Coverage of Rapid Whole Genome Sequencing (rWGS) for Critically Ill Children

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The journey to diagnosis for a rare disease patient is arduous.

- Averages almost 5 years and evaluation by over 7 specialists.\(^1\,^2\)

Rapid WGS is a clinical diagnostic tool used in high acuity neonatal and pediatric settings when time to result is critically important to manage and treat a patient.

- rWGS investigates genetic changes that are causing a child’s health condition \textit{right now} (not looking for risks of other disease that may occur later in life) and returns results to providers in a matter of days.

Growing evidence base demonstrates clinical and cost effectiveness of rapid whole genome sequencing in high acuity care settings.

\begin{itemize}
  \item \textbf{43\%}\(^3\) Diagnostic Yield
  \item \textbf{40\%}\(^4\) Diagnostic Yield
  \item \textbf{39\%}\(^5\) Diagnostic Yield
  \item \textbf{31\%}\(^3\) Change in Medical Care
  \item \textbf{38\%}\(^4\) Change in Medical Care
  \item \textbf{27\%}\(^5\) Change in Medical Care
\end{itemize}

Across the country, States are taking action to improve access to rWGS for critically ill children.
Pursuit of Pediatric precision medicine: coverage of rapid whole genome sequencing (rWGS)

Significant value for both patients and Medicaid if the program provides coverage and reimbursement of rWGS in high acuity clinical settings.

- Initial focus should be on critically ill children (high acuity ICU setting & disease of unknown etiology).
- MI saw 47 claims in first full year of implementation.

**Benefits of rWGS**

- Fewer days in the hospital: 513
- Fewer major surgeries: 11
- Fewer invasive diagnostic tests: 16
- $2.55 M Net savings realized from CA rWGS pilot.
- $2.88 M Net savings realized from FL rWGS pilot.

**Outcomes**

- rWGS has consistently demonstrated aggregate and per person cost savings as compared to standard of care.
- Rapid WGS yields twice as many diagnoses than usual care (including other molecular testing) in the ICU.
  - An estimated 15% of diagnoses made with WGS would have been missed using whole exome sequencing.

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3 3 4
513 11 16

Outcomes of using rWGS to diagnosis critically-ill children in CA ICUs
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Clinical Impact of rWGS

One day, Hudson was a healthy 13-month-old headed for fun at daycare. The next day, he was headed for admission to Rady Children's Hospital with vocal cords so swollen, he could hardly breathe.

Using rWGS, he was diagnosed with a riboflavin transporter deficiency and given a simple vitamin treatment.

Sebastiana was not yet 24-hours old when she was admitted to the Rady Children’s Hospital neonatal intensive care unit. She was unable to eat due to seizures and was rapidly failing. She was two days old when whole genome sequencing was ordered.

Diagnosed with KCNQ2 Ohtahara Syndrome, the youngest child to be diagnosed with this condition, and given an appropriate anti-seizure medication for her condition.

Economic impact of early diagnosis and intervention

Sebastiana: diagnosed on day of life 6, given an appropriate ASM and spent only 18 days in the hospital.

Comparatively, a patient seen 2 years earlier with same diagnosis: Epilepsy panel diagnoses him on day of life 42 and spent 59 days in the hospital.

Difference in hospital costs: $181,481
Trajectory of care for children with rare and undiagnosed diseases

- Hospitalization for critically ill child with suspected genetic condition
- Initial diagnostic testing for suspected genetic condition
- Follow-up diagnostic testing (if necessary)
Trajectory of care for children with rare and undiagnosed diseases

Hospitalization → Follow-up diagnostic testing

Standard of care testing

Hospitalization → Follow-up diagnostic testing

WGS

10 YEARS
Expected number of children who will receive rapid whole genome sequencing (rWGS) in Connecticut

Assume 127 children with Medicaid get rWGS in CT state (annually) (early adopters)

50% rapid testing at Rady Children’s Institute for Genomic Medicine
63 children tested at Rady
Expected cost per patient: $12,297

50% testing at other labs in US
63 children tested at other labs
Expected cost per patient: $9,746

Expected average cost of testing: $11,022 per case
## Inpatient costs: initial diagnostic testing costs

<table>
<thead>
<tr>
<th>Costs of the diagnostic pathway</th>
<th>Current standard of care testing</th>
<th>Rapid Whole Genome Sequencing (rWGS)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Diagnostic yield</td>
<td>9%</td>
<td>50%</td>
</tr>
<tr>
<td>Initial testing costs</td>
<td>$2,466</td>
<td>$11,022</td>
</tr>
</tbody>
</table>

**Current Medicaid coverage**

- Part of the all-inclusive DRG payment for inpatient services
- Not covered
Future scenario: What if we had a Medicaid DRG carve-out payment for rWGS?

