December 16, 2024

The Honorable Chiquita Brooks-LaSure
Administrator
Centers for Medicare & Medicaid Services
7500 Security Boulevard
Baltimore, MD 21244

Delivered electronically

Dear Administrator Brooks-LaSure,

On behalf of the undersigned organizations that represent a diverse and broad community of patient advocates, laboratory professionals, providers, and manufacturers we write to respectfully request that the Centers for Medicare and Medicaid Services (CMS) provide guidance to states on Medicaid coverage for pediatric genetic tests for rare diseases under the Federal Early and Periodic Screening, Diagnostic, and Treatment (EPSDT) Program. We appreciate CMS' recent effort to provide best practices for states to ensure they can adhere to EPSDT requirements;[1] however, these general best practices are not sufficient to eliminate the obstructive administrative and medical necessity burdens faced by clinicians and underserved families when trying to access genetic testing and genomic sequencing services.

Half of patients with rare conditions are children, and approximately 70 to 80 percent of rare diseases are due to genetic causes[2,3]. The diagnostic odyssey that patients endure is riddled with burdens for them and their families. In the *National Economic Burden of Rare Disease* study conducted by the EveryLife Foundation for Rare Diseases, researchers found patients wait an average of 6.3 years for a diagnosis and receive an average of 17 medical interventions, including emergency visits and out-of-state-specialists, from the time symptoms began until a diagnosis was determined.[4] Without a diagnosis, patients may not have access to the most appropriate and comprehensive treatment plan for their unique health needs, which can profoundly and unnecessarily impact their prognosis.

EPSDT requirements are a cornerstone of the Medicaid program and are intended to provide robust health care, diagnostic services, treatment, and other measures for eligible children. Despite the importance of genetic and genomic testing and the benefits included as a part of the EPSDT program, it is well understood among the rare disease community that there are gaps in providing this needed testing for children. In 2021, the GAO recognized that the lack of accessible diagnostic tests complicates the diagnosis of rare diseases.[5] The report describes studies that demonstrate that payers are denying coverage for testing deeming it "not medically

[1] https://www.medicaid.gov/federal-policy-guidance/downloads/sho24005.pdf
[2] https://www.nature.com/articles/s41431-019-0508-0
[3] https://pubmed.ncbi.nlm.nih.gov/31023718/
[4] Yang G, Cintina I, Pariser A, Oehlrlein E, Sullivan J, Kennedy A. The national economic burden of rare disease in the United States in 2019. Orphanet J Rare Dis. 2022;17:163
[5] https://www.gao.gov/products/gao-22-104235

necessary" or "experimental" despite the test providing information vital to making an accurate diagnosis. GAO also noted that experts shared that prior authorization for testing can delay care. This is particularly problematic for children with diseases where time is of the essence. Considering the high prior authorization denial rates and that most providers and patients do not pursue the appeals process, it is possible that prior authorization is also creating barriers to accessing genetic testing for Medicaid beneficiaries.[6][7] Many states have made it clear that genetic testing is indeed covered; however, there are a great deal of disparities among states, with a number providing no coverage at all for genetic testing for rare diseases.[8] This is concerning because the EPSDT benefit covers necessary diagnostic services to correct or ameliorate defects whether or not such services are covered under the state plan.

Scientific understanding of the role of genetic testing in clinical care has evolved considerably, and the lack of clear guidance to Medicaid programs that genetic diagnostic testing is included under the EPSDT benefit contributes to disparate access to these services across the United States. Moreover, it is essential that patients have access to all types of genetic and genomic testing, including single gene tests, multigene panels, whole exome sequencing, and whole genome sequencing. We believe determining which test is most appropriate based on the clinical situation is best left up to patients and their physicians. Single gene tests are often the most appropriate choice when a patient's clinical presentation is associated with a single, well-known gene. They are also lower-cost and typically have a shorter turnaround time, allowing for a rapid diagnosis. In other cases, a multi-gene panel may be used when there are a number of well-known genes associated with a patient's clinical presentation. Whole exome and whole genome testing are best suited for cases when it is not apparent or known which genes may be involved, and the patient has complex clinical presentations or multiple diagnoses. As a result, the undersigned organizations respectfully request that the agency clarify, through guidance, that evidence-based genetic and genomic tests, including single gene, multigene, whole exome, and whole genome sequencing tests, are a covered benefit under the EPSDT program.

Thank you for considering this request. Patients with rare diseases and their families deserve affordable access to these critical tools, and we appreciate the steps the agency has taken to recognize their importance. Please do not hesitate to reach out to Cynthia A. Bens at cbens@personalizedmedicinecoalition.org or Brett Kopelan at brett@debra.org if you have any questions or if we can be of any further assistance.

Sincerely,

AiArthritis
Alexion AstraZeneca Rare Disease
The Alex Manfull Fund
Alliance to Solve PANS & Immune-Related Encephalopathies (ASPIRE)

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[6] https://www.gimjournal.org/article/S1098-3600(21)01452-0/fulltext#:~:text=Prior%20authorization%20request%20outcome,%3C%200.001;%20Table%203).&text=Reasons%20for%20prior%20authorization%20request,CPT%20current%20procedural%20terminology.
[7] https://www.ncbi.nlm.nih.gov/pmc/articles/PMC10584034/
[8] https://geneticspolicy.nccrcg.org/medicaid-coverage/

Association for Molecular Pathology
AstraZeneca
Assurance Health Data
Avery's Hope
BDSRA Foundation
Biocom California
Biomarker Collaborative
Child Neurology Foundation
Chondrosarcoma Foundation
Cure CMD
CureLGMD2i Foundation
debra of America
DNAnexus
Dravet Syndrome Foundation
Eli Lilly and Company
EveryLife Foundation for Rare Diseases
Exon 20 Group
GeneDx
GO2 for Lung Cancer
Harbor Precision Genomics Consulting
HCU Network America
H. Lee Moffitt Cancer Center
ICAN, International Cancer Advocacy Network
Illumina, Inc.
Institute for Gene Therapies
Labcorp
Little Hercules Foundation
Look. Foundation
The Louisa Adelynn Johnson Fund for Complex Disease
MET Crusaders
MJB Lab Services
MLD Foundation
Muscular Dystrophy Association
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