

Angie Purvis, PhD at Baby Genes, uses VarSeq for Newborn Screening

It is standard practice for newborns to be screened for genetic diseases before leaving the hospital to assess for serious medical conditions that, left untreated, can cause lifelong health problems or in some cases, early death. What is not standard is which and how many conditions are screened for in each state. And in many states, the number of conditions screened simply isn't enough.

This means there are children all over the United States who are not being screened for serious genetic diseases they may have. By the time parents or physicians recognize the condition, serious, sometimes irreparable damage may already have been done. Baby Genes, a CLIA certified clinical laboratory formed in 2013 in Colorado, is trying to change this.

The mission at Baby Genes is to save lives by standardizing the approach to newborn screening (NBS) with a more comprehensive genetic sequencing-based test that screens for more conditions and provides more actionable information.

Using next-generation sequencing assays focused on NBS, Baby Genes is able to provide parents and physicians with screening results quickly. An earlier diagnosis means that a treatment plan can come together sooner and more innocent lives can be saved.

Dr. Angie Purvis, Associate Laboratory Director and Clinical Research Lead at Baby Genes, has been a part of the development of this test. She emphasized the importance of a more comprehensive test that provides better information. She states, "We are really looking at this from the perspective of, if this was my baby, what kind of information would I want?"

Of course, I want the best information available. And that's what we are trying to give people."

The current standard for newborn screening uses biochemical testing for various genetic, metabolic and rare diseases. Purvis explained how Baby Genes wants to revolutionize the testing to look at newborns earlier on for genetic diseases. The earlier these rare disorders are detected, the better the outcome for the affected newborns. The idea of harnessing the power of genetics is ideal in this process as it provides a faster, more comprehensive test over waiting for slower biochemical tests to be run, which can save more lives.

And this is Baby Genes' main emphasis; to take newborn screening out of the biochemical lab and put it into a new millennium of genomics. Using next-generation sequencing, Dr. Purvis and the rest of the team have created a genetic test that includes full exon sequencing of the genes associated with various conditions and diseases. Next-generation sequencing allows the team at Baby Genes

QUICK FACTS

Name: Dr. Angie Purvis

Title: Associate Laboratory Director and Clinical Research Lead

Company: Baby Genes

Location: Golden, CO

Service Provided: Newborn Screening for genetic diseases

Data: Next-generation sequencing assays

Golden Helix Software: VarSeq



to determine if a newborn has a mutation that is clinically linked to one of these genetic disorders. Golden Helix's VarSeq software has been instrumental in the development of this test. "Not only has it been valuable to speed up the process of sifting through large, complex genomic data sets, but also for initial quality control", said Dr. Purvis.



Dr. Angie Purvis

Many Hematology/Oncology diseases are caused by mutations in specific genes, so early on Purvis found she had a high interest level in genetic disorders.

Upon graduating, Dr. Purvis was hired at a diagnostic company that focused on proteomics work in oncology which, provided

her with an introduction into diagnostics. Purvis quickly developed a passion for diagnostics as she saw the potential and current innovation in the field.

When the opportunity arose for her to work at Baby Genes, Dr. Purvis was excited to put her diagnostic and pathology background to work improving something as fundamental as newborn screening.

Dr. Purvis completed her PhD in Biochemistry at Washington University School of Medicine at the Howard Hughes Medical Institute laboratory. A great deal of human genome sequencing work was being done at the time, and Purvis was able to participate in it through integrated programs.

Her exposure to the work on the Human Genome Project sparked an interest in the genetic field. She followed up with post doctoral work in Hematology/Oncology at Washington University.

The goal of a standardized NBS is to take a genetic approach and make it accessible. This requires two major factors be met to fulfill the Public Health Program's

demands; one is to be low cost, and two is to be quick. So far, Baby Genes has made significant progress in developing a faster test.

The team has shortened the turn-around time to only 48-96 hours from the time the sample arrives at the laboratory to the return of the genetic data back to physicians. "We've found VarSeq to be user-friendly and highly rational. It really simplifies our entire process and makes it go much faster." Now, Baby Genes is working on lowering the cost of this procedure to make it available to everyone.

Ultimately, a newborn screening panel that is comprehensive and standardized can save more lives. As the challenges of creating a cost-effective and a quick testing process are overcome, the accessibility of a genetic screening approach to more parents and physicians increases.

About Golden Helix

Golden Helix has been delivering industry leading bioinformatics solutions for the advancement of life science research and translational medicine for over 16 years. Our innovative technologies and analytic services empower scientists and healthcare professionals at all levels to derive meaning from the rapidly increasing volumes of genomic data produced from micro-arrays and DNA sequencing. With our solutions, hundreds of the world's top pharmaceutical, biotech, and academic research organizations are able to harness the full potential of genomics to identify the cause of disease, improve the efficacy and safety of drugs, develop genomic diagnostics, and advance the quest for personalized medicine. Golden Helix products and services have been cited in over 800 peer-reviewed publications.