<table>
<thead>
<tr>
<th>Standard of care testing</th>
<th>Rapid Whole Genome Sequencing (rWGS)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Incremental Medicaid reimbursement for testing (above traditional DRG payment)</td>
<td>+$0</td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
</tbody>
</table>
## Index Hospitalization Expenditures and Savings - rWGS

<table>
<thead>
<tr>
<th>Expenditures and Savings by rWGS (Estimated Annual)</th>
<th>Rapid Whole Genome Sequencing (rWGS)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Testing during inpatient admission (Per Patient)</strong></td>
<td></td>
</tr>
<tr>
<td>Supplemental Payment (Annual)</td>
<td>$11,022</td>
</tr>
<tr>
<td>Cost Savings Realized on Avoided Payments by Medicaid (Savings associated with care avoided due to rapid diagnosis and intervention)</td>
<td>($6,015)</td>
</tr>
<tr>
<td>Upfront Budget Impact (Annual)</td>
<td>$5,007</td>
</tr>
</tbody>
</table>

Total Annual Net Expenditure for Upfront Testing Cost = $5,007 * 127 = $635,889
## Index Hospitalization Expenditures and Savings - rWGS

<table>
<thead>
<tr>
<th>Expenditures and Savings by rWGS (Estimated Annual)</th>
<th>CT Medicaid</th>
<th>Federal CMS</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Assumptions, per patient:</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Upfront Expenditure = $11,022 per test</td>
<td></td>
<td></td>
</tr>
<tr>
<td>FMAP = 50%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Supplemental Payment (Annual)</td>
<td>$5,511.00</td>
<td>$5,511.00</td>
</tr>
<tr>
<td>Cost Savings Realized on Avoided Payments by Medicaid (Savings associated with care avoided due to rapid diagnosis and intervention)</td>
<td>($3,007.50)</td>
<td>($3,007.50)</td>
</tr>
<tr>
<td><strong>Budget Impact (Annual)</strong></td>
<td>$2,503.50</td>
<td>$2,503.50</td>
</tr>
</tbody>
</table>

CT Medicaid Portion of Upfront Testing Cost = $2,503.50 * 127 = $317,945
Trajectory of diagnostic testing: 10- years

Hospitalization → Follow-up diagnostic testing

Follow-up diagnostic testing → Standard of care testing

Hospitalization → Follow-up diagnostic testing

10 YEARS
## Downstream diagnostic testing costs over 10-years

<table>
<thead>
<tr>
<th>10-year costs of the diagnostic pathway</th>
<th>Downstream testing costs over 10 years</th>
</tr>
</thead>
<tbody>
<tr>
<td>Downstream diagnostic testing costs following standard of care testing</td>
<td>$4596 per child</td>
</tr>
<tr>
<td>Downstream diagnostic testing costs following rWGS</td>
<td>$186 per child</td>
</tr>
<tr>
<td>Downstream cost savings associated with rWGS</td>
<td>($4410) per child</td>
</tr>
<tr>
<td>Downstream total cost savings associated with rWGS (for 127 children)</td>
<td>($560,070) total</td>
</tr>
</tbody>
</table>

Notes: (1) costs reflect total costs of testing over 10-years, and not Medicaid-specific costs. (2) only diagnostic testing costs are included in estimate, not associated hospitalizations and provider visit costs.
## Summary

Downstream cost savings associated with rWGS over 10 years (total for 127 children), due to diagnostic tests avoided. ($560,070)

<table>
<thead>
<tr>
<th>Testing and services</th>
<th>Medicaid expenditures and savings (per child)</th>
<th>Medicaid budget impact – all funds (for 127 children)</th>
<th>Medicaid budget impact – CT state funds (for 127 children)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Upfront testing costs</td>
<td>$11,022</td>
<td>$1,399,794</td>
<td>$699,897</td>
</tr>
<tr>
<td>Cost savings in index hospitalization</td>
<td>($6,015)</td>
<td>($763,905)</td>
<td>($381,953)</td>
</tr>
<tr>
<td><strong>Net upfront budget impact</strong></td>
<td><strong>$5,007</strong></td>
<td><strong>$635,889</strong></td>
<td><strong>$317,945</strong></td>
</tr>
</tbody>
</table>
Questions & Discussion
Cited Sources


