AUTOSOMAL RECESSIVE DISEASES

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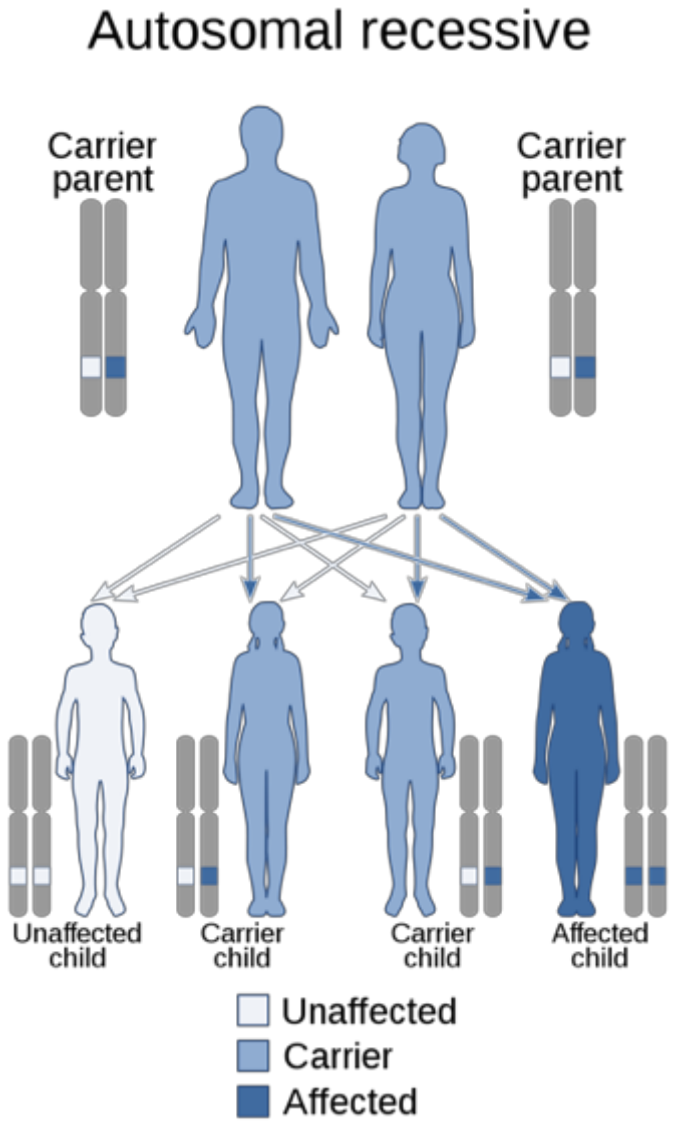
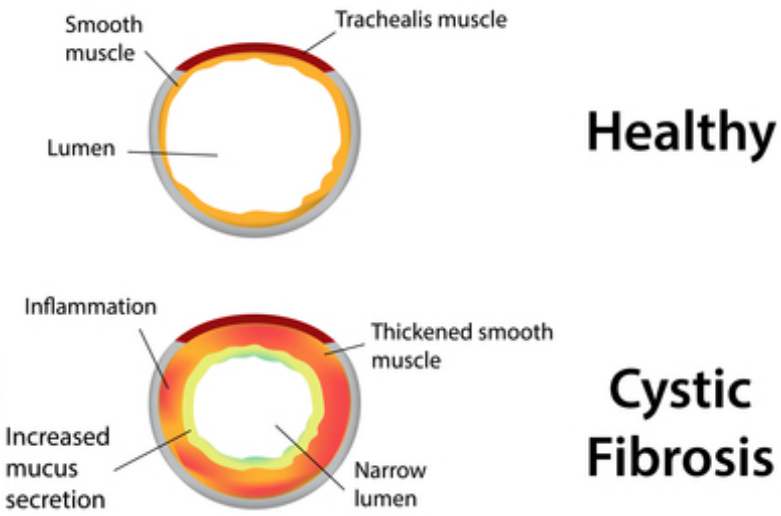
INTRODUCTION

- An autosomal recessive disorder means two copies of an abnormal gene must be present in order for the disease or trait to develop.
- Mutations (or changes in the DNA that codes for a gene) have occurred over time in different parts of the world. Anyone can carry virtually any type of recessive gene; however, there may be certain ethnic groups more likely to carry certain recessive genes, because of where the mutation originated.
- People with only one defective gene in the pair are called carriers. These people are most often not affected with the condition. However, they can pass the abnormal gene to their children.

