## EXTENDED RESEARCH SOLUTIONS <br> (FROM JOURNAL PAGES 18 AND 19)

A family tree is shown below. The genotypes of the grandparents regarding a recessive genetic mutation (a) that gives rise to a genetic disorder are given.


1. What percentage chance does the child have of inheriting the disorder? Of being an asymptomatic carrier? Of not carrying the mutation at all? Show your work below.

Grandparents 1:

|  | $\boldsymbol{A}$ | $\boldsymbol{a}$ |
| :---: | :---: | :---: |
| $\boldsymbol{A}$ | AA | Aa |
| $\boldsymbol{A}$ | AA | Aa |

## Grandparents 2:

|  | $\boldsymbol{A}$ | $\boldsymbol{a}$ |
| :---: | :---: | :---: |
| $\boldsymbol{A}$ | AA | Aa |
| $\boldsymbol{a}$ | Aa | aa |

The father has a $\mathbf{5 0 \%}$ chance of being $\boldsymbol{A} \boldsymbol{A}$, and $\mathbf{5 0 \%}$ chance of being Aa. The mother has a $\mathbf{2 5 \%}$ chance of being AA, 50\% chance of being Aa, and $25 \%$ chance of being aa. This creates six possible crosses resulting in the child. Carrying the probabilities through each gives these results:

Possibility 1 (50\% x 25\% = 12.5\%):

|  | $\boldsymbol{A}$ | $\boldsymbol{A}$ |
| :---: | :---: | :---: |
| $\boldsymbol{A}$ | AA | AA |
| $\boldsymbol{A}$ | AA | AA |

$100 \% \times 12.5 \%=12.5 \% \boldsymbol{A} \boldsymbol{A}$

Possibility 3 (50\% x 25\% = 12.5\%):

|  | $\boldsymbol{A}$ | $\boldsymbol{A}$ |
| :---: | :---: | :---: |
| $\boldsymbol{a}$ | Aa | Aa |
| $\boldsymbol{a}$ | Aa | Aa |

$100 \% \times 12.5 \%=12.5 \% ~ A a$

Possibility 2 (50\% x 50\% = 25\%):

|  | $\boldsymbol{A}$ | $\boldsymbol{A}$ |
| :---: | :---: | :---: |
| $\boldsymbol{A}$ | AA | AA |
| $\boldsymbol{a}$ | Aa | Aa |

$50 \% \times 25 \%=12.5 \%$ AA
$50 \% \times 25 \%=12.5 \%$ Aa
Possibility 4 (50\% x 25\% = 12.5\%):

|  | $\boldsymbol{A}$ | $\boldsymbol{a}$ |
| :---: | :---: | :---: |
| $\boldsymbol{A}$ | AA | Aa |
| $\boldsymbol{A}$ | AA | Aa |

$50 \% \times 12.5 \%=\mathbf{6 . 2 5 \%} \boldsymbol{A} \boldsymbol{A}$
$50 \% \times 12.5 \%=6.25 \% ~ A a$

Possibility 4 (50\% x 50\% = 25\%):

|  | $\boldsymbol{A}$ | $\boldsymbol{a}$ |
| :---: | :---: | :---: |
| $\boldsymbol{A}$ | AA | Aa |
| $\boldsymbol{a}$ | Aa | aa |

$25 \% \times 25 \%=6.25 \% \boldsymbol{A} \boldsymbol{A}$
$25 \% \times 50 \%=12.5 \% \boldsymbol{A a}$
$25 \% \times 25 \%=6.25 \%$ aa

Possibility 6 (50\% x 25\% = 12.5\%):

|  | $\boldsymbol{A}$ | $\boldsymbol{a}$ |
| :---: | :---: | :---: |
| $\boldsymbol{a}$ | Aa | aa |
| $\boldsymbol{a}$ | Aa | aa |

