Genetic Counseling

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What is Genetic Counseling?

Genetic counseling involves a process of communication and education that addresses:

- Diagnosis, management and prognosis
- Estimation and communication of risks
- Explaining reproductive options

Steps in genetic counseling

- Diagnosis - based on accurate family history, medical history, examination and investigations
- Mode of inheritance and risk of developing/transmitting the disease
- Communicating the information to the families so that they make their own decisions in view of their risk and family goals and their ethical and religious standards
- Management and prognosis
- Options available for dealing with the risks
- Maintaining contact with families and follow up

Genetic Counseling

- Should be non-directive, non-judgmental
- Should provide accurate scientific and comprehendible information so that families take their own decisions (Autonomy)

Scope of genetic counseling

1. Counseling affected individuals and families having affected children with a congenital disorder
2. Premarital counseling
3. Prescreening counseling
4. Preconception counseling
5. Counseling regarding consanguinity

Number 1 is usually offered by a genetics specialist

Numbers 2-5 can be offered by trained primary health care providers

An individual who seeks genetic counseling is known as a *consultand*

The consultand should be provided with information that enables him or her to understand:

1. The medical diagnosis and its implications in terms of prognosis and possible treatment.
2. The mode of inheritance of the disorder and the risk of developing and/or transmitting it.
3. The choices or options available for dealing with the risks.

Genetic Counselor

- Genetic counselors work as members of a health care team, providing information and support to families with congenital disorders.

Role of genetic counselor

- Diagnose
- Explain risks of recurrence , prognosis and management
- Explain possible methods for prevention, including prenatal screening and diagnosis, and preimplantation diagnosis

- NOT DIRECTIVE

Role of genetic Counselor

Having established the diagnosis and discussed the risk of occurrence/recurrence, the counselor is then obliged to ensure that the consultands are provided with all the information necessary for them to make their own informed decisions. This should include details of all the choices open to them. For example, if relevant, the availability of prenatal diagnosis should be discussed, together with details of the techniques, limitations and risks associated with the various methods employed.

Risk assessment

- Quantification
- Qualification

Risk quantification

- Most prospective parents will be familiar to some degree with the concept of risks, but not everyone is comfortable with probability theory and the alternative ways of expressing risk, such as in the form of a percentage. Thus, for example, a risk of 1 in 4 can be presented as 25%.
- Consistency and clarity are important if confusion is to be avoided.
- It is essential to emphasize that a risk applies to each pregnancy and that chance does not have a memory. For example, the fact that parents have just had a child with an autosomal recessive disorder (recurrence risk equals 1 in 4) does not mean that their next three children will be unaffected.

Risk qualification

- The factor which most influences parents when deciding whether or not to have another child is the nature of the long-term burden, or severity, associated with a risk rather than its precise numerical value. A 'high' risk of 1 in 2 for a mild problem such as an extra digit (polydactyly) is not considered a major problem, whereas a 'low' risk of 1 in 25 for a disabling or fatal condition is not accepted. For example, a woman who grew up watching her brother develop Duchenne muscular dystrophy and subsequently die from the condition aged 20 years, may not risk having children even if there is only a 1% chance that she is a carrier.
- Other factors, such as whether a condition can be successfully treated, whether it is associated with pain and suffering, and whether prenatal diagnosis is available, will all be relevant to the decision-making process.

Communication

- The ability to communicate is essential in genetic counseling.
- Communication is a two-way process. The counselor should provide information, and has to listen to the fears and worries of the family.
- A readiness to listen is a key attribute for anyone involved in genetic counseling, as is an ability to present information in a clear, sympathetic and appropriate manner.

Some tips for risk communication (NCHPEG).

