EPIGENETIC IMMUNE CELL QUANTIFICATION - A NOVEL APPROACH TO EARLY DETECTION OF PRIMARY IMMUNODEFICIENCY FROM DRIED BLOOD SPOTS

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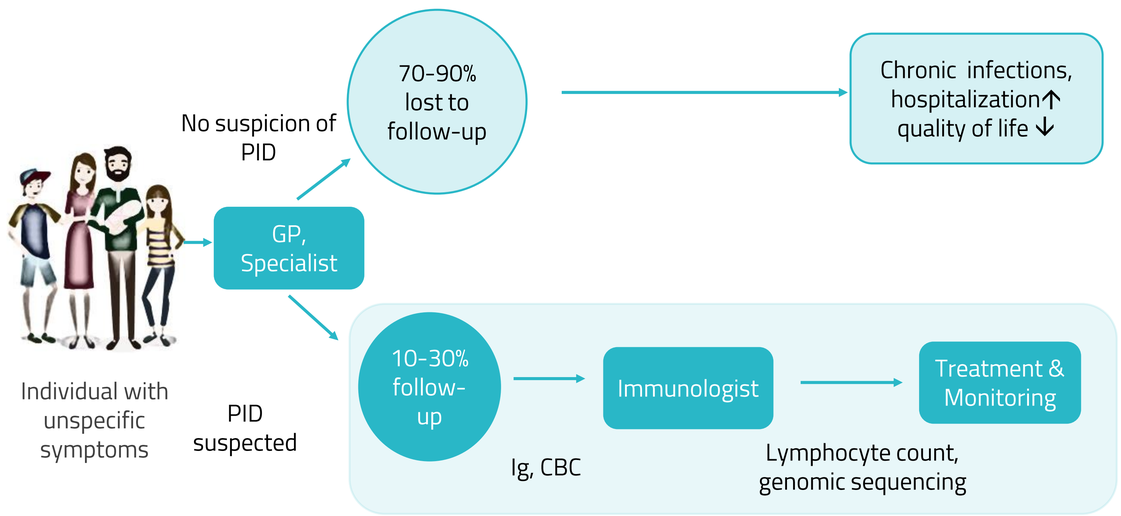
The Problem

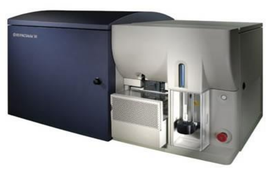
Many PID Patients are Diagnosed Too Late



Current Standard for Early Detection & Monitoring of PID Patients is Time Consuming and Invasive

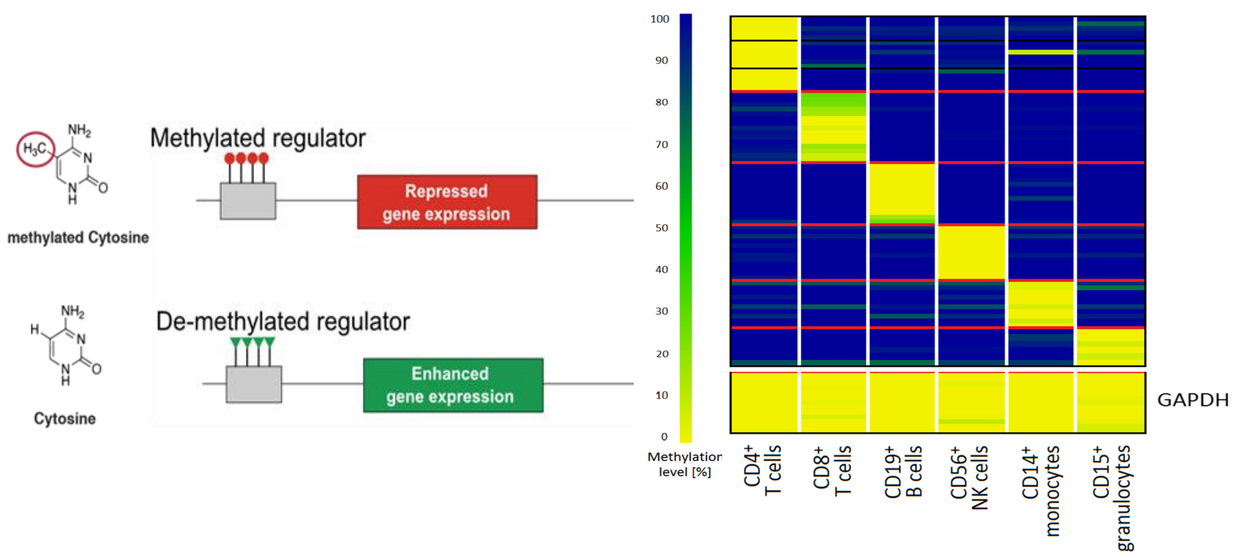
Flow Cytometry Requires a Fresh Blood Draw

