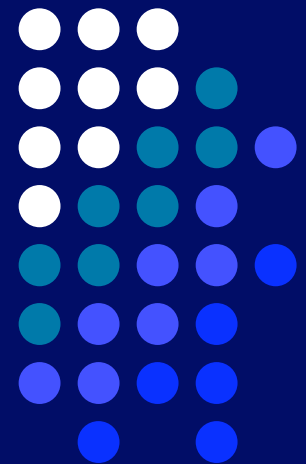


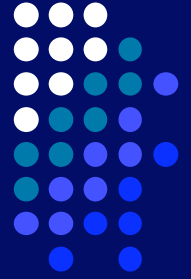
# Tay-Sachs disease

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Binta Wague & Naa-akomaah Yeboah  
Dr. Brennan – Professor McMahon  
Bronx Community College  
Bioinformatics  
2008

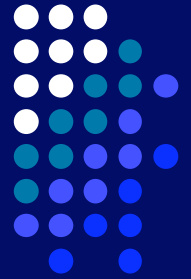


# Bioinformatics

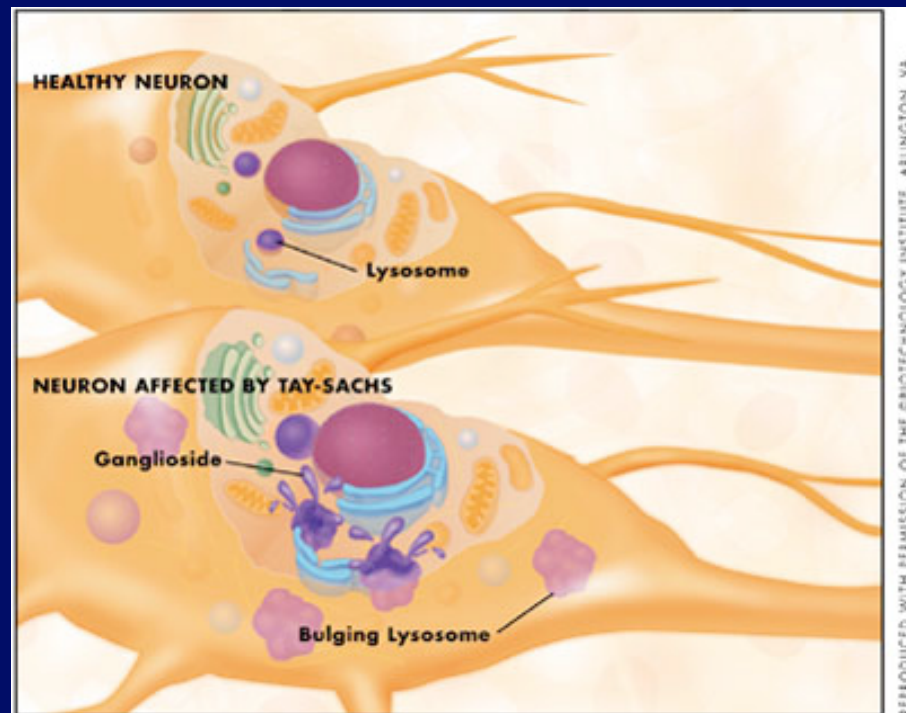


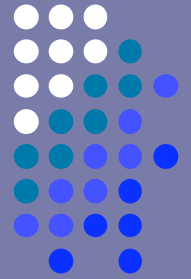
- **Bioinformatics** is the field of science in which biology, computer science, and information technology merge to form a single approach. In bioinformatics Computers are used to store biological information.
- Gene banks are for preserving genetic material and sequences. The bank is responsible for collection, preservation and evaluation of organisms.
- BLAST® stands for (Basic Local Alignment Search Tool). It is a search program that is used to compare sequence databases.

# What is Tay-Sachs disease ??



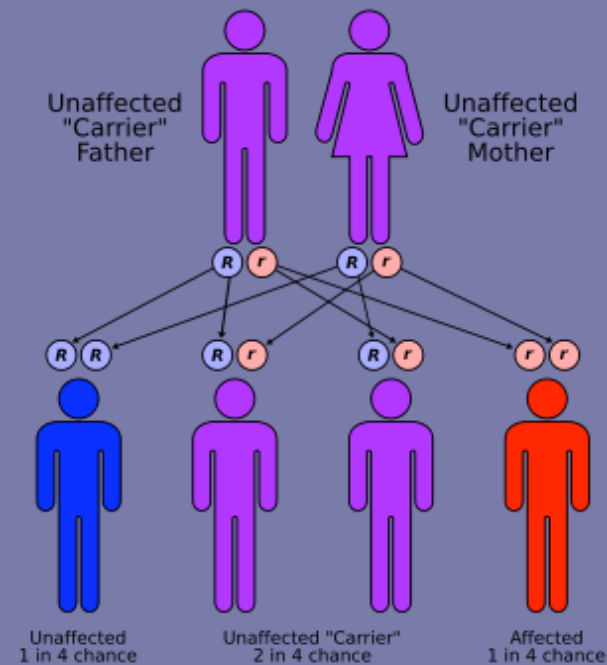
- Tay-Sachs disease is a rare, recessive disorder
- It causes destruction of the central nervous system (nerve cells in the brain and spinal cord).