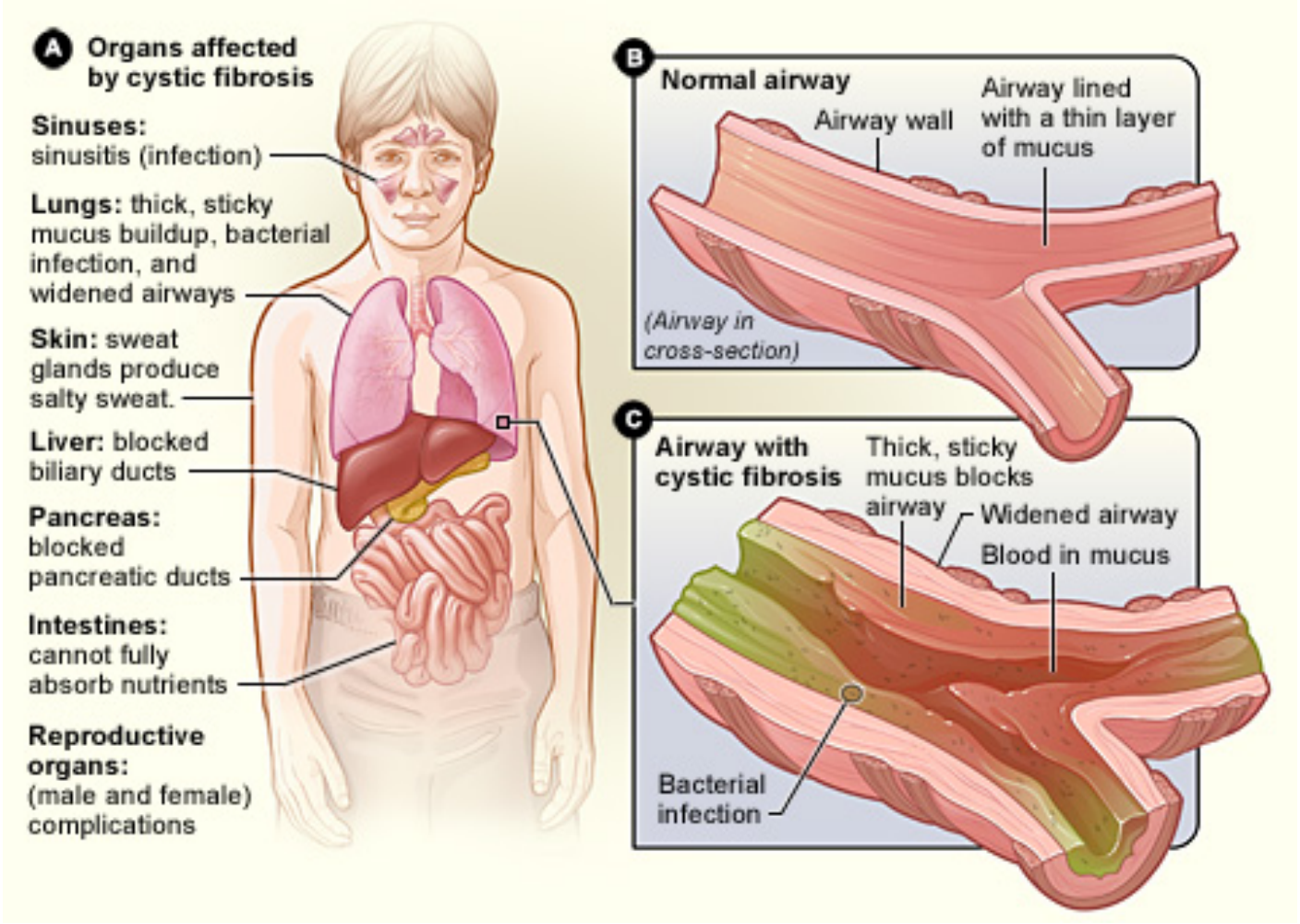


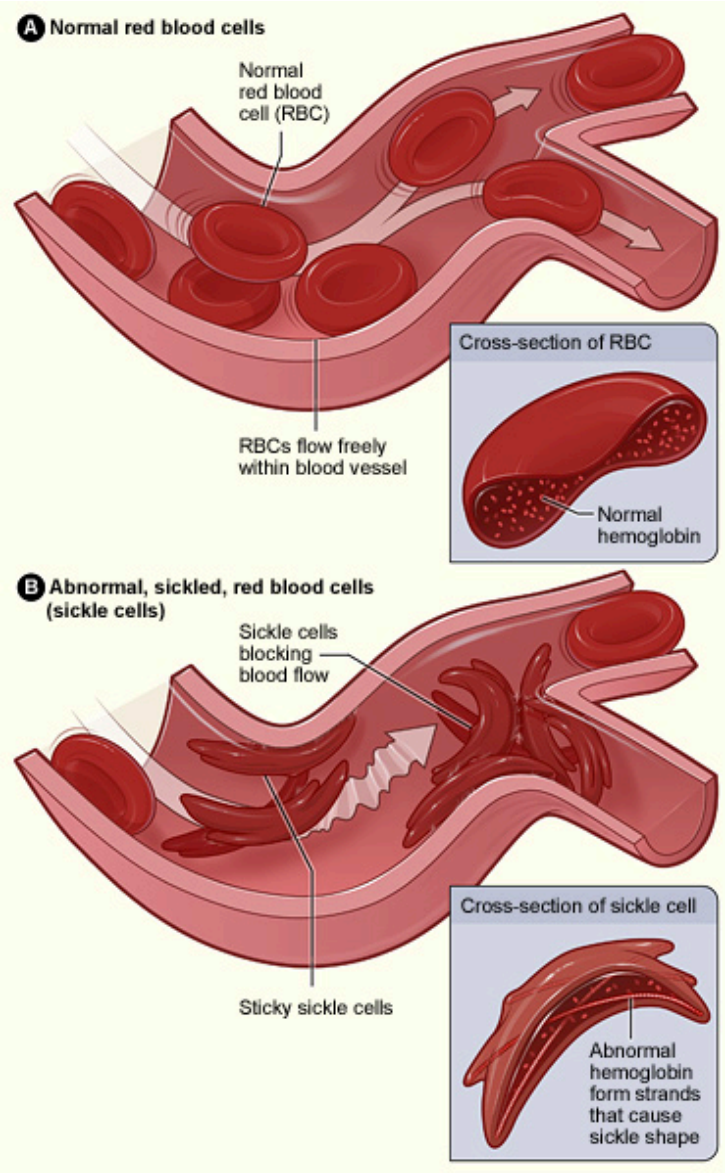
- If you are born to parents who carry the same autosomal recessive change (mutation), you have a 1 in 4 chance of inheriting the abnormal gene from both parents and developing the disease. You have a 50% (1 in 2) chance of inheriting one abnormal gene.

CYSTIC FIBROSIS



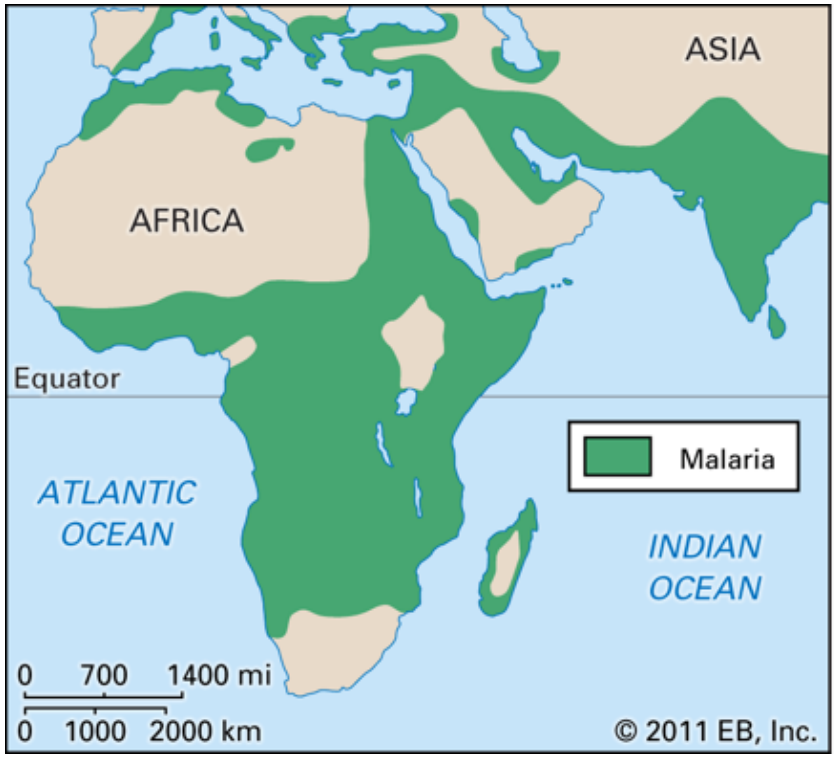
- Is one of the most common inherited single gene disorders in Caucasians.
- People with CF secrete abnormal body fluids, including unusual sweat and a thick mucus which prevents the body from properly cleansing the lungs. The mucus interrupts the function of vital organs and leads to chronic infections.
- Classic CF also involves the pancreas and causes decreased absorption of essential nutrients. Life expectancy has improved, but, ultimately, death most often occurs from respiratory failure. Other people with variants of CF may have only lung involvement, sinusitis, or infertility.


