Pick the Risk: The Polygenic Pedigree Challenge

Activity Overview

Participants are challenged to track and record the passage of colored pom poms (representing genes) through generations of a family using a pedigree. Participants learn that common chronic diseases (such as heart disease) run in families and are caused by the combined action of multiple genes.

Learning Objectives

- An inherited trait can be determined by one or by many genes.
- All humans have the same genes, but each inherits slightly different forms or "flavors" of each gene.
- Many common diseases (such as heart disease) run in families and have a genetic component.
- Most common diseases are caused by the combined action of multiple genes and environmental factors.
- An individual's risk of developing a common disease is estimated by looking at siblings, parents and grandparents in a family medical history.

Logistics

Time Required

- Activity Time: 30 - 45 minutes
- Prep Time: 30 minutes

Materials

Copies of participant pages, colored pencils or crayons, disposable cups, colored pom poms

Prior Knowledge Needed

Genes are passed from parents to offspring and contribute to observable physical characteristics. Pedigrees are used to track genetic information.

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Instructions

To engage participants in this topic:

- Compare the prevalence of rare genetic disorders caused by a single gene such as cystic fibrosis (1 in 10,000) with the prevalence of more common diseases such as heart disease (1 in 3).
- Ask the participants: Do common diseases like heart disease, diabetes, or colon cancer have a genetic component?
- Explain that most common diseases do have a genetic component and tend to run in families. However, common diseases differ from rare genetic disorders in that they are usually not caused by defects in a single gene. Rather, they result from the combined effects of multiple genes and environmental factors. Thus, they are called multifactorial diseases.
- Explain that because more than one gene is involved in most common diseases, the inheritance of a common disease is not predictable.
- Information found in a family health history and recorded on a pedigree is used to estimate an individual's genetic risk (low, medium, or high) of developing a common disease.

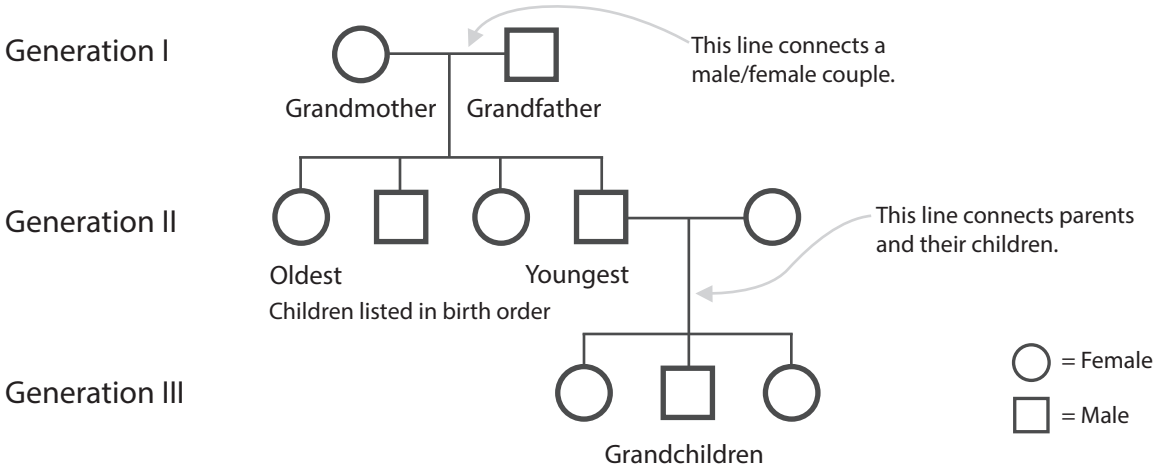
Quantities

Per Group of 2

- One copy of participant pages
- 2 - 3 disposable cups
- Colored pencils or crayons
- Colored pom poms (5 different colors: at least 10 red, 2 yellow, 1 each of orange, green, blue)

Begin activity:

- Explain that the following activity will explore how common "polygenic" diseases (in this case, heart disease) are inherited.
- Invite participants to find a partner with whom they will work to complete the activity; pass out the participant pages and other materials.
- Review the symbols and structure used for a pedigree:



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- Invite participants to begin by following the instructions found on pages P-1 and P-2; each pair of participants should complete the pedigree analysis and answer the questions that follow.

