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

Tara Lavelle, PhD
Assistant Professor
Tufts Medical Center
Boston, MA

Madison Arenchild, MPH
Manager, Policy and Payer Engagement
Rady Children's Institute for Genomic Medicine
San Diego, CA

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Pursuit of Pediatric precision medicine: coverage of rapid whole genome sequencing (rWGS)

TuftsMedicine
ALEXION AstraZeneca Rare Disease
Rady Children's Institute Genomic Medicine

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 *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.

43%[3]
Diagnostic Yield

31%[3]
Change in Medical Care

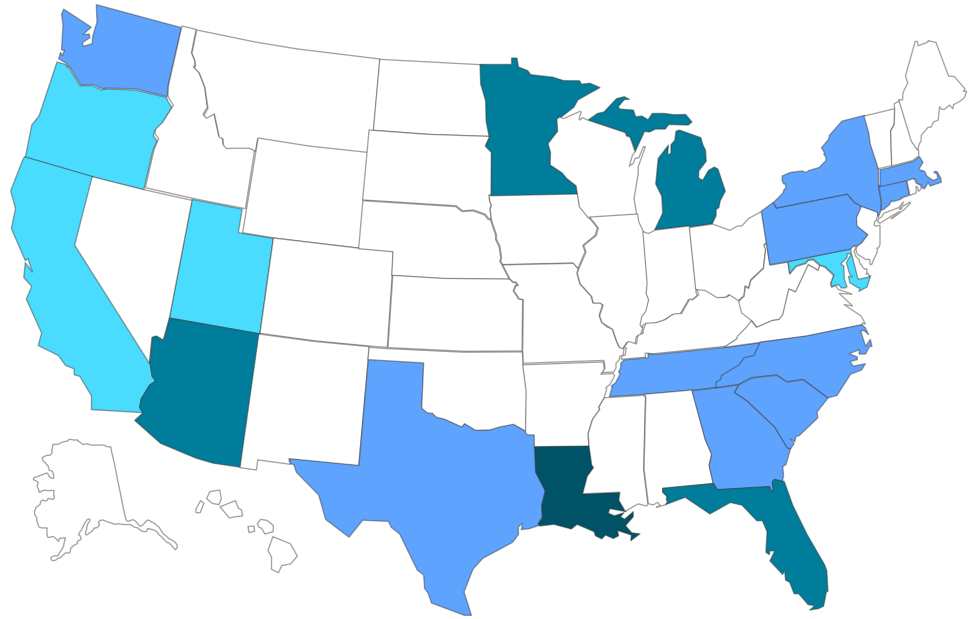
40%[4]
Diagnostic Yield

38%[4]
Change in Medical Care

39%[5]
Diagnostic Yield

27%[5]
Change in Medical Care

Across the country, States are taking action to improve access to rWGS for critically ill children.



LEGEND

- Approved policy
- Separate payment by Medicaid
- Private and public plan requirement
- Policy under review or legislation in progress

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.

$2.55 M [3]

Net savings realized from CA rWGS pilot.

$2.88 M [4]

Net savings realized from FL rWGS pilot.

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 [6]

- An estimated 15% of diagnoses made with WGS would have been missed using whole exome sequencing. [7]

rWGS has immediate impacts on care [3]

513	Fewer days in the hospital	**11**	Fewer major surgeries	**16**	Fewer invasive diagnostic tests	Outcomes of using rWGS to diagnosis critically-ill children in CA ICUs

Clinical Impact of rWGS

25% of patients experience change in clinical management due to results

97% of Families found rWGS helpful regardless of result

15-20% have resulting change in health outcome

77% of Surveyed Clinicians Found rWGS useful

$14K Per patient cost reduction on inpatient care when rWGS is provided as a first-tier test.

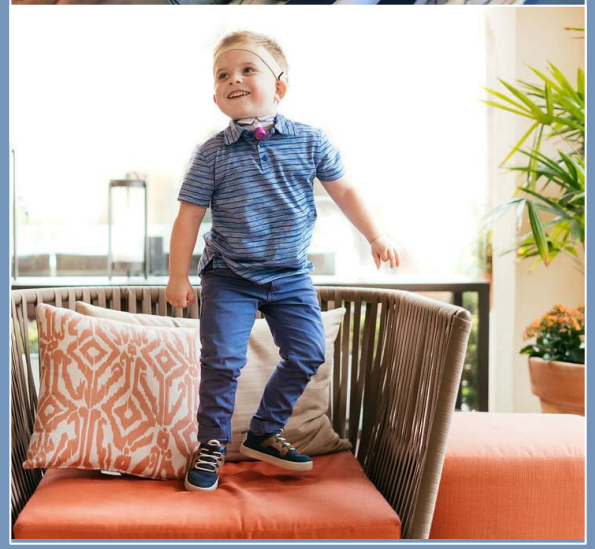
<u>Economic impact of early diagnosis and intervention</u>

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

Hudson



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.

