Genetic Disorders Responses (from the web links, main source indicated for each disorder)

1. Cri du Chat (https://www.genome.gov/19517558)

a) Part of the small arm of chromosome 5 is deleted in the genome of individuals with this disorder.

b) Phenotype can vary but may include some or all of these traits:

- high-pitched cat-like cry
- mental retardation
- delayed development
- distinctive facial features, small head size (microcephaly), widely-spaced eyes (hypertelorism)
- low birth weight and weak muscle tone (hypotonia) in infancy
- difficulty with language
- feeding difficulties
- delays in walking,
- hyperactivity
- scoliosis
- significant retardation
- serious organ defects and other life-threatening medical conditions (although most individuals with cri du chat syndrome have a normal life expectancy)

2. Down Syndrome (http://learn.genetics.utah.edu/content/disorders/chromosomal/down/)

a) Individuals with Down Syndrome have an extra copy (3 total) of chromosome 21.

b) Phenotype:

- distinct facial features: a flat face, a small broad nose, abnormally shaped ears, a large tongue, and upward-slanting eyes with small folds of skin in the corners
- increased risk of developing a number of medically significant problems, including respiratory infections, gastrointestinal tract obstruction (blocked digestive tract), leukemia, heart defects, hearing loss, hypothyroidism, and eye abnormalities
- moderate to severe intellectual disability; children with Down syndrome usually develop more slowly than their peers and have trouble learning to walk, talk, and take care of themselves
- decreased life expectancy

3. Edward Syndrome (Trisomy 18) (http://ghr.nlm.nih.gov/condition/trisomy-18)

a) Individuals with Trisomy 18 have an extra copy (3 total) of chromosome 18.
b) Phenotype may include:

- slow growth before birth and low birth weight
- heart defects and abnormalities of other organs that develop before birth
- small, abnormally shaped head
- a small jaw and mouth
- clenched fists with overlapping fingers
- several life-threatening medical problems with low probability of survival past the year of life
- severe intellectual disability

4. Klinefelter Syndrome (https://ghr.nlm.nih.gov/condition/klinefelter-syndrome)

a) Chromosome 46: Extra sex chromosomes (most commonly an extra X [XXY]; rarely two or even three X chromosmes)

b) Phenotype may include:

- small testes with decreased testosterone production
- delayed or incomplete puberty
- breast enlargement (gynecomastia)
- reduced facial and body hair
- infertility
- effects on genital morphology
- taller body height
- increased risk of developing breast cancer and a chronic inflammatory disease called systemic lupus erythematosus
- learning disabilities, delayed speech and language development

5. Patua Syndrome (Trisomy 13) (https://ghr.nlm.nih.gov/condition/trisomy-13)

a) Individuals with Trisomy 18 have an extra copy (3 total) of chromosome 13.

b) Phenotype may include:

- severe intellectual disability
- physical abnormalities in many parts of the body
- heart defects
- brain or spinal cord abnormalities
- very small or poorly developed eyes (microphthalmia)
- extra fingers or toes

- cleft lip with or without a cleft palate
- weak muscle tone (hypotonia)
- many infants die within their first days or weeks of life

6. Philadelphia Chromosome (https://en.wikipedia.org/wiki/Philadelphia_chromosome)

a) Patients with the Philadelphia Chromosome have a shortened chromosome 22, because part of their chromosome 9 has been translocated to chromosome 22.

b) Phenotype: Philadelphia Chromosome leads to the development of blood cancers (most commonly Chronic myeloid leukemia (CML)). The *ABL1* gene, normally found on chromosome 9, is translocated to chromosome 22 where it gets chronically turned on by the control elements that regulate the *BCR* gene (which was already located on chromosome 22). This combo of these two genes (*BCR-ABL1*) produces a protein that causes cells to divide continuously.

7. Turner Syndrome (http://ghr.nlm.nih.gov/condition/turner-syndrome)

a) Females with a missing X chromosome or who have a structurally altered X chromosome have Turner Syndrome.

b) Phenotype may include:
- short stature
- early loss of ovarian function
- not undergoing puberty without hormone therapy
- infertility
- extra folds of skin on the neck (webbed neck)
- a low hairline at the back of the neck
- puffiness or swelling (lymphedema) of the hands and feet
- skeletal abnormalities
- kidney problems
- heart defects
- developmental delays, nonverbal learning disabilities, and behavioral problems