Common Misconception

Participants may think that all heritable traits (and genetic disorders) are caused by a single gene and exhibit dominant or recessive patterns of inheritance. But more commonly, traits result from the combined action of many genes and environmental factors. Such multifactorial traits can exhibit varied and complex patterns of inheritance that are not easy to predict.

Group discussion:

- Point out that this activity differs from reality in the following ways:
  - » The number of genes contributing to a polygenic disease is usually not known.
  - » The number of genes carried by parents or offspring that can increase heart disease risk is not known.
  - » Environmental factors can also vary an individual's risk of developing multifactorial diseases.
- Discuss the following questions as a group, comparing and contrasting the Pedigree results obtained by each pair:
  - » The grandfather in this family was a "high risk" individual. How many of his children were either medium or high risk individuals? How many of his grandchildren were either medium or high risk individuals? (Answers will vary.)
  - » Did the number of "medium risk" and "high risk" individuals decrease or increase over subsequent generations? Why do you think that happened? (Answer: Decrease. Risk of inheriting heart disease from an affected individual (suchas a grandparent) decreases through the generations because it is unlikely that all of the necessary risk factors [genes] will be passed down to less closely related family members.)
  - » In this activity you were able to label family members as having a low, medium or high risk of developing heart disease. In reality, do you think it might be difficult to predict an individual's risk of developing heart disease? Why? (Answer: An individual's risk of developing heart disease IS difficult to predict because of the reasons outlined under the first group discussion point.)
  - » If a parent is diagnosed with heart disease, does that mean the children will have it also? Defend your answer. (Be sure to include the key word "risk" in your answer.) (Answer: Not necessarily. But because heart disease does have a genetic component, children of an affected parent have an increased risk of developing heart disease relative to the population at large.)
- Emphasize that these are a few of the reasons why heart disease and other common diseases are so complex, and why the inheritance pattern for such diseases are difficult to predict.
- Therefore, individuals are placed in general categories (high, medium, or low risk groups) based on features from their family health history that correlate with a certain probability of developing a disease.
  - » For example, it is said that an individual who has a parent (or possibly a grandparent) with heart disease may be "at risk" and should take steps to protect themselves.
- Describe the important features to identify in a family health history (see chart at http://learn.genetics.utah.edu/units/health), including what is meant by a "close" relative.

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» Siblings, parents, and possibly grandparents, are informative when assessing an individual's risk of developing a common disease and need be included in a family health history. It is unlikely that all of the same risk factors (genes) will be present in less closely related family members.

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- Discuss why each feature in the chart indicates an individual may be at increased risk for developing heart disease.
  - » Each feature indicates that the family has accumulated more risk factors (genetic or environmental). Therefore, an individual in this family is more likely to develop disease.
- Discuss behaviors and choices that can reduce an individual's risk of developing heart disease.
- Conclude the discussion by reminding participants that genetic susceptibility does not mean an individual will inevitably inherit a disease. Positive lifestyle changes and healthy living can reduce genetic risk dramatically. That is why it is so important to know your family health history. If you know you are "at risk" you can take steps to protect yourself.

Credits

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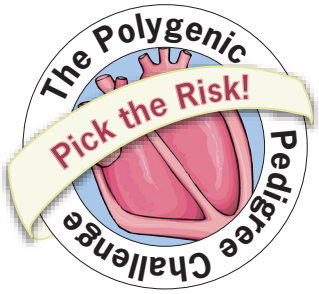

Eres un investigador que está averiguando sobre las enfermedades coronarias. Sabes que existen 6 genes en los seres humanos que pueden contribuir al riesgo de enfermedades coronarias. Todos los seres humanos tienen estos 6 genes, pero podemos heredar formas y "sabores" apenas distintos de estos genes. Tu desafío es rastrear y registrar el traspaso de estos 6 genes (indicados con los pompones de colores) de generación en generación en una familia utilizando la genealogía. Luego, deberás predecir qué miembros de la familia son más propensos a desarrollar una enfermedad coronaria.

Sigue estos pasos para completar el desafío.

Parte 1:

1. Consigue un árbol genealógico en blanco, crayones o lápices de colores, 2 vasos desechables y los pompones de colores que te dará tu maestro.

2. Escoge un vaso, el cual representará a la abuela. Coloca dentro del vaso 5 pompones rojos (5 genes "normales" que NO contribuyen a las enfermedades coronarias) y 1 pompón azul (1 gen que aumenta el riesgo de una enfermedad coronaria).