A Organs affected by cystic fibrosis
Sinuses: sinusitis (infection)
Lungs: thick, sticky mucus buildup, bacterial infection, and widened airways
Skin: sweat glands produce salty sweat.
Liver: blocked biliary ducts
Pancreas: blocked pancreatic ducts
Intestines: cannot fully absorb nutrients
Reproductive organs: (male and female) complications
B Normal airway
Airway wall
Airway lined with a thin layer of mucus
(Airway in cross-section)
C Airway with cystic fibrosis
Thick, sticky mucus blocks airway
Widened airway
Blood in mucus
Bacterial infection



SICKLE CELL DISEASE (SCD)

- SCD is an abnormality in the oxygen carrying protein, Haemoglobin in erythrocytes. SCD occurs when a person inherits two abnormal copies of the ß-Globin gene, found on chromosome 11, from each parent.
- People diagnosed with SCD have atleast one of their ß-Globin subunit in Haemoglobin A replaced to Haemoglobin S (Hgb S). In sickle cell anaemia (common form of SCD), both ß-Globin subunits are replaced by Hgb S.
- A person who only carries one copy of the abnormal gene has Sickle Cell Trait, they are not severely symptomatic.
- This change to Hgb S causes the erythrocytes to be rigid and sickle shaped. causing loss of elasticity of the cells, and therefore loss of ability to pass through small capillaries easily causing vessel occlusion and ischaemia. An attack can be induced by stress, temperature changes and high altitudes.
- SCD occurs more commonly in people who live in tropical regions or who have ancestors form this region, where malaria is or was common. People with sickle cell trait confer a heterozygote advantage, they show less severe symptoms when infected with malaria.


