



GENETIC COUNSELLING

Bertram Henderson
Div. Human Genetics
Universitas Hospital
FPA/UFS

GENETIC COUNSELLING

ASHG definition 1975

Genetic counselling is a communication and education process which deals with the human problems associated with the occurrence, or the risk of occurrence of a genetic disorder in a family. This process involves an attempt by appropriately trained persons to help the individuals or family to:

GENETIC COUNSELLING

- (1) Understand the *medical* facts, including the *diagnosis*, probable cause and available *management*;
- (2) Appreciate the *genetics* and risk of recurrence;
- (3) Understand the *options* for dealing with the risk of recurrence;
- (4) Choose an appropriate *course of action* and act upon the choice and
- (5) Make the best possible *adjustment*.

GENETIC COUNSELLING

NSGC definition 2006

Genetic counselling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. This process integrates the following:

GENETIC COUNSELLING

- Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.
- Education about inheritance, testing, management, prevention, resources and research.
- Counselling to promote informed choices and adaptation to the risk or condition.

SCENARIOS

- Pre test counselling – screening
- diagnostic
- Post test counselling
- After the termination of an abnormal fetus

COMMUNICATION

Sending, receiving & interpreting messages

- Verbal and non-verbal
- Cognitive level
- Emotional level
- Behavioural level

EFFECTIVE COMMUNICATION

Specific messages - verbal and non-verbal

- Practice active listening
 - Words (experience, feelings, behaviours)
 - Non-verbal (body language, facial expressions, tone of voice)

BARRIERS TO COMMUNICATION

- Interruptions
- Excessive questions
- Judgemental
- Criticizing
- Name-calling
- Impatience
- Lack of respect
- Assumptions
- Your needs
- Unresolved issues
- Medical jargon
- Advising
- Disempowering
- Control discussion
- Inadequate listening

STEPS IN GENETIC COUNSELLING


- **D**efine the problem
- **I**nform
- **A**llow the people to make their own decisions
- **S**upport

REQUIREMENTS of the counsellor

- Knowledge of the disorder (read up)
- Communication skills
- Empathy
- Self understanding (no personal baggage)
- Time
- Place
- Plan of action
- Respect for the family and their decisions

REQUIREMENTS of the counsellee

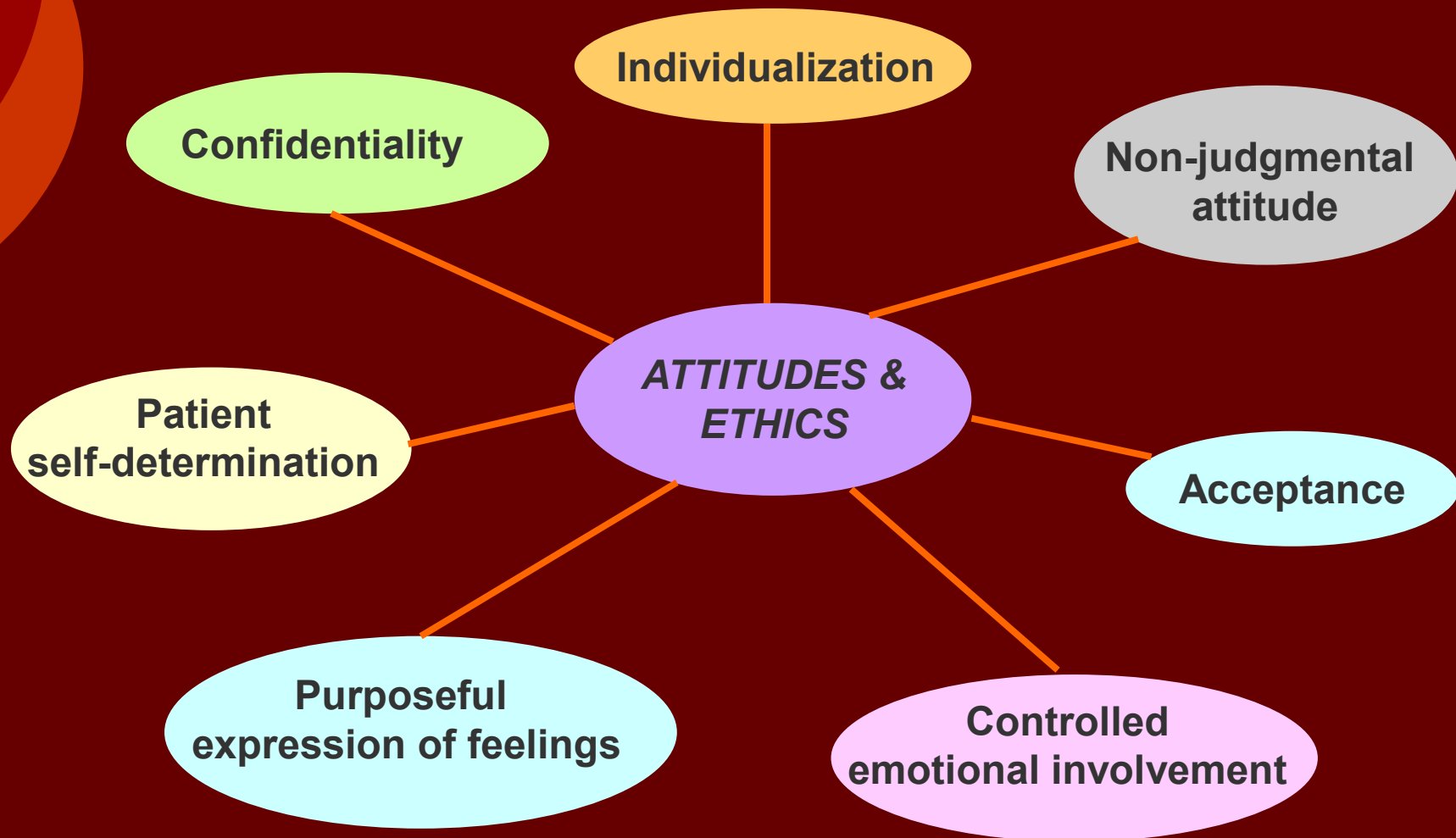
- Honest information
- Privacy
- Be part of the treatment
- Information in language they understand
- Time
- Be allowed to move at own tempo
- Allowed to ask questions

