

Catherine E. Cottrell, Elizabeth Varga, Susan Vear, Tara Lichtenberg, Kristen Leraas, Kathleen Schieffer, Katherine Miller, Vincent Magrini, Amy Wetzel, Daniel Koboldt, Ben Kelly, James Fitch, Patrick Brennan, Gregory Wheeler, Peter White, Ashita Dave-Wala, Devon Dishman, When your child needs a hospital, everything matters.[®] Lauren Shoemaker, Ruthann Pfau, Jonathan Finlay, Jeffrey Leonard, Diana Osorio, Mohamed S. AbdelBaki, Nicholas Yeager, Bhuvana Setty, Annie Drapeau, Nilay Shah, Selene Koo, Daniel Bouè, Christopher R. Pierson, Julie Gastier-Foster, Richard K. Wilson, Elaine R. Mardis.

Introduction

The Institute for Genomic Medicine developed a translational protocol to evaluate the genomic landscape of cancer and hematologic disease in a focused, patient-specific manner.

Medical Provide

Completes

Nomination Form

in REDCap

Nomination Form

Nomination Accepted

Patient and Family

Approached for

Consent

Consented

Germline Samples Obtained from

Patient and Family

and

Disease Involved

Specimen Acquired Explanation Given

to Provider

Family/Patient

Undecided or

Declined

Nomination

Declined

Not

Consented

Analytes Extracted

for All Samples

Received

Parental Samples May Be Used for Confirmation

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Central to the protocol is a multifunctional REDCap database designed to streamline workflows including enrollment, consenting, specimen selection, assay methodology, and documentation of results/ LAN S patient metadata. Patient enrollment begins following completion of a provider survey by a nominating \mathbf{V} clinician. Once reviewed by the study team, an eligible patient/family is approached for consent. Following enrollment, genomic profiling methodologies are employed.

Studies of precision medicine can:

- Refine diagnosis
- Inform prognosis
- Determine eligibility for clinical trials & targeted therapeutics
- Detect germline disease predisposition

Current Status



Design and Implementation of a Comprehensive Genomic Profiling Protocol for Rare and Refractory Pediatric Cancer and Hematologic Disease

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Figure 5. Initial REDCap [™] provider survey
Figure 5. Initial REDCap TM provider survey



I REDCap[™] workflow

Cap is being used to manage protocol flows and data both internal to IGM by other key project collaborators n NCH (e.g. heme/onc and pathology). currently the central hub for data ction and enables communication it patient status within the workflow.





In this patient, IGM tumor profiling refined diagnosis, ultimately allowing for tailored treatment and improved management.

Overall Study Results

In total, 53 patients have been nominated for enrollment onto the protocol with 47 (87%) deemed appropriate for inclusion. To date, 36 patients have consented, and of those IGM has sequenced 33 patient cases. Resulting analyses have allowed for refinement of diagnosis, improved understanding of prognosis, implementation of targeted therapies, as well as counseling and follow-up in these children and families.