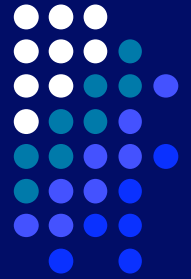




# Who gets Tay-Sachs?

- Since Tay-Sachs is a recessive disorder you need two alleles to get it.
- It is mostly associated with Ashkenazi Jews, and people in the middle east.

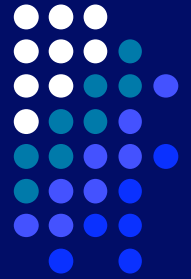




# What are the symptoms?

- The symptoms for Tay-Sachs vary on whether you get it in infancy or in adulthood.
- The much more rare form of Tay-Sachs appear in adults which is called adult-onset Tay-Sachs disease.

# What are the symptoms? Cont'd



- Symptoms in infants

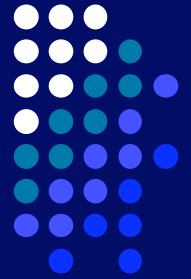
- slowing down of development
- weakening of muscles
- skills such as turning over, sitting or crawling are lost.

Other symptoms may occur as the disease progresses.

- seizures
- inability to swallow
- mental retardation

- Symptoms in adults

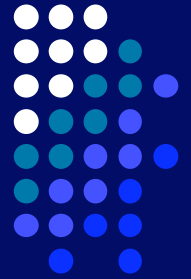
- muscle weakness
- ataxia (muscle coordination loss)
- problems with movement
- mental illness
- speech problems



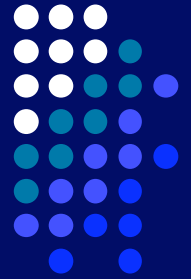
# The causes of Tay-Sachs

- The cause of Tay-Sachs disease is a genetic mutation that is passed on from parent to offspring.
- This defect is located on the HEXA gene. You can find this gene on the 15<sup>th</sup> chromosome.

