KING SAUD UNIVERSITY
COLLEGE OF MEDICINE
FOUNDATION BLOCK

Lecture 5:
Genetic counseling

HUMAN GENETICS 433

Lecture objectives

- Understand the principle steps of **genetic counseling.**
- Understand unique features of genetic counseling in **Arabic/Islamic communities**.
- Be familiar with the general application of Hardy-Weinberg principle

Color index:

Red → important

Yellow → notes

Green → explanation

If you have any questions please contact us:
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What is genetic counseling?

It is a process of communication and education which addresses concerns related to the development and/or transmission of a hereditary disorder

Steps of genetic counseling

- Diagnosis
- Risk assessment
- Communication
- Discussion of options
- Long term contact & support

Where do Genetic counselors work?

- The majority work at:
  - University medical centers.
  - Private or public hospitals.
- Some:
  - Work in laboratories.
  - Coordinate research studies.
  - Are employed by the state.
  - Work in private industry.

Consultant (the person who asks advice in a consultation):

1) Seeks counseling.
2) Information to understand.
3) Reach their own fully informed decisions without pressure or stress.

Counselor:

1. The diagnosis, prognosis, & possible treatment
2. The mode of inheritance & the risk of developing/ transmitting
3. The choices/options available

Steps of genetic counseling:

i. Diagnosis: based on accurate family history, medical history, examination and investigation.
ii. Risk assessment
iii. Communication
iv. Discussion of options
v. Long-term contact and support

i-Establishing the Diagnosis:

1.History:

detailed information about the patient's family history (3-generations family tree)

2.Examination

3.Investigation:

chromosome and molecular studies

referral to specialists in other fields (e.g. neurology and ophthalmology)

ii-Calculating and presenting the risk (risk assessment)

Calculation of the recurrence risk:
Can be **straightforward** (Mendelian inheritance).
Or..Can be **much more complex**, due to many factors, for example:
delayed age of onset

iii-Communication:

Be Consistent & clear to avoid confusion

- Genes are made up of DNA molecules, which are the simplest building blocks of heredity.
- They're grouped together in specific patterns within a person's chromosomes, forming the unique "blueprint" for every physical and biological characteristic of that person.
- Example : there is a risk of 1 in 4 to have affected childe; that means : 25% chance to get an affected child .

- **Emphasize that a risk applies to each pregnancy and that chance does not have a memory.**

Example: a couple has a child with an autosomal recessive disorder (recurrence risk equals 1 in 4).
That means that:

✗*A*- their next three children will be unaffected.

✓*B*- Each of their future children will have a recurrence risk of 1 in 4.

- **Emphasize the good side of the coin.**

Example: If a couple is faced with a probability of 1 in 25 that their next baby will have a neural tube defect, the counselor should tell them that:

✗*A*- there 1 chance out of 25 that their next baby will be affected.

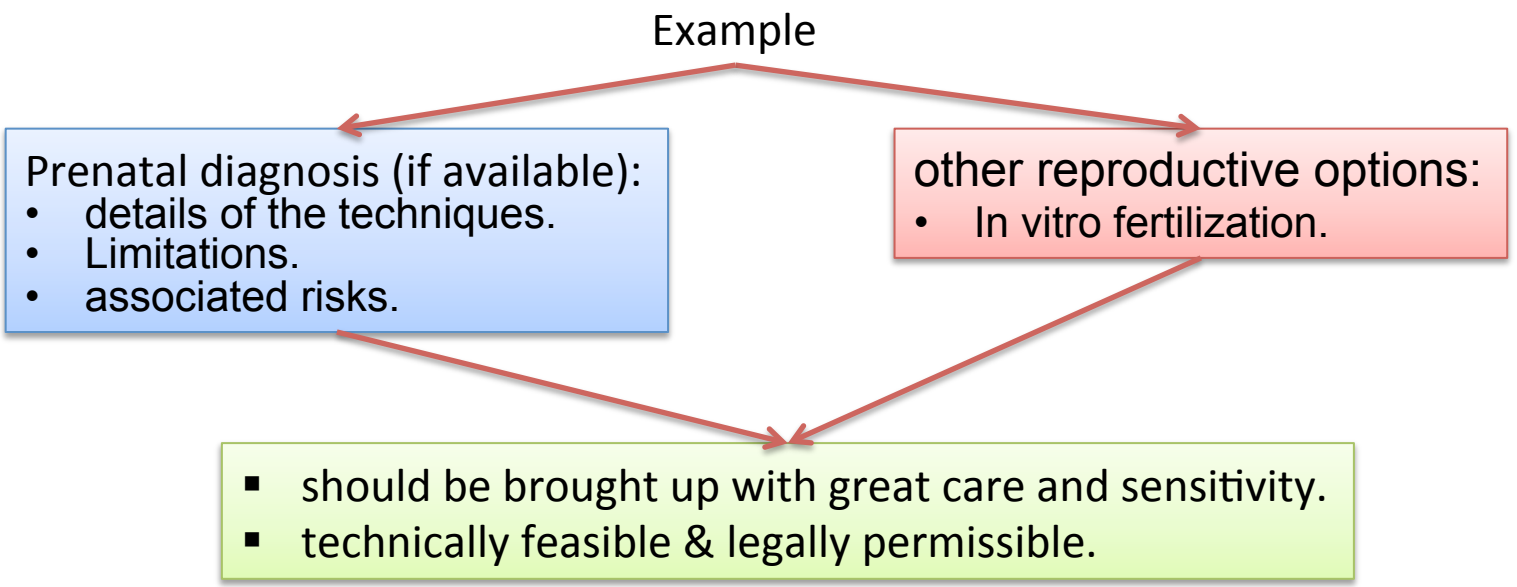
✓*B*- there are 24 chances out of 25 that their next baby will not be affected.

