Breast Cancer
Mode of Inheritance: mix of genetics and environment, multifactorial
Genes: BRCA1, BRCA2, p53, ATM, CHEK2, Her2neu, and PTEN/MMAC1 some of which are in chromosomes 13 and 17
Occurrence: 1 in 9 women, 257,800 new cases per year, 1,500 cases in men-39,000 women and 400 men will die this year
Prevention or Cure: none– though early detection is more likely when monthly Breast Self Exams are conducted and when women annually receive mammograms—many treatments are available that may allow patients to go into remission but no true “cure”
Symptoms: tumor in breast often felt as a hard lump or seen through mammography
Types: ductal carcinoma and lobular carcinoma (both invasive and noninvasive in situ cancers)
Treatments: surgery, radiation, hormone therapy, chemotherapy, biological therapy
Life Expectancy: dependent on stage of cancer at diagnosis—may be months or normal

Dwarfism
Definition: a genetic condition resulting in short stature (adults of 4’10” height or less)—effects depend on the type & severity of dwarfism
Mode of Inheritance: achondroplasia is autosomal dominant in chr. 4 and 5, dysostosis is due to SLC26A2 gene mutation in chr. 5, spondyloepiphyseal dysplasia congenita (SED) due to COL2A1 gene mutation in chr. 12, also types due gene defects in chr. 19 and 5
Occurrence: 1 in 10,000 births
Prevention or Cure: none
Types: there are 940 types of dwarfism and dysplasia—known most common is Achondroplasia (50-80% of cases), dysostosis (1 in 110,000 births), SED (1 in 95,000 births) and Turner Syndrome is also considered a type of dwarfism
Symptoms: vary with type of dwarfism—some kinds are lethal, some are milder than others
Treatment: no treatment for actual dwarfism but a variety of treatments to curb other effects related to dwarfed condition
Life Expectancy: majority have normal life span

Albinism
Definition: little to no pigment in eyes, hair, and skin due to reduced production of melanin caused by genetic defects
Mode of Inheritance: Type 1 in chr. 11 (tyrosinase gene defect), Type 2 in chr. 15 (defect in P gene), Type 3 in chr. 9 and Hernansky-Fudlack syndrome in chr. 10 all autosomal recessive; ocular albinism is X-linked
Occurrence: 1 in 17,000 in U.S.
Prevention or Cure: none
Symptoms: lack of pigment, eye problems, and skin cancers (if they do not use sunscreen and other precautions)
Types: Type 1 (tyrosinase-related), Type 2 (some pigment) & Type 3 ocucutaneous albinisms, Hernansky-Fudlack syndrome in chr. 10, all autosomal recessive, ocular albinism is X-linked
Treatments: sunscreen, opaque clothing, visual rehabilitation, optical aids
Life Expectancy: normal though Hernansky-Fudlack syndrome type’s lives may be shortened by lung disease & other problems

Alzheimer Disease
Definition: form of dementia that attacks nerve cells in the cerebral cortex, basal forebrain, and hippocampus—affects control of thought, memory, and language
Mode of Inheritance: genetic or sporadic—Familial AD is autosomal dominant and due to gene mutations in chromosomes 1, 14, and 21 while Sporadic AD is due to mutations of the three forms of the apoE gene found in chromosome 19
Occurrence: up to 4 million Americans—10% of cases are Familial
Cure or Prevention: none
Symptoms: forgetfulness, getting lost, forgetting how to do simple tasks, trouble speaking, writing, reading, understanding, wandering, anxious or aggressive
Treatments: vitamin E, drugs tacrine (Cognex), donepezil (Aricept), rivastigmine (Exelon), or galantamine (Reminyl); testing use of NSAIDs, gingko biloba, and estrogen as possible treatments
Life Expectancy: 8 to 10 years after diagnosis, though some may live 20 years with the disease
Duchenne Muscular Dystrophy
Definition: most common and severe type of MD
Mode of Inheritance: X-linked recessive inheritance or new mutation—genotypes may result from point mutations, frameshift mutations, deletions, or duplications
Occurrence: 1 in 3,500 boys
Prevention or Cure: none
Symptoms: deficient in the protein dystrophin, waddling gait, frequent falls, difficulty with steps & rising from ground, some mental retardation, lose ability to walk & reduced abilities with hands & feet
Treatments: Prednisone, Deflazacort, Oxandrolone, surgical insertion of spinal rod, night splints on ankles, orthopedic appliances, physical therapy, surgical release of contractures (when short muscles around joints cause abnormal positioning)
Life Expectancy: 20 years +/- 3 years

