

Family history



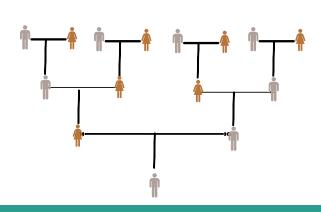
Red flags that suggest that an individual (or his/her/their family) may be at increased risk for a genetic condition.

The best way to identify red flags is by taking a family history (in addition to a personal health history).



Multiple affected family members (with the same or related disorder)

b<u>reast and ovarian cancer</u> i<u>ron overload and diabetes</u> <u>very high cholesterol</u>





Earlier age of onset of disease (symptom) than typically expected

- May demonstrate genetic predisposition in an individual who is more susceptible to environmental risk factors
 - e.g. <u>pre-menopausal breast cancer</u>;
 premature ovarian failure before age 40
 (<u>fragile X syndrome</u> carrier)



e.g. breast cancer in a person assigned male at birth





Presence of disease in the absence of other precipitating factors

sudden unexplained death in an athletic 20-year-old; diabetes mellitus (hereditary hemochromatosis or myotonic dystrophy)

Ethnicity

Some <u>genetic conditions</u> are more common in those of certain ethnicities.

e.g. Tay-Sachs disease in <u>Ashkenazi</u> Jewish individuals;

hemoglobinopathies (thalassemia, sickle cell anemia) in individuals of Mediterranean, African, Middle Eastern and South East Asian ethnicity





Consanguinity

Generally defined as a union between two individuals related as second cousins or closer

 Higher than average chance for both members of a couple to be carriers of the same autosomal recessive condition

