Tay-Sachs Disease
- Description: disorder causing buildup of fatty substances in brain and nerve cells eventually resulting in death by age 5
- Mode of Inheritance: autosomal recessive
- Occurrence: 1 of 30 of Central and Eastern European Ashkenazi Jewish descent
- Symptoms: mild motor weakness, occasional eye twitches and progressively worsens to loss of motor skills, convulsions, blindness, paralysis and death
- Treatment: use of medication to treat symptoms
- Types: acute (infantile), sub acute (juvenile), chronic (adult)
- For More Information: http://www.familyvillage.wisc.edu/lib_tays.htm

Parkinson Disease
- Description: malfunctioning of brain cells, usually affecting cells that control movement
- Mode of Inheritance: in most cases appears to be autosomal dominant
- Occurrence: ~50,000 Americans diagnosed each year
- Symptoms: tremors, rigidity of limbs and muscles, general lack of movement, dementia, depression, decreased sense of smell, problems controlling heart rate and blood pressure
- Treatment: use of medication to treat symptoms
- Types: early onset (before age 50), late onset (50 or older)

Colon Cancer
- Description: disease in which malignant cells form in the tissues of the colon
- Mode of Inheritance: autosomal dominant; new mutations linked to chromosomes 2 and 5
- Stages: 0—innermost lining of the colon only
1—second and third layers of the colon
2—outside colon in nearby tissue but not in lymph nodes
3—in lymph nodes but not other parts of the body
4—found in other parts of the body
- Treatment: surgery, chemotherapy, radiation therapy depending on stage

An Insight To Common Genetic Diseases
Prepared by Jessica Kabool in fulfillment of GN493 student project Spring 2003
Turner Syndrome
- Description: chromosomal disorder of females, due to having only one X chromosome
- Mode of Inheritance: improper separation of chromosomes in gamete formation
- Occurrence: 1 of 2500 live female births
- Symptoms: short stature, lack of sexual development at puberty, webbed neck, heart defects, kidney and other abnormalities
- Treatment: growth hormone, estrogen replacement therapy

Hemophilia
- Description: genetic bleeding disorder caused by a shortage of clotting Factors VIII or IX
- Mode of Inheritance: X-linked recessive
- Occurrence: mostly affects men; in rare cases women are affected
- Symptoms: bleeding into joints or muscles causing pain or swelling, abnormal bleeding after injury, easy bruising, frequent nose bleeds, blood in urine
- Treatment: replacement of clotting factors
- Types: Hemophilia A (lack of clotting Factor VIII), Hemophilia B (lack of clotting Factor IX)
- For More Information: http://www.hemophilia.org

Diabetes
- Description: condition in which the pancreas produces insufficient insulin essential in controlling blood sugar levels
- Mode of Inheritance: some autosomal genes increase susceptibility but environment is also a factor
- Occurrence: affects 17 million Americans
- Symptoms: frequent urination, extreme thirst and dry mouth, weight loss, increased hunger
- Treatment: monitoring blood sugar levels, taking insulin, well-balanced diet, regular exercise, weight control
- Types: Type I: the pancreas does not produce insulin
  Type II: the pancreas does not produce enough insulin or does not respond to insulin

Phenylketonuria (PKU)
- Description: lack of phenylalanine hydroxylase to convert phenylalanine to tyrosine causes a buildup of toxic ketones
- Mode of Inheritance: autosomal recessive mutation in chromosome 12
- Occurrence: 1 of 13,500-19,000 births
- Symptoms: musty smell to skin, hair and urine, vomiting and diarrhea, irritability, skin problems
- Treatment: special diet with reduced phenylalanine
- For More Information: http://www.pkunews.org

Lupus
- Description: an autoimmune disease that can cause inflammation, pain and tissue damage throughout the body
- Mode of Inheritance: specific autosomal gene defects adversely affecting the immune system, cell functions and hormones; thought to be linked to chromosomes 1 and 6
- Occurrence: increases for family history, use of certain medications, females, African Americans and Asians, people age 15-40
- Treatment: self-care and medication based on severity of symptoms
- For More Information: http://www.lupus.org