24h

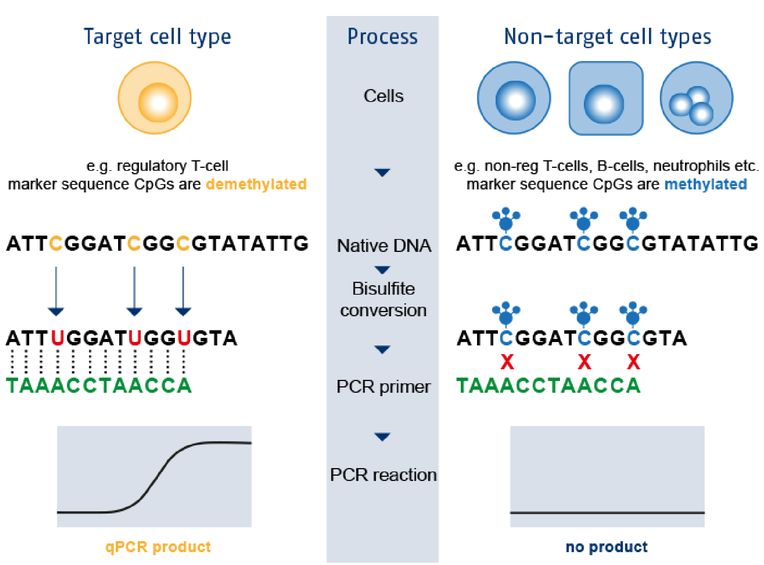


The Approach

Identification of Immune Cell-Specific DNA De-Methylation

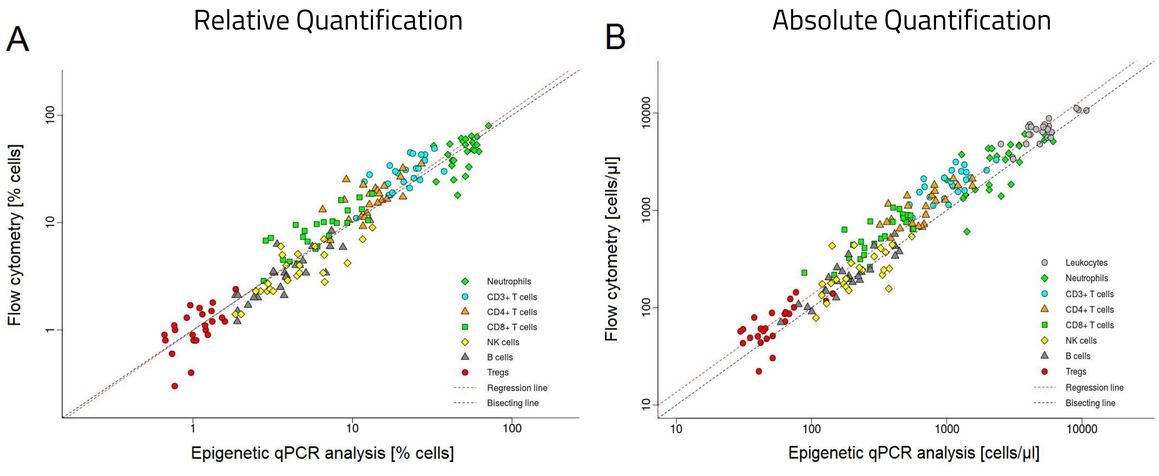


Epigenetic qPCR Allows Quantification of Immune Cells



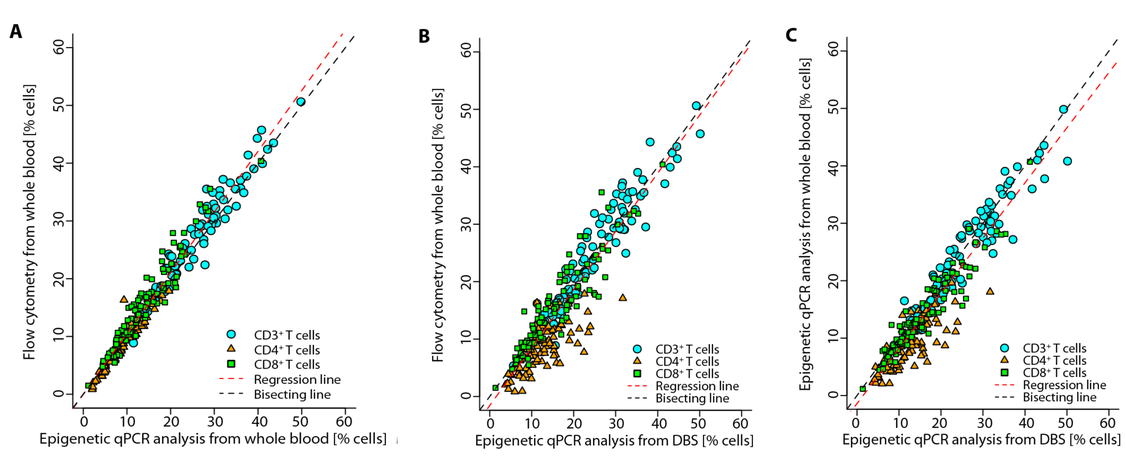
The Performance

Epigenetic Immune Cell Quantification is Equivalent to Flow Cytometry



Comparison of immune cell quantification by flow cytometry and epigenetic qPCR. Immune cells from blood samples of 25 independent donors were measured with flow cytometry (y axis) and epigenetic qPCR (x axis). (A) Relative immune cell counts are shown as percentage of total leukocytes. (B) Absolute immune cell counts are shown as cell number per microliter of whole blood. The regression line is depicted in red as computed from all data points, and the black line indicates the bisectrix.

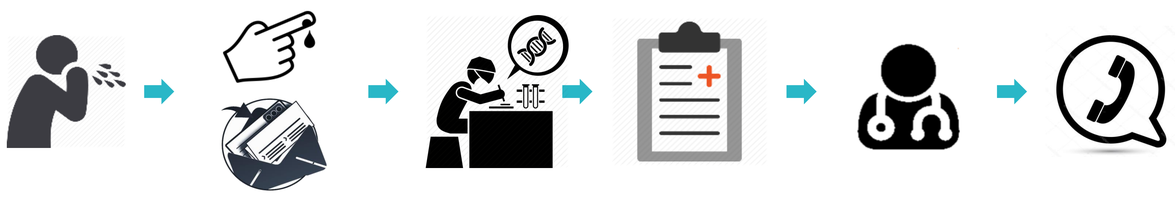
Epigenetic Immune Cell Quantification Works on Dried Blood Spots



Method comparison of T cell subsets in an HIV+ cohort. Samples were analyzed from 97 HIV+ subjects. Relative counts of CD3+, CD4+, and CD8+ T cells in percentage of total nucleated cells determined by (A) flow cytometry and epigenetic qPCR in liquid whole blood, (B) flow cytometry as in liquid blood and epigenetic qPCR from DBS, and (C) comparison of epigenetic qPCR from liquid blood and DBS. Data are presented as scatterplots. The regression line is depicted in red as computed from all data points, and the black line indicates the bisectrix.

The Solution

Epigenetic Immune Cell Quantification Allows Home Sampling and Easy Sample Logistics



Epigenetic Immune Cell Quantification Could Also Be Used for Expanded PID Newborn Screening

PIDs successfully identified in Newborn DBS samples using epigenetic immune cell quantification:

- Severe Combined Immunodeficiency (SCID)
- X-Linked Agammaglobulinemia (XLA)
- Immunodysregulation polyendocrinopathy enteropathy X-linked (IPEX) syndrome
- Severe Congenital Neutropenia (SCN)

Literature

Baron U, et al. Epigenetic immune cell counting in human blood samples for immunodiagnostics. Sci Transl Med. 2018 Aug 1;10(452)

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