ASIA
AFRICA
Equator
Malaria
ATLANTIC OCEAN
INDIAN OCEAN
0 700 1400 mi
0 1000 2000 km
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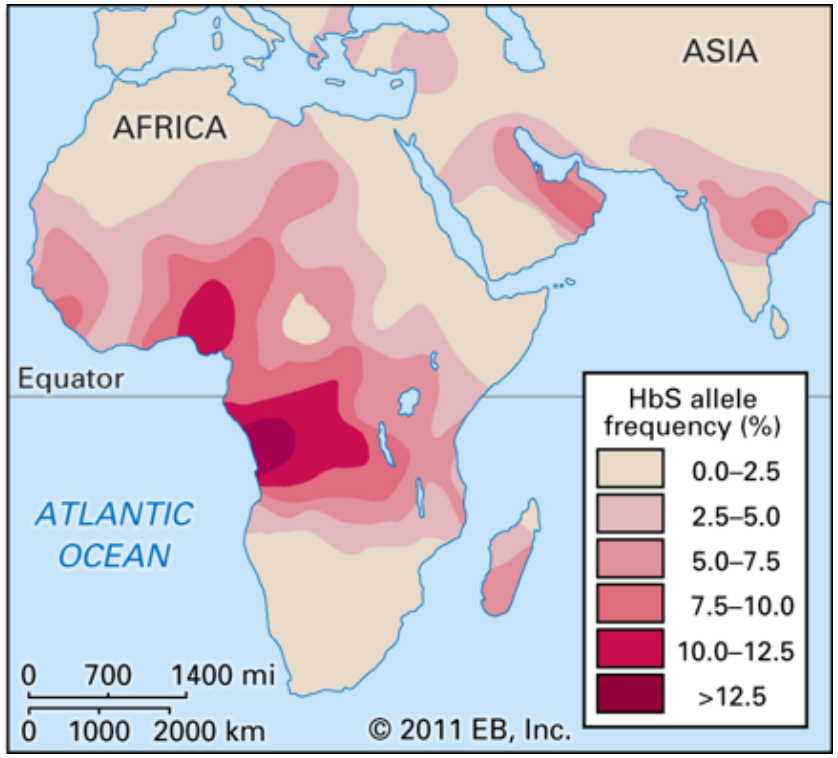
ASIA
AFRICA
Equator
HbS allele frequency (%)
0.0–2.5
2.5–5.0
5.0–7.5
7.5–10.0
10.0–12.5
>12.5
ATLANTIC OCEAN
0 700 1400 mi
0 1000 2000 km
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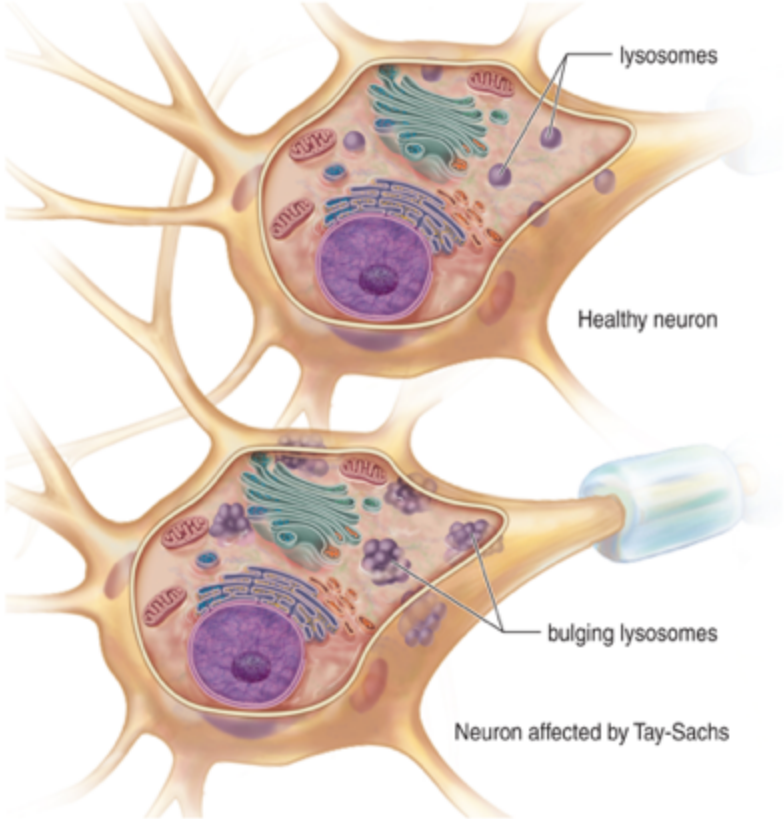
TAY-SACHS DISEASE



- Is a rare and fatal genetic disorder that causes progressive damage to the nervous system.
- The disease is characterized by destruction of nerve cells (neurons) in the brain and spinal cord.
- Caused by a genetic mutation in HEXA gene on chromosome 15, which codes for hexosaminidase A enzyme. The recessive autosomal mutation which leads to disruption in the enzyme and build up of the molecule GM2 ganglioside within cells in the CNS.

- There are 3 forms of the disease: Infantile, juvenile and late-onset.
- the most common form of this disease is infantile Tay-Sachs disease, which becomes noticeable at around 3-6 months. The symptoms present are the baby has a loss in ability to crawl, sit or turn over.
- However as the diseases progresses the symptoms get worse, to seizures, inability to move, hearing loss, blindness and inability to swallow.
- initial testing involves an enzyme assay to measure the activity of hexosaminidase.
- Patients positive for Tay-Sachs disease have a decrease in total hexosaminidase enzyme activity.

THANK YOU FOR LISTENING