PhenX Toolkit

Data Collection Worksheet

Please Note: The Data Collection Worksheet (DCW) is a tool to aid integration of a PhenX protocol into a study. The PhenX DCW is not designed to be a data collection instrument. Investigators will need to decide the best way to collect data for the PhenX protocol in their study. Variables captured in the DCW, along with variable names and unique PhenX variable identifiers, are included in the PhenX Data Dictionary (DD) files.

This section will help us understand what information people need to know in order to understand their sequencing results. Before you begin, you should know that we are using the term "gene variant" to mean a version of a gene. Sometimes two people have the same version of a gene (they have the same gene variant), and other times two people have different versions of a gene (they have different gene variants).

Below is a list of statements. They are either true or false. For each statement:

- Select True if you think it is true
- Select False if you think it is false
- Select Don't know/uncertain if you are not sure or don't know.

Please answer all of the questions. Don't worry if you do not know the right answers. We do not expect you to answer all of these correctly.

Information About Genes

1. Genes are made of DNA.

    [ ] True

    [ ] False

    [ ] Don't know/uncertain

2. Genes affect health by influencing the proteins our bodies make.

    [ ] True

    [ ] False

    [ ] Don't know/uncertain

3. All of a person's genetic information is called his or her genome.

[ ] True

[ ] False

[ ] Don’t know/uncertain

4. A person’s genes change completely every 7 years.

[ ] True

[ ] False

[ ] Don’t know/uncertain

5. The DNA in a gene is made of four building blocks (A, C, T, and G).

[ ] True

[ ] False

[ ] Don’t know/uncertain

6. Everyone has about 20,000 to 25,000 genes.

[ ] True

[ ] False

[ ] Don’t know/uncertain

Genes and health

7. Gene variants can have positive effects, harmful effects, or no effects on health.

[ ] True

[ ] False

[ ] Don’t know/uncertain

8. Most gene variants will affect a person’s health.

[ ] True

[ ] False

[ ] Don’t know/uncertain

9. Everyone who has a harmful gene variant will eventually have symptoms.

[ ] True

[ ] False

[ ] Don’t know/uncertain

10. Some gene variants have a large effect in health, while others have a small effect.

[ ] True

[ ] False

[ ] Don’t know/uncertain

11. Some gene variants decrease the chance of developing a disorder.

[ ] True

[ ] False

[ ] Don’t know/uncertain

12. Two unrelated people with the same genetic variant will always have the same symptoms.

[ ] True

[ ] False

[ ] Don’t know/uncertain

How genes are inherited in families

13. Genetic disorders are always inherited from a parent.

[ ] True

[ ] False

[ ] Don’t know/uncertain

14. If only one person in the family has a disorder, it can’t be genetic.

[ ] True

[ ] False

[ ] Don’t know/uncertain

15. Everyone has a chance for having a child with a genetic disorder.

[ ] True

[ ] False

[ ] Don’t know/uncertain

16. A girl inherits most of her genes from her mother, while a boy inherits most of his genes from his father.

[ ] True

[ ] False

[ ] Don’t know/uncertain

17. A mother and daughter who look alike are more genetically similar than a mother and daughter who do not look alike.

[ ] True

[ ] False

[ ] Don’t know/uncertain

18. If a parent has a harmful gene variant, all of his or her children will inherit it.

[ ] True

[ ] False

[ ] Don’t know/uncertain

19. If one of your parents has a gene variant, your brother or sister may also have it.

[ ] True

[ ] False

[ ] Don’t know/uncertain

Whole exome sequencing

20. Whole exome sequencing can find variants in many genes at once.

[ ] True

[ ] False

[ ] Don’t know/uncertain

21. Whole exome gene sequencing will find variants that cannot be interpreted at the present time.

[ ] True

[ ] False

[ ] Don’t know/uncertain

22. Whole exome sequencing could find that you have a high risk for a disorder even if you do not have symptoms.

[ ] True

[ ] False

[ ] Don’t know/uncertain

23. Your whole exome sequencing may not find the cause of your disorder, even if it is genetic.

[ ] True

[ ] False

[ ] Don’t know/uncertain

24. The gene variants that whole exome sequencing can find today could have different meanings in the future as scientists learn more about how genes work.

[ ] True

[ ] False

[ ] Don’t know/uncertain

25. Whole exome sequencing will not find any variants in people who are healthy.

[ ] True

[ ] False

[ ] Don’t know/uncertain

Scoring:

1. Code all responses as having been answered correctly (=1) or incorrectly (=0). Items marked as “don’t know” are scored as incorrect (=0).

- Items that are correct if endorsed as “True”: Q1, Q2, Q3, Q5, Q6, Q7, Q10, Q11, Q15, Q19, Q20, Q21, Q22, Q23, Q24
- Items that are correct if endorsed as “False”: Q4, Q8, Q9, Q12, Q13, Q14, Q16, Q17, Q18, Q25

2. Sum scores across items to create a score from 0 to 25.

Protocol source: https://www.phenxtoolkit.org/protocols/view/310401