Evelyn

Using rWGS, Evelyn was diagnosed very early on with KAT6b-OHDO syndrome. Her diagnoses allowed her physicians to research all symptoms and conditions related to her syndrome. They used this research as a guide to provide Evelyn with the best care plan suited to her needs. Without this genome project, Evelyn would not be home recovering and living her best life.

Kayde

Kayde was born with low muscle tone and was very floppy as an infant. Thanks to rWGS being available in the NICU, we were able to find out about his genetic difference. Since then, he has been set up with all the right specialists and therapies to help make him strong. This test could find more babies similar to Kayde and help provide answers for the best possible care. All babies and families should have access to quick answers and any early prevention.

Hazel

When Hazel was born at Helen DeVos Children’s Hospital, she had a dark purple hue to her skin. She spent 13 days in the NICU with feeding difficulties and transient tachypnea, a breathing disorder sometimes seen in newborns. After a few tests, it was discovered that Hazel had several heart defects and needed further evaluation to figure out the root cause of her defects. Thanks to a rWGS made available through Project Baby Deer, in a matter of days, results showed that Hazel had Noonan’s Syndrome. Hazel’s doctors were able to determine a successful treatment plan and we were able to find peace of mind, finally knowing what was wrong. Today, Hazel is a happy, content little girl with a smile that lights up the room.

Adrian

We felt so lucky to get such quick answers for Adrian’s genetic condition and felt very supported by the genetics department from afar. Because of rWGS, we were able to make a plan early on to best support her and get answers about our future fertility.

*Each story is told by the parents of each child.
INCLUSION CRITERIA

› Inpatient at a MI project site
› <18 years old
› Meets one of the following criteria:
  • Admitted to a critical care unit OR
  • Admitted to another high-acuity in-patient unit and is suspected of having a genetic diagnosis
› Meets one of the following criteria:
  • Within 1 week of admission OR
  • Within 1 week of development of an abnormal response to standard therapy for an underlying condition

EXCLUSION CRITERIA

Patients whose clinical course is entirely explained by:
› Infection or sepsis with normal response to therapy
› Isolated prematurity
› Isolated unconjugated hyperbilirubinemia
› Hypoxic ischemic encephalopathy with clear precipitating event
› Previously confirmed genetic diagnosis that explains the clinical condition (e.g. have a positive genetic test)
› Isolated transient neonatal tachypnea
› Trauma
› Meconium aspiration

PILOT SITE | # OF CHILDREN WHO RECEIVED RWGS | CHILDREN DIAGNOSED (DIAGNOSTIC RATE) | CHILDREN WHOSE CARE WAS CHANGED (CHANGE IN MANAGEMENT RATE)
--- | --- | --- | ---
Beaumont Health – Dearborn | 3 | 2 (67%) | 2 (67%)
Beaumont Health – Royal Oak | 6 | 0 (0%) | 0 (0%)
Beaumont Health – Troy | 1 | 1 (100%) | 0 (0%)
Bronson Methodist Hospital | 15 | 6 (40%) | 7 (47%)
Children’s Hospital of Michigan | 10 | 4 (40%) | 1 (10%)
Helen DeVos Children’s Hospital | 45 | 18 (40%) | 11 (25%)
Sparrow Hospital | 9 | 4 (44%) | 3 (33%)

TOTALS | 89 | 35 (39%) | 24 (27%)

PRELIMINARY RESULTS OF RAPID WHOLE GENOME SEQUENCING (rWGS):
› 39% of rapid genomes resulted in a diagnosis confirmed by the treating physician
› 27% of children had changes to their care as a result of rWGS
› Oldest PBD patient: 17 years of age
› Youngest PBD patient: 1 day old
› Race: White - 72%; Black or African-American - 9%; Asian - 3%; Other - 9%; Unknown - 7%
› Ethnicity: Non-Hispanic - 43%; Hispanic - 11%; Other - 7%; Unknown - 39%
› Sex: Female - 36%; Male - 64%

THESE CHANGES LED TO:
› Avoided between 95 and 214 inpatient hospital days
› Multiple avoided surgeries and procedures (including lung biopsy, tracheostomy, muscle biopsy, and skin biopsy)
› Appropriate medications prescribed based on genetic diagnosis
› Initiation of a heart transplant

PROJECT SAVINGS*: NET BENEFIT PER PATIENT DUE TO REDUCTION IN HOSPITALS DAYS AND MAJOR PROCEDURES

$2,842 per patient for a total of $252,938

*as of 11/30/21

For more information, contact: keystone@mha.org
# CASES WITH ECONOMIC SAVINGS

## SITE 1

| Case 1 | Avoided skin biopsy |

## SITE 2

| Case 6 | 7-42 hospital days avoided |
| Case 11 | 7 hospital days avoided |
| Case 14 | Avoided lung biopsy, Avoided transfer to another hospital for lung biopsy |

## SITE 3

| Case 1 | Stopped medication, 2 hospital days avoided |
| Case 4 | 2 hospital days avoided |
| Case 5 | Cancelled interventional radiology, cerebral angiogram; added medication |
| Case 10 | Avoided muscle biopsy |
| Case 18 | 14-28 hospital days avoided |
| Case 19 | 14-28 hospital days avoided |

## SITE 4

| Case 2 | Tracheostomy avoided, 28-84 hospital days avoided |
| Case 3 | 7 hospital days avoided |
| Case 4 | 14 hospital days avoided |

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