3. Escoge un vaso, el cual representará al abuelo. Coloca dentro del vaso 3 pompones rojos (3 genes "normales" que NO contribuyen a las enfermedades coronarias), 1 pompón naranja, 1 verde y 1 amarillo (3 genes que aumentan el riesgo de una enfermedad coronaria).

4. En el árbol genealógico, registra los colores de los pompones presentes en la abuela y en el abuelo, rellenando los círculos en blanco con los crayones o lápices de colores.

5. Con los ojos cerrados, mezcla y retira al azar 3 pompones de cada abuelo (6 pompones en total). Éstos representan la información genética heredada a su primer hijo.

Recuerda: ¡No hagas trampa! Debe ser al azar.

6. En el árbol genealógico, colorea la combinación de pompones que se transmiten al primer hijo.

7. Regresa los pompones al abuelo correspondiente. (Devuélvele los genes de la abuela a la abuela y los del abuelo al abuelo.)

8. Repite los pasos 5 a 7 para cada hijo indicado en el árbol genealógico.

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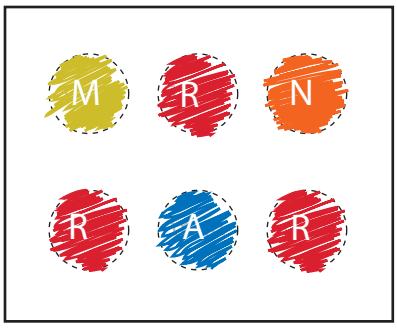
9. Diagnóstico:

Observa los 6 genes susceptibles a las enfermedades coronarias de cada individuo (la combinación de los pompones coloreados).

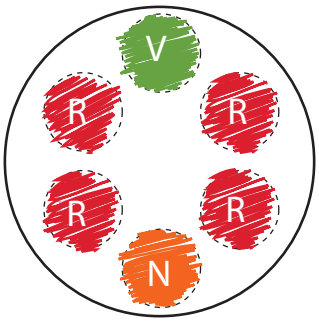
Recuerda: Las formas o "sabores" de un gen que aumentan el riesgo de una enfermedad coronaria son:

amarillo(M): naranja(N): verde(V): azul(A):

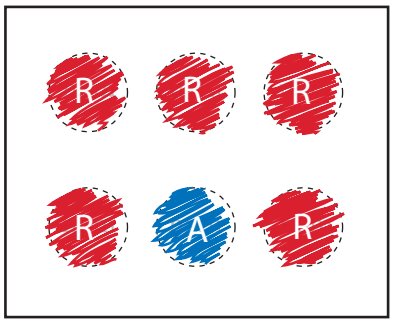
Cataloga a cada individuo del árbol genealógico con riesgo bajo, medio o alto, según este cuadro:



Riesgo alto



Riesgo medio



Riesgo bajo

Parte 2:

10. Uno de los hijos se casa con un individuo con "riesgo bajo". Coloca dentro de un vaso 5 pompones rojos y 1 pompón amarillo para representar los 6 genes susceptibles a las enfermedades coronarias de este individuo.

11. En el árbol genealógico, colorea la combinación de pompones (o genes) que lleva cada uno.

12. La pareja tiene hijos. Con los ojos cerrados, mezcla y retira al azar 3 pompones de cada padre (6 pompones en total). Éstos representan la información genética heredada a su primer hijo.

13. En el árbol genealógico, colorea la combinación de pompones que han transmitido a su hijo.

14. Regresa los pompones al padre correspondiente.

15. Repite los pasos 12 a 14 para determinar la formación genética de los otros dos hijos indicados en el árbol genealógico.

