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January 30, 2023

The Honorable Chiquita Brooks-LaSure
Administrator
Centers for Medicare & Medicaid Services
Department of Health and Human Services
7500 Security Blvd
Baltimore, MD 212441

RE: Request for Information – Essential Health Benefits
CMS-9898-NC

Dear Administrator Brooks-LaSure:

Haystack Project appreciates the opportunity to respond to the Center for Medicare & Medicaid Services' (CMS') Request for Information (RFI) related to Essential Health Benefits (EHB) under the Patient Protection and Affordable Care Act (the ACA).

Haystack Project is a 501(c)(3) non-profit organization enabling rare and ultra-rare disease advocacy organizations to highlight and address systemic access barriers to the therapies they desperately need. We strive to amplify the patient and caregiver voice in disease states where unmet need is high, and treatment delays and inadequacies can be catastrophic. Our core mission is to evolve health care payment and delivery systems, spurring innovation and quality in care toward effective, accessible treatment options for Americans living with rare or ultra-rare conditions. Haystack Project is committed to educating policymakers and other stakeholders about the unique circumstances of extremely rare conditions with respect to product development, commercialization, and fair access to care.

Haystack Project recognizes that the health insurance marketplaces created under the ACA are critical to achieving the goal of equitable, affordable access to quality health care for all Americans. For individuals and families living with an extremely rare disorder, access to meaningful, comprehensive coverage can be a literal lifeline. We applaud CMS for proactively engaging with stakeholders to assess the real-world experience of patients with respect to the scope of benefits, care affordability, and access to the items and services they need to meet their health goals.

Benefit Descriptions in EHB-Benchmark Plan Documents

CMS noted that the various states describe their benchmark plans differently, particularly with respect to the level of specificity on the items and services included in or excluded from coverage. Haystack Project agrees that it would be unreasonable, and of little benefit, to require that states describe their EHB benchmark plans by delineating covered benefits and exclusions with detailed precision. We recommend a balanced approach that ensures sufficient detail to enable the states and CMS to evaluate the generosity of coverage within the benchmark plans and update the scope of benefits to respond to coverage gaps. We also recommend that CMS ensure that EHB are not only defined, but implemented, to address the unique health care needs of individuals with rare conditions.

Approximately 7,000 rare diseases have been identified to date, 90-95% of which have no FDA approved treatment. Cumulatively, rare diseases affect approximately 30,000,000 or 1 in 10 individuals in the U.S. Rare disease patients face substantial challenges from symptom emergence through treatment or management of their condition. These patients:

- See an average of 4.2 primary care physicians and 4.8 specialists before receiving an accurate diagnosis.
- Make an average of 2.4 out-of-state trips related to their diagnosis.
- Visit an emergency room an average of 3.7 times and - are hospitalized an average of 1.7 times for reasons related to their rare disease prior to diagnosis.
- Face a heightened risk of misdiagnosis.
- Have a very limited set of clinicians with disease-specific expertise, making it difficult for many patients to identify an experienced provider within their network, or even their state.
- Disproportionately rely on off-label use of treatments indicated for more common conditions to address disease symptoms and/or progression. Due to disease rarity, these off-label uses are seldom included within compendia.

Individually, these access challenges can present inconveniences, frustration, and delays in receiving care. Cumulatively, they can present an overwhelming burden for patients and their families. We recommend that CMS require that benchmark plan descriptions provide clarity on:

- Scope of coverage for routine care costs (and other expenses) associated with participation in clinical trials.
- Availability of coverage for medically necessary services that are not available within a plan's network and/or state.
- Inclusion of disease-specific specialist consultations with treating providers within the set of covered benefits.
- Coverage for home nursing services, the type of nursing provider is within the scope of benefits, and whether the payment provided for such services is in amount equal to or greater than the average hourly wage for each covered nursing provider.

