What do I mean when I say “Bayes allows us to easily update our information?”

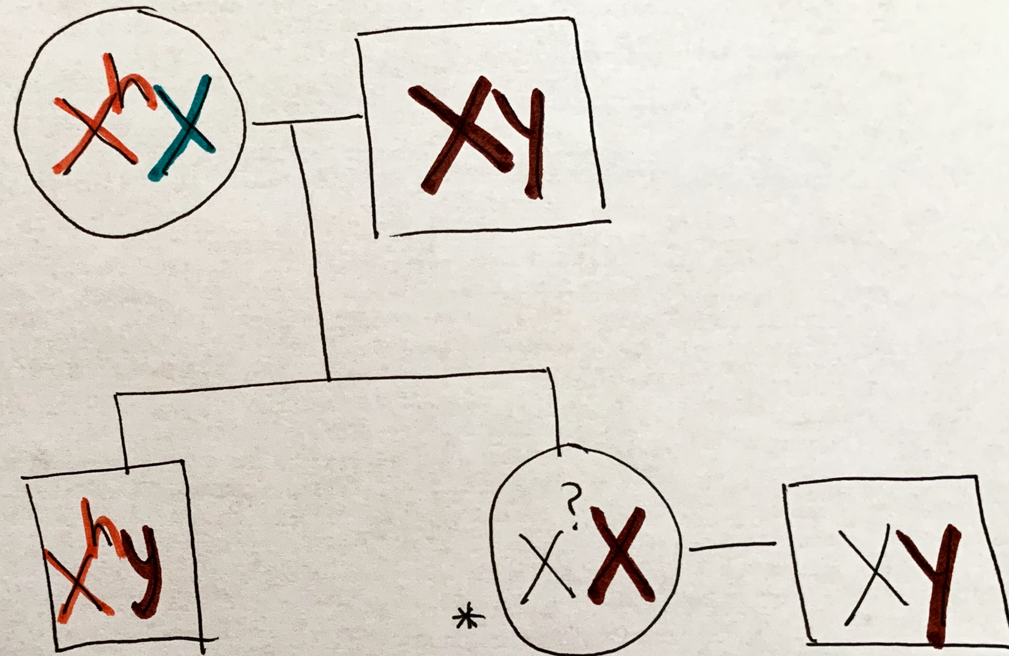
* Before offspring:

Probability of being carrier:

$$P(\theta=1) = \frac{1}{2}$$

Probability of not being carrier:

$$P(\theta=0) = \frac{1}{2}$$



- Woman is unaffected by Hemophilia but she has a brother who is affected by Hemophilia
- Hemophilia allele is located on the X chromosome
- Hemophilia is a recessive trait
- Their father is unaffected and their mother is phenotypically unaffected (but she must be a carrier)

Since the woman has a brother with the disease, she can be a carrier for the recessive allele or she may have inherited normal X from her mother (we know that she inherited the normal X from her father because he is unaffected and therefore must have a normal X)

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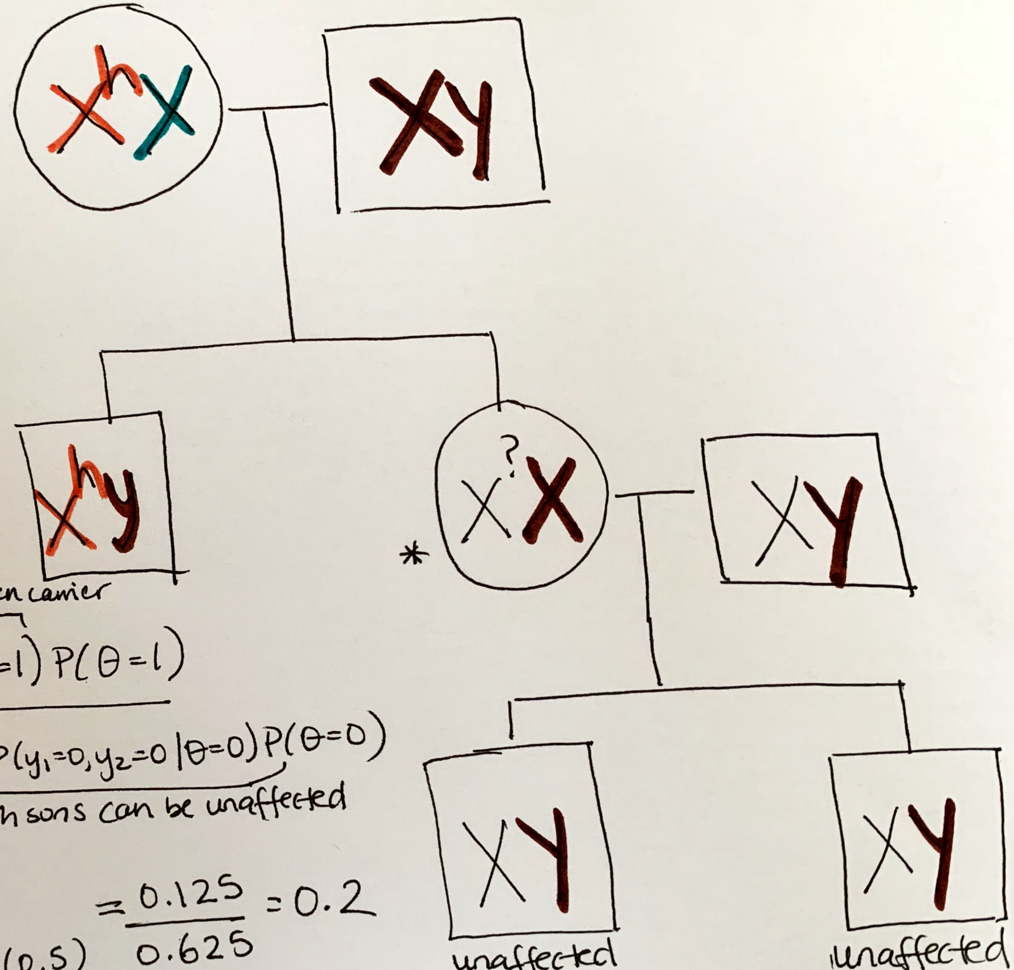

