Name: _____ School: _____

Date: _____ District: _____

Patterns of Inheritance
10th Grade

The Goldstein family is of Ashkenazi Jewish descent and recently experienced the tragic death of their youngest child, Sarah, who was diagnosed with Tay Sachs disorder. Tay Sachs is a genetic disorder resulting from a mutation on chromosome 15. This mutation causes a fatty substance to build up in the nerve cells of the brain, resulting in a deterioration of brain function that ultimately leads to paralysis and death before the age of 5. A person with Tay Sachs disease lacks a protein (enzyme) called hexosaminidase A, which is necessary for breaking down certain fatty substances.

After Sarah's death, the family decides to consult a genetic counselor to better understand the implications for the rest of the family.

1. The genetic counselor recommended that the entire family be tested for Tay Sachs.

   What does the counselor want to learn given that the brothers and sister are over the age of 5 and are healthy?

© 2009 Delaware Department of Education

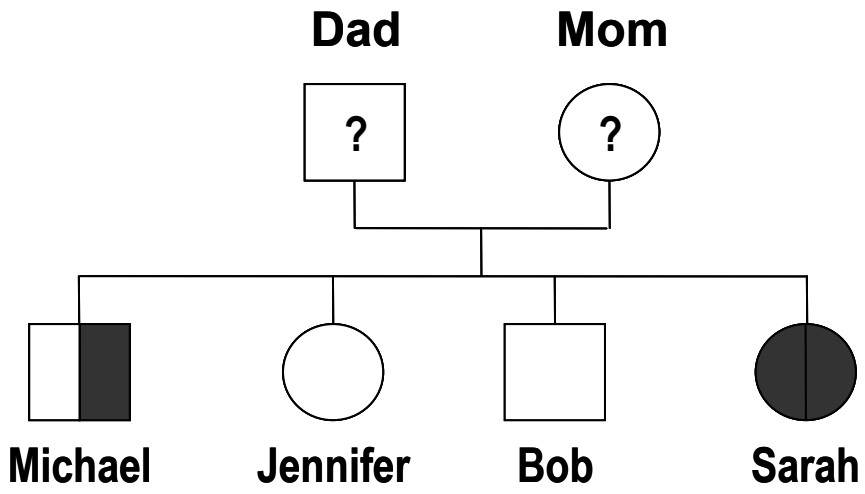
Genetic Testing Results

Figure 1 – DNA Sequences

DNA Sequence on Chromosome 15

Siblings	First Allele	Second Allele	Age
Michael	AGACTATCA	AGACTACCA	18
Jennifer	AGACTACCA	AGACTACCA	12
Bob	AGACTACCA	AGACTACCA	7
Sarah	AGACTATCA	AGACTATCA	Died – age 3

Figure 2 – Goldstein Family Pedigree



© 2009 Delaware Department of Education

2. The genetic screening for Tay Sachs provided the evidence in Figures 1 and 2.
   a. Circle the specific mutations in the DNA sequence (Figure 1) that is responsible for Sarah's disorder.
   b. Identify the autosomal pattern of inheritance in which Tay Sachs is passed from one generation to the next using the evidence in Figures 1 and 2.

3. Identify both Mom and Dad's genotypes based on the information provided in Figures 1 and 2 and in the family history.

   Mom _____ Dad _____

4. Investigate how Sarah's DNA sequence resulted in the absence of the protein necessary to break down fatty substances in the brain by using Figure 1.
   a. In the process of transcription, DNA is copied into mRNA.

      Identify the messenger RNA strand that is transcribed from Sarah's DNA sequence.

      Identify the messenger RNA strand that is transcribed from Jennifer's DNA sequence.

© 2009 Delaware Department of Education

Figure 3 – Codon Amino Acid Chart

Source: www.en.wikipedia.org

		2nd base			
		U	**C**	**A**	**G**
1st base	**U**	UUU (Phe/F)Phenylalanine UUC (Phe/F)Phenylalanine UUA (Leu/L)Leucine UUG (Leu/L)Leucine	UCU (Ser/S)Serine UCC (Ser/S)Serine UCA (Ser/S)Serine UCG (Ser/S)Serine	UAU (Tyr/Y)Tyrosine UAC (Tyr/Y)Tyrosine UAA Ochre (*Stop*) UAG Amber (*Stop*)	UGU (Cys/C)Cysteine UGC (Cys/C)Cysteine UGA Opal (*Stop*) UGG (Trp/W)Tryptophan
	C	CUU (Leu/L)Leucine CUC (Leu/L)Leucine CUA (Leu/L)Leucine CUG (Leu/L)Leucine	CCU (Pro/P)Proline CCC (Pro/P)Proline CCA (Pro/P)Proline CCG (Pro/P)Proline	CAU (His/H)Histidine CAC (His/H)Histidine CAA (Gln/Q)Glutamine CAG (Gln/Q)Glutamine	CGU (Arg/R)Arginine CGC (Arg/R)Arginine CGA (Arg/R)Arginine CGG (Arg/R)Arginine
	A	AUU (Ile/I)Isoleucine AUC (Ile/I)Isoleucine AUA (Ile/I)Isoleucine AUG (Met/M)Methionine, *Start*[1]	ACU (Thr/T)Threonine ACC (Thr/T)Threonine ACA (Thr/T)Threonine ACG (Thr/T)Threonine	AAU (Asn/N)Asparagine AAC (Asn/N)Asparagine AAA (Lys/K)Lysine AAG (Lys/K)Lysine	AGU (Ser/S)Serine AGC (Ser/S)Serine AGA (Arg/R)Arginine AGG (Arg/R)Arginine
	G	GUU (Val/V)Valine GUC (Val/V)Valine GUA (Val/V)Valine GUG (Val/V)Valine	GCU (Ala/A)Alanine GCC (Ala/A)Alanine GCA (Ala/A)Alanine GCG (Ala/A)Alanine	GAU (Asp/D)Aspartic acid GAC (Asp/D)Aspartic acid GAA (Glu/E)Glutamic acid GAG (Glu/E)Glutamic acid	GGU (Gly/G)Glycine GGC (Gly/G)Glycine GGA (Gly/G)Glycine GGG (Gly/G)Glycine

b. Referring to Figure 3, explain how Sarah's DNA sequence results in her inability to break down fatty substances in the brain (Tay Sachs disorder).

© 2009 Delaware Department of Education

5. Years later, Michael (Sarah's oldest brother) and his new bride, Rebecca, began to plan for a family. Rebecca was tested for the presence of the Tay Sachs mutation because of Michael's family history.

   a. Predict the probability of each genotype and phenotype in the offspring if Rebecca IS carrying a mutation for Tay Sachs.

Genotype	Probability	Phenotype	Probability

   b. Predict the probability of each genotype and phenotype in the offspring if Rebecca IS NOT carrying a mutation for Tay Sachs.

Genotype	Probability	Phenotype	Probability

© 2009 Delaware Department of Education

6. Using the information in the introduction, explain why Tay-Sachs would affect more males than females if it was a sex-linked disorder.

© 2009 Delaware Department of Education