- Sources used for determining whether a particular treatment is medically necessary, and availability of reconsideration and appeals processes that utilize clinical guidelines and/or disease-specific expert opinion (rather than relying solely on labeled and compendia-listed uses).
- Cost sharing (tiers, and use of copayment or coinsurance) and conditions of reimbursement (prior authorization, step therapy protocols, quantity limits) for specialty drugs, including orphan drugs.
- Existence of any plan procedural delays in covering newly-approved treatments. Many plans will delay coverage for up to a year due to the need to perform "formulary review" – this delay can be catastrophic (and unnecessary) for patients without alternative FDA-approved treatment options.
- Covered newborn screening tests.
- Pediatric care benefits, including availability of coverage for genetic testing in children with serious, undiagnosed health conditions.
- Existence of a distinct pediatric services benefit class and/or specific exclusions for children with special health care needs.

Typical Employer Plans

CMS has requested stakeholder input on typical employer plans and how the scope of benefits currently available in a typical plan may differ from those offered when the Agency defined "typical employer plan" in 2014. Increased flexibility in coverage for clinician services through telemedicine was driven by necessity during the COVID-19 Public Health Emergency (PHE). The appropriate use of telehealth services can offer value to patients, providers, and plans so long as the decision on remote versus face-to-face care is made within the clinician/patient relationship based on the patient's condition, needs and preferences. Increased inclusion of telehealth services within covered health benefits has:

- Allowed physicians to use telehealth to supervise rural non-physician providers;
- Permitted practitioners to satisfy direct supervision requirements virtually using real-time, interactive audio and video technology.
- Included use of audio-only devices for patients without access to audiovisual capabilities and permitted patients to access telemedicine from their homes.
- Increased coverage for "wearables" and other patient monitoring technologies to provide key patient information that may not be ascertained from periodic in-person visits.

Haystack collected information from its ultra-rare patient groups on challenges patients faced in obtaining high-quality care through telemedicine. While many patients noted that they found it easier to access a specialist through telehealth, only 8% of surveyed patients stated that they were satisfied with their ability to receive injectable medications within the home setting despite increased coverage for this service among commercial insurers. Respondents generally viewed telehealth as vital through the PHE, and useful as an adjunct to in-person care thereafter. We also heard from patients and caregivers regarding the barriers individuals with

hearing and/or visual impairments face in seeking remote care. Usher Syndrome, for example, is a very rare (approximately 25,000 US patients) inherited disease causing combined hearing loss and vision loss from retinitis pigmentosa. For these patients, it is essential that remote care includes access to an American Sign Language (ASL) interpreter if they have sufficient remaining vision, or a tactile sign interpreter if they do not.

Throughout the COVID-19 Public Health Emergency (PHE), Haystack collected information from patient groups on challenges patients faced in obtaining care while maintaining social distancing to minimize exposure to the coronavirus. Specifically, we heard from patients and caregivers regarding the barriers that individuals with hearing and/or visual impairments face in seeking care through telemedicine and expect that those barriers similarly impede eligibility determinations, enrollment, and meaningful access to providers. Usher Syndrome, for example, is a very rare (approximately 25,000 US patients) inherited disease causing combined hearing loss and vision loss from retinitis pigmentosa. For these patients, it is essential that remote care includes access to an American Sign Language (ASL) interpreter if they have sufficient remaining vision, or a tactile sign interpreter if they do not. We urge CMS to consider incorporating sufficient add-on codes to telemedicine options, on a permanent basis, to enable payment for services of an ASL or tactile sign interpreter and facilitate data collection on access to these services.

Medical Evidence and Scientific Advancement

Individuals with rare conditions face disproportionate challenges, including high out-of-pocket costs, in receiving the care they need when they need it.

CMS expressed an interest in stakeholder feedback on how changes in medical evidence or scientific advancement generally could inform CMS' health equity and nondiscrimination efforts with regards to EHB. As noted above, rare disease patients and their families face significant burdens in accessing the right care at the right time. In 2021, the Government Accountability Office (GAO) complied a report to Congress entitled “RARE DISEASES: Although Limited, Available Evidence Suggests Medical and Other Costs Can Be Substantial” in collaboration with EveryLife Foundation for Rare Diseases and the National Organization for Rare Disorders. The report assessed the challenges rare disease patients face accessing diagnostic and treatment services as well as the personal and economic costs associated with treatment delays. Among its many findings, the GAO found that rare disease patients are unable to access specialists due to geography or failure to receive a referral for follow-up care at initial symptoms, and often progress to more severe disease states by the time they receive an accurate diagnosis. Forty-one percent of rare disease patients also receive at least one misdiagnosis, and many are treated for a condition they did not have, and approximately 7 percent of rare disease patients reported that they were given a false psychological/psychiatric diagnosis that further impeded

and delayed their treatment.[1] Making access to appropriate experts through telemedicine could make a meaningful difference in addressing some of these findings.