- Use the term "chance" instead of "risk," because "chance" connotes less of a value judgement of whether the outcome is good or bad.
- Provide the risk in different formats; for example, use both a percentage and a ratio (eg: 25% or 1 in 4).
- Consider also using natural numbers, which many people find more understandable than percentages and ratios. For example, "If there were 100 people in a room with the same chance you have, 25 of them would develop the illness."
- Give the chance for the "good" outcome as well as the "bad" outcome. For example, "While the chance is 25% for xyz to happen, there is a 75% chance that xyz won't happen."
- Put risk in context by comparing an individual's risk to the baseline risk. For example, "In the general population, 1 out of 11 women get breast cancer. In women with a family history like yours, on average 1 out of 8 get breast cancer."

Discussing options

- Carrier testing
- Prenatal diagnosis
- Preimplantation diagnosis

Discussing Options

- There are issues that should be approached with great care and sensitivity. For some couples the prospect of prenatal diagnosis followed by selective termination of pregnancy is unacceptable, whereas others view this as their only means of ensuring that any children they do have will be healthy.
- Whatever the personal views of the counselor, the consultands are entitled to knowledge of prenatal diagnostic procedures that are both technically feasible and locally permissible.

In a genetic counseling setting

- Often the couple will be extremely upset when first made aware of a genetic diagnosis in their child, and it is very common for guilt feelings to set in, with scrutiny of every event during the pregnancy
- Genetic counselors need to take into account the complex psychological and emotional factors that can influence the counseling dialog and the delivery of distressing information.
- The setting should be agreeable, private and quiet, with ample time for discussion and questions.
- When possible, technical terms should be avoided or, if used, fully explained.
- Questions should be answered openly and honestly, and if a definite diagnosis cannot be reached, it is important to say so.

Genetic counselors are sometimes asked what they would do if placed in the consultand's position.

- Generally it is preferable to avoid being drawn into expressing an opinion, opting instead to suggest that the consultand try to imagine how he or she might feel in the future having pursued each of the available options. This approach, sometimes referred to as 'scenario-based decision counseling', provides individuals with an opportunity for careful reflection. This is particularly important if one of the options under consideration involves a potentially irreversible reproductive decision such as sterilization. There is a well-established maxim that it is the consultands and not the counselors who have to live with the consequences of their decisions and, indeed, consultands should be encouraged to make the decision that they can best live with- the one that they are least likely to regret.

Follow up

- A counseling session can be so intense that the amount and accuracy of information retained on follow-up at a later date can be very limited.
- A letter summarizing the topics discussed at a counseling session is ideally sent to the family afterwards.
- The family could be contacted at a later date by a member of the counseling team thereby providing an opportunity for clarification of any confusing issues and for further questions to be answered.

Ethical issues in genetic counseling

- **Autonomy:** the couple should take their own decision
- **Informed choice:** the decision is based on the information given by he counselor
- **Informed consent:** The individual or couple should give their informed consent for any investigation
- **Confidentiality**

Main Services Offered by a Medical Genetics Centre

- Genetic counselling to families reporting to the centre
- Laboratory services for the diagnosis of genetic disorders (cytogenetics, molecular genetics, biochemical tests)
- Training of health professionals on the principles and practice of medical genetics
- Research
- Development of community genetics services for the control of genetic disorders in the community

Genetic counseling at the primary health care level

Genetic counseling approaches appropriate at the primary health care level

- Preconception counseling information
- Screening and pre-screening counseling for premarital carrier detection programs and neonatal screening
- Post-screening counseling for couples not at risk and referral of at risk couples
- Identification and referral of individuals and families at increased genetic risk to specialized centers
- Counseling related to consanguinity and referral when indicated

Role of primary health care worker in basic community genetics

- Taking and recording a basic genetic family history, including pedigree construction for large consanguineous families
- Knows how to detect families with genetic risks and where and when to refer them
- Knows the basic ethical principles and techniques of genetic counseling
- Offers preconception counseling
- Offers prescreening counseling, and knows how to detect at risk couples or individuals and with appropriate referral to special centers
- Knows the advantages and disadvantages of consanguineous marriages
- Knows the common genetic disorders in the community
- Knows how to access appropriate information materials for patients, families and the general public
- Knows the existing specialist services and patient and family support organizations
- Knows the genetic programs that would be introduced on the national level