Communication is a two-way process

As a genetic counselor, be ready to:

- Listen
- Present information in a clear, sympathetic and appropriate manner
- take into account the complex psychological and emotional factors
- Offer an opportunity for further discussion and long-term support
- Create a network of genetic nurse counselors keeping genetic registers
- Offer contact with “Patient support groups”

iv-Discussion of options

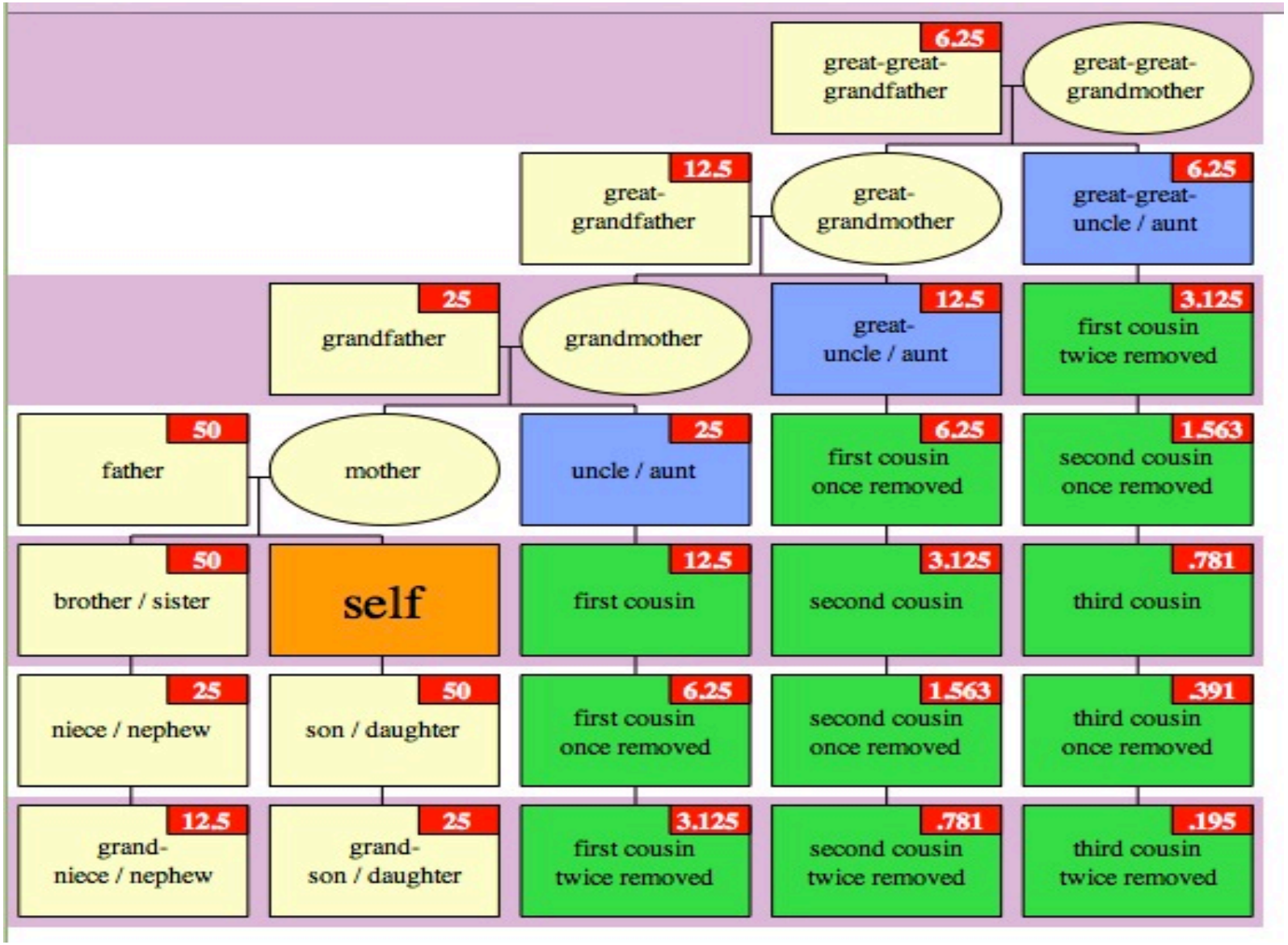


Unique features of genetic counseling in Arabic/Islamic communities:

- Consanguineous marriage (parents are related) is customary in the Middle East and parts of South Asia.

Population of children studied*	% of parents related	Prevalence of recessive disorders
Northern European	0.4	0.28%
British Pakistani	69	3.0 -3.3%

Proportion of nuclear genes shared as a function of degree of relationship



While discussing the options

The availability op prenatal diagnosis & other reproductive options should be :

- Brought up with a great care and sensitivity
- Religious & legally permissible
- Technically feasible

The frequency of alleles (The Hardy-Weinberg Principle)

- Mathematical relationship between allele frequencies and genotype frequencies.
- The ***frequency of genotypes*** between individual mating can be predicted using the ***Punnett square.***
