Nicklaus Children's Hospital | Project Baby Manatee

Background

This program has provided diagnoses of rare genetic diseases in critically ill children who would otherwise go on a "diagnostic odyssey".

Timely diagnosis can help avoid unnecessary tests and procedures, possibly preventing further irreversible harm and unnecessary cost.

Inclusion: critically ill children from the NICU, PICU, CICU with poorly defined diseases of undetermined, possibly genetic causes.

The ultimate vision is to make this technology available to the ICU patient population during their hospital stay and have it be covered by payors.

Key Results

- Completed rWGS on 82 children and families
- Provided diagnoses for 40 children and families (49%)
- Led to change in care for 36 patients (44%)
- Diagnosed 58 rare genetic conditions
- Offered referrals for mental health counseling to 18 parents experiencing elevated levels of depression
- Saved $16,300,862 ($198,791 per patient)
- Diagnostic odyssey shortened from 250 to 96 days on average

Changes in Clinical Management

(based on a positive or negative diagnosis)

1 cardiac surgery avoided

8 children started diet therapy for a metabolic condition

9 patients had their medication adjusted

1 lung transplant avoided

1 fundoplication surgery performed

13 patients had lab tests or procedures performed based on the genetic result

9 patients had follow-up with specialists

> I would love it if…the insurance companies would support this because there are truly kids who need this and the parents can't pay for this. —Nicklaus Children's Hospital parent

Mental Health Assessment

- 28% of caregivers scored above the cutoff for depression
- 43% scored above the cutoff for anxiety during patient enrollment.
- These parents were offered referrals for assistance
- Caregivers expressed gratitude to the project because it helped them to take steps to address their mental health

Case Study

- 15-year-old boy with previous genetic diagnosis of 13q33 microdeletion
- Patient had seizures with increasing frequency that were unrelated to his previous genetic diagnosis and was admitted to the PICU
- rWGS® diagnosed pathogenic variant in CARKD gene, explained his other symptoms
- At time of enrollment, his mother reported symptoms of severe depression and suicidal ideation
- Mother was referred to a mental health specialist
- Mother thanked the team for assistance that she received, as she recognized that taking care of her mental health would have a direct impact on her son's health and well-being
- The rWGS result led to the mother's understanding that her son's condition was caused by a genetic disease, not by her, and this helped dispel the mother's guilt.

Impact of Rapid Whole Genome Sequencing

The introduction of genome sequencing in some of the most vulnerable of children had a profound impact on three key dimensions of healthcare

1. Beneficial changes in clinical management by providing timely diagnostic and prognostic information.
2. Improved healthcare experience for families by providing mental health support for parents, reducing uncertainty and empowering them to make informed medical decisions.
3. Lowering the cost of care by reducing unnecessary tests, procedures, and time spent in the hospital.

Return of Results

20%
of patients received UrWGS

Average turnaround time for ultra-rapid diagnoses:

2.5 days to preliminary report

6 days to final report

1 day for physician to return results to the family

80%
of patients received rWGS

Average turnaround time for rapid diagnoses:

4 days to preliminary report

8 days to final report

1.4 days for physician to return results to the family

Length of Stay

48
DAYS
On average, length of stay was 48 days for the 50 patients

2-297
Range was 2-297 days

1
WEEK
rWGS has shortened LOS for one patient by a week