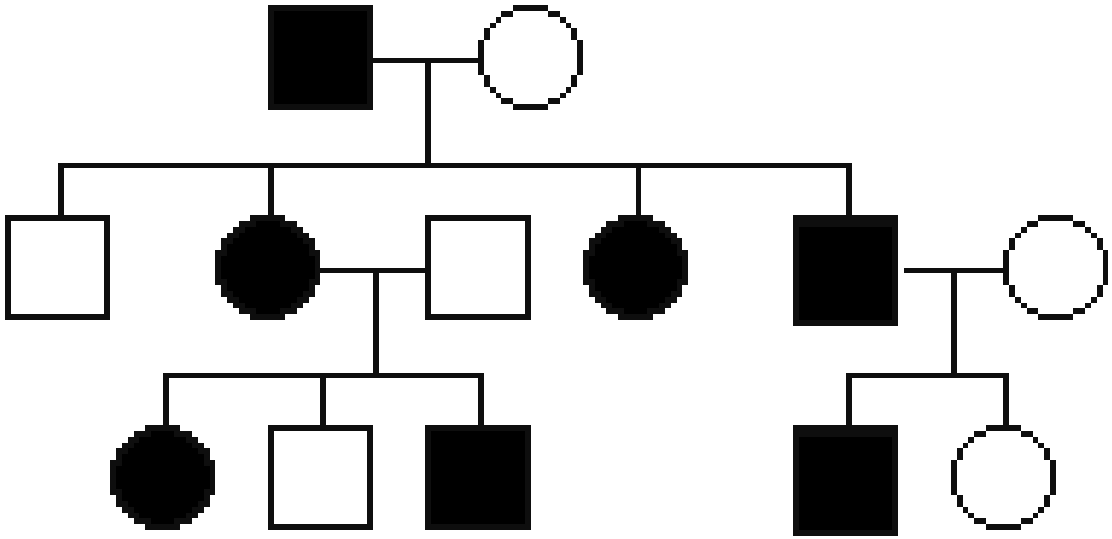
Premarital counseling regarding consanguinity at the primary health care level

- Detailed family history should be taken to recognize any congenital disorder
- Ask about any known inherited disease in the family, history of birth defects, hearing or vision impairment, mental retardation ,any unexplained neonatal or infant deaths and failure to thrive among infants
- If a genetic disorder is suspected in the family, refer the couple to a specialized genetic counseling clinic
- If there is no known inherited disorder in the family, first cousin marriages are given a risk for birth defects of double the population risk (for example instead of 3%, it becomes 6%)

Preconception counseling to maximize the chances of a couple to have a healthy baby

1. Preconception nutritional supplementation for example proper intake of folic acid can minimize the risks for neural tube and other defects
2. Prevention and treatment of maternal infections (Rubella, TORCH, Syphilis)
3. Diagnosis of maternal Rh status
4. Information on risks of advanced maternal age at conception
5. Avoidance of teratogenic drugs and chemicals
6. Cessation of smoking and alcohol intake
7. Family planning
8. Monitoring of maternal health before and during pregnancy (diabetes, hypertension, epilepsy, hyperthermia)

