Unit II – Problem 4 – Genetics: Genetic Counseling and Anticipatory Guidance

- **How would you define genetic counseling?**
  - It is a communication process between you (the doctor) and the family in which you explain and clarify the risks which are associated with a specific genetic disease and the risk of occurrence of that disease in their children. In addition, you will provide available options to deal with such a risk (e.g. prenatal screening, prenatal diagnosis through chorionic villus sampling or aminiocentesis and preimplantation diagnosis which is done with In-Vitro-Fertilization).
- **When do you need counseling or who are the people which you will counsel?**
  - Counseling of affected individuals and families which have children with congenital disorders.
  - Premarital counseling فحص ما قبل الزواج إجباري في مملكة البحرين
  - Preconception counseling (accounting the risk of having an affected child).
  - Counseling regarding consanguinity (which increases the risk of inheritance of autosomal recessive disorders).
- **Who is the genetic counselor?**
  - Any medical professional who is professionally qualified to provide genetic counseling (such as medical Geneticist) while the individual who is seeking for genetic counseling is known as consultand.
  - **Where would you find genetic counselors?**
    - ✓ Adult and pediatric clinics.
    - ✓ Prenatal genetic clinics.
    - ✓ Speciality clinics.
    - ✓ Public health genetic programs.
    - ✓ Human genetics research.
    - ✓ Public policy.
    - ✓ The biotechnology industry.
    - ✓ Human genetics education.
- **What are the stages of genetic counseling?**

History and pedigree construction	The affected individual who caused the consultand to seek advice is called the proband (which you must collect a full history about his condition and any other affected members in the family of the consultand).
Examination	-
Diagnosis (which must be accurate)	This is done through specific genetic investigations which include (karyotyping, DNA diagnosis… etc).
Counseling	Both parents should be counseled but notice that it is inappropriate to counsel after recent bereavement or after the initial shock of a serious diagnosis. Genetic counseling should be: non-directive, non-judgemental (you don't tell the parents what to do and you don't force them on something which they don't want) and respecting autonomy.

- **Misconceptions about heredity:**
  - Some think if the disorder is not present in other family member then it is not genetic.
  - Some think that genetic diseases cannot be treated or managed.
  - Some think that mental and physical stress of the mother while she is pregnant will cause malformations in the baby.

- **Pitfalls in genetic counseling:**
  - Incorrect or incomplete diagnosis.
  - Genetic heterogeneity.
  - Non-penetrance.
  - Variable expression.
  - Gonadal mosaicism.
- **Recurrence risk (quantification):**
  - If one of the parents is affected with an autosomal dominant disorder while the other is unaffected the risk of them to have and affected child in 50% in each pregnancy. Notice that there might be incomplete penetrance or variable expression.
  - If both parents are carriers of an autosomal recessive disorders the risk of them having an affected child is 25% in each pregnancy.
  - **In X-linked recessive traits:**
    - ✓ For each carrier (female):
      - ❖ 50% of her boys will be affected.
      - ❖ 50% of her daughters will be carriers.
    - ✓ If an affected male reproduce then all of his sons will be normal while all of his daughters will be obligate carriers for the disease.
- **Factors which are relevant to decision making process:**
  - The nature of the long-term burden.
  - The severity of the disorder.
  - If the condition can be successfully treated.
  - If the condition is associated with pain and suffering.
  - If prenatal genetic diagnosis is available.
  - If pre-implantation genetic diagnosis is available.
- **What are the common indications for genetic counseling?**
  - Previous child with multiple congenital anomalies or mental retardation.
  - Family history of a hereditary condition.
  - Advanced maternal age (for example, it is associated with increased risk of having a baby affected with Down syndrome).
  - Consanguinity (parents are relatives).
  - Teratogen exposure (which can cause congenital malformations).
  - Repeated pregnancy loss (which is usually due to aneuploidy).