- The frequency of particular ***alleles*** based on frequency of a phenotype within a population can be calculated by the ***Hardy-Weinberg principle.***

- For normal allele (A) : the frequency in the population is p
- For the mutant allele (a): the frequency in the population is q
- Because there are assumed to be only 2 alleles, p + q = 1

The frequency of:

the homozygote AA = p2

the heterozygote Aa = 2pq

the mutant homozygote aa = q2

	p	q
p	pxp	pxq
q	pxq	qxq

$$p^2 + 2pq + q^2 = 1$$

Explanatory slide:

Keeping in mind that the Hardy-Weinberg equations are: $p + q = 1$,
$p^2 + 2pq + q^2 = 1$ lets take an example:
The ability to role the tong is a dominantly inherited feature i.e. people with (RR,Rr) genotype can role their tongs and people with (rr) genotype can't.
-**In a population 16% cannot role their tongs what is the frequency of Rr(heterozygous)?**

q^2=16%=0.16
q=0.4
p= 1-q =0.6
Since $p^2+2pq+q^2=1$
and since 2pq represents the heterozygous
then 2pq=2(0.4)(0.6)=0.48

Ans: The frequency of Rr(heterozygous) = 0.48 = 48%

- Remember that

P+q=1, So:

✓ P=1-q

✓ q=1-p

For a population to be in Hardy-Weinberg equilibrium, the following conditions must be met:

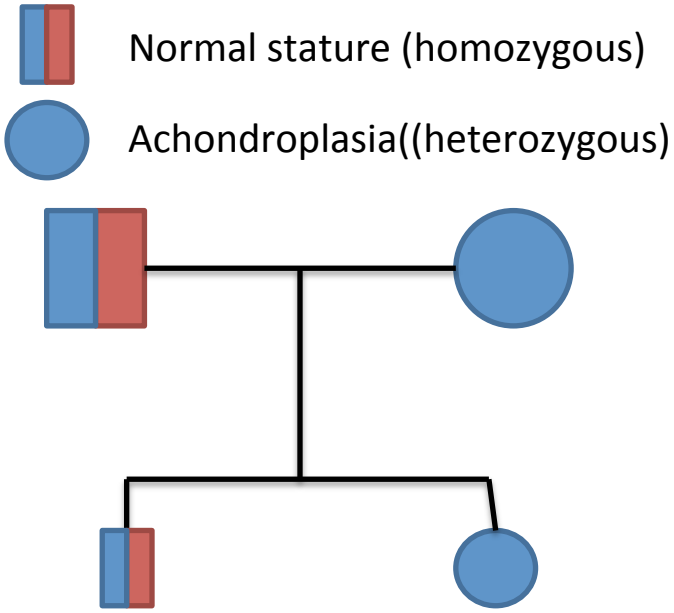
1. Random mating
2. Constant mutation rates
3. Large population sizes
4. Absence of migration

