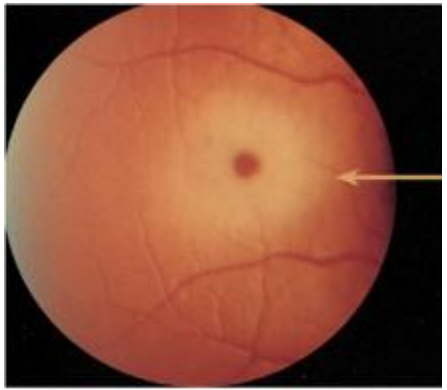
Diseases and Disorders

Tay-Sachs, Alzheimer's, Parkinson's, Down Syndrome, Leprosy, Tuberculosis, HIV/AIDS, and Cystic Fibrosis

Tay-Sachs

- Mutation in Hexa gene
  - Problem producing enzyme Hexosaminidase-A:
  - buildup of GM2 gangliosides, waste in brain cells
- Lysosome failure, leads to loss of motor skills, potential blindness or deafness
- Nervous system cannot sustain life
  - Life expectancy: 5 years
- Seen in Eastern European Jews
- "Cherry-red" spot in macula in center of retina
- Named for Tay and Sachs

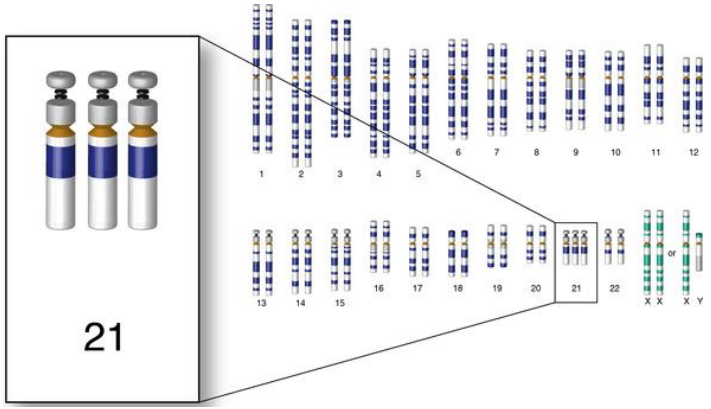


Tay-Sachs

Question: The 1278insTATC mutation is the most common cause of this disease, and is thought to have originated circa 800 AD in central Europe. That mutation causes the accumulation of sialylated glycosphingolipids in the brain, owing to a defect in hexosaminidase A, as contrasted with Sandhoff disease, in which hexosaminidase B is also affected. Thus, people with this disease are unable to properly hydrolyze gangliosides in their lysosomes. It presents with a characteristic red macula and usually leads to death by age 4. For 10 points, name this incurable genetic disease that is common in Ashkenazi Jews.

ANSWER: Tay-Sachs disease [or GM2 gangliosidosis; prompt on "gangliosidosis"]

Down Syndrome



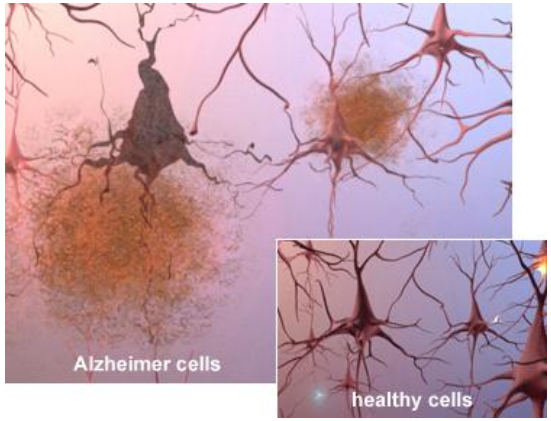
- A genetic chromosome 21 disorder causing developmental and intellectual delays.
  - Most diagnosed have *mild* to *moderate* cognitive or intellectual diability
- abnormal cell division results in extra genetic material from chromosome 21
  - Trisomy 21: nondisjunction of chromosome 21
- Symptoms include Brushfield spots (light dots on the iris), epicanthic folds of the eye, and are at greater risk to heart or thyroid disease (immune deficiency)
- Increased development of Alzheimer's, due to overexpression of APP and SOD1
- 20% of sufferers also suffer from transient leukemia