16. Cataloga a cada individuo agregado al árbol genealógico con riesgo bajo, medio o alto.

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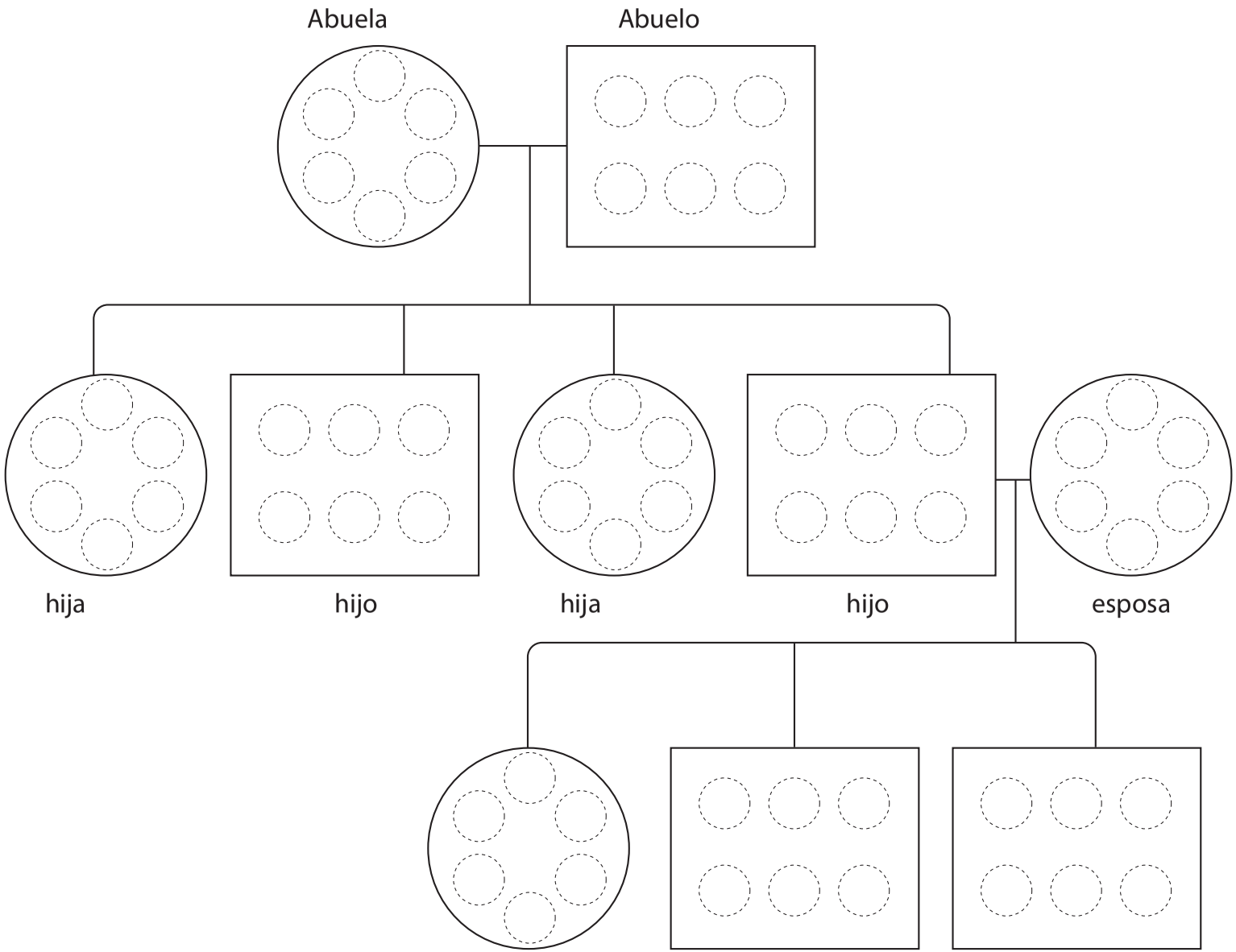
el árbol genealógico



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Responde estas preguntas:

1. El abuelo de esta familia era un individuo con "riesgo alto". ¿Cuántos de sus hijos eran individuos con riesgo medio o alto? ¿Cuántos de sus nietos eran individuos con riesgo medio o alto?

2. ¿El número de individuos con "riesgo medio" y con "riesgo alto" disminuyó o aumentó a través de las generaciones? ¿Por qué crees que sucede?

3. En esta actividad has podido calificar a los miembros de esta familia con riesgo bajo, medio o alto de desarrollar enfermedades coronarias. En realidad, ¿crees que sería difícil predecir el riesgo de un individuo de desarrollar una enfermedad coronaria? ¿Por qué?

4. Si se diagnostica a unos de los padres con una enfermedad coronaria, ¿eso significa que sus hijos también la padecerán? Argumenta tu respuesta. (Tu breve respuesta debe incluir la palabra clave "riesgo".)

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