Genetic Counseling in Achondroplasia

- It is inherited in an autosomal dominant manner.
- Homozygous achondroplasia is a lethal condition.
- > 80% of achondroplasia cases have parents with normal stature i.e.: new gene mutation.
- Such parents have a low risk of having another child with achondroplasia.
- Prenatal molecular genetic testing is available.

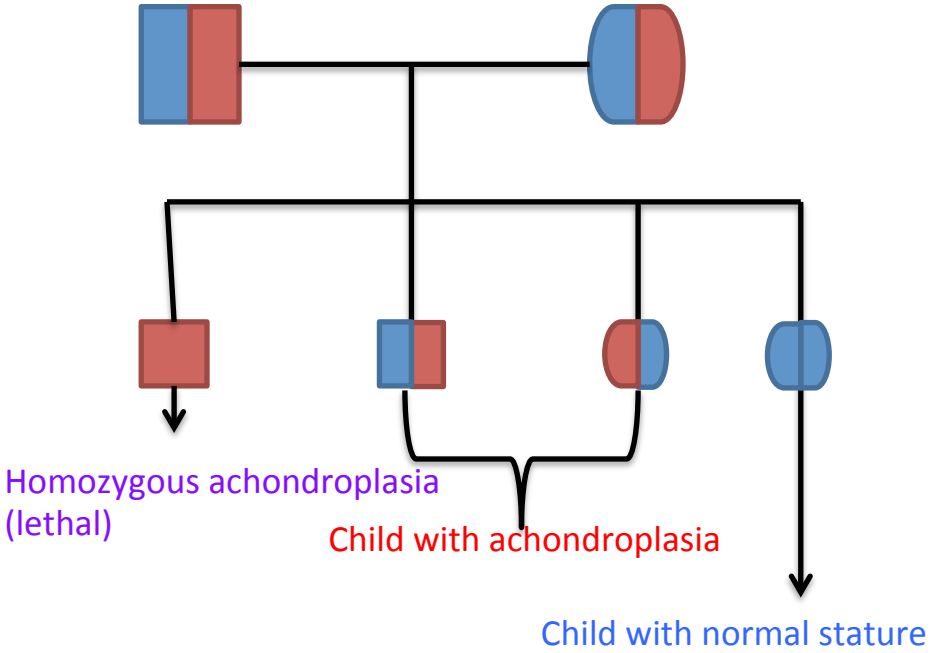
Genetic counseling cases

- An individual with achondroplasia who has a reproductive partner with normal stature has a 50% risk in each pregnancy of having a child with achondroplasia.



When both parents have achondroplasia, the risk to their offspring of having:

- normal stature: 25%
- achondroplasia: 50%
- homozygous achondroplasia (lethal): 25%



MCQs

1) Which of the following is the Hardy-Weinberg equation:

A. $p^2 + 2pq + q^2 = 1$
B. $p^2 + 2pq + q^2 = 2$
C. $p^2 + 2pq - q^2 = 1$

2) The following are all the condition of the Hardy-Weinberg equation except:

1. Random mating
2. Presence of Immigration
3. Constant mutation rates
4. Large population sizes

3) How much genes do you share with your first cousin:

A. 50%
B. 25%
C. 6.25%
D. 12.5%

4) In which step is prenatal diagnosis is discussed:

A. Diagnosis
B. Communication
C. Discussion of options
D. Risk Assessment

5) Which of the following is true about homozygous achrondoplasia

A. Extreme short stature
B. Just like heterozygous achrondoplasia
C. Lethal
D. Child having Normal stature

Answer Key

1.A 2.B 3.D 4.C 5.C

And by that we finish genetics for the foundation block

GOOD LUCK.

Human genetics team

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