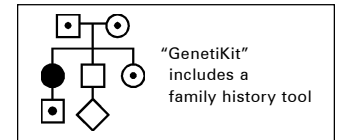




## General Pearls

1. Take a family history: 3 generations (both sides) →
2. Update it regularly
3. Pay attention to the unusual:
  - early age of onset
  - lack of other precipitating factors
  - multiple affected related individuals
  - occurrence of the condition in one sex when it usually occurs in the opposite sex
  - multiple congenital anomalies
  - uncommon conditions or diseases
4. Ask about specific diseases in family members, depending on the concern in question
5. See the family as your patient and realize that a disease in one or more members may affect the risk for another



## Specific - Prenatal and Pediatric - Look for:

1. Maternal age  $\geq 35$ : increased risk for chromosome abnormalities
2. Pregnancy history:
  - infertility
  - 3 or more spontaneous abortions
  - stillbirths
  - childhood deaths

} consider chromosome testing
3. Consanguinity: is the couple related or from the same isolated community?
4. Family history of: birth defects, mental retardation, stillbirths or childhood deaths, chromosome disorders e.g. Down syndrome, severe childhood conditions such as muscular dystrophy, cystic fibrosis
5. Potential patterns of inheritance e.g. affected males linked through females may indicate X-linked inheritance
6. Ethnic background - consider carrier screening for:
  - Hemoglobinopathies (thalassemia and sickle cell disease) in couples whose ethnicity is from Mediterranean, African, Middle Eastern or SE Asian countries
  - Tay-Sachs, Canavan, familial dysautonomia in couples of Ashkenazi Jewish background
  - Tay-Sachs in couples of French Canadian background

## Specific - Adult - Look for:

1. Features in the family history:
  - very early onset of disease in patient or close relative
  - relatively early onset in 2 or more family members
2. Pattern recognition in disease syndromes with multiple conditions caused by one gene e.g. BRCA1 and breast, ovarian and prostate cancer
3. Unusual neurological signs and symptoms, especially if progressive and early onset e.g. ataxia, weakness, dementia