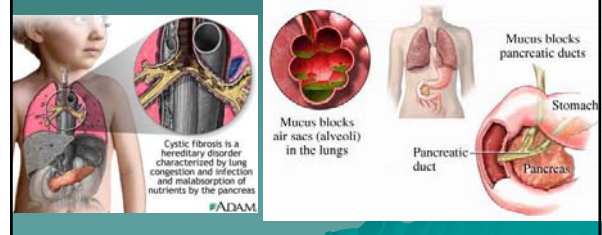


Human Genetic Disorders

Inherited Disorders

Cystic Fibrosis

1. Causes excess of mucus in lungs & digestive tract

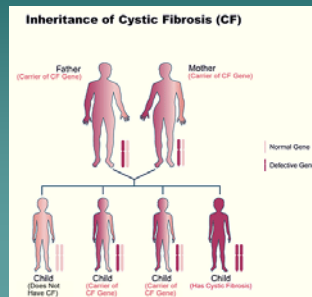


Cystic Fibrosis

2. How it is inherited:

recessive,
autosomal

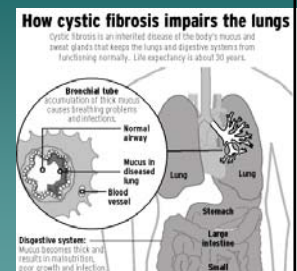
- ☛ Both Mom & Dad can be carriers



Cystic Fibrosis

3. Symptoms:

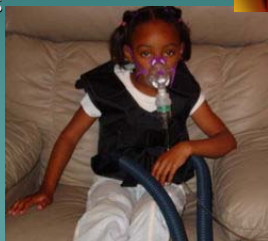
- ◆ Breathing problems
- ◆ Problems with digestion
- ◆ Possible malnutrition
- ◆ Salty tasting skin



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



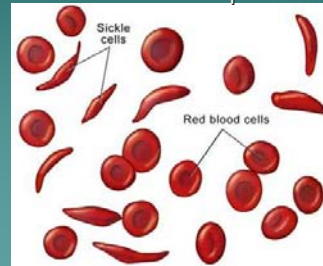
Cystic Fibrosis

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

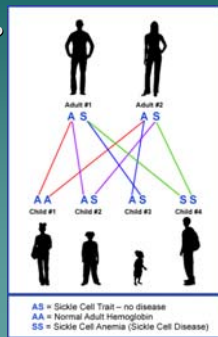


Sickle-cell Anemia

2. How is it inherited?

↳ Codominant disorder

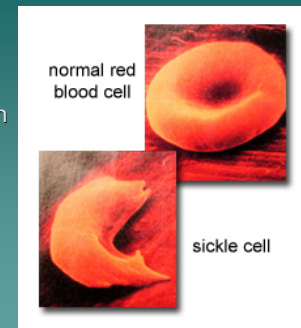
What does co-dominant mean?
Both genes always show



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

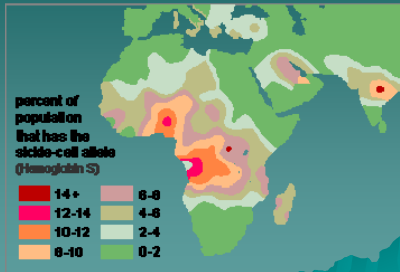
- ◆ Risk of blood-borne pathogens via transfusion
- ◆ High risk of disease
- ◆ Normal life possible with proper precautions & treatment



Sickle-cell Anemia

6. Distribution / Frequency

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



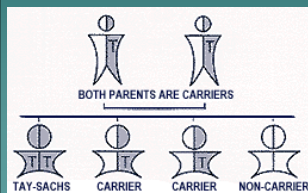
Tay Sachs' Disorder

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 dvlpmnt. slows down
- ◆ gradual loss of motor skills & mental f(x)s.
- ◆ blindness, deafness, mental retardation
- ◆ usually fatal by age five.

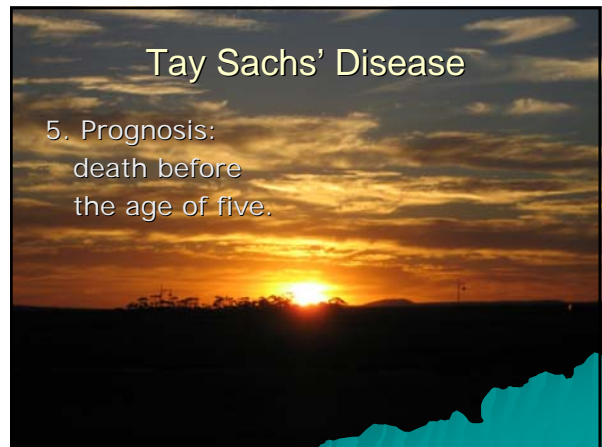
Tay Sachs' Disease

4. Treatment: none



Tay Sachs' Disease

5. Prognosis: death before the age of five.



Tay Sachs' Disease

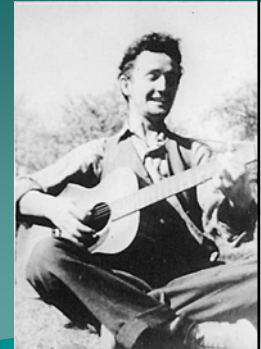
6. Distribution / Frequency

- 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

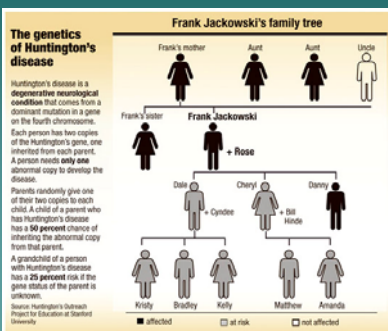


Woody Guthrie, folksinger

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

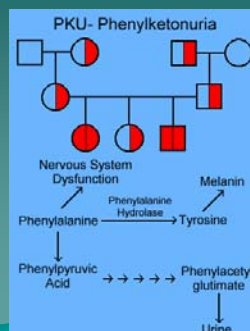
Phenylketonuria

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 within two weeks of birth in Colorado)
- ❖ With low phenylalanine diet,
no mental retardation



PKU

6. Distribution / Frequency

- ◆ 1/15,000 births
- ◆ All ethnic groups



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