TO: Trump Transition Team
FROM: Genomic Answers for Children's Health Alliance
RE: Ensuring Children with Rare Disease Have Medicaid Coverage for Genomic Sequencing

The Genomic Answers for Children's Health Alliance is a public policy alliance of organizations working together to ensure that any child with a rare disease or suspected genetic disorder enrolled in Medicaid has timely access to genomic sequencing, especially whole genome sequencing.[1] We understand that President-Elect Trump's Administration is expected to prioritize health policy reforms that put patients first, seek to keep individuals healthy rather than only treating illnesses when they arise, and minimize wasteful spending in government programs to ensure that taxpayer dollars are spent efficiently. We also know that, although the Medicaid program is expected to spend about $600 billion federal dollars in FY 2025,[2] the Medicaid program can both underpay for some needed interventions and overpay in other cases. In the interest of improving care and boosting health for the most vulnerable while being cost-effective with Medicaid program funds, **the Alliance asks that the incoming Administration use its regulatory authority under Title XIX to promulgate rulemaking that requires state Medicaid programs to cover genome sequencing, especially whole genome sequencing (WGS), for children with a suspected rare disease or genetic disorder.**

In the United States, millions of children with a rare disease or genetic condition are enrolled in Medicaid as their primary or secondary health insurance coverage. Overall, about 30 million Americans have a rare disease, and more than half of those Americans are children.[3] Tragically, of the estimated 7,000 to 10,000 rare diseases, only 5 percent have an FDA-approved specific treatment.[4]

Regardless of what type of rare disease a child may have, the first step to getting care the child needs is getting an accurate diagnosis of the disease. However, under current law, the majority of state Medicaid programs are failing children and their families by not providing coverage and reimbursement for genomic sequencing for children with a suspected rare disease or genetic disorder. While today a slight majority of state Medicaid programs cover whole exome sequencing (another form of genetic testing) and a minority cover whole genome sequencing, a national standard for Medicaid coverage of whole genome sequencing would help children with rare disease, their families and caregivers, and the clinicians who provide medical care for them.

Today, too often, children with genetic disorders may go years without a diagnosis, potentially receiving unnecessary tests or procedures, and utilizing care in ways that could be more informed and efficient if the child had a diagnosis. Research shows that delays in diagnosis increase Medicaid spending and can increase program expenditures. In fact, one study estimated that the economic burden of rare disease in the U.S.—counting direct and indirect costs—totaled nearly $1 trillion in just

[1] https://gachalliance.org/about-us/
[2] https://www.cbo.gov/system/files/2024-06/51301-2024-06-medicaid.pdf
[3] https://www.gao.gov/products/gao-22-104235
[4] https://ncats.nih.gov/sites/default/files/NCATS_RareDiseasesFactSheet.pdf

one year.[5] As Medicaid is the largest insurer for American children, this study found that caring for children with rare diseases result in 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 a period of searching for a diagnosis (commonly called a "diagnostic odyssey"), Medicaid may pay for children with rare diseases to have unnecessary tests and procedures. Without precision testing, children may receive the wrong diagnosis or experience delays in getting effective care. Tragically, 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]

Rather than wait months or years for a correct diagnosis, genomic sequencing—in a single test—provides a definitive diagnosis in up to 50 percent of children in *days or weeks.*[7] Despite the robust evidence showing the efficacy of genomic sequencing,[8] state Medicaid coverage policies are mixed.[9] Even when states cover genomic sequencing, coverage may be inadequate in different ways. For example, coverage may only be provided to children when they are in inpatient settings, coverage may be "coverage" in name only when included in a hospital payment bundle without a corresponding increase in the bundle's reimbursement (*e.g.*, a DRG), or children who are beyond a certain age may be ineligible for coverage regardless of their symptoms and medical need.

Omitting coverage for genomic sequencing means that many state Medicaid programs are spending their money inefficiently and wastefully. It is telling that many commercial insurers, who often pioneer efficiencies in health care coverage and payment, are covering whole genome sequencing today.[10] Commercial insurers are keenly aware that WGS often leads to cost savings—one literature review found that health care costs were reduced by **more than $14,000 per child tested** with WGS.[11] Medicaid coverage of WGS would bring the program in line with a best practice that is becoming commonplace in the commercial insurance sector.

Limiting coverage for genomic sequencing also means that it is likely underutilized in our country—limiting the amount of genomic data we generate and use and unnecessarily slowing the biomedical innovation of our best medical researchers and scientists. This puts our country at a competitive disadvantage and puts us on a path where the cures and treatments of the future will be developed in other countries, and not made in America.

As a collection of stakeholders across the rare disease community, as a matter of putting patients first, prioritizing wellness, and basic fairness for the families of children with rare diseases, we believe every state Medicaid program should provide coverage of and reimbursement for genomic

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[5] https://ojrd.biomedcentral.com/articles/10.1186/s13023-022-02299-5
[6] https://everylifefoundation.org/delayed-diagnosis-study/#about-study
[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. It can also be used to advance future treatments as families partner with medical researchers who, over time, discover key insights into characterizing new diseases and developing treatments for diseases.
[8] For a compilation of sources regarding the cost-effectiveness and efficacy of genomic sequencing, see https://gachalliance.org/data-and-evidence/.
[9] https://radygenomics.org/clinical-genome-services/payer-policy-advocacy/
[10] See information on UnitedHealthcare's WGS coverage policies here: https://www.optum.com/en/business/insights/health-care-delivery/optum-evidence-engine.html and Aetna's announcement of WGS coverage here: https://www.aetna.com/content/dam/aetna/pdfs/olu/officelink-updates-september-2024-olu.pdf.
[11] https://www.nature.com/articles/s41525-024-00404-0

sequencing for any child who has a suspected rare disease or genetic condition, when ordered by a clinician and deemed medically necessary. A number of other Congressional policymakers agree. Indeed, improving access to genomic sequencing and genetic tests has been the subject of bipartisan legislation in the House and Senate during a number of past Congresses. However, the Trump Administration does not need to wait on Congress. The Trump Administration could use its Medicaid regulatory authority to specify under the *Early and Periodic Screening, Diagnostic, and Treatment* (EPSDT) Medicaid benefit that genomic sequencing, especially whole genome sequencing, is a medically-necessary service when ordered by a clinician for any child with a suspected rare disease or suspected genetic disorder who is enrolled in Medicaid. Such genomic sequencing should be covered and reimbursed without delay. Such an action would clearly be within CMS' regulatory authority and legally justifiable—states are allowed to restrict coverage of EPSDT services, but only for individual medical necessity, if the treatment is experimental, or if there are more cost-effective alternatives.[12] For children that meet clinical guidelines, WGS is medically necessary and cost-effective—there are significant amounts of data attesting to this.[13]

Taking this stance would be in line with the Trump Administration's priority to implement policies that put patients and families first and spend our tax dollars wisely in a key federal program.

Thank you for your time and attention to this important matter. If you have additional questions, please contact Josh Trent, Advisor to the Genomic Answers for Children's Health Alliance, at Josh.Trent@LeavittPartners.com.

Sincerely,

The Genomic Answers for Children's Health Alliance

[12] https://www.medicaid.gov/medicaid/benefits/downloads/epsdt-coverage-guide.pdf
[13] https://gachalliance.org/data-and-evidence/