Children's MINNESOTA

The Kid Experts™

March 26, 2024
House Health Finance and Policy Committee

Dear Chair Liebling and Committee Members,

As a clinical geneticist and Medical Director of Genomic Medicine at Children's Minnesota, I'm writing in support of HF3330 which would require coverage for rapid whole genome sequencing (rWGS) for acutely ill children. rWGS is a now well-established diagnostic molecular study that detects genetic disorders in critically ill children with unusual clinical presentations who may benefit from prompt treatment and relevant evaluations.

rWGS has quickly become the clinical standard of care for inpatients suspected to have a genetic disorder, given its higher diagnostic yield versus other, more limited clinical tests, fast turnaround time (up to about 14 days), and growing medical literature supporting its clinical utility. Unfortunately, payer coverage has remained behind, effectively limiting access to testing, and potentially contributing to increased morbidity and mortality of this high-risk inpatient population.

rWGS is currently being used in our neonatal intensive care unit, pediatric intensive care unit, and cardiovascular critical care unit. Pre- and postnatal genetic counseling is offered to all families. Of those patients who undergo rWGS, about one third are found to have a primary genetic diagnosis which may help provide a specific prognosis, initiate appropriate interventions, and potentially reduce length of stay and admission costs. It is important to note that a negative result is also helpful, as it may allow ongoing care to continue without further invasive clinical interventions which are sometimes initiated if a genetic diagnosis is suspected. It has been estimated that around 20% of total infant deaths in the United States are due to an early onset disorder with a genetic basis. More significantly, about 10-25% of children admitted to a neonatal intensive care unit may have a monogenic disorder, which may be missed due to the lack of defining features in early age and lack of insurance coverage for inpatient genetic studies. Our neonatology program takes care of more than 2300 babies each year. Accordingly, up to 575 of those neonates would benefit from rWGS. Currently, only two percent of candidate patients admitted to the neonatal intensive care unit can access rWGS primarily due to limitations in insurance coverage. This lack of access leads to delays in decision-making from the relevant inpatient teams and families when approached about the opportunity to participate in rWGS.

Please consider approving coverage for rWGS as we strive to offer care to critically ill children in Minnesota.

Sincerely,

Marcelo Vargas, MD
Medical Director
Genomic Medicine
Children's Minnesota

MINNESOTA
RARE DISEASE ADVISORY COUNCIL

March 15, 2024

House Health Finance and Policy Committee
Chair Tina Liebling and Ranking Member Joe Schomacker
Minnesota House
100 Rev. Dr. Martin Luther King Jr. Blvd.
Saint Paul, MN 55155

Chair Tina Liebling and Ranking Member Joe Schomacker,

I am writing on behalf of the Minnesota Rare Disease Advisory Council to express support for HF3330 which would expand coverage of rapid whole genome sequencing (rWGS) of critically ill children admitted to the ICU/NICU. The Minnesota Rare Disease Advisory Council (RDAC) is a state agency whose mission is to improve diagnosis and care for the 1 in 10 Minnesotans living with a rare disease.

The FDA defines a rare disease as a disease that affects fewer than 200,000 people in the US. There are reported to be as many as 7,000 to 10,000 rare diseases, and the total number of Americans living with a rare disease is estimated to be between 25-30 million individuals. In addition, the majority of rare diseases are genetic in origin and are of pediatric onset. The complexity and heterogeneity of rare diseases combined with their low prevalence leads to extreme difficulties in establishing a diagnoses for practitioners, with the results being an average delay of 7-8 to diagnosis and 2-3 misdiagnosis[1].

In addition to the immense stress that a desperate search for a diagnosis places on families, the delay in diagnosis also increases costs to the health system significantly due to multiple visits/testing that do not result In a diagnosis, the administration of inappropriate treatments because of misdiagnosis, and the increase in hospitalizations due to an unmanaged condition. A recent NIH-led pilot study[2] examined claims data and determined that cost of rare disease care is significantly higher for those with rare diseases compared to their non-disease age matched counterparts (Florida Medicaid claims data Indicated a PPPY cost ranging from $8,812 to $140,044 versus $2,211). While the reasons for the higher cost are complex, one of the cited contributing factors is the delay to diagnosis.

The advances in genetic testing hold significant potential to both reduce the diagnostic Odessey as well as the cost of care. Since more than 85% of the roughly 10,000 rare diseases are genetic in origin, genetic testing such as rWGS is a particularly effective tool in more timely diagnosis. Multiple studies done across the United States such as those conducted at Rady Children's Hospital[3] have demonstrated that incorporating rapid testing into NICUs for children with a suspected rare disease both improves diagnosis and clinical outcomes and reduces the cost of care.

We urge you to support HF3330 to ensure that children living with a complex and life-threatening condition can receive the diagnosis they deserve.

Sincerely,

Erica Barnes

[1] ShireReport-1.pdf (globalgenes.org)

[2]NIH Study Suggests People with Rare Diseases Face Significantly Higher Health Care Costs | National Center for Advancing Translational Sciences

[3] Project Baby Bear: Rapid precision care incorporating rWGS in 5 California children's hospitals demonstrates improved clinical outcomes and reduced costs of care - ScienceDirect