Genomic Answers FOR Children's Health ALLIANCE

August 2, 2024

The Honorable Diane DeGette
2111 Rayburn House Office Building
Washington, D.C. 20515

The Honorable Larry Bucshon, M.D
2313 Rayburn House Office Building
Washington, D.C. 20515

Dear Representatives DeGette and Bucshon,

Thank you for the chance to respond to your June 6, 2024, Request for Information related to seeking improvements to H.R. 6000, Cures 2.0.[1] The Genomic Answers for Children's Health Alliance is pleased to respond to your questions. **The Genomic Answers for Children's Health Alliance is a multi-sector alliance on a mission to ensure that any child with a rare disease or suspected genetic disorder who is enrolled in Medicaid has timely access to genomic sequencing, especially whole genome sequencing.**[2] We also seek to ensure that patients and their families can share the sequencing results with medical researchers, to help aid in the discovery and development of new treatments for rare diseases.

The Alliance agrees with you that the *21st Century Cures Act* (P.L. 114-255), which Congress enacted on an overwhelmingly bipartisan basis, has been tremendously successful in accelerating scientific understanding, spurring innovation, and bringing new treatments to patients. We are excited about your bipartisan efforts to start a new chapter in Congress' efforts to build on the legacy of 21st Century Cures.

As you know, in the U.S., an estimated 30 million Americans have a rare disease. More than half of those Americans are children.[3] There are estimated to be between 7,000 and 10,000 rare diseases and 95 percent have no specific FDA-approved treatment.[4] One study estimated that the economic burden of rare disease—counting direct and indirect costs—totaled nearly $1 trillion in the U.S. in just one year.[5] Rare diseases consume billions of dollars in Medicaid program expenditures each year.

Research shows that it too often takes children with rare diseases several years and many visits to different doctors to get a correct diagnosis. During this "diagnostic odyssey," children with rare diseases might have unnecessary tests and procedures, receive the wrong diagnosis, and experience delays in getting effective care. Because of the time it can take to get a diagnosis, patients may have fewer effective treatment options when they finally receive a diagnosis. This also means that many children with rare diseases experience irreversible damage as the disease progresses. One study estimated that the avoidable economic costs associated with the diagnostic odyssey were more than $500,000 per patient.[6]

[1] https://degette.house.gov/media-center/press-releases/reps-degette-bucshon-seek-stakeholder-input-next-generation-cures-bill
[2] https://gachalliance.org/about-us/
[3] https://www.gao.gov/products/gao-22-104235
[4] https://ncats.nih.gov/sites/default/files/NCATS_RareDiseasesFactSheet.pdf
[5] https://ojrd.biomedcentral.com/articles/10.1186/s13023-022-02299-5
[6] https://everylifefoundation.org/delayed-diagnosis-study/#about-study

The mapping of the human genome and advances in medical technology have made shortening the diagnostic odyssey a reality for many Americans. In particular, genomic sequencing can help children receive an accurate diagnosis in days. The speed of genomic sequencing to get to a diagnosis is nothing short of revolutionary, compared to the waiting, uncertainty, and challenges undergone by many patients prior to receiving sequencing.[7]

We appreciate that Section 407 of H.R. 6000, the *Cures 2.0 Act* (117th Congress), included a policy designed to expand access to genetic testing and genomic sequencing. The policies relevant to whole genome sequencing in the original version of Section 407 are as follows:

- The policy in the legislation reflected input from multiple members of Congress—including Reps. Swalwell (D-CA) and Peters (D-CA)—and was substantially similar to H.R. 5989 in the 117th Congress, the *Precision Medicine Answers for Kids Today Act*.[8]
- Section 407 of H.R. 6000 would establish a demonstration program for up to 15 state Medicaid programs to cover genetic and genomic testing for certain individuals under the age of 21 (or a lower age, if the state chooses) and for former foster youth under the age of 26.
- The Centers for Medicare & Medicaid Services would be required to report on overall Medicaid coverage of such services and issue guidance for states on ways to increase coverage for children.
- The bill also included a National Academy of Medicine study to assess the impact of this guidance as well as the general utility of genetic and genomic testing.

