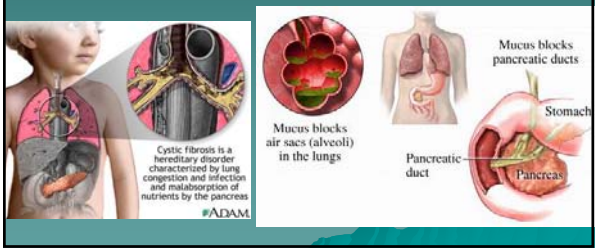
Human Genetic Disorders

Inherited Disorders

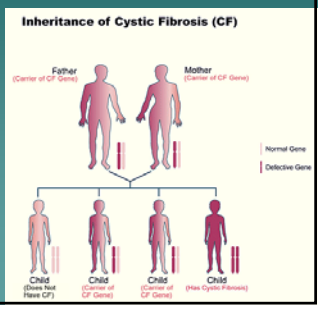
Cystic Fibrosis

1. Causes excess of mucus in lungs & digestive tract



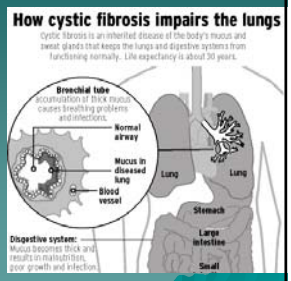
Cystic Fibrosis

2. How it is inherited:
   recessive, autosomal
   - Both Mom & Dad can be carriers



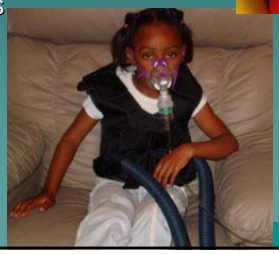
Cystic Fibrosis

3. Symptoms:
   - Breathing problems
   - Problems with digestion
   - Possible malnutrition
   - Salty tasting skin



Cystic Fibrosis

4. Treatment
   - Lung treatments
   - Antibiotics



Cystic Fibrosis

5. Prognosis = predicting the outcome of having a disease
   - With proper treatment, CF patients can enjoy a normal life-span

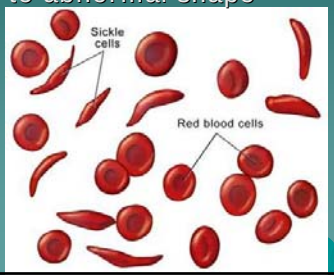
Cystic Fibrosis

6. Distribution / Frequency
   = how common it is in a population

- 1/2000 Caucasians
- Less common in other ethnic groups

Sickle-cell Anemia

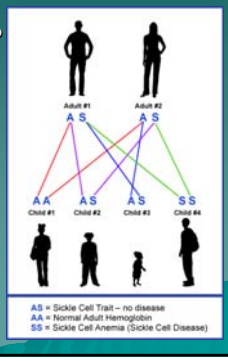
1. Red blood cells get stuck in vessels due to abnormal shape



Sickle-cell Anemia

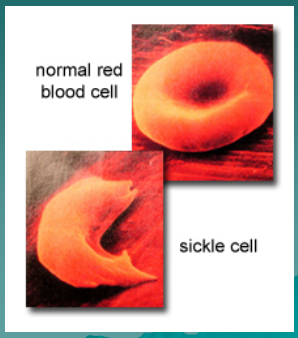
2. How is it inherited?
   - Codominant disorder

What does co-dominant mean? Both genes always show



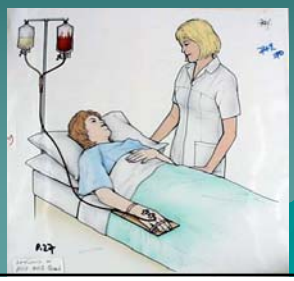
Sickle-cell Anemia

3. Symptoms:
- Pain
- Fever
- Severe chest pain
- Fatigue
- Weakness
- Tissue damage
- Possible brain damage
- Possibly fatal



Sickle-cell Anemia

4. Treatment:
- Transfusions
- Pain killers



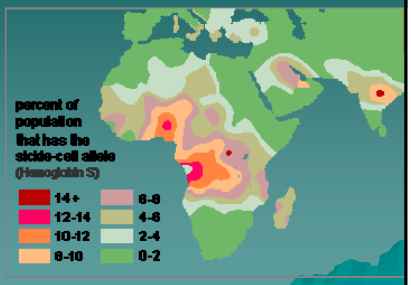
Sickle-cell Anemia

5. Prognosis
- Risk of blood-borne pathogens via transfusion
- High risk of disease
- Normal life possible with proper precautions & treatment

