September 04, 2013

NIH Awards up to $25M over Five Years to Teams Testing Genome Sequencing in Newborn Screening

By a GenomeWeb staff reporter

NEW YORK (GenomeWeb News) — Four research teams across the US have been awarded a total of $5 million in fiscal year 2013 under a new Genomic Sequencing and Newborn Screening Disorders program jointly funded by the Eunice Kennedy Shriver National Institute of Child Health and Human Development and the National Human Genome Research Institute.

NIH plans to fund the program at $25 million over five years.

The four pilot projects will examine whether sequencing newborns' genomes or exomes can provide useful medical information beyond what is already delivered by current newborn screening. They will study technical, clinical, and ethical aspects of genomics research in newborns, and its potential to improve newborn healthcare.

"We are at a point now where powerful new genome sequencing technologies are making
it faster and more affordable than ever to access genomic information about patients," said NHGRI Director Eric Green in a statement. "This initiative will help us better understand how we can appropriately use this information to improve health and prevent disease in infants and children."

In a teleconference today announcing the awards, NICHD Director Alan Guttmacher said that among the questions the program attempts to answer are how a baby's privacy will be protected and what the appropriate consent process will be; where newborn data will be stored and who will have access to it; what conditions should be included in a newborn sequencing test; what criteria will be used to determine a positive result; what the
follow-up for positive screening results should be; what doctors should tell parents about what was found; how much sequencing-based newborn screening will cost; and what its overall impact on healthcare and other spending will be.

The funding has been awarded to principal investigators Robert Green and Alan Beggs at Brigham and Women's Hospital and Boston Children's Hospital; Stephen Kingsmore at Children's Mercy Hospital; Robert Nussbaum at the University of California, San Francisco; and Cynthia Powell and Jonathan Berg at the University of North Carolina at Chapel Hill.

Green and Beggs' team will make genomic data available to parents and doctors throughout infancy and childhood and study the impact and usefulness of the results. They will also compare the response of parents with sick and healthy children, and the differences between receiving genomic results and conventional newborn screening results. In addition, they will develop a process for reporting results to doctors.

Kingsmore's group will look at the benefits and risks of using genomic sequencing in newborns in a neonatal intensive care unit, a strategy it has already explored in a small number of individuals. The researchers also aim to reduce the turnaround time to 50 hours to make the test comparable to existing newborn screening tests, and to study the parents' perception of receiving the results, and how it may change over time.

Nussbaum's project will focus on the potential of exome sequencing both for screening for disorders that are currently tested for and others that are not, and to assess the value of the additional information. It will also develop a participant protection framework for genomic sequencing during infancy and explore legal issues related to genome analysis as part of newborn screening.

Powell and Berg will lead a team that will sequence the exomes of healthy infants as well as those with known genetic conditions, and plan to identify the best ways to return results to doctors and parents. Their study, which will include multicultural families, will also develop a tool to help parents understand what the results mean and examine challenges that doctors may face in using the new technology.