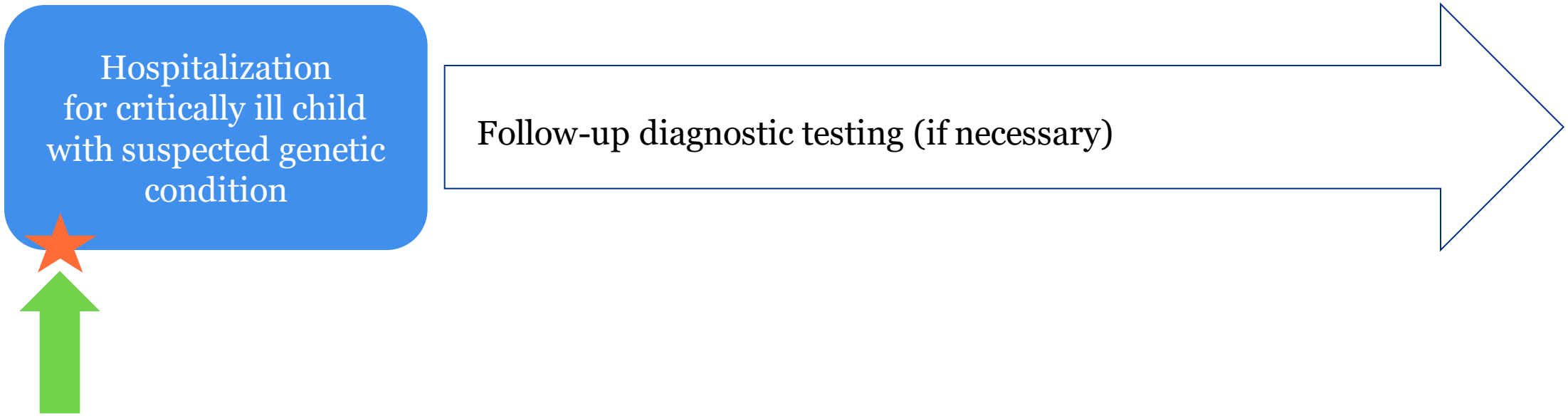
Using rWGS, he was diagnosed with a riboflavin transporter deficiency and given a simple vitamin treatment.

