**Question 1**

James and Rene come to a genetic counselor because they are deciding whether to have another child or adopt. They want to know the probability that their un-born child will have cystic fibrosis.

Consider the Bayesian network for cystic fibrosis. We consider a person's phenotype variable to be "observed" if the person's phenotype is known. Order the probabilities of their un-born child having cystic fibrosis in the following situations from smallest to largest: (1) No phenotypes are observed (nothing clicked), (2) Jason has cystic fibrosis, (3) Sandra has cystic fibrosis.

(1), (3), (2) Y

(1), (2), (3)

(3), (2), (1)

(2), (1), (3)

(3), (1), (2)

**Question 2**

James never knew his father Ira because Ira passed away in an accident when James was a few months old. Now James comes to the genetic counselor wanting to know if Ira had cystic fibrosis. The genetic counselor wants your help in determining the probability that Ira had cystic fibrosis. Consider the Bayesian network for cystic fibrosis. We consider a person's phenotype variable to be "observed" if the person's phenotype is known. Order the probabilities of Ira having had cystic fibrosis in the following situations from smallest to largest: (1) No phenotypes are observed (nothing clicked), (2) Benjamin has cystic fibrosis, (3) Benjamin and Robin have cystic fibrosis.

(3), (2), (1)

(3), (1), (2)

(2), (1), (3)

(1), (3), (2) Y

(1), (2), (3)

**Question 3**

Recall that, for a trait with 2 alleles, the CPD for genotype given parents' genotypes has 27 entries, and 18 parameters were needed to specify the distribution. How many parameters would be needed if the trait had 3 alleles instead of 2?

Answer for Question 3

5\*6\*6 = 180

**Question 4**

You will now gain some intuition for why decoupling a Bayesian network can be worthwhile. Consider a **non-decoupled** Bayesian network for cystic fibrosis with **3 alleles** over the pedigree that was used in section 2.4 and 3.3. How many parameters are needed to specify all probability distributions across the entire network?

Answer for Question 4

**Question 5**

Now consider the **decoupled** Bayesian network for cystic fibrosis with **3 alleles** over the pedigree that was used in section 2.4 and 3.3. How many parameters are needed to specify all of the probability distributions across the entire network?   
  
**Hint**: A child cannot inherit an allele that is not present in either parent, so there aren't as many degrees of freedom here as there might be without that context-specific information.

Answer for Question 5

**Question 6**

Consider the **decoupled** Bayesian network for cystic fibrosis with three alleles that you constructed in section 3.3. We consider a person's gene copy variable to be "observed" if the person's allele for that copy of the gene is known.

James and Rene are debating whether to have another child or adopt a child. They are concerned that, if they have a child, the child will have cystic fibrosis because both of them have one F allele observed (their other gene copy is not observed), even though neither of them have cystic fibrosis. You want to give them advice, but they refuse to tell you whether anyone else in their family has cystic fibrosis. What is the **probability** (NOT a percentage) that their unborn child will have cystic fibrosis?

Answer for Question 6

0.4672

**Question 7**

Consider a Bayesian network for spinal muscular atrophy (SMA), in which there are multiple genes and 2 phenotypes.

Let *n* be the number of genes involved in SMA and *m* be the maximum number of alleles per gene. How many parameters are necessary if we use a table CPD for the probabilities for phenotype given copies of the genes from both parents?

*O*(*m*2*n*)

*O*(*n*)

Depends on the phenotype

*O*(*mn*) Y

**Question 8**

Consider the Bayesian network for spinal muscular atrophy (SMA), in which there are multiple genes and two phenotypes.

Let *n* be the number of genes involved in SMA and *m* be the maximum number of alleles per gene. How many parameters are necessary if we use a sigmoid CPD for the probabilities for phenotype given copies of the genes from both parents?

*O*((*mn*)2)

*O*(*n*) Y

*O*(*mn*)

Depends on the phenotype

**Question 9**

Consider genes A and B that might be involved in spinal muscular atrophy. Assume that A has 2 alleles *A*1 and *A*2, and B has 2 alleles, *B*1 and *B*2. Which of the following relationships between A and B can a sigmoid CPD capture?

Allele *A*1 and allele *B*1 make a person more likely to be have SMA when both of these alleles are present, but neither affect SMA otherwise. Y

Allele *A*1 and allele *B*1 make a person equally more likely to have SMA, but when both are present the effect on SMA is the same as when only one is present.

Allele *A*1 makes a person more likely to have SMA, while allele *B*1 independently makes a person less likely to have SMA. Y

Neither gene A nor gene B contribute to SMA.

Alleles *A*1 and *B*1 each independently make a person likely to have SMA.

When the alleles are *A*1 and *B*2 or *A*2 and *B*1 the person has SMA; otherwise the person does not have SMA. Y

Gene A contributes to SMA, but gene B does not contribute to SMA and thus does not affect the effects of gene A on SMA. Y

**Question 10**

Consider the Bayesian network for spinal muscular atrophy that we provided in spinalMuscularAtrophyBayesNet.net. We consider a person's gene copy variable to be "observed" if the person's allele for that copy of that gene is known.

Now say that Ira and Robin come to the genetic counselor because they are debating whether to have a biological child or adopt and are concerned that their child might have spinal muscular atrophy. They have some genetic information, but because sequencing is still far too expensive to be affordable for everyone, their information is limited to only a few genes and to only 1 chromosome in each pair of chromosomes.

Order the probabilities of their un-born child having spinal muscular atrophy in the following situations from smallest to largest: (1) No genetic information or phenotypes are observed (nothing clicked), (2) Ira and Robin each have at least 1 M allele, (3) Ira and Robin each have at least 1 M allele and at least 1 B allele.

(2), (1), (3)

(1), (3), (2)

(3), (2), (1)

(1), (2), (3) Y

(3), (1), (2)

**Question 11**

Consider the Bayesian network for spinal muscular atrophy that we provided in spinalMuscularAtrophyBayesNet.net.

No longer interested in finding out whether his father had cystic fibroisis, James comes to the genetic counselor with another question: Did his father have spinal muscular atrophy? The genetic counselor now wants your help in figuring this out. This time, however, James has other information for you: both he and Robin have spinal muscular atrophy.

What is the **probability** (NOT a percentage) that Ira had spinal muscular atrophy?

0.3541