After having two unaffected sons:

(Bayes)

$$P(\theta=1 \mid y_1=0,y_2=0)=\frac{\overbrace{P(y_1=0,y_2=0 \mid \theta=1)}^{\text{Two unaffected when carrier}}P(\theta=1)}{\underbrace{P(y_1=0,y_2=0 \mid \theta=1)P(\theta=1)+P(y_1=0,y_2=0 \mid \theta=0)P(\theta=0)}_{\text{all ways both sons can be unaffected}}}$$

$$=\frac{(0.25)(0.5)}{(0.25)(0.5)+1\cdot(0.5)}=\frac{0.125}{0.625}=0.2$$

- WITH EXTRA INFORMATION (2 unaffected sons), the probability that mom is a carrier has gone from **0.5 → 0.2**
- NOW what happens if mom has one more unaffected son?

Based on the pedigree and the known inheritance mechanism
P(woman being carrier) =P(Θ=1)= 0.5
P(woman not being carrier) =P(Θ=0)= 0.5

P(woman being carrier | two sons are unaffected)
= P(two unaffected sons and carrier)/P(all ways unaffected sons)

$$P[\Theta=1 \mid y_1=0, y_2=0] = \frac{P(y_1=0, y_2=0 \mid \Theta=1) * P(\Theta=1)}{P(y_1=0, y_2=0 \mid \Theta=1) * P(\Theta=1) + P(y_1=0, y_2=0 \mid \Theta=0) * P(\Theta=0)}$$

$$P[\Theta=1 \mid y_1=0, y_2=0] = \frac{0.25 * 0.5}{0.25 * 0.5 + 1 * 0.5} = \frac{0.125}{0.625} = 0.2$$

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$$P[\Theta = 1 \mid y_1 = 0, y_2 = 0] = \frac{0.25 * 0.5}{0.25 * 0.5 + 1 * 0.5} = \frac{0.125}{0.625} = 0.2$$

We have now updated our prior probability of the woman being a carrier from a starting prior of 0.5 to 0.2!

If she went on to have a **third unaffected son**, this would provide additional evidence and would continue to change the woman's probability of being a carrier:

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If she went on to have a **third unaffected son**, this would provide additional evidence and would continue to change the woman's probability of being a carrier. Note: we would use our new updated prior probability of 0.2 (instead of the original prior of 0.5):

$$P[\Theta = 1 \mid y_1 = 0, y_2 = 0, y_3 = 0] = \frac{P(y_1 = 0, y_2 = 0, y_3 = 0 \mid \Theta = 1) * P(\Theta = 1)}{P(y_1 = 0, y_2 = 0, y_3 = 0 \mid \Theta = 1) * P(\Theta = 1) + P(y_1 = 0, y_2 = 0, y_3 = 0 \mid \Theta = 0) * P(\Theta = 0)}$$

$$P[\Theta = 1 \mid y_1 = 0, y_2 = 0, y_3 = 0] = \frac{0.5 * 0.2}{0.5 * 0.2 + 1 * 0.8} = 0.111$$