Sebastiana

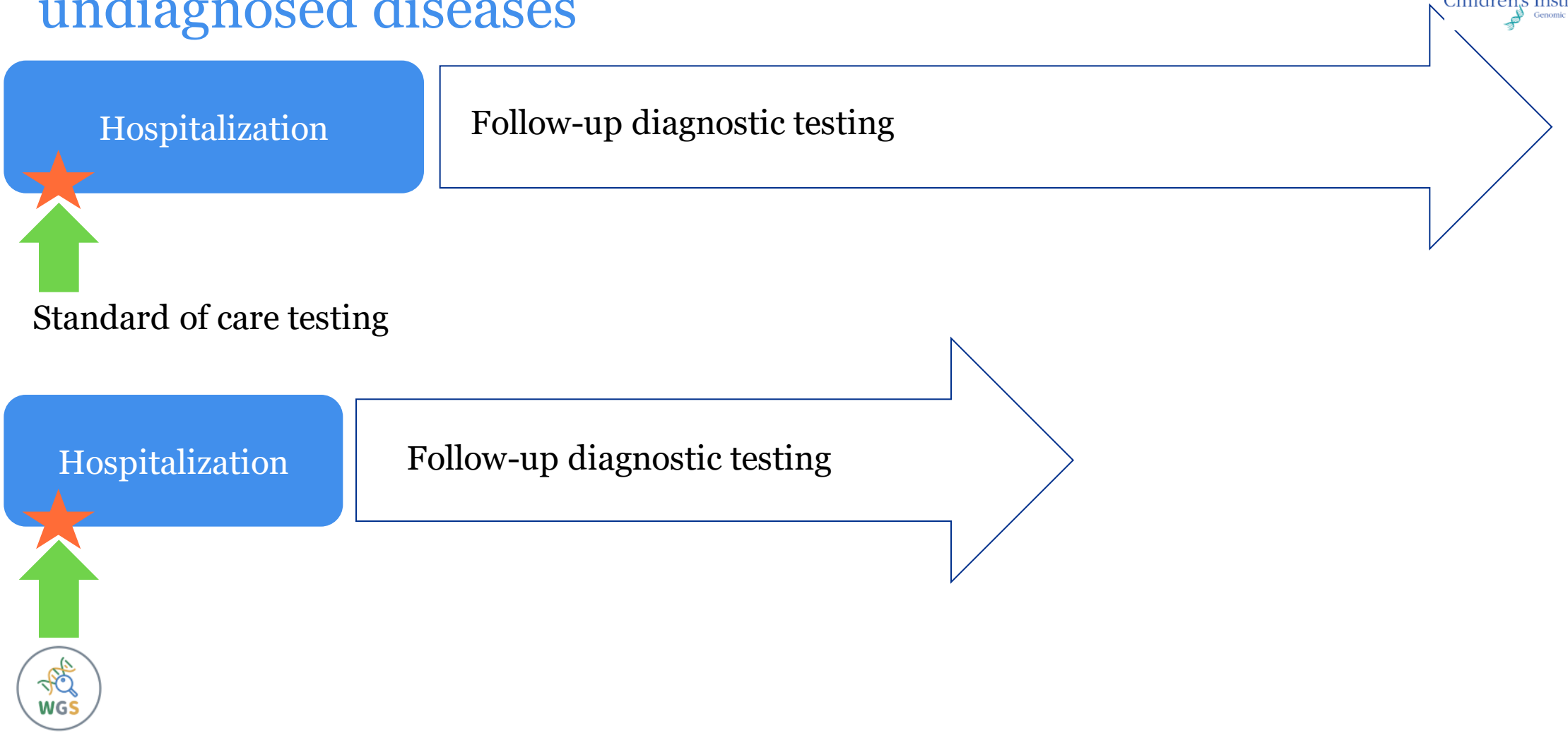
Sebastiana was not yet 24-hours old when she was admitted to the Rady Children's Hospital neonatal intensive care unit. She was unable to eat due to seizures and was rapidly failing. She was two days old when whole genome sequencing was ordered.

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

Trajectory of care for children with rare and undiagnosed diseases



Trajectory of care for children with rare and undiagnosed diseases



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

50% testing at other labs in US

63 children tested at Rady

Expected cost per patient:

$12,297

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

Costs of the diagnostic pathway	Current standard of care testing	Rapid Whole Genome Sequencing (rWGS)
Diagnostic yield	9%	50%
Initial testing costs	$2,466	$11,022
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?

	Standard of care testing	Rapid Whole Genome Sequencing (rWGS)
Incremental Medicaid reimbursement for testing (above traditional DRG payment)	+$0	+$11,022 Supplemental payment to the DRG for inpatient rWGS.

Index Hospitalization Expenditures and Savings - rWGS

Expenditures and Savings by rWGS (Estimated Annual)	Rapid Whole Genome Sequencing (rWGS)
Testing during inpatient admission (Per Patient)	
Supplemental Payment (Annual)	$11,022
Cost Savings Realized on Avoided Payments by Medicaid (Savings associated with care avoided due to rapid diagnosis and intervention)[8]	($6,015)
Upfront Budget Impact (Annual)	$5,007