Marfan Syndrome
Definition: disorder of connective tissue that affects many organ systems, including the skeleton, lungs, eyes, heart and blood vessels
Mode of Inheritance: autosomal dominant—due to dominant gene in chromosome 15, mutation in Fibrin-1 (FBN1) gene or may be caused by new mutation (25-33% of cases)
Occurrence: 1 in 20,000 births
Prevention or Cure: none
Symptoms: deficient in the protein dystrophin, waddling gait, frequent falls, difficulty with steps & rising from ground, some mental retardation, lose ability to walk & reduced abilities with hands & feet
Treatments: Prednisone, Deflazacort, Oxandrolone, surgical insertion of spinal rod, night splints on ankles, orthopedic appliances, physical therapy, surgical release of contractures (when short muscles around joints cause abnormal positioning)
Life Expectancy: 20 years +/- 3 years

Sickle Cell Anemia
Definition: inherited blood disease from defective hemoglobin (HbS)—sickle shaped red blood cells
Mode of Inheritance: autosomal recessive in chr. 11—must be inherited from both parents to be expressed, if transmitted from one parent only it results in sickle cell trait, due to gene mutation thousands of years ago
Occurrence: 1 in 500 African-American births, 1 in 1000-1400 Hispanic-American births, 72,000 Americans affected and 2 million have sickle cell trait—most common among people with ancestors from sub-Saharan Africa, Spanish speaking regions, Saudi Arabia, India, and Mediterranean countries
Prevention or Cure: none
Symptoms: chronic anemia, periodic pain, cells get stuck in narrow blood vessels and block blood flow, eye problems, strokes, yellowing, infections, delayed growth
Treatments: pain relievers, blood transfusions, penicillin when young, anticancer drug hydroxyurea, etc.
Benefits: sickle cell trait carrier is resistant to malaria
Life Expectancy: 55 years +/- 25 years

Cystic Fibrosis
Definition: abnormally thick, sticky mucus is produced by the body’s epithelial cells—prevents enzymes needed for digestion from reaching intestines, clogs airways of the lungs blocking the flow of air—affects urinary tract, liver, & reproductive tract
Mode of Inheritance: autosomal recessive—double dose of the defective gene (one from each parent) in chr. 7
Occurrence: 1 in 31 Americans is a carrier, 30,000 Americans affected
Prevention or Cure: none
Symptoms: very salty-tasting skin; persistent coughing, wheezing or pneumonia; excessive appetite but poor weight gain; and bulky stools
Treatments: chest physical therapy for breaking up mucus, antibiotics, enriched diet, replacement vitamins and enzymes, currently testing gene therapy treatment
Life Expectancy: 30 years +/- 15 years

Down Syndrome
Mode of Inheritance: 99% are sporadic, 1% are inherited (translocation)—all due to an extra chr. 21
Types of Trisomy 21: in 94% of cases all cells have extra chromosome 21, 4% Translocation (part of extra 21 attached to another chromosome), and 2% Mosaic (some cells have extra 21)
Occurrence: 1 in 750 live births
Prevention or Cure: none, though younger mothers are less likely to have babies with Down Syndrome
Symptoms: mental retardation, cognitive disability, developmental delays—common physical traits include epicanthal folds of the eyes, flattened Bridge of the nose, a single palmar crease, and decreased muscle tone
Life Expectancy: reduced 10-20 years

Huntington Disease
Definition: progressive degeneration of brain tissue in the basal ganglia that affects the ability to walk, talk, think, and reason—onset generally 40-45 years of age but can be as early as teens or as late as 70
Mode of Inheritance: autosomal dominant in chromosome 4 due to repeats of CAG in gene—the more repeats, the more severe the case—juvenile HD thought to be inherited from father
Occurrence: 1 in 10,000 people in US
Prevention or Cure: none
Symptoms: depression; mood swings; forgetfulness; clumsiness; involuntary twitching; lack of coordination; loss of concentration and short-term memory; deterioration of walking, speaking, and swallowing—may vary for juvenile HD
Treatments: Botulinum toxin injections, nutrition alteration, medications to lessen symptoms
Life Expectancy: dependent on age of onset—death often from complications such as choking, infection, or heart failure—juvenile HD generally progresses more rapidly than adult-onset HD

Life Expectancy: 20 years +/- 3 years