In addition to the difficulties rare disease patients face in receiving a diagnosis, identifying an in-network specialist with disease-specific expertise, and identifying a treatment plan to manage disease symptoms and progression, individuals insured through employer-sponsored coverage are increasingly confronted with noncoverage when an FDA-approved therapy becomes available. Self-insured and large employer plans have leveraged benefit flexibilities (e.g., determining that specific prescription drugs are "nonessential" health benefits) to deny patient access to what may be the only therapeutic option to reduce disease burden and/or slow disease progression. This can be catastrophic for families impacted by a rare condition with a treatment that is deemed to be a non-essential health benefit. We urge CMS to revisit this policy and either revoke it or otherwise ensure that impacted patients have access to the treatment they need at a cost they can afford.

Education and outreach to primary care providers could improve the real-world health care experience for rare disease patients and substantially reduce health inequities in these patient populations.

In addition, individuals with rare diseases and conditions often suffer from disease symptoms for 5-9 years and see an average of 7 specialists before obtaining a diagnosis. For people of color who suffer from a rare disease, access to diagnostic and treatment services can be even more challenging. Underrepresentation in research and clinical trials make timely diagnosis and adequate treatment an elusive goal for underserved populations. Primary care clinicians are on the front line for these patients, making referrals to specialists based on the patient's symptoms, assessing progression or changes in symptoms, and sharing their patients' frustration. Clinicians and patients often focus on managing symptoms without considering the possibility of a rare or ultra-rare disease.

Haystack urges HHS to engage with the rare disease community to focus on primary care outreach strategies that (1) emphasize the fact that 1 in 10 U.S. patients suffers from a rare disease and (2) outline actionable recommendations that primary care clinicians should follow based on symptom persistence, combinations of symptoms, and other factors. In addition, HHS should increase provider-level awareness of initiatives like the Common Fund's Undiagnosed Diseases Network (UDN). This National Institutes of Health initiative is operated through the Office of Strategic Coordination as a research study to improve the level of diagnosis of rare and undiagnosed conditions.

The UDN nationwide network of clinicians and researchers should be a known, go-to resource for primary care clinicians unable to identify the cause of chronic, persistent, or episodic patient symptoms. Haystack urges HHS to assess the extent to which UDN participants to date are

[1] GAO Report. GAO-22-104235, RARE DISEASES: Although Limited, Available Evidence Suggests Medical and Other Costs Can Be Substantial

representative of patient populations with respect to racial, ethnic, sex/gender, socioeconomic, and geographic factors. We also CMS and HHS to engage primary care providers, including those practicing in rural and underserved urban areas, to facilitate increased participation in UDN.

Rare disease patients face significant, disproportionate barriers to the services needed to diagnose, treat, and manage their condition(s).

The GAO report discussed above found that rare disease patients face a lack of availability or accessibility to diagnostic tests. Even when confirmatory diagnostic testing is available, it is often not accessible due to reimbursement hurdles. Since most rare diseases are without an FDA-approved treatment, some payers decline coverage due to lack of "medical necessity." Patients and their clinicians seeking a definitive diagnosis to enable a treatment plan (on- or off-label therapies) face significant paperwork burdens, including prior authorization and appeals processes.

Haystack Project's outreach efforts have revealed several areas of concern to patients that, if adequately addressed, could close care gaps and reduce health inequities.

- Patients face uncertainties in accessing off-label treatments used within the standard-of-care due to limited inclusion of rare disease considerations in the compendia that payers generally rely on. The rarer the disease, the less likely it is that medically accepted treatments will be published in compendia. Patient access programs are not generally available since a manufacturer offering free or discounted drug in this patient population would face off-label promotion scrutiny and potential liability. This leaves patients with few options unless they receive care from a provider willing to navigate the reconsiderations and appeals processes. Patients in underserved communities are less likely to have access to these clinicians.

