TEACHING THE GENOME GENERATION

Module Introduction

The Jackson Laboratory
Leading the search for tomorrow's cures

NIH National Institutes of Health
Turning Discovery Into Health
SEPA SCIENCE EDUCATION PARTNERSHIP AWARD
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Newborn Genomic Sequencing in the Future

Decades into the future, genomic sequencing will likely be a routine part of healthcare. Knowledge of gene variants associated with disease risk or outcomes can help individuals make decisions about not only their healthcare, but also their overall lifestyle.

Imagine it is the year 2120, whole genome sequencing for newborns has been the standard of care for 40 years. While still in the delivery room, a few of the newborn baby's cells are collected for DNA analysis. Parents who opt for this testing will receive an electronic *Newborn Genomics Report* (NGR) detailing the gene variants associated with a range of diseases or conditions that were detected. The report also includes lifetime risk for developing specific diseases, as well as a description of the lifestyle factors that could increase or decrease the risk for disease and, additionally, any genomic-based treatments developed to treat disease based on the presence of a specific gene variant. The NGR is updated periodically as new information becomes available and treatments are developed. The NGR is part of the baby's official medical record.

Cancer Genomics

Cancer is a genetic disease. Changes in DNA sequence in specific genes contribute to the onset and development of cancer. Cancer is a disease that results when cells in the body grow out of control and form a mass called a tumor. Only about 5–10% of cancer cases are **hereditary** and associated with an inherited gene variant, and therefore, most cancers are **sporadic** and arise from DNA sequence changes that occurs in body cells. Sequence changes are called mutations and mutations create gene variants. Changes in DNA sequence arise from errors in DNA replication or from environmental exposure such as to UV radiation or carcinogens like those found in cigarette smoke. Not all gene variants lead to cancer, but those in a set of important genes are more frequently associated with cancer.