

Presentation Notes from Genetics Projects

<p style="text-align: center;">Down Syndrome</p> <ol style="list-style-type: none"> 1. -Slanted eyes, broad feet/hands, short height, crease on palm, low IQ, loose muscle tone, flat nose - Heart, eyes, and ears are affected 2. Error during meiosis → 3 chromosomes 21 instead of a pair (Trisomy) 3. physical therapy, special education treat eye, ear, and heart problems community programs 	<p style="text-align: center;">Tay-Sachs Disease</p> <ol style="list-style-type: none"> 1. low muscle tone - Eventual paralysis vision difficulty - Eventual blindness Brain function affected - Seizures 2. Error of HEX-A gene on chromosome 15 3. Gangliosides (fatty tissues) builds up in brain Physical therapy, seizure medications and Feeding tubes
<p style="text-align: center;">Cystic Fibrosis</p> <ol style="list-style-type: none"> 1. Excess mucus forms → respiratory and digestive systems Salt levels are not controlled 2. Error of CFTR gene on chromosome 7 3. Drainage tubes, Clap on back Good nutrition, Antibiotics, Steroids 	<p style="text-align: center;">Muscular Dystrophy</p> <ol style="list-style-type: none"> 1. 9 Types - Duchenne is most common → affects skeletal and heart muscles 2. Error of DMD gene on X chromosome → lack of muscle protein dystrophin 3. Muscle wastes away → exercise, physical therapy, surgery
<p style="text-align: center;">Hemophilia</p> <ol style="list-style-type: none"> 1. Blood does not clot properly → Missing clotting factors 2. Error of F8/F9 genes on X chromosome 3. Blood transfusions of missing factors or frozen plasma that's been thawed Low contact sports - they bruise easily 	<p style="text-align: center;">Sickle Cell Anemia</p> <ol style="list-style-type: none"> 1. Red Blood cell becomes sickle shaped → swelling in joints, pain → Dizziness, headaches, jaundice 2. Error of HBB gene on chromosome 11 3. Bone marrow transplants Hydroxyurea Antibiotics Pain Relievers

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<p style="text-align: center;">Huntington's Disease</p> <ol style="list-style-type: none"> 1. Damaged brain cells, speech and swallowing difficulty, muscle control irregular, memory weak, emotions hard to control 2. Error of the DNA structure - normal CAG repeats 7-35 - HD has 36-120 CAG repeats on chromosome 4 3. Antiepileptic and Antidepressant medications Physical therapy, speech therapy 	<p style="text-align: center;">Turner's Syndrome</p> <ol style="list-style-type: none"> 1. Swollen hands/feet, short height, lack of puberty, droopy eyelids, flat chest, infertility (no ovaries) 2. Error during meiosis Missing X-chromosome or damaged X-chromosome → missing SRY gene 3. Estrogen replacement Growth hormone replacement Diets - watch for diabetes Egg donor to have children
<p style="text-align: center;">Marfan Syndrome</p> <ol style="list-style-type: none"> 1. Affects eyes, skin, lungs, bones and muscles → connective tissue concern Aorta of Heart enlarged or damaged 2. Error of FBN1 gene on chromosome 15 3. Annual evaluations of back and heart, eye exams Medications for circulatory system Surgery if needed 	<p style="text-align: center;">Frederick Sanger</p> <ol style="list-style-type: none"> 1. Born Aug. 13, 1918 Retired 1985 2. Sequencing of amino acids for various proteins and insulin 3. Advances in treatment for diabetes Better understanding of structure and function of many proteins
<p style="text-align: center;">Rosalind Franklin</p> <ol style="list-style-type: none"> 1. Born July 25, 1920 Died April 16, 1958 ovarian cancer 2. X-ray picture showing basic dimensions and structure of DNA → Helical shape 3. → work confirmed by Watson & Crick Lead to Human Genome Project Better understanding of the mechanism of many genetic disorders 	<p style="text-align: center;">James D. Watson and Francis Crick</p> <ol style="list-style-type: none"> 1. Born April 6, 1928 Born June 8, 1916 Died July 28, 2004 2. Nobel prize in Physiology for DNA structure 3. Lead to understanding of how DNA replicates, how proteins are made and the Human Genome Project where genes were mapped out.

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<p style="text-align: center;">Hugo de Vries</p> <ol style="list-style-type: none"> 1. Born Feb. 16, 1848 Died May 1, 1935 Netherlands Dutch Botanist and Geneticist 2. Mutation Theory → how mutations can lead to new species 3. → Rediscovered Mendel's Laws of Heredity Lead to deeper understanding of Charles Darwin's Theory of Evolution 	<p style="text-align: center;">Barbara McClintock</p> <ol style="list-style-type: none"> 1. Born June 16, 1902 Died Sept. 2, 1992 Hartford, CT 2. Mobile genetics - "Jumping genes" - used maize or corn Nobel Prize for Physiology 3. Advanced understanding of how variation develops in a species Lead to advances in evolution
<p style="text-align: center;">George Beadle</p> <ol style="list-style-type: none"> 1. Born Oct. 20, 1903 Died June 9, 1989 2. "One gene - One enzyme" theory Nobel Prize for Chemistry 3. Earned several other awards Advanced understanding of how genes control chemical reactions in body. 	<p style="text-align: center;">Ian Wilmut</p> <ol style="list-style-type: none"> 1. Born July 7, 1944 Born/Raised in England 2. Cloning Process - Dolly the Sheep 1997 - Frozen embryo - calf named Frosty 1993 - 1st clones were Megan/Morag (lambs) 1996 3. Can lead to advances in treatments for diseases → make disease resistant species, for organ transplants
<p style="text-align: center;">Thomas H. Morgan</p> <ol style="list-style-type: none"> 1. Born Sept. 25, 1866 Died Dec. 4, 1945 Lexington, Kentucky 2. Father - Charlton H. Morgan BS 1886 University of Kentucky Morphology, Physiology, Embryology 3. Chromosome Theory - linear linkage of genes on chromosomes - some genes are linked only to the sex chromosomes where they can affect one gender more than the other. 	<p style="text-align: center;">Phenylketonuria</p> <ol style="list-style-type: none"> 1. Delayed intellectual ability Delayed development Behavioral, Emotional, Psychiatric problems Seizures lead to neurological problems 2. Hyperactivity Stunted Growth Tremors 3. Defective PAH gene on chromosome 12 Can't produce enzyme that breaks down phenylalanine Maintain strict diets - no dairy, no meat, no eggs - no phenylalanine (amino acid) - low protein