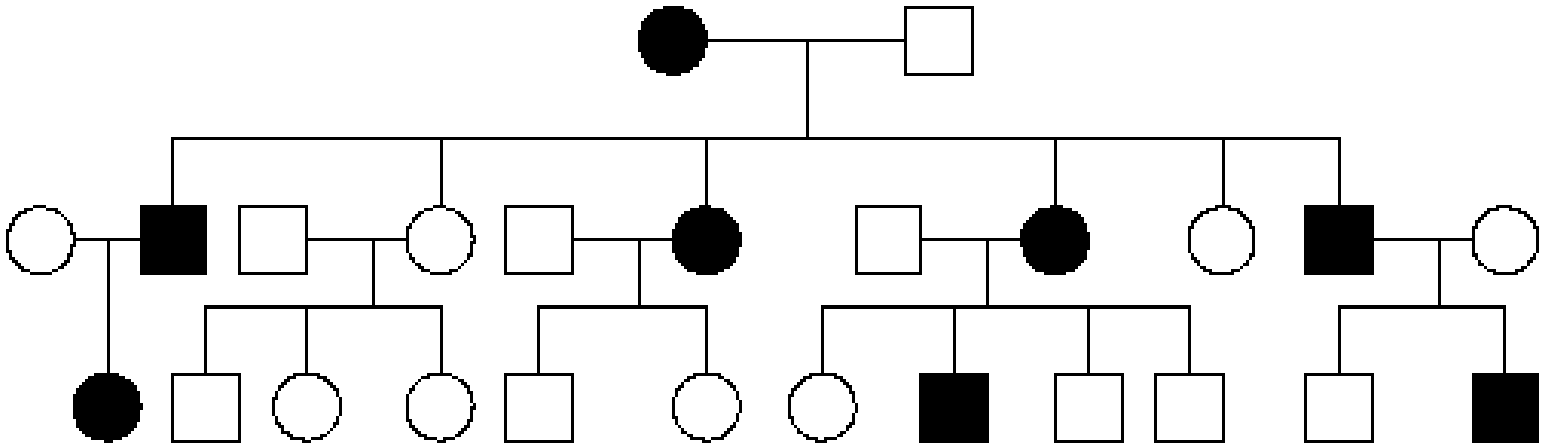
What is the mode of inheritance in the following pedigrees?



Solution: 1

Most probably this pedigree shows a condition that is inherited in an autosomal dominant manner because there are affecteds in all generations (vertical transmission), both males and females are affected, and an affected father has an affected son which excludes X-linked dominant inheritance