Down Syndrome

Question: People with this condition may display small light-colored dots on the iris, known as Brushfield spots. Heart defects and poor muscle tone are apparent in infants, and most adults with this disorder display symptoms of Alzheimer's disease by age 50. Rare mosaic and translocation varieties of it exist, though this disease is usually caused by nondisjunction in meiotic gamete production. Epicanthic folds and a single transverse crease on the palm are common, and it can be detected by karyotyping. This disease can cause hearing loss and is more likely with increased maternal age. For 10 points, name this chromosomal disorder often resulting in developmental delays, also known as trisomy 21.

ANSWER: Down's syndrome [accept trisomy 21 before it's mentioned]

Alzheimer's



- Loss of brain tissue that detroys mental funtions
- Most common form of senile dementia
- Old hypothesis : caused by a reduced synthesis of acetylcholin
    - So proposed treatment incresed acetylcholine production
- Largest genetic risk caused by E4 variant of Apoliopoproten E
- Buildup of beta-amyloid deposits and hyperphosphorilation of tau proteins
    - Pieces of protein that build up
- Plaques and tangles of brain result in cell death and tissue loss
    - Leads to atrophy of prefrontal cortex
- Accelerates process of forgetting memories

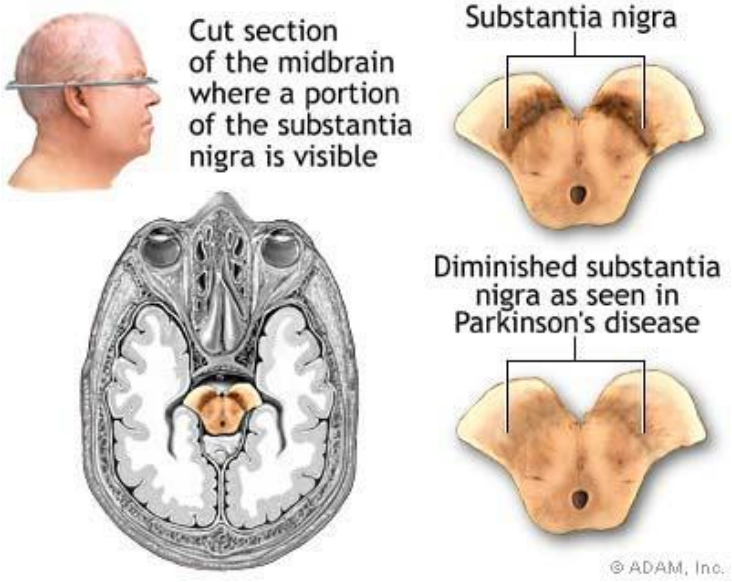
Alzheimer’s

Question: The E4 allele of the ApoE gene is the largest genetic risk factor for this disease. According to one theory it is caused by abnormalities in tau proteins. Another theory states it is caused by beta amyloid deposits which would relate this disease to (*) prion related disorders like Creutzfeld-Jakob disease. Another hypothesis states that it is caused by decreased production of acetylcholine. Patients of this disease exhibit plaques and tangles in their brains along with a loss of long term memory. For 10 points, name this most common form of dementia, named for a German psychiatrist.

ANSWER: Alzheimer's disease [prompt on AD; prompt on dementia]

Parkinson's

- A chronic and progressive disorder of the central nervous system that affects movement, often including tremors.
  - Nerve cell damage causes decrease in dopamine levels
    - Dopamine: neurotransmitter controls reward and pleasure centers of brain
  - Accumulation of protein alpha synuclein in Lewy bodies (abnormal collections of protein), causing damage to the substantia nigra
  - Linked to systematic defects in complex I of mitochondrial inner membrane
  - Characteristic symptom: bradykinesia (slowness of movement)
  - Commonly treated with L-dopa



Parkinson’s

Question: Abnormal alpha-synuclein proteins known as Lewy bodies were first discovered in patients suffering from this disease. This disease is caused by damage to the pars compacta of the substantia nigra, which is part of the basal ganglia. This disease results in less active dopamine pathways in the brain, which is why it is sometimes treated with L-dopa. For 10 points, name this neural disease that results in a loss of motor skill, often causing tremor and rigidity in muscles.