$12.5 \% \times 50 \%=6.25 \% ~ A a$
$12.5 \% \times 50 \%=6.25 \%$ aa

We now compile the probabilities for each result from each cross:

$$
\begin{aligned}
& 12.5 \%+12.5 \%+6.25 \%+6.25 \%=\mathbf{3 7 . 5 \%} \boldsymbol{A} \boldsymbol{A} \\
& 12.5 \%+12.5 \%+6.25 \%+12.5 \%+6.25 \%=\mathbf{5 0 \%} \boldsymbol{A} \boldsymbol{a} \\
& 6.25 \%+6.25 \%=\mathbf{1 2 . 5 \%} \boldsymbol{a} \boldsymbol{a}
\end{aligned}
$$

In other words, the child has a $12.5 \%$ chance of inheriting the disorder, a $50 \%$ chance of being an asymptomatic carrier, and a $37.5 \%$ chance of not carrying the mutation at all.

Please note that simply counting the three genotypes and dividing by the total number of genotypes is not a valid method, as it assumes that all six possible crosses are weighted equally, which is not the case. Doing so on this question will coincidentally yield the correct answer, but would lead to false results on the next question.

## 2. You discover that those with the disorder rarely live beyond the age of 5 years. How does this change the above percentages? Show your work below.

Any "aa" results in the parents can be ignored, as they would not live long enough to reproduce. Thus, the mother now has $\sim 33.3 \%$ (1 in 3) chance of being $A A$ and $\sim 66.7 \%$ (2 in 3) chance of being Aa. The father's percentages remain the same. This leads to four possible crosses:

Possibility 1 (50\% x $\sim 33.3 \%=\sim 16.7 \%)$ :

|  | $\boldsymbol{A}$ | $\boldsymbol{A}$ |
| :---: | :---: | :---: |
| $\boldsymbol{A}$ | AA | AA |
| $\boldsymbol{A}$ | AA | AA |

$100 \% \mathrm{x} \sim 16.7 \%=\sim 16.7 \% \boldsymbol{A} \boldsymbol{A}$

Possibility 2 (50\% x $\sim 66.7 \%=\sim 33.3 \%)$ :

|  | $\boldsymbol{A}$ | $\boldsymbol{A}$ |
| :---: | :---: | :---: |
| $\boldsymbol{A}$ | AA | AA |
| $\boldsymbol{a}$ | Aa | Aa |

$50 \% \mathrm{x} \sim 33.3 \%=\sim \mathbf{1 6 . 7 \%} \boldsymbol{A} \boldsymbol{A}$
$50 \% \mathrm{x} \sim 33.3 \%=\sim 16.7 \%$ Aa
Possibility 4 (50\% x $\sim \mathbf{6 6 . 7 \%}=\sim \mathbf{3 3 . 3 \%}$ ):

|  | $\boldsymbol{A}$ | $\boldsymbol{a}$ |
| :---: | :---: | :---: |
| $\boldsymbol{A}$ | AA | Aa |
| $\boldsymbol{a}$ | Aa | aa |

$25 \% \mathrm{x} \sim 33.3 \%=\sim 8.3 \% A A$
$50 \% \mathrm{x} \sim 33.3 \%=\sim 16.7 \%$ Aa
$25 \% \mathrm{x} \sim 33.3 \%=\sim 8.3 \%$ aa

Compiling the probabilities as before:
$\sim 16.7 \%+\sim 16.7 \%+\sim 8.3 \%+\sim 8.3 \%=\mathbf{5 0 \%} \boldsymbol{A A}$
$\sim 16.7 \%+\sim 8.3 \%+16.7 \%=\sim 41.7 \% ~ A a$
$\sim 8.3 \%$ aa
This means that the child now has about an $8.3 \%$ (1 in 12) chance of inheriting the disorder (and thus likely dying young), about a $41.7 \%$ ( 5 in 12) chance of being an asymptomatic carrier, and a 50\% chance of not carrying the mutation at all.

Notice at this point that counting each possible genotype and dividing by the total number of genotypes would yield incorrect results, as the four possible crosses are not weighted equally.

