Autosomal Dominant Inheritance

In determining whether a pattern of autosomal dominant (AD) inheritance is present, several issues may need to be considered:

Pedigree Characteristics of Autosomal Dominant Inheritance

- Each child of an affected person has a 50% risk of inheriting the gene mutation.
- Males and females are equally likely to be affected.
- The condition is seen in sequential generations, affecting 50% of individuals in each generation on average.

Pitfalls in Recognizing Autosomal Dominant Inheritance

- **Incomplete penetrance.** Some people who have the gene mutation do not show the clinical effects.
- **Penetrance limited to one gender.** For example, when prostate cancer risk is inherited in an autosomal dominant manner, women who inherit the mutation are not affected; they can, however, pass the mutation on to their sons.
- **Variable expressivity.** The gene mutation has variable clinical manifestations: the disorder may range from mild to severe; or a range of different complications may occur among people with the mutation.
- **New mutation.** An affected person may be the first person in the family with the condition, due to a mutation arising for the first time in sperm, egg, or embryo
- **Germline mosaicism.** A new mutation may arise in testis or ovary, resulting in an unaffected parent transmitting the condition to two or more children