ESTIMATION OF CARRIER FREQUENCY IN POPULATION

A , a   A allele exist in p frequency.
a " " " q "

then

♀ \ ♂	A(p)	a(q)
A(p)	AA p^2	Aa pq
a(q)	Aa pq	aa q^2

$(p+q)^2 = AA : Aa : aa$
$p^2 : 2pq : (q^2)$ → affected individual

Autosomal recessive (AR) disease

Get q by taking square root of q^2

$q^2 = x$
$q = \sqrt{x}$

$\because (p+q)^2 = 1 \quad \therefore \quad p+q = \sqrt{1} = 1$

$p + \sqrt{x} = 1$
$p = 1 - \sqrt{x} = y$

Now we have value of both p & q and now for frequency of carrier we could calculate 2pq.

Q) This incidences 1/10,000 in population. What is carrier frequency

$q^2 = 1 \times 10^{-4}$
$q = 10^{-2} \; \left(\frac{1}{100}\right)$
$p = 1 - 10^{-2} = 1 - .01$
$= .99 \; \left(\frac{99}{100}\right)$

~~2pq = 2x~~

$2pq = 2 \times 0.99 \times .01$ or $\left(2 \times \frac{99}{100} \times \frac{1}{100}\right)$
$= .0198 = 1.98 \times 10^{-2}$ or $\left(\frac{198}{10,000}\right)$
$= \frac{198}{10,000} = \frac{1}{50.5} = \frac{1}{51}$

$198 \,|\, 10000 \,|\, 50.5$
990
1000

Q) $2pq = \frac{1}{50}$ — (1) $(p+q)^2 = 10,000$
$\therefore p+q = 100$ — (2)

from (1)
$2pq = \frac{1}{50}$
$q = \frac{1}{100p}$

putting value of q in eq- (2)

$p + \frac{1}{100p} = 100$
$\frac{100p+1}{100p} = 100$
$100p + 1 = 10,000p$
$9900p = 1$

or $p = \frac{1}{9900}$ Answer

Q) q value of males in the case of a x linked recessive (XLR) ... $q = \frac{1}{12}$ find the value of p.

H.W. Binomial probability

$\therefore p + q = 1$

$\therefore p + \frac{1}{12} = 1$

or $p = 1 - \frac{1}{12}$

$p = \frac{11}{12}$

Carrier frequency $= 2pq$

$= \cancel{2} \times \frac{11}{\cancel{12}_{6}} \times \frac{1}{12}$

$= \frac{11}{72}$ Answer

Q) Rr × Rr

1^{st} round $= 3/4$

2^{nd} round $= 3/4$

3^{rd} wrinkled $= 1/4$

4^{th} wrinkled $= 1/4$

5^{th} round $= 3/4$

in sequence

$\boxed{\text{Total frequency}}$ = Product of all these frequencies

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Q) Dd × Dd

~~Dd~~ × Dd × dd.

autosomal recessive

Autosomal recessive

parents are heterozygous

Probability of 1^{st} child to be normal $= 3/4$

2^{nd} " $= 3/4$

3^{rd} diseased/affected $= 1/4$

when seq. is not followed → $\boxed{(a+b)^3 = a^3 + b^3 + 3a^2b + 3ab^2}$

↓

Bn. Th.

$3a^2b$ $\quad 3 \times \frac{3}{4} \times \frac{3}{4} \times \frac{1}{4} = \frac{27}{64}$ Ans

(p) (p) (q)

4^{th} child to be affected $= 1/4$

when followed just multiply simply → Particular probability to be happen of an event in an sequential order then $\frac{3}{4} \times \frac{3}{4} \times \frac{1}{4}$ (for 3 children) $= \frac{9}{64}$

~~for~~ $\frac{3}{4} \times \frac{3}{4} \times \frac{1}{4} \times \frac{1}{4}$ (for 4 children) $= \frac{9}{256}$

$4 \times \frac{3}{4} \times \frac{3}{4} \times \frac{1}{4} = \frac{27}{64}$

$(a+b)^4$ → A couple, heterozygous for cystic fibrosis (AR) were to have 4 children, what would be the probability that they will have normal & affected children whether all 4 will be normal or all 4 will be affected or what are the other possibilities?

$a^4 + 4a^3b + 6a^2b^2 + 4ab^3 + b^4$

Dd × Dd

affected = dd (1/4)

normal = Dd & DD (3/4)

In view of Normal Mendelian Law out of 4 childrens, 3 should be normal and 1 should be affected (as per 3 : 1 ratio of Mendelian

But actual cond. & possibility may vary.

5- Different possibilities for occurance of normal & affected child (provided they have 4 children)

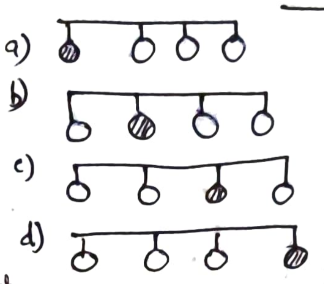
① All 4 will be normal and no one will be affected = 4 : 0 $\left(\frac{81}{256}\right)$
② 3 " " " " 1 will be affected = 3 : 1 $\left(\frac{27}{256}\right)$ (4...)
③ ~~3~~2 " " " " 2 " " " = 2 : 2 $\left(\frac{9}{256}\right)$
④ 1 " " " " 3 " " " = 1 : 3 $\left(\frac{3}{256}\right)$
⑤ 0 " " " and all 4 " " " = 0 : 4 $\left(\frac{1}{256}\right)$

In case, where sequence not followed, there are only 2 categories – one normal and other affected.

Out of all above combinations, what is the probability 3 will be normal and one will be affected.

$4 \times \frac{3}{4} \times \frac{3}{4} \times \frac{3}{4} \times \frac{1}{4} = \frac{27}{64}$

Possibility for ② nd condition.



4 different ways of occurance of the combination of 3 normal : 1 affected.

$\frac{3}{4} \times \frac{3}{4} \times \frac{3}{4} \times \frac{3}{4}$

$6 \times \frac{3}{4} \times \frac{3}{4} \times \frac{1}{4} \times \frac{1}{4}$

$4 \times \frac{3}{4} \times \frac{1}{4} \times \frac{1}{4} \times \frac{1}{4}$

$\frac{1}{4} \times \frac{1}{4} \times \frac{1}{4} \times \frac{1}{4}$

Summary

used for making all possibility of order

Normal	:	Affected	=	
4	:	0	=	$1 \times \left(\frac{3}{4}\right)^4$
3	:	1	=	$4 \times \left(\frac{3}{4}\right)^3 \times \left(\frac{1}{4}\right)^1$
2	:	2	=	$6 \times \left(\frac{3}{4}\right)^2 \times \left(\frac{1}{4}\right)^2$
1	:	3	=	$4 \times \left(\frac{3}{4}\right) \times \left(\frac{1}{4}\right)^3$
0	:	4	=	$1 \times \left(\frac{1}{4}\right)^4$

NOTE: If seq. is not given then all ways are considered, if seq or particular no. is specified then other possibilites are not considered

e.g. If a couple having one child, the probability of being affected = normal Mendian multiplication

" 2 children and if they are telling what is probability of either being affected = normal Mendian

if couple tells out of their children what is probability of 1st child of being affected (out of 2) → go to binomial.

2nd " " " " → " "

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