**Genetic Disorder Counseling**

Patients are referred to a genetic counselor for many reasons. This can include:

* There is a genetic condition that happens to be in either partners family.
* There is a child with a learning difficulty or developmental delay, or has health problems and the Doctor thinks there is a genetic link.
* Either parent has a genetic disorder that can be passed on to children.
* A test was done during pregnancy and it shows that there is an increased risk the baby has a genetic condition.
* The mother has had a miscarriage or stillbirth.
* Particular types of cancer have occurred in several close relatives.
* The Doctor believes that a genetic specialist may be able to provide you with additional information about current health conditions.

Individuals that have some of these risk factors seek Genetic counselors. Genetic counselor jobs include:

* Explaining tests available to help a family confirm a diagnosis
* Providing information about the condition and how it is passed on
* Discussing the risk of having a genetic disorder and the risk of it being passed on
* Talking about ways of living with the condition and the medical, psychological and social support available.
* Answer any question about the disorder.

**Overview:**

In this activity, you will take on the role of a genetic counselor. You will first need to research a genetic disorder and create a brochure about the disorder. This material will provide information on a given genetic disorder that can be used to inform patients and provide support to individuals that have this genetic disorder in the family. Then you will help a hypothetical family determine their risk for the disorder, and their risk of passing on the trait to any children they may have.

**Part 1:**

Directions: Look at the list of genetic disorders. Chose one that interests you and use the following questions AS A GUIDE to help you research and fully understand the disorder.

* What other names are there for this disorder? (Any common names?)
* What body system(s) are affected by this disorder?
* What causes the disorder? (What kind of mutation. If it is Non-disjunction, don’t use it as it will not work for part 3 of this activity)
* What gene or chromosome is affected by this disorder?
* Are there prenatal tests for this disorder?
* What are the symptoms, and what age do we typically see the onset of the symptoms?
* What population is affected? Can anyone be a candidate for this disorder? Are some populations more at risk?
* How is it inherited? (Sex-linked, Autosomal, Recessive, Dominant)
* What kind of medical assistance will the affected person need? Will further assistance be necessary as the person ages? What is the prognosis for an individual with this genetic disorder?
* Are there any treatments or cures?
* Can this individual have children? Will the children be affected?
* What is the current status of research on this disorder? Is there a cure coming soon? What types of studies are being done? (Stem cell? Gene therapy?)

**Note Guide (this in no way is a complete list of questions/ information, simply a guide). You will need a works cited page, so make sure you are listing the resources you used!**

1. What is the name of the genetic disorder? Are there other names for this disorder?
2. What system or part of the body does it affect? How does it affect it?
3. What gene and/or chromosome is mutated in this disorder? How is it mutated/ changed? (insertion, deletion, non-disjunction).
4. Is this mutation dominant or recessive? How is it inherited?
5. What are the symptoms of the disorder?
6. What are the possible treatments for this disorder? What research is being done for future treatment? (gene therapy etc.)
7. What is the prognosis for someone who has tested positive for this genetic disorder?
8. Does this disorder affect specific populations of people more so than other populations?
9. What is the percentage or ratio of the population suffering from this disorder?
10. What are at least two support groups in Albuquerque/ New Mexico that can lend support in case of a positive diagnosis? (Include all contact information and a brief description of the support group).

Additional Notes:

Resources used:

**Part 2:**

Create an informative Brochure to give to families that are faced with this genetic disorder.

Include all of the information from your research as well as at least TWO informative visuals for your “clients”. Make sure that your educational material is easy to read, informative and that the information is accessible to a wide audience. For this activity assume that your audience are adults with a typical high school science background. However, make sure you explain complex concepts. You also need to make it visually appealing (After all, no one will read the information if it’s boring! ☺)

**Part 3:**

Now that you have thoroughly researched your genetic disorder, you are going to take on the role of a genetic counselor.

1. On a separate piece of paper, create a hypothetical pedigree for a family that has this genetic disorder in the family. There must be at LEAST 3 generations, and there must be at LEAST 15 individuals in the family. When you are creating your pedigree, keep in mind which two individuals in the pedigree you are “counseling”.
* In your pedigree, you MUST represent the mode of inheritance correctly (sex-linked/ autosomal and dominant/ recessive).
* Must include the genotypes of all of the individuals in the pedigree.
* Must include a punnett square for the individuals that you are “counseling” to inform them of the likelihood of having offspring with the disorder or the likelihood of them having the disorder. (You are NOT telling them what decision to make about having children, or getting further genetic testing, you are simply giving them information so they can make a decision).
* Must include a summary of the findings from the pedigree. Include any punnett squares for support of your findings. This summary must clearly show that you understand the mode of inheritance, and the inheritance patterns of the disorder.

**Genetic Disorders:** (You are not limited to this list, but if the condition is a result of a chromosomal mutation, it will be difficult to do Step 3 of this activity.)

Achondroplasia

Adrenoleukodystrophy

Albinism

Alzheimers Disease

Beta-Thalassemia

Breast Cancer

Cystic Fibrosis

Fragile X

Hemophilia

Marfan Syndrome

Muscular Dystrophy

Neurofibromatosis

Phenylketonuria

Prader-Willi

Severe Combined Immunodeficiency

Sickle Cell

Tay-Sachs

Hemochromatosis

Huntington Disease

**Brochure/ Pedigree Rubric:**

4-Excellent 3- Very good 2-Acceptable 1-Lacking 0-Missing

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| --- | --- | --- |
| **Section of Brochure** | **Score** | **Comments** |
| **Overall (x1)**Colorful, organized, grammer/ spell checked | 4 3 2 1 0 |  |
| **Works Cited (x1)**At least **4** sources referenced and cited on a separate page from the brochure | 4 3 2 1 0 |  |
| **Brochure Information**: **(x10)**Clear understanding of the genetic disorder. Accurate info. Includes: * Name
* System
* Gene/ Chromosome affected
* Mutation
* Dom/ Reces
* Symptoms
* Treatment
* Future research
* Prognosis
* Pop affected
* 2 support gps
* Additional info
 | 4 3 2 1 0 |  |
| **Visuals (x2)**At least two USEFUL visuals that help explain the condition. | 4 3 2 1 0 |  |
| **Pedigree (x5)*** Straight edge used
* Labeled correctly
* Genotypes correct
* Mode of inheritance clear and accurate
* Demonstrates a complete understanding of the inheritance of the disorder
 | 4 3 2 1 0 |  |