Sickle-cell Anemia

6. Distribution / Frequency

- 1/100,000 world population
- 1/400 African Americans

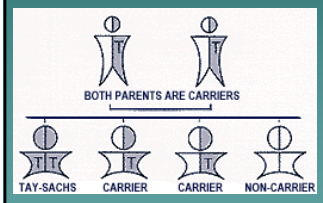


Tay Sachs' Disorder

1. General description:
   - baby normal at birth
   - an enzyme is lacking, causing progressive deterioration of central nervous system
   - fatal by age 5

Tay Sachs' Disorder

2. How it is inherited: autosomal recessive



Every person of Jewish, Irish, French-Canadian or Cajun heritage should be tested for Tay-Sachs

Tay Sachs' Disease

3. Symptoms:
   - @ about six months dvlpmt. slows down
   - gradual loss of motor skills & mental f(x)s.
   - blindness, deafness, mental retardation
   - usually fatal by age five.

Tay Sachs' Disease

4. Treatment:
   none

Tay Sachs' Disease

5. Prognosis:
   death before the age of five.

Tay Sachs' Disease

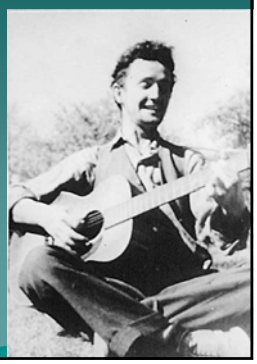
6. Distribution / Frequency

1/27 Jews in the U.S.
1/27 Cajuns
1/50 Irish Americans

RandomSanDiego.com

Huntington's Disease

1. General description:
   - Neurological disorder
   - Loss of mental faculties & physical control

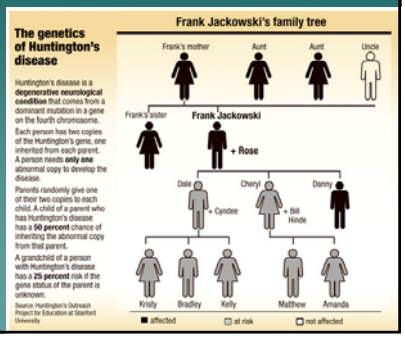


Woody Guthrie, folksinger

Huntington's Disease

2. How it is inherited:

Autosomal dominant



Huntington's Disease

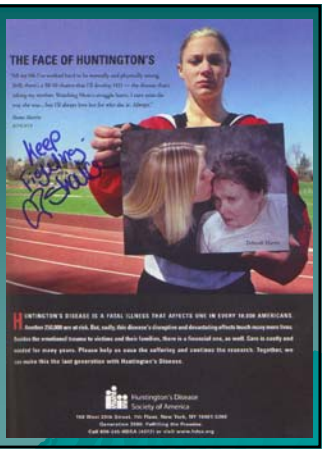
3. Symptoms: develop btwn. 35-40 yrs. old
   - Depression
   - Personality changes
   - Mood swings
   - Memory loss
   - Involuntary movements

Huntington's Disease

4. Treatment:
   - None
   - Meds. to manage symptoms

Huntington's Disease

5. Prognosis:
   - Deterioration of nervous system
   - Fatal within 15 years of onset
   - Negative effects on family



Huntington's Disease

6. Distribution / Frequency

1/10,000 Americans
30,000 Americans have it
250,000 Americans at risk

PKU

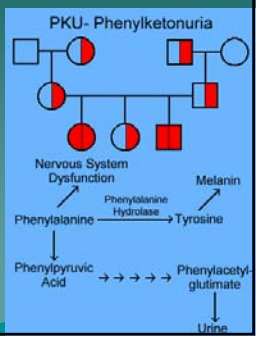
Phenylketonuria

1. General description:

- Body chemistry disorder
- Causes mental retardation if not treated
- Cannot break down the amino acid phenylalanine.