ANSWER: Parkinson's Disease

“Florida Warning: Armadillos Are Spreading Leprosy”

Leprosy

- Greek: “Makes skin scaly”
- Hansen’s disease
- Caused by Myobacterium genus
- Causes skin lesions: macules and papules
- Type 1 or 2 helper T cells dominate immune system’s response, determine symptoms
- Test looks for Phenolic glycolipid 1 antibodies
- Pathogen attacks Schwann Cells
    - Anciently treated with : chaumugra oil, venom from cobras, frogs, scorpions, bee stings, now with MDT and dapsone
    - Only animals affected are humans and armadillos

Leprosy

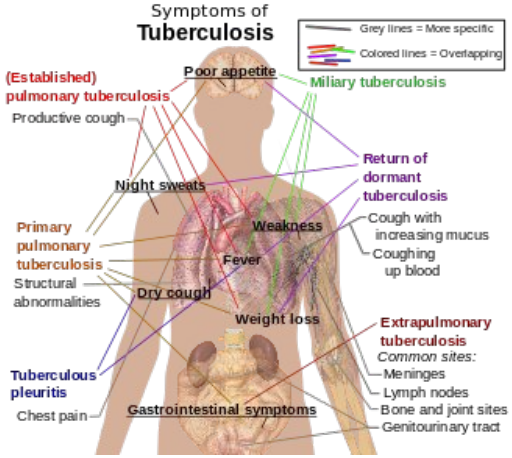
Question: The main treatment for this disease, often referred to as MDT, contains both clofazimine and rifampicin.The Ridley-Jopling system classifies this disease, which can also be classified into paucibacillary and multibacillary forms. This disease can be treated with Dapsone and is known for causing macules and papules, two different types of skin lesions. Caused by the Myobacterium genus, for 10 points, name this disease sometimes referred to as Hansen's disease whose severity and highly contagious nature have led sufferers of it to be quarantined to namesake colonies.

ANSWER: leprosy

Question: The pathogen responsible for this disease can bind the laminin-2-dystroglycan complex, allowing it to attack Schwann cells. One test for disease looks for phenolic glycolipid I antibodies. The symptoms of this disease are determined by whether type one or type two helper T cells dominate the response. This disease can infect only one (*) animal besides humans, armadillos. For 10 points, name this disease which includes skin lesions, whose sufferers used to be exiled into namesake colonies.

ANSWER: leprosy [or Hansen's disease]

Tuberculosis



- An infectious bacterial disease characterized by the growth of nodules (tubercles) in the tissues, especially the lungs.
- Also known as Consumption
- Caused by Mycobacterium tuberculosis
- Causes Ghon's complex: lesion seen on lungs, calcified focus of infection
- Blood test is QFT-G which checks for presence of interferon gamma
- Can spread through nervous system

Mimi, Violetta, and Edmund Tyrone all die from tuberculosis!

Tuberculosis

Question: This disease produces calcified scars called the Ghon complex at its initial site of infection. It causes Rasmussen's aneurysm, and it can spread to the neck in scrofula or to the spine in Pott's disease. Caused by a Gram-positive bacterium commonly treated with rifampin, it is targeted by the BCG vaccine. It causes necrotizing granulomas and is caused by a Mycobacterium infection. A test for this disease involves a PPD injected under the skin. Its symptoms include night sweats, bloody phlegm, and chronic cough. For 10 points, name this bacterial lung infection formerly known as consumption.

