Genetic counselling

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Genetic counselling - definition

- **The process by which patients or relatives at risk of a disorder that may be hereditary are advised of the consequences of the disorder, the probability of developing or transmitting it and of the ways in which this may be prevented, avoided or ameliorated.** Harper 1981

- **An educational process that seeks to assist affected and/or at risk individuals to understand the nature of the genetic disorder, its transmission and the options open to them in the management and family planning.** Kelly 1986

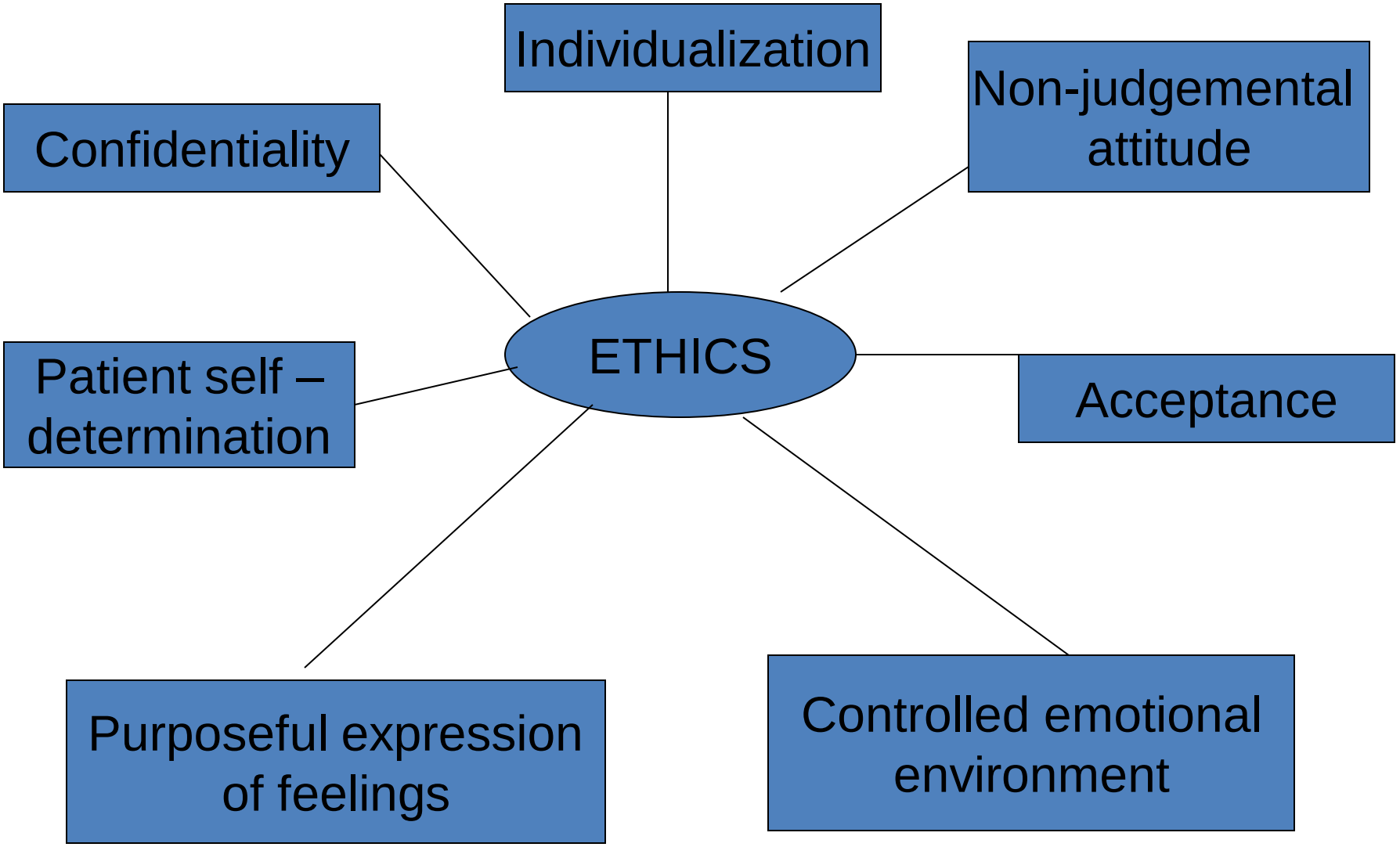
Aims of Genetic Counselling (WHO)

- 1. To comprehend the medical facts, diagnosis, prognosis and management regarding their condition
- 2. To appreciate the genetics of the disorder and risk of recurrence
- 3. To understand the options for dealing with the recurrence risk
- 4. To choose the appropriate course of action and carry out their choice
- 5. To make the best possible adjustment to the disorder

Procedure of Counselling

- Obtain history (family and ethnic information)
- Advise patients of the genetic risks to them and other family members
- Offer genetic testing or prenatal diagnosis
- Outline the various treatment or management options for reducing the risk

NB: Non-directive counselling


Individualization
Confidentiality
Non-judgemental attitude
ETHICS
Patient self – determination
Acceptance
Purposeful expression of feelings
Controlled emotional environment

Important aspects of counselling

- Autonomy
- Respect
- Educate
- Empower
- Support
- Advocate
- Facilitate

Counselling gives insight into:

- The individual
- Family values
- Cultural beliefs

Who can do counselling?

- Any person in the medical field who has enough knowledge.

  Doctors: Medical (Pediatricians and Obstetricians), Clinical Geneticists

  Nursing: Genetic nurses

  Genetic counselors

- Condition: They must know enough about the field

Purpose of Genetic counselling

- To provide information and support to families

1. Comprehend the medical facts including the diagnosis, probable course and management
2. Understand the way hereditary contributes to the disorder and the risk of recurrence
3. Understands the options for dealing with the risk of recurrence
4. Identify the values, beliefs, goals and relationships affected by the risk for a hereditary disease
5. Choose the course of action that seems the most appropriate to them
6. Make the best possible adjustments to the disorder or to the risk of recurrence by providing supportive counseling to families and making referrals to support services

Clinical case scenarios

- New born baby with Down syndrome
- Mother 16 weeks pregnant with a baby with a neural tube defect
- Five year old boy with Duchene muscular dystrophy
- 18 year old girl with a family history of Huntington's disease (mother just recently died from the condition)
- Mother who abuse alcohol and illicit drugs referred to you to evaluate her baby

“One gains knowledge when information is broken down and assimilated into the personality. Until this is done, information is like a tool which is useless because the person does not know how to handle it. Learning is not simply a matter of acquiring information. The learned person knows how to apply this information to life, especially to his own life. He has related it to his feelings and has integrated it with his experience”

Lowen 1970