ثانوية الامام الصدر الرسمية – صور

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Risk Calculation

Always start by going back to remember and be sure about the mode of inheritance of the studied disease.

Autosomal recessive

Conditions and probabilities:

case A: parents are both normal:

1- both should be heterozygous

2- the child should inherit the allele of the disease from each of them.

Probability of a normal individual to be heterozygous:

i - a normal individual having an affected parent or child is 100% heterozygous; P = 1

ii - a normal individual having an affected sibling has the probability 2/3 to be heterozygous; P = 2/3

iii - a normal individual with no family history, the probability of heterozygotes in the population should be given.

example: for cystic fibrosis, P = 1/20

Probability of a child to inherit the allele of the disease from a heterozygous parent is ½ (the heterozygous parent produces two different equiprobable types of gametes.

Risk = P (father to be heterozygote) × P (mother to be heterozygote) × P (child to inherit 2 alleles of the disease; one from each parent)

X-Linked recessive

Conditions and probabilities

case A: parents are both normal:

- the mother should be heterozygous
- the child should inherit the X^d from the mother and Y from the father.

Probability of the mother to be heterozygous:

- a normal mother having an affected parent or child is 100% heterozygous; P = 1
- a normal mother having an affected sibling has the probability ½ to be heterozygous; P = ½
- a normal mother with no family history, the probability of heterozygotes in the population should be given.

Probability of a child to inherit X^d from a heterozygous mother is ½ (the heterozygous mother produces two different equiprobable types of gametes.

Probability of a child to inherit Y chromosome from the father is ½ .

Risk = P (mother to be heterozygote) × P (child to inherit X^d from the mother) × P (child to inherit Y from the father).

- in case it is sure that the child is a boy, then the probability to inherit Y becomes 1.
- The risk for the couple in this case (both parents are normal) to have an affected girl is zero since she inherits X carrying the normal dominant allele from her father.

Risk Calculation

Autosomal recessive

Conditions and probabilities:

case B: one parent is affected and the other is normal:

1- the normal parent should be heterozygous

2- the child should inherit the allele of the disease from both of them.

Probability of a normal individual to be heterozygous

is as mentioned before: P = 1, or P=2/3, or given.

Probability of a child to inherit the allele of the disease from a heterozygous parent is ½ (the heterozygous parent produces to different equiprobable types of gametes.

Probability of a child to inherit the allele of the disease from the affected parent is 1. (the affected parent should be homozygous to express the recessive allele of the disease, thus produces only one type of gametes carrying the allele of the disease).

Risk = P (normal parent to be heterozygote) × P (child to inherit an allele of the disease from the normal parent if heterozygote) × P (child to inherit the allele of the disease from the affected parent).

X-Linked recessive

Conditions:

case B: the father is affected and the mother is normal:

- the mother should be heterozygous
- the child should inherit the allele of the disease from the mother (X^d). (in this case, the child will be affected whatever he/she inherit from father).

Probability of the mother to be heterozygous is mentioned before:

in this case, the risk = probability of the mother to be heterozygote × probability of the child to inherit X^d from the mother which is ½ .

Let's consider that the mother has affected sibling and both of her parents are normal in this case she has a probability ½ to be heterozygote and ½ to transmit X^d to her child.

Risk = ½ × ½ = ¼

Clarification: the child must inherit X^d from the mother and Y from the father OR X^d from both.

P(to inherit Y from the father) = ½

P(to inherit X^d from the father)= ½

Risk = ½ × ½ × ½ + ½ × ½ × ½ = 1/8 + 1/8 = ¼

Risk = P(mother to be heterozygote) × P(child to inherit X^d from the mother) **OR** Risk = P (mother to be heterozygote) × P (child to inherit X^d from the mother) × P (child to inherit X^d from father) + probability (mother to be heterozygote) × probability (child to inherit an X^d from the mother) × P (child to inherit Y from father)

Autosomal dominant:

X-Linked dominant:

It is sufficient for an individual to have the disease if he/she inherits at least one allele of the disease.

A normal parent has a probability zero to transmit the allele of the disease since he/she should be homozygotes to express the recessive normal allele n//n.

An affected parent can transmit the allele of the disease with a probability:
- ½ if he/she is heterozygote
- 1/1 if he/she is homozygote.

Case A: both parents are affected and each has a normal parent or child
in this case the parents are obligatorily heterozygotes, the probability of each of them to transmit the allele of the disease is ½.

The child will be affected if he/she inherits: the allele of the disease from the mother (½)and the normal allele from the father (½) or the normal allele from the mother (½) and the allele of the disease from the father (½) or the allele of the disease from the mother (½) and the father (½)
Risk = ½ × ½ + ½ × ½ + ½ × ½ = ½ + ½ + ½ = ¾ .

P (normal mother to transmit X^D) = 0 since he/she should be homozygotes X^n // X^n to express the recessive normal allele.
P (normal father to transmit the allele of the disease) = 0 … Genotype X^n//Y

An affected mother can transmit X^D with a probability:
- ½ if he/she is heterozygote
- 1/1 if he/she is homozygote.

P (affected father to transmit X^D) = ½ … Genotype X^D//Y

Case A: both parents are affected and each has a normal parent or child
In this case the mother is obligatorily heterozygotes, the probability that she transmits X^D is ½.
The father is X^D// Y, can transmit X^D or Y, with a probability ½ for each.

The child will be affected if he/she inherits: the X^D from the mother (½) and Y from the father (½) or the X^n from the mother (½) and X^D from the father (½) or X^D from the mother (½) and the father (½)
Risk = ½ × ½ + ½ × ½ + ½ × ½ = ½ + ½ + ½ = ¾ .

All girls will be affected in this case, Risk to have an affected Girl is zero

Risk to have an affected son is ½; The son will be affected if he/she inherits: the allele of the disease from the mother (½) and Y from the father (1)

Autosomal dominant:

X-Linked dominant:

Case B:
Both parents are normal ; that is, homozygote for the normal allele, the probability that each transmit the allele of the disease is zero
Risk = zero

Case C: one of the parents is affected with a normal parent or / child and the other parent is normal

In this case the affected parent is obligatorily heterozygote, the probability that he/she transmits the allele of the disease is ½.

The normal parent is homozygote n//n, has no effect in determining the genotype of the offspring since he/ she has only the recessive allele.

The child will be affected if he/she inherits the allele of the disease from the affected parent with a probability = (½) since the children will express the allele they inherit from the affected parent.
Risk = 1/2

Case C: The mother is affected with a normal parent or child / and father is normal
In this case the mother is obligatorily heterozygote, the probability that he/she transmits X^D is ½.

The father is X^n // Y, has no effect in determining the genotype of the offspring since he/ she has only the recessive allele.

The child will be affected if he/she inherits X^D from the mother with a probability = (½) since the children will express the allele they inherit from the affected parent.
Risk = ½

To have an affected son: Risk = ½ (inherits X^D mother (½) and Y from the father (1)

To have an affected girl, Risk = ½ (inherits X^D from the mother (½) and X^n from the father (1/1)