Medical Genetics

Sheet: 11 - Risk Assessment

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Risk assessment

In this sheet, we are going to solve questions that will demonstrate the idea of risk assessment, take a minute to solve the question by your own and then look at the answer.

Question 1: what is the probability that if a couple has 4 children all will be males?

Answer :-

[Probability of Male = 1/2]

All males means (the first child is male, the second child is male, the third child is male and the fourth child is male).

Probability (all males) = 1/2 x 1/2 x 1/2 x 1/2 = 1/16

For the same question: what Is the probability that the 4 children all will be females?

Answer :-

[Probability of female = 1/2]

All females means (the first child is female, the second child is female, the third child is female and the fourth child is female).

Probability (all females) = 1/2 x 1/2 x 1/2 x 1/2 = 1/16

Again, for the same question: what Is the probability that the 4 children all will be the same gender?

Answer :-

The same gender means that the 4 children are Either (4 females **OR** 4 males)

Probability (same gender) = 1/2 x 1/2 x 1/2 x 1/2 + 1/2 x 1/2 x 1/2 x 1/2 = 2/16=1/8

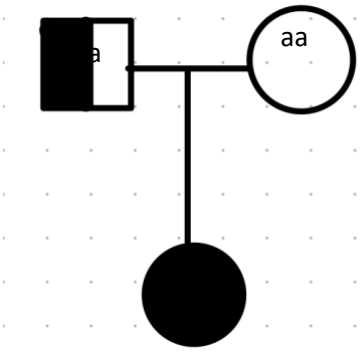
note : when we use AND ; It means that we multiply the probabilities, whereas when we use OR we add the probabilities .

Question 2: A women has a father who died of Huntington's disease, what Is the probability that she will develop the symptoms of the disease?
(For these kinds of questions draw a pedigree)

Answer :-

* Remember Huntington's disease is Autosomal dominant disease.

* Use **A = Dominant allele, a = Recessive allele** for the disease.



-Regarding the pedigree

Remember that we assume that the affected father Is heterozygous **(Aa)**, also we assume that the partner (the mother here) Is not affected unless stated otherwise **(aa)**.

-Using Punnett square :-

Dad / Mom	***A***	***a***
a	***Aa***	***aa***
a	***Aa***	***aa***

Probability (that the female child will develop the symptoms of the disease) =2/4= 1/2

Question 3: A couple are both tested and found to be carriers of the cystic fibrosis gene, If they have 2 children :-

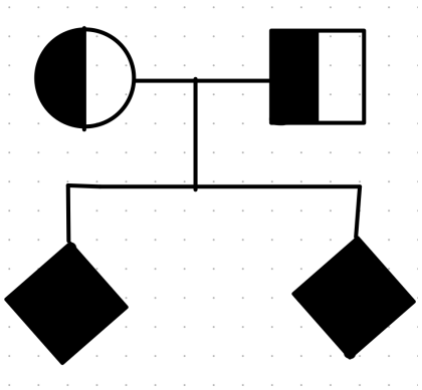
A- what Is the chance that both children will be affected by cystic fibrosis?

Answer :-

* Remember that cystic fibrosis Is autosomal recessive disease.

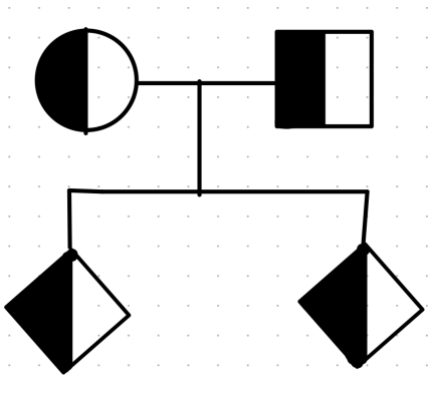
* Use **A= dominant allele, a = recessive allele.**

Dad / Mom	***A***	***a***
A	***AA***	***Aa***
a	***Aa***	***aa***



Probability (that both children will be affected by cystic fibrosis, the first child is affected AND the second child is affected) = 1/4 x 1/4 = 1/16

B-what Is the chance that both will be carriers of cystic fibrosis?



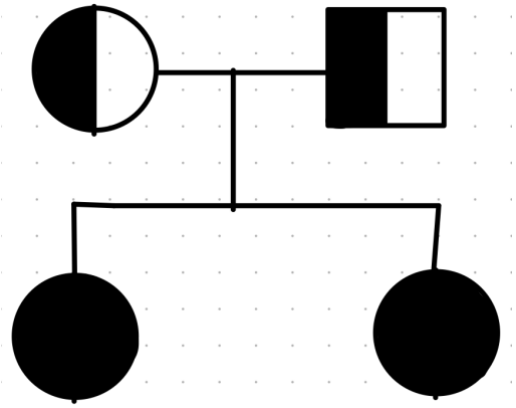
Dad / Mom	***A***	***a***

A	***AA***	***Aa***
a	***Aa***	***aa***

Probability (that both children are carriers of cystic fibrosis, the first child Is a carrier AND the second child Is a carrier) = 1/2 x 1/2 = 1/4

C- what Is the chance that they will have 2 girls that are both affected by cystic fibrosis?

