



POSNA

The Core Curriculum

Sex-linked recessive disorders

Objectives

1. Define sex-linked recessive
2. Describe the features of the pedigree of a sex-linked recessive disorder
3. List 2 sex-linked recessive disorders commonly accompanied by orthopaedic problems

Discussion

Sex-linked recessive disorders are of course carried on the X chromosome. As a recessive gene, phenotypic expression is evident only in the absence of the allelic unaffected X gene, another way of saying it can only be seen in males. All daughters of an affected male will be carriers, since they must acquire X genes from both parents. Males do not pass the gene to their sons. A carrier's sons have a 50-50 chance of acquiring the affected gene.

Duchenne and Becker muscular dystrophy are now recognized as sex-linked recessive disorders originating from different mutations of the same gene. Carrier females may be identified by having elevated creatine phosphokinase levels. Another disorder with orthopaedic implications is classic hemophilia.

References

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