PKU

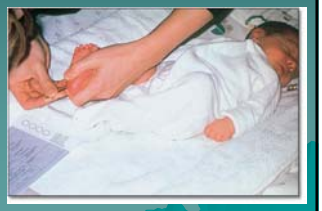
2. How it is inherited:
Autosomal recessive



PKU

3. Symptoms:

- Lose interest in surroundings
- Irritability
- Restlessness



PKU Test
(blood taken from baby's heel)

PKU

4. Treatment:
Special diet in first 3 weeks of life

PKU

5. Prognosis

- Early detection important
  (Testing required with in two weeks of birth in Colorado)
- With low phenylalanine diet, no mental retardation

PKU

6. Distribution / Frequency

- 1/15,000 births
- All ethnic groups

Bibliography

1. Image of cystic fibrosis http://www.nlm.nih.gov/medlineplus/ency/images/ency/fullsize/18135.jpg retrieved 12/3/07
2. Inheritance of CF http://www.nhlbi.nih.gov/health/dci/images/cf_graphic_1.gif retrieved 12/3/07
3. How CF impairs lungs http://www.ocregister.com/newsimages/news/2004/11/09CysticFibrosis.g.gi retrieved 12/3/07
4. How CF blocks pancreatic duct http://www.medicalook.com/diseases_images/cystic_fibrosis.jpg retrieved 12/3/07
5. Lung treatment http://goldbamboo.com/images/content/4083-250px-cftreatmentvest2-cystic-fibrosis.jpg retrieved 12/3/07
6. Image of antibiotics http://www.enviroblog.org/pills-red-and-yellow-antibiotics-closeup-2-AJHD.jpg retrieved 12/3/07
7. Normal vs. sickled RBCS http://www.dartmouth.edu/~toxmetal/images/sicklecell.jpg retrieved 12/3/07
8. Inheritance of sickle cell http://www.dukehealth.org/HealthLibrary/AdviceFromDoctors/YourChildsHealth/sicklecelldisease/chartHuman Genetic Disorders 07.ppt Human Genetic Disorders 07.ppt retrieved 12/3/07
9. Sickle vs normal RBC photo http://bsw-uiuc.net/tutorials_current/Sickle_Cell_Anemia/SC2001/intro_files/image002.jpg retrieved 12/3/07
10. Blood transfusion http://www.dkimages.com/discover/previews/940/22323.JPG retrieved 12/3/07
11. Sickle cell child with family http://www.stlouischildrens.org/SITE_IMAGES/sicklecell1.jpg retrieved retrieved 12/3/07
12. Girl with flowers **www.wepsicklecell.org/pages/about.html** retrieved 12/3/07
13. Sickle cell distribution http://anthro.palomar.edu/synthetic/images/map_of_sickle_cell_frequencies.gif retrieved 12/3/07
14. Tay Sachs' patient http://www.curetay-sachs.org/gpx/dakota_260b.jpg retrieved 12/4/07
15. Tay Sachs' inheritance http://www.jewishgeneticscenter.org/images/graph.gif retrieved 12/3/07
16. Testing for TSD **www.tay-sachs.org/taysachs.php** retrieved 12/3/07
17. TSD inheritance **www.tay-sachs.org/taysachs.php** retrieved 12/3/07

Bibliography

18. TSD symptoms http://pinker.wjh.harvard.edu/photos/cape_cod/images/Sunset%20on%20Pilgrim%20Beach.jpg retrieved 12/3/07
19. TSD prognosis **blogs.salon.com/0001970/categories/sunsets/** retrieved 12/3/07
20. Huntington's inheritance http://www.dailyherald.com/graphics/jackowfam.jpg retrieved 12/3/07
21. Huntington's prognosis http://www.shanamartin.com/images/HD%20Pics/HDSAad.jpg retrieved 12/3/07
22. Easter lily http://www.aspca.org/toxicplants/images/TN_IMG01455_300X300Y.jpg retrieved 12/3/07
23. PKU Inheritance http://cas.bellarmine.edu/tietjen/Ethology/Ethol06.gif retrieved 12/3/07
24. PKU diet http://www.myspecialdiet.com/Files%5CProds%5CImages/PKU3.jpg retrieved 12/3/07
25. Diet soda http://www.uthscsa.edu/mission/ArticleData/2006-01/images/diet_soda.jpg retrieved 12/3/07
26. Image of child http://www.growingkids.co.uk/images/809.jpg retrieved 12/3/07
27. PKU Test http://openlearn.open.ac.uk/file.php/2389/SK195_6_002i.jpg retrieved 12/3/07
28. Woody Guthrie http://www.historycooperative.org/journals/jah/87.3/images/exr_4_f1.jpg retrieved 12/3/07