Epigenetic immune cell diagnostic tool helps detect diseases in newborns not currently identified

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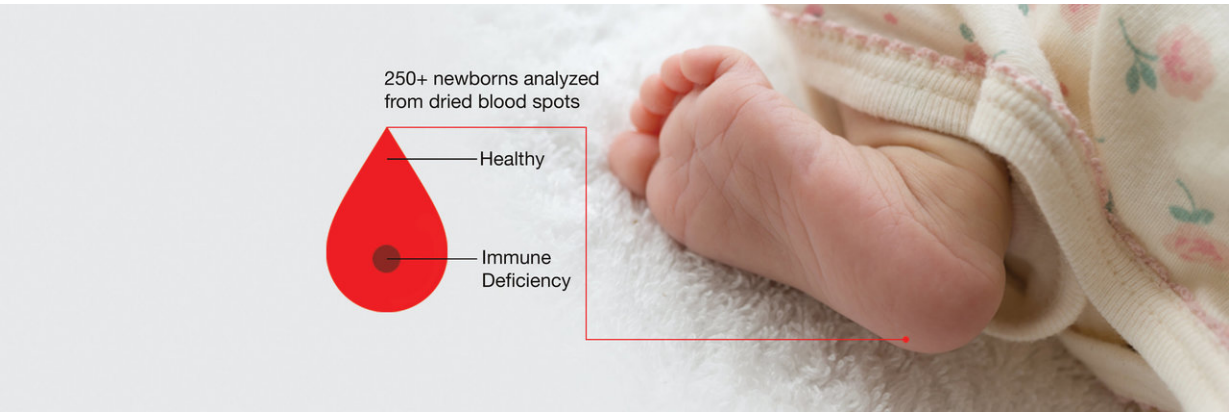

Novel epigenetic immune monitoring tool - Epiontis ID - shows promise in detecting severe immune diseases not currently identified in newborn screenings. Credit: Precision for Medicine

A novel diagnostic approach using epigenetic immune monitoring to screen newborns for inherited diseases could expand the number of life-threatening immune deficiencies identified in newborns. This method could enable treatment to begin soon after birth, promising improved disease outcomes and survival. The research is published in the August issue of the journal, *Science Translational Medicine*.

Newborns are routinely screened for inheritable diseases by analyzing

dried blood spots (DBS) from blood taken from a heel-prick. Among the more than 300 known primary immune deficiencies (PIDs), only Severe Combined Immunodeficiencies (SCID) are detected at birth with the current technology used to analyze the DBS. The novel alternative approach, using epigenetic quantitative real-time PCR (qPCR) assays was shown in the study to successfully detect a larger number of PIDs including, not only SCID, but others, such as X-linked agammaglobulinemia (XLA); Immune dysregulation, polyendocrinopathy, enteropathy, X-linked (IPEX) syndrome; and severe congenital neutropenia (SCN), severe diseases that often clinically manifest within months of birth.

"While further research is needed, these initial results are very encouraging as they provide early evidence that this epigenetic technology could eventually be a newborn screening method that would identify primary immune diseases that are currently very difficult to detect," said Rosa Bacchetta, M.D., one of the study investigators and an Associate Professor, Department of Pediatrics, Stanford School of Medicine. "Typically, children are not diagnosed until they're sick and are showing symptoms at which point there already is organ impairment and infections that make effective treatment much more difficult. It would be highly advantageous if we could identify these diseases at birth and begin treatment soon after birth, prior to the development of symptoms."

Improving Care for HIV Patients

The advantages that this epigenetic technology has in quantifying immune cells from dried blood was also shown to have additional positive implications for the treatment of HIV infections. Current methods for counting immune cells, such as those done with flow cytometry, require fresh or well-preserved blood which is especially challenging for monitoring HIV patients in limited-resource areas where

lab facilities may be far away from where a patient lives and is treated. The research found that immune cell counts could be effectively done using the epigenetic technology on a dried blood sample, which can then be communicated back to the patient's health practitioner for proper medication adjustments.

"This technology is simple and inexpensive to use, making it especially promising for improving the care of HIV patients where monitoring blood cell counts is critical to proper treatment but extremely difficult to do in parts of the developing world where storing and transporting liquid blood is not feasible, said Sven Olek, Ph.D., a study investigator and managing director of Epiontis GmbH, a German subsidiary of Precision for Medicine that developed Epiontis ID, the epigenetic qPCR used in the research. "Since a drop of the patient's blood can be placed on a piece of paper and mailed into a lab for analysis, this technology eliminates a patient's need to travel, often long distances, to a facility to have blood drawn. This increases the probability that the patient's immune cell counts will be more consistently monitored."

Study Details

Epigenetic quantitative real-time PCR (qPCR) assays were developed for analysis of human leukocyte subpopulations. Whole blood from 25 healthy donors, 97 HIV positive patients, as well as DBS from 250 healthy newborns and 24 newborns with primary immunodeficiencies were analyzed.

Concordance between flow cytometric and epigenetic data for neutrophils, B, NK, and CD3 positive T cells, CD8 positive T cells, CD4 positive T cells, and FOXP3 positive T regulatory cells was evaluated demonstrating substantial equivalence between epigenetic quantitative PCR analysis and flow cytometry. Epigenetic qPCR achieved both relative and absolute quantification.

Applied to DBS, epigenetic immune cell quantification was shown to identify newborns suffering from various primary immunodeficiencies.

More information: U. Baron el al., "Epigenetic immune cell counting in human blood samples for immunodiagnostics," *Science Translational Medicine* (2018). stm.sciencemag.org/lookup/doi/ … scitranslmed.aan3508

Provided by Precision for Medicine

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