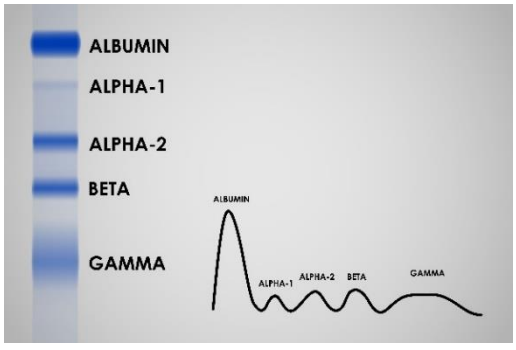
A Look inside the Lab: Gel Electrophoresis
Using Gel Electrophoresis to Test for Medical Conditions in Newborns

Your Aunt Michelle has had a baby! Your mom picks you up after school to go to the hospital to meet your new cousin. You are so excited that the elevator up to the maternity ward seems to take forever. After washing your hands, you find your aunt's room. She is smiling as you enter the room, holding a tiny bundle in her arms. She welcomes you over to meet the baby. You marvel at his impossibly tiny features and fingers! After a few minutes of visiting and taking some pictures, a nurse knocks on the door and enters the room. She asks your aunt for her permission to take a blood sample from the baby. You wonder aloud what the blood sample is for, and how could the nurse even get blood from such a tiny baby? The nurse tells you she will collect a few drops of the baby's blood from a heel-prick. As you are thinking about that, you hear her tell your aunt that it is important to get the blood for a newborn screening panel. The panel is a series of tests that help doctors test for certain medical conditions in an infant. Your aunt agrees to the blood draw, and as the nurse gets the sample, she explains that the blood tests in the panel screen for certain genetic, endocrine, and metabolic disorders. Examples of conditions screened for include sickle cell disease (SCD), congenital hypothyroidism (low thyroid hormone levels), and cystic fibrosis (CF). As the nurse leaves the room with the blood sample, you can't help but wonder: how can just a few drops of blood provide all this information?

Every year, millions of babies in the United States are screened for serious and life-threatening medical conditions. Newborn screening programs help medical professionals test for more than 35 different conditions. Screening programs differ by state; however, national guidelines are developed by the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC). The ACHDNC creates the Recommended Uniform Screening Panel (RUSP), a list of 35 primary and 26 secondary conditions.

In addition to the blood test that Aunt Michelle's baby underwent in the story, newborns are also given a hearing test and a pulse oximetry test before being released. The pulse oximetry test measures the amount of oxygen in the blood. It allows doctors to be sure the baby's lungs are working well.

The blood test is sent to a laboratory for analysis. Gel electrophoresis is one tool used by the lab to screen the sample for certain genetic disorders.



Gel electrophoresis is a laboratory technique used to separate DNA or proteins based on size in a gel medium. At the completion of a gel electrophoresis experiment, a visual banding pattern emerges on the gel. The banding patterns from the newborn sample and a control sample are compared to look for differences between the two samples. With this technique, researchers can quickly screen newborns for several genetic diseases caused by abnormalities in the structure of a person's hemoglobin. Genetic mutations in the DNA that code for hemoglobin impact its protein shape and electrical charge. These changes result in different band patterns when compared to the control sample of hemoglobin. If irregularities are detected in the newborn screening tests, the medical team can do additional tests or start treatment. Early diagnosis can help families successfully manage or treat conditions, improving the quality of life for their child.