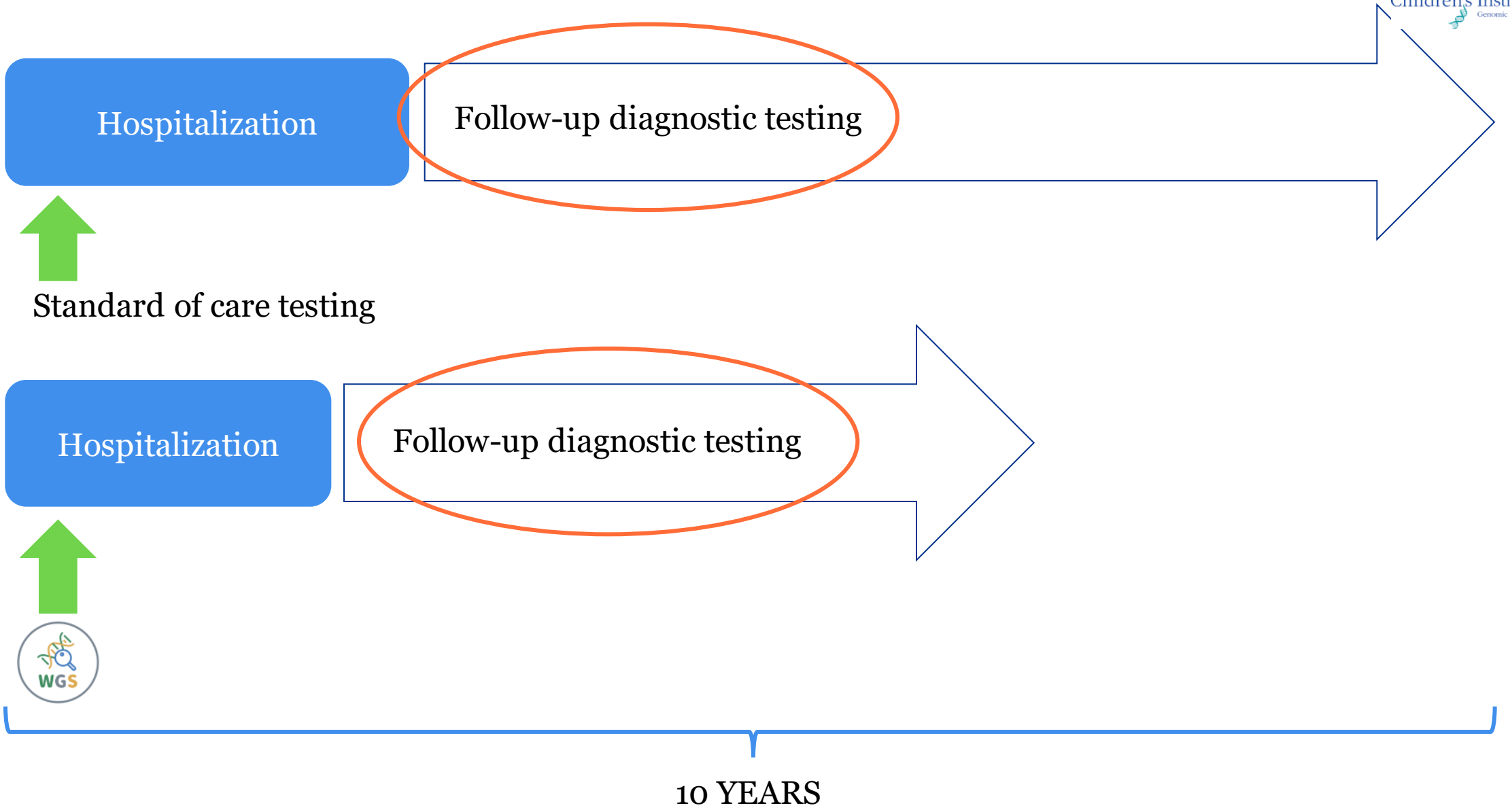
Total Annual Net Expenditure for Upfront Testing Cost = $5,007*127 = $635,889

Index Hospitalization Expenditures and Savings - rWGS

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

CT Medicaid Portion of Upfront Testing Cost = $2,503.50 * 127 = $317,945

Trajectory of diagnostic testing: 10- years



Downstream diagnostic testing costs over 10-years

10-year costs of the diagnostic pathway	Downstream testing costs over 10 years
Downstream diagnostic testing costs following standard of care testing	$4596 per child
Downstream diagnostic testing costs following rWGS	$186 per child
Downstream cost savings associated with rWGS	($4410) per child
Downstream total cost savings associated with rWGS (for 127 children)	($560,070) total

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

Testing and services	Medicaid expenditures and savings (per child)	Medicaid budget impact – all funds (for 127 children)	Medicaid budget impact –CT state funds (for 127 children)
Upfront testing costs	$11,022	$1,399,794	$699,897
Cost savings in index hospitalization	($6,015)	($763,905)	($381,953)
Net upfront budget impact	**$5,007**	**$635,889**	**$317,945**
Downstream cost savings associated with rWGS over 10 years (total for 127 children), due to diagnostic tests avoided.			($560,070)

Questions & Discussion

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