The Genomic Answers for Children's Health Alliance agrees with you that Congressional action is needed to require Medicaid coverage and payment of genomic sequencing for infants and children up to age 21 who are suspected to have a genetic condition or a condition of unknown origins. Coverage for this entire population (i.e., coverage that is not unduly restricted by subgroup) is urgently needed.[9] Critically, in conversations with the Alliance, CMS has communicated that it does not currently believe that the agency has the authority to provide directive guidance to state Medicaid programs such that every state would cover and reimburse for whole genome sequencing for the target population. This necessitates Congressional action in order to provide Medicaid coverage and sufficient reimbursement for these services.

Your Request for Information asked stakeholders to identify additional reforms that are needed, so we are happy to provide comment on genomic sequencing policy. We urge you to adopt the following specific policy reforms related to your interest in Section 407 of Cures 2.0.

---

[7] In the cases in which genomic sequencing may not yet yield a definitive diagnosis based on our current scientific understanding, genomic sequencing still informs clinical care. Sequencing can rule out thousands of diseases and give clinicians stronger ideas about how to care for children with that rare disease. Even when sequencing does not yield a diagnosis today, it can often be used to help find an answer in the future as families partner with medical researchers who, over time, discover key insights into characterizing new diseases and developing treatments for diseases. The NIH has noted, "about 80 percent of these rare disorders are genetic in origin" and virtually every human ailment has some basis in our genes, so genomic medicine has the capacity to revolutionize the discovery and development of new treatments over time.

[8] https://www.congress.gov/bill/117th-congress/house-bill/5989

[9] Today, a majority of state Medicaid programs cover whole exome sequencing, which has been helpful to many patients. While a minority of states currently cover whole genome sequencing, more state Medicaid programs are covering this sequencing each year. While our comments center on effectuating Medicaid coverage and reimbursement of whole genome sequencing, we believe whole exome sequencing plays an important role in the diagnostic odyssey. Whole exome sequencing (WES) involves sequencing all the pieces of an individual's DNA that provide instructions for making proteins. These pieces of DNA are known as exons and are found within genes. Because changes in the exons are what cause most known diseases, this method can be an efficient way to identify changes that cause disease. Whole genome sequencing (WGS) is more comprehensive than whole exome sequencing. Research has shown that some genetic disorders are caused by DNA changes that occur outside of the exons. Because of this, whole genome sequencing is useful as this type of testing can identify changes in any part of the genome.

1. **Congress should require Medicaid coverage for whole genome sequencing for the target population and reimburse at a state's regular Federal Medical Assistance Percentage (FMAP).** Section 407 of Cures 2.0 specifies a 100 percent FMAP, which would mean that the federal government would pay the entire cost of the service. While we appreciate this is well-intended to boost coverage within the construct of the demonstration in the legislation, there are very few bipartisan precedents for a 100 percent FMAP. Given the need to enact this policy as soon as possible, we support regular FMAP if it is politically necessary.

2. **Congress should require Medicaid coverage and reimbursement for whole genome sequencing for the target population be a permanent requirement for Medicaid programs in each of the 50 states, Washington, D.C., and the territories.** Section 407 of Cures 2.0 establishes temporary coverage of sequencing in 15 states, followed by CMS-issued guidance on ways for states to improve access to sequencing and testing. We understand the multi-state demonstration is intended to generate insights that will inform CMS guidance. However, as a matter of fairness and equity, any child with a disease of suspected genetic origins who is enrolled in Medicaid and in need of whole genome sequencing should have access to it—regardless of their state of residence. In addition, there is already an extensive body of evidence indicating the clinical effectiveness of whole-genome sequencing, both in experimental and in real-world settings, which eliminates the need for a demonstration project to generate evidence.[10,11,12,13]

Finally, for your convenience, we are including with our submission a collection of published studies and peer-reviewed literature that illustrates the merits and cost-effectiveness of whole genome sequencing.

We appreciate your consideration of our input and look forward to working with you as you work to iterate on Cures 2.0 to develop and advance bipartisan legislation that will improve access to sequencing, testing, care, and treatment for millions of patients across the country.

Sincerely,

The Genomic Answers for Children's Health Alliance

www.gachalliance.org

[10] https://pubmed.ncbi.nlm.nih.gov/34089648/
[11] https://pubmed.ncbi.nlm.nih.gov/25473036/
[12] https://pubmed.ncbi.nlm.nih.gov/25937001/
[13] https://pubmed.ncbi.nlm.nih.gov/28567303/