**Tay-Sacs**

Genetic Disorder writing prompt

In the simplest situation, a recessive allele is a genetic variant that has no effect on the phenotype of an individual if a normal (dominant) allele is present. However, when both alleles at a given locus are recessive, a variant phenotype will occur, producing a recessive genetic disease if the function of that genetic locus is required for normal health. Recessive genetic diseases often involve genes that encode (synthesis) enzymes, and the recessive alleles are often null alleles (no enzyme activity produced). For these reasons, a recessive allele may be present in a family for many generations before one of the heterozygote happens to have children with another heterozygote and an affected (recessive) homozygote offspring is born (McConkey 2004).

Mary and Joseph are a recently married couple that want to start a family. They have some concern about the potential of a fatal genetic disorder that may be in their respective families. Joseph’s father had a brother that died in infancy from a little known disorder called Tay-Sacs. Mary’s brother had developed symptoms in his twenties of an unsteady gait, some neurological deterioration and “cherry-red” spots in the eyes, which may also indicate the presence of Tay-Sacs. Mary’s Great Grandmother, on her mother’s side, also had an infant sister that was undiagnosed, but family letters indicate the child had convulsions and seemed to be paralyzed at 4 months of age. Mary and Joseph have gone to their doctor about their concern and had a genetic test done to determine if they “carry” the recessive allele for Tay-Sacs.

Mary and Joseph’s genetic report did strongly indicate that they may each be a Tay-Sacs *carrier.* Their Doctor has referred them to a renowned genetic counselor so that they can make an informed decision about starting their family. In Mary and Joseph’s case, mathematically (punnett) we can determine that there is a 25% chance of having an offspring with Tay-Sacs. However according to *Mendelian Genetics* and gametic potential it may not be that simple. Most couples believe that the 25% refers to the fact that only 1 in 4 of their potential offspring could inherit the disorder. Unfortunately it doesn’t work that way.

Note:

Couples who are carriers of a Tay-Sachs gene or those who may be at increased risk due to ethnic background or family history may want to consult a genetic counselor. These health professionals help families understand what is known about the causes of a birth defect and the chances of the birth defect occurring in a pregnancy. They also help guide families through the testing process. Genetic counselors can provide referrals to medical experts and appropriate support groups in the community. Genetic counseling is available at most large medical centers and teaching hospitals. To find a genetic counselor in their area, individuals can ask their health care provider or contact the [National Society of Genetic Counselors](http://www.nsgc.org/).



**Task:**

As the renowned Genetic Counselor, you are asked to counsel Mary and Joseph. You will need to outline and explain to them the reason that gametes could always carry the recessive gene. You will also have to outline how and why an amniocentesis test would be required in the first trimester.

* You will complete a report to Mary and Joseph so that they can make an informed decision on starting their family.
* The report will define what Tay-Sacs is and the symptoms and outcomes of the genetic disorder.
* You will need to explain and graphically indicate the Mendelian Laws and how gametes are produced through meiosis.
* The use of a Family Tree (pedigree chart) will be used to indicate the inheritance of the recessive alleles.
* Lastly you will need to outline the procedure in getting the genetic material through amniocentesis in order to determine if the fetus does or does not have the recessive alleles for Tay-Sacs.

**Procedure (outline):**

**You will need to use a full page to (rough draft) each section of this outline**

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| --- |
| **Research Section on Tay-Sacs;**                                                <http://www.marchofdimes.com/professionals/681_1227.asp>                                             <http://www.ninds.nih.gov/disorders/taysachs/taysachs.htm>                                                <http://www.ygyh.org/tay/whatisit.htm>                                                                  <http://www.ncbi.nlm.nih.gov/disease/Tay_Sachs.html>                                                    <http://video.google.com/videoplay?docid=2005163388543825547&q=Tay-Sachs+Disease&hl=en> |
| **Explanation of Mendelian Laws, Meiosis and Punnett graphics to explain how a couple could conceivably have all offspring with Tay-Sacs** |
| **Mary and Joseph’s Family Tree (Pedigree):**  http://www.nsgc.org/About/FamilyHistoryTool/DrawYourFamilyTree/tabid/227/Default.aspx |
| **How Amniocentesis works: (include images)**  [**http://www.babycenter.com/0\_amniocentesis\_327.bc**](http://www.babycenter.com/0_amniocentesis_327.bc)  [**http://www.webmd.com/baby/guide/amniocentesis**](http://www.webmd.com/baby/guide/amniocentesis)  **http://www.google.com/images?hl=en&sugexp=ldymls&xhr=t&q=amniocentesis&cp=4&rlz=1I7GGLL\_en&wrapid=tljp129641721966006&um=1&ie=UTF-8&source=univ&ei=w8FFTf\_WGpOssAO4ju2xCg&sa=X&oi=image\_result\_group&ct=title&resnum=4&sqi=2&ved=0CEMQsAQwAw&biw=1276&bih=627** |

**The completed paper will also need to include all rough draft notes and outlines in order to be considered complete**

**Completed finished paper will be on top**

**due date; \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_**

**Rough drafts are attached next---**

**Information on how to write a killer report;**

**(Abstract section not required)**

[**http://www.wikihow.com/Write-a-Report**](http://www.wikihow.com/Write-a-Report)

[**http://geog.arizona.edu/~comrie/geog230/report.htm**](http://geog.arizona.edu/~comrie/geog230/report.htm)

references:

McConkey, Edwin H. Ph.D . 2004. How the Genome Works. Jones and Bartlett Publishers