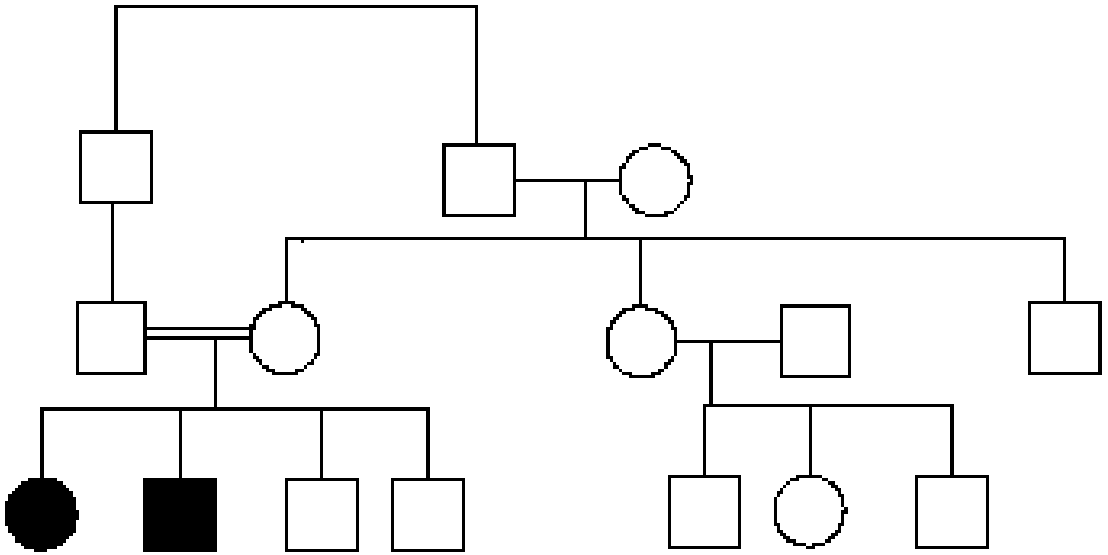
2



Solution: 2

Most probably autosomal dominant

3

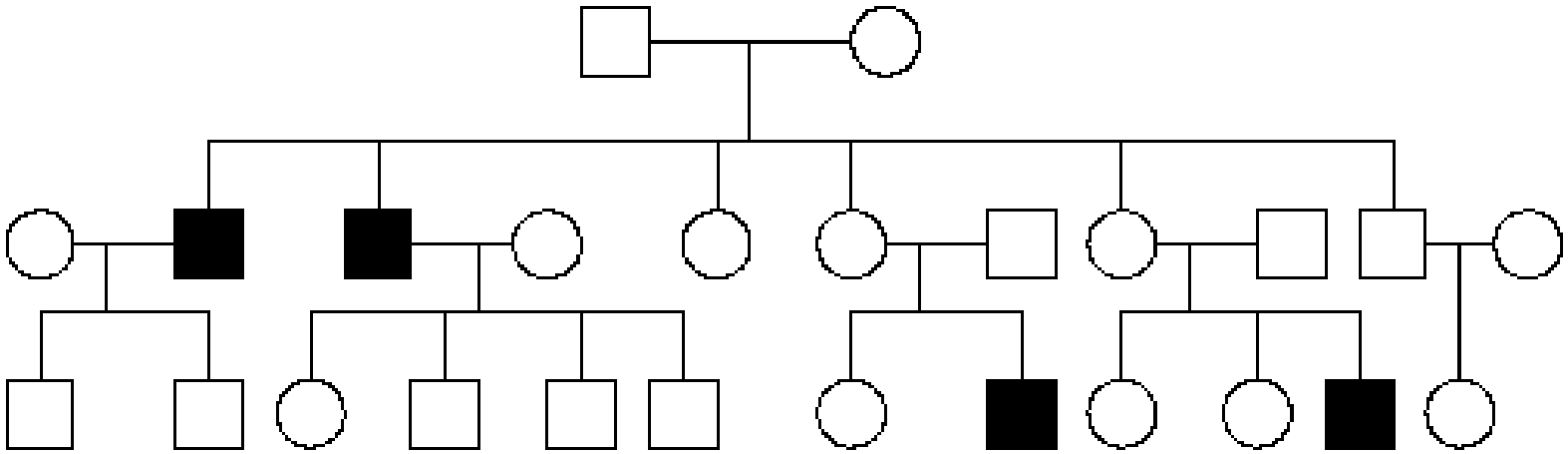


Solution:3

The pedigree here shows affecteds in the same generation while the parents are not affected (horizontal transmission), and are consanguineous (related as shown by the double line drawn between them). The condition is affecting both males and females.

Most probable mode of inheritance is autosomal recessive.