Dad / Mom	***A***	***a***
A	***AA***	***Aa***
a	***Aa***	***aa***



Probability (that both children are affected girls, the first child Is an affected girl AND the second child Is an affected girl) = 1/4 x 1/2 x 1/2 x 1/4 = 1/64

* Note we multiply the probability to have an affected child (1/4) by (1/2) because the question has specified the gender.

Question 4: The ability to taste the chemical (PTC) Is determined by a single gene In humans with the ability to taste given by the dominant allele (T) and inability to taste by the recessive allele (t). Suppose two heterozygous tasters (Tt) have a large family.

A-Predict the proportion of their children who will be tasters and non-tasters. Use a Punnett square to illustrate how you make these predictions.

Dad / Mom	***T***	***t***
T	***TT***	***Tt***
t	***Tt***	***tt***

Proportion of their children who will be tasters = 3/4 (75%)

Proportion of their children who will be non-tasters = 1/4 (25%)

B- what Is the likelihood that their first child will be a taster? What Is the likelihood that their fourth child will be a taster?

Dad / Mom	T	t
T	TT	Tt
t	Tt	tt

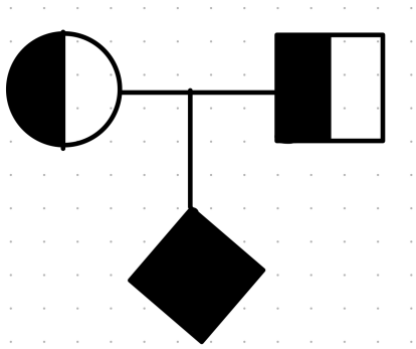
Probability (that their first child will be a taster) = 3/4

Probability (that their 4th child will be a taster) = 3/4

* Remember that the probabilities of each pregnancy Is **Independent** from the probabilities of the previous pregnancies.

Question 5: For a Caucasian couple with no family history, what Is the chance they will have a child with CF (Cystic fibrosis)? Knowing that caucasian population carrier frequency Is 1/25.

Dad / Mom	A	a
A	AA	Aa
a	Aa	aa



Answer :-

* Remember that Cystic fibrosis Is an autosomal recessive disease.

* use **A= Dominant allele, a = recessive allele.**

- In this question It doesn't tell us that the couple tested for CF neither if one or both of them are affected, but the red note mentioned In the question gives us a clue (carrier frequency of 1/25; means that out of 25 people one will be carrier).

-Remember that we assume that the couple aren't affected unless otherwise Indicated, so for these unaffected couple to have an affected child **(aa)** they must be at least both heterozygous **(Aa,Aa)**, to pass on the affected allele.

So, now we have three probabilities =

Probability for the mom to be a carrier AND Probability for the dad to be a carrier AND Probability to have an affected child = 1/25 x 1/25 x 1/4 = 1/ 2500

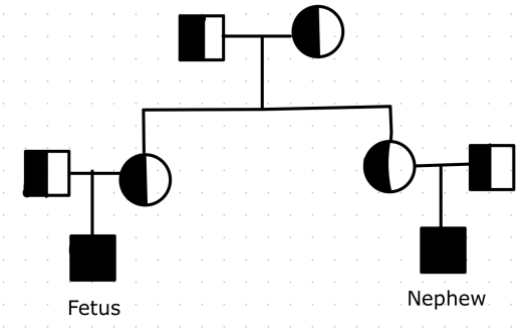
Question 6: Couple with diagnosis of Cystic fibrosis in mother's nephew, what Is risk to fetus (to be affected by CF)? Carrier frequency Is 1/25

* Remember that Cystic fibrosis Is an autosomal recessive disease.

*Use **A= dominant, a= recessive.**

- Here, when there Is a family history (In the mother's side) we discard the carrier frequency for her.

-So lets draw the pedigree first :-



1-Regarding the nephew (nephew means: a son of one's brother or sister) :-

-He Is an affected child (aa), each of his parents must have the affected allele (a).

- Regarding his father, we assume he Is heterozygous (Aa)

-Regarding his mother (the sister of the Fetus's mother) we assume she Is heterozygous not affected; because It Is not mentioned in the question that she Is affected (Aa).

2- Regarding the grandparents, the affected allele (a) present In the nephew's mother must be passed on from **one** of her parents **(Either the mom or the dad but not from both)**, so the probability that the grandfather will be a carrier Is **1/2** and the probability that the grandmother will be a carrier Is **1/2.**

3- Regarding the Fetus :-

-He Is an affected child (aa), each of his parents must have the affected allele.

-Regarding his father, we assume that he Is heterozygous (Aa).

-Regarding his mother we assume that she Is heterozygous (Aa) , now the probability that she will be a carrier Is = (the probability that she Inherited the mutant allele from her father **OR** from her mother) **1/2** (the odds that the father Is a carrier) **x 1/2** (the odds to pass the mutant allele from the father to his daughter) **+ 1/2** (the odds that the mother Is a carrier) **x 1/2** (the odds to pass the mutant allele from the mother to her daughter) **= 1/2**

*** NOTE: if my niece or nephew are affected with a recessive disease the odds that I will be a carrier is ½ .**

So, what is the probability that the fetus is affected (the probability that the mother is a carrier AND the probability that the father is a carrier AND the probability that the fetus is affected) = **1/2 x 1 /25 x 1/4 =1/200**

Good luck