxia families demonstrate autosomal recessive patterns. Another important non-orthopaedic condition is cystic fibrosis. Consanguinity increases the chances of offspring with autosomal recessive disorders.

#### **References**

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