- 
-
- Be offered hope but not inappropriate expectations
 - Express own feelings
 - Allowed to proceed with the mourning process

PRINCIPLES IN GENETIC COUNSELLING

- **NON-DIRECTIVE**
- **AUTONOMY**
- **PROVIDING SUPPORT**

BASIC ATTITUDES AND ETHICS





PSYCHOSOCIAL ASPECTS

Understanding and appreciating the psychological effects of genetic disease to ensure more effective communication

FAMILY DYNAMICS

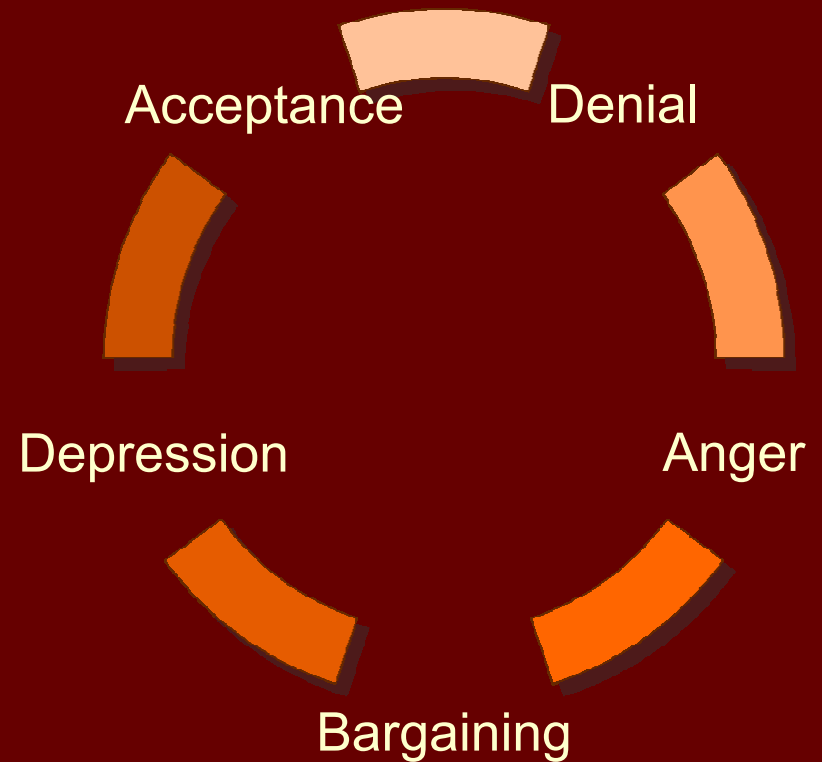
A family is a dynamic system of interacting individual personalities – whatever affects the individual affects the family

- **Family life cycle**
- **Family interaction**
- **Family functions**

BEREAVEMENT

Kubler-Ross 1970

- Denial
- Anger
- Bargaining
- Depression
- Acceptance



BREAKING BAD NEWS

1. The physical setting ought to be private, with both physician and patient comfortably seated. Minimize distractions and interruptions.
2. Decide who should be present, and let the patient also decide - studies show that different patients have widely varying views on what they would want.
3. Find out what the patient knows and what their general level of understanding is. We can assess the patient's emotional state and respond appropriately.

BREAKING BAD NEWS

4. Sometimes it is appropriate to find out how much the patient wants to know. Do they want to see photos of a very abnormal baby? This can help establish a two-way communication.
5. Inform the patient carefully and gently providing small amounts of information at a time.
6. Use appropriate language, few patients understand medical.

BREAKING BAD NEWS

7. Stop from time to time and check that they are following and understand, be prepared to repeat.
8. Answer questions honestly.
9. Most patients appreciate written information.
10. End with an invitation to return if they have further questions.