4

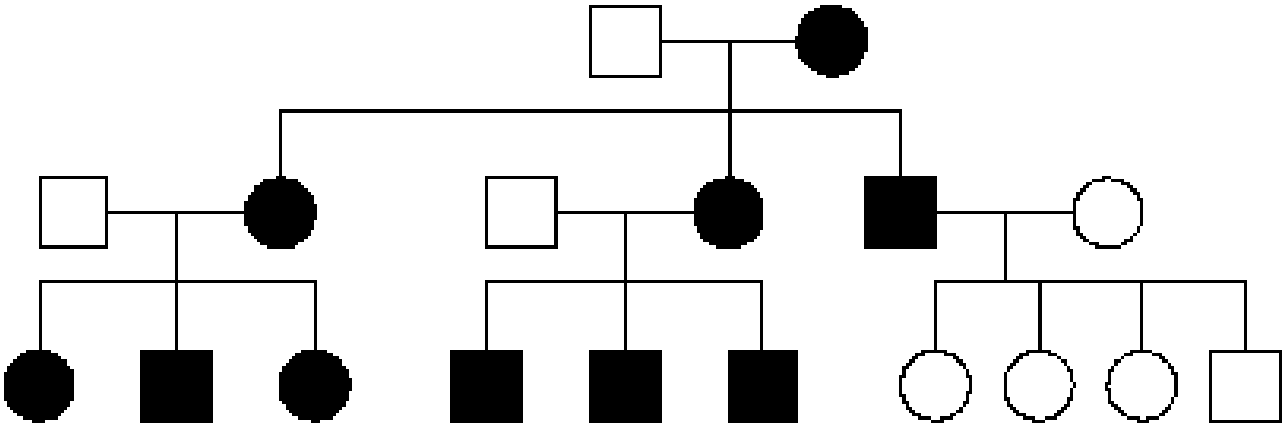


Solution :4

In this pedigree, the affecteds are all males and they are linked together by unaffected females (a female has her brothers and sons affected).

This pedigree shows a condition that is inherited as an X-linked condition.

5



Solution:5

This pedigree shows that all children of an affected female are affected, while children of the affected male are not affected.

It points to a maternal transmission: mitochondrial inheritance. Mitochondria are in the cytoplasm and only maternal cytoplasm from the ovum is passed to the zygote.

Risk calculation

Examples

1

Mr X is affected by Leber's hereditary optic neuropathy (LHON), characterized by rapid optic nerve death, leading to bilateral blindness. Subsequent molecular studies reveal the causative mutation in a mitochondrial gene. Mr X has two daughters. What is the probability that both daughters have inherited this mutation?

A) Virtually 0%

B) Approximately 25%

C) Approximately 38%

D) Approximately 50%

E) Approximately 75%

Solution:1

Since the causative gene is mitochondrial, it means that it can be passed to the children only from the mother and not from the father.

Correct answer is A

2

Mr E is a 28-year-old man with a younger brother who died from type 2 Gaucher disease, an autosomal recessive lysosomal storage disorder that is fatal by the age of 2 years. What is the probability that Mr E is a heterozygous carrier for the Gaucher mutation?

A) 0.25

B) 0.33

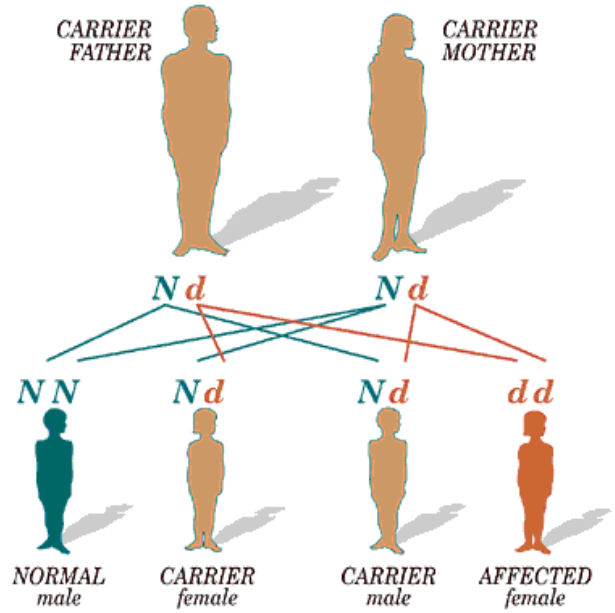
C) 0.50

D) 0.67

E) 0.75

Solution:2

When a child is affected by an autosomal recessive condition, usually both parents are carriers of the abnormal allele.



Look at the children of this couple, $\frac{1}{4}$ is affected (dd), 3 look unaffected and among them 2/3 are carriers (Nd), and 1/3 is not a carrier (NN)

Answer is D which is 0.67 or 2/3

Conclusions
Genetic counseling involves:

- Medical diagnosis, prognosis and treatment
- Mode of inheritance and risk of recurrence
- Options available for dealing with the risks
- Communicating the information to the couple so that they make their own decisions and reproductive options in view of their risk and family goals and their ethical and religious standards
- Should be non-directive, non-judgemental