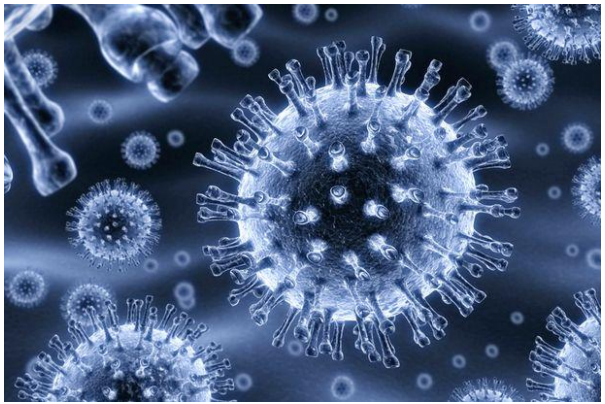
ANSWER: tuberculosis or MTB

HIV/AIDS

- A disease resulting in severe loss of the body's cellular immunity, greatly lowering the resistance to infection and malignancy.
- Caused by HIV: retrovirus (specifically, a lentivirus) that destroys CD4 plus T-cells
    - T-cells: type of lymphocyte, subtype of a white blood cell, central to immune system
- HIV infection usually aspymptomatic until progresssion to AIDS
- Swelling of lymph nodes, weight loss, fever, night sweats, fatigue, recurring infection
- Incresed susceptibility to Kaposi's Sarcoma
    - Lesion causing cancer in soft tissues

HIV

- Duplicates,
- attacks human t-cells,
- breaks down immune system



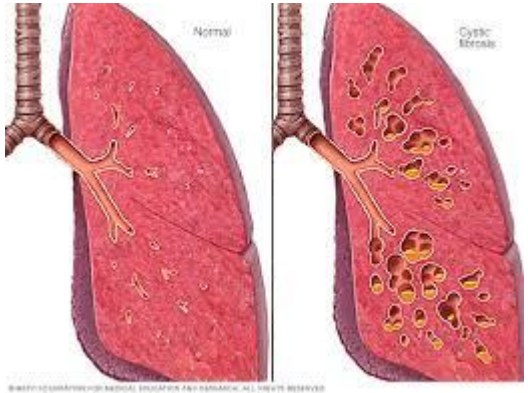
HIV/AIDS

Question: AZT was the first drug used to inhibit the replication of this lentivirus. Its macrophage-tropic "1" strain hurts the host indirectly by attacking CD4-plus "helper" T-cells, which usually protect against diseases like (*) Kaposi's sarcoma. First discovered in 1981, it can be transmitted by reusing needles or through unprotected sex. For 10 points, name this as-yet uncured retrovirus that causes AIDS.

ANSWER: HIV or H(uman) I(mmunodeficiency) V(irus) (prompt on "AIDS" or "acquired immune deficiency syndrome" before "lentivirus" is read)

Cystic Fibrosis

- An inherited life-threatening disorder that damages the lungs and digestive system.
- Affects cells that produce mucus, sweat, and digestive juices
- causes fluids to become thick and sticky, plug up tubes, ducts, and passageways.
- Symptoms can include cough, repeated lung infections, inability to gain weight, and fatty stools
- G551D mutation
- Males infertile
- Common in United Kingdom



Cystic Fibrosis

Question: A form of this disease particularly prevalent in the United Kingdom is caused by the G551D mutation and can be treated with Ivacaftor. Male sufferers of this disease are typically infertile because this disease results in the congenital absence of the vas deferens. In patients with this disease, a missing phenylalanine molecule caused by a deletion at the 508th residue results in a misfolded CFTR protein, which fails to conduct chloride ions across the cell membrane. This disease causes a blockage of ducts in the pancreas and patients suffering from this disease have salty sweat. For 10 points, name this disease in which an accumulation of thick mucus causes frequent lung infections.

ANSWER: cystic fibrosis [or CF; or mucoviscidosis

Mutation in hexa gene, no hexosaminidase enzyme, too much brain waste, ton of damage, early death

Cherry Red Spot

Eastern European Jews

Abnormal cell division: nondisjunction

(trisomy 21)

Brushfield spots, epicanthic eye folds

Risk of heart failure, possible development of Alzheimers

Less acetylcholine

Build up of amyloid- beta and tau proteins

Plaques and tangles

Parts of brain waste away

Memory loss

Less dopamine

Build up of alpha synuclein in Lewy bodies

Damage to substantia nigra

Bradykinesia

Pathogen attacks Schwann Cells

Causes macules and papules

Hansen’s disease

Colonies

Poor Armadillos

Mycobacterium tuberculosis

Ghon’s complex (calcified lung scars)

Consumption

Thick body fluids (digestic juices, sweat, mucus)

HIV virus attacks t-cells

immunodeficiency

More subject to infection, cancers (Karposi Sarcoma)