GEN-EQUIP

GENETICS EDUCATION FOR PRIMARY CARE

Red flags for clinical practice - guidance on indicators that your patient may have a genetic condition

General red flags for clinical practice

One or more of these red flags that may indicate a high genetic risk in your patient:

- The patient is unusually young to have the condition
- The condition does not commonly occur in patients of this sex
- There is an absence of typical environmental factors that are usually associated with the condition
- There is a strong family history of the condition
- The presentation is more severe than usual
- The patient is affected bilaterally in paired organs.

Red flags regarding cancer

Red flags for suspicion of an inherited breast cancer

- Breast cancer and/or ovarian cancer occurring in multiple relatives who are biologically related to each other
- Cancers occurring at an unusually younger age than typical in the general population (for example breast cancer occurring before the age of 40 years)
- Association of both breast and ovarian cancer in the same patient
- Bilateral breast cancer in one individual
- Breast cancer in a male.

Red flags for suspicion of an inherited bowel cancer

- Colorectal cancer in multiple family members who are biologically related to each other
- Colorectal cancer occurring at an unusually younger age than typical in the general population
- Cancers such as endometrial cancer (which is associated with colorectal cancer in some cancer syndromes) in family members or the patient
- Multiple primary cancers arising separately at different sites in the colon.

Red flags regarding cardiac disease

Red flags for suspicion of an inherited cardiac condition

- A patient who reports the sudden, unexplained death of a previously healthy young relative
- Family history of palpitations, arrhythmias, congestive heart failure at a young age, syncope or cardiac arrest
- A young patient with unexplained palpitations, syncope or seizures
- A patient reporting that a biological family member has been diagnosed with an inherited cardiac condition.

Red flags regarding reproduction and pregnancy

General information

- When there is a pregnancy, time is limited to provide the couple with counselling and, if they wish prenatal testing
- If either parent has a known genetic disorder, or if there is a known genetic disorder in the family, further specific genetic counselling and investigation may be required before any prenatal testing can be done
- The genetic department will need to know about the pregnancy as early as possible, preferably before week 10
- If a couple are planning a pregnancy, it is best to refer them to the genetics department before they conceive if at all possible.

Red flags for suspicion of a recessive condition that might affect the fetus

Any couple has a risk of having a child with an autosomal recessive condition. The chance of both parents being carriers of the same condition is increased when:

- There is a family history of the condition in one or both sides of the family
- The mother and father are biologically related (consanguineous relationship)
- The mother and father belong to an ethnic group with high risk for certain conditions
- There is a high number of carriers in the population to which the parents belong
- The mother and father come from a small population that has had little addition of genes with those from outside that population.

Red flags for suspicion of a genetic condition in a baby or child

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- The infant/ child has signs of developmental delay – in motor skills, speech and/or cognition,
- The infant/child has multiple congenital abnormalities
- The child has similar problems to other family members
- The infant/child has unusual physical (dysmorphic) features (or features very different from either parent)
- The child/infant has failure to thrive, or overgrowth
- The child/infant has a small or large head
- There is a history of recurrent spontaneous miscarriage in the family
- The infant/child has a combination of unusual problems.