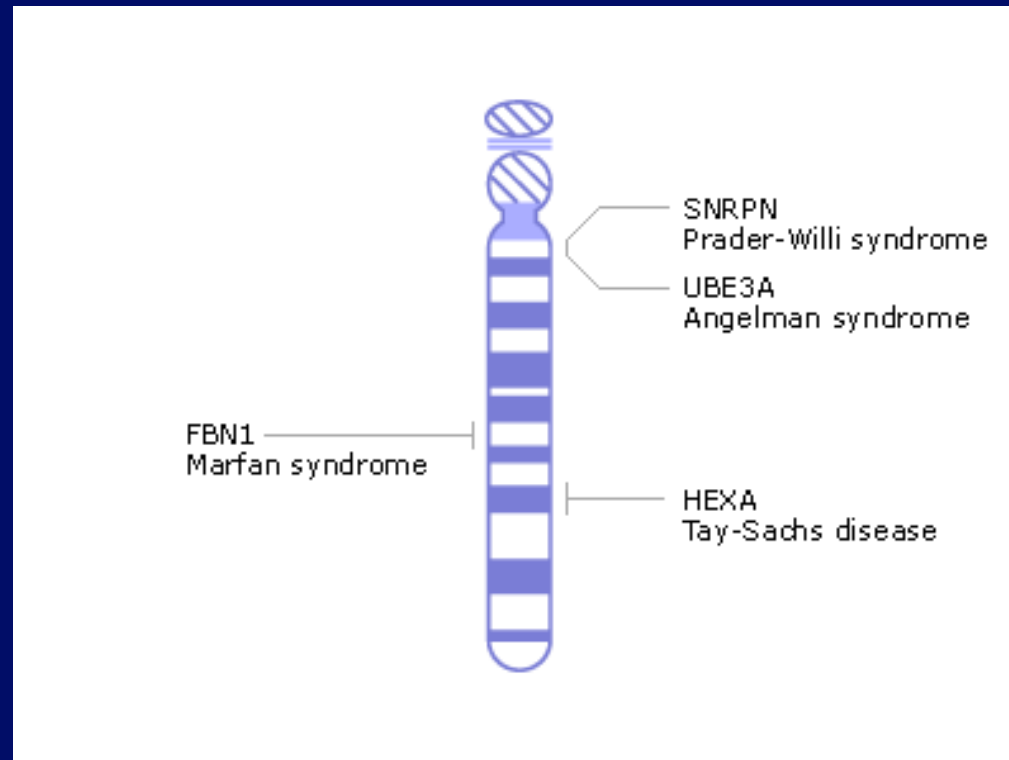
# The causes of Tay-Sachs Cont'd



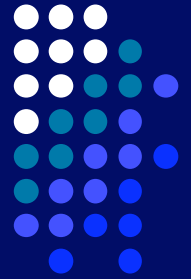
- The HEXA gene gives instructions for making part of an enzyme called beta-hexominidase A. This enzyme plays a critical role in the central nervous system.
- This enzyme breaks down a fatty substance known as GM2 ganglioside. Mutations in the HEXA gene disrupt the activity of the enzyme, preventing the breakdown of the GM2 ganglioside. As a result this can cause an accumulation to a toxic level in the brain and spinal cord.



# Where is it located?



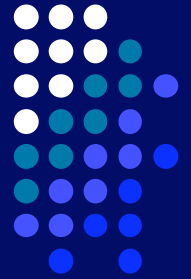
# Human Hexa Gene



Frame 3

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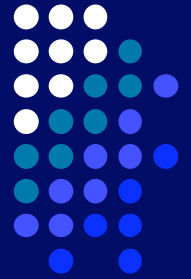
# Mouse Hexa Gene



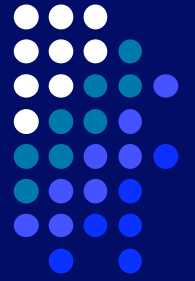
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# How is Tay-Sachs diagnosed?

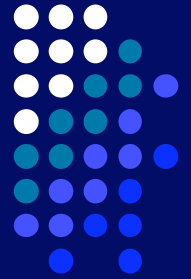


- In diagnosing Tay-Sachs the doctor will ask questions about symptoms and family history.
- Many tests will be recommended including a blood test.
- The blood test measures the activity of beta-hexosaminidase A to identify patients and carriers of the disease.
- It looks for changes or mutations in the gene that codes for HEXA.



# Treatment for Tay-Sachs

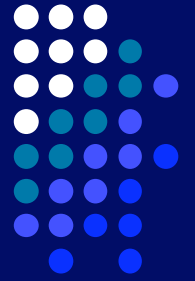
- There is no cure for Tay-Sachs nor anyway of slowing down the progression of the disease.
- The treatments control the symptoms.
- Treatments include:
  - medications
  - proper nutrition and hydration



# Treatment for Tay-Sachs cont'd

- anticonvulsant medicine can be given to control seizures.
- In helping individuals to eat, food should be cut into pieces or softened to prevent choking and ease swallowing.
- Individuals with Tay-Sachs are at special risk for dehydration.

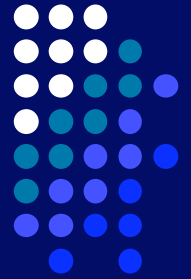




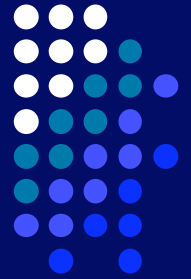
# Future

- In the future we would like to work with Tay-Sachs patients and see differences and similarities between gene sequences.
- We would also like to compare gene sequences of Tay Sachs patients with people without tay-sachs.

# 2009 Work



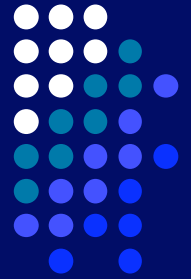
- Nutrition and healthy living group
- We will look at different neighborhoods and look at different factors that make up that neighborhood such as race, population, wealth, and other factors.
- We will also find relationships between obesity and other factors such as eating habits and availability of a healthy product.
- We will also search different farmers markets, corner stores, and groceries and conduct surveys which will ask questions like age ranges, medical history and food intake.



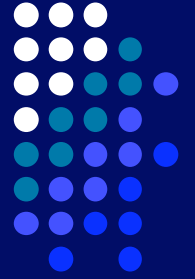
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- [http://www.mathworks.com/access/helpdesk/help/toolbox/bioinfo/index.html?/access/helpdesk/help/toolbox/bioinfo/ug/fp35834dup12.html&http://www.google.com/search?hl=en&q=tay+sachs+sequence+a&rlz=1R2ADSA\\_enUS333&aq=f&oq=&aqi=](http://www.mathworks.com/access/helpdesk/help/toolbox/bioinfo/index.html?/access/helpdesk/help/toolbox/bioinfo/ug/fp35834dup12.html&http://www.google.com/search?hl=en&q=tay+sachs+sequence+a&rlz=1R2ADSA_enUS333&aq=f&oq=&aqi=)

# Acknowledgements



- Dr. Thomas Brennan
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- Dr. Sat Bhattacharya
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- The audience



Any  
Questions?