### 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 amniocentesis 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.
- 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?

<table>
<thead>
<tr>
<th>Stage</th>
<th>Description</th>
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<tbody>
<tr>
<td>History and pedigree construction</td>
<td>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).</td>
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<td>Examination</td>
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<tr>
<td>Diagnosis (which must be accurate)</td>
<td>This is done through specific genetic investigations which include (karyotyping, DNA diagnosis... etc).</td>
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<tr>
<td>Counseling</td>
<td>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.</td>
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### 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 an 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 disorder 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).