- Receiving care in the home through telemedicine is often the best option for low-income and rural patients and their families. Social determinants of health (SDOH) can, however impede availability of this option due to lack of broadband internet capabilities and financial impediments to maintaining reliable housing and utilities.

- We suspect that the unduly lengthy journey from emergence of symptoms to diagnosis is even longer for patients in communities of color and other underserved populations. Unfortunately, our member organizations do not have the data to quantify those inequities or identify clear causative factors.

- In rare disease patients, subtle changes in disease symptoms and/or progression could have profound impacts on longer-term outcomes. Encouraging plans to deploy wearables, monitors, and layperson friendly medical equipment would enhance remote monitoring capabilities and provide key patient information that may not be ascertained from periodic in-person visits,

Newborn screening programs in vary among states and do not consistently reflect advances in diagnosing and treating conditions impacting newborn development and health.

Haystack Project urges CMS to consider an inter-agency effort to increase access to newborn screening services and reduce the variability in covered screening among the states. The Health Resources and Services Administration (HRSA) maintains the Recommended Uniform Screening Panel (RUSP) which lists disorders that the Secretary recommends for inclusion in state newborn screening (NBS) programs. The RUSP update process is supported by recommendations from the Advisory Committee on Heritable Disorders in Newborns and Children.

Evidence from state NBS programs that include testing for metabolic disorders suggests that mandatory screening can reduce health disparities.[2] Unfortunately, state variability in breadth of NBS programs, testing protocols, and costs to families varies considerably. Spinal Muscular Atrophy, for example, is a genetic disease affecting the central nervous system, peripheral nervous system, and voluntary muscle movement. It is the most common genetic cause of mortality in infants. The first disease-modifying therapy (Spinraza (nusinersen)) received Food and Drug Administration (FDA) approval on December 26, 2016; in May 2019, FDA subsequently approved Zolgensma (onasemnogene abeparvovac-xioi), the first gene-replacement therapy for a neuromuscular disease. Both treatments have demonstrated potential to slow, stop, or even reverse the devastating symptoms of SMA and can be given in individuals who are not yet symptomatic. HRSA did not include testing for SMA on the RUSP until 2018. The majority of state NBS programs continue to exclude SMA testing despite the fact that two disease modifying therapies are now available.

States also vary considerably in their testing protocols. Most simply specify that the NBS testing is performed before discharge. Maryland, however, specifies that one NBS blood sample be collected when the baby has had 24 hours of feeding. A second sample is collected when the baby is between 10 and 14 days old. The website outlining Maryland's NBS program notes that "the two screen system has been supported by the fact that about 10% of congenital hypothyroidism is identified by testing the second (subsequent) screen." The two-screen system is also necessary for accurate testing for cystic fibrosis (CF).

We urge CMS and HHS to ensure that robust newborn screening (including genetic testing) is available in all states and includes testing for rare and ultra-rare conditions that can be identified and addressed through treatments (on- and off-label) as well as those for which families will likely benefit from caregiver resources. Haystack recommends that CMS and HHS:

[2] Brosco JP, Grosse SD, Ross LF. Universal state newborn screening programs can reduce health disparities. JAMA Pediatr. 2015 Jan;169(1):7-8.

- Consider implementing a "carrots and sticks" approach to allocating funding from the Centers for Disease Control and Prevention (CDC) based on the extent to which state NBS programs
    - include all RUSP tests,
    - expand NBS testing to a larger set of conditions for which treatments, including dietary regimens, can reduce disease symptoms or progression, and
    - conduct testing in a manner likely to secure valid results for all tests on the panel (e.g., timing of sample, subsequent testing).

- Consult with the rare disease advocacy community on development of a supplemental informed consent document that would give new parents of infants diagnosed with a condition the option of being connected with an advocacy organization or other entity sponsoring a patient registry and/or connecting patients with care and support resources.

- Develop, in consultation with disease-specific experts, a uniform set of disease-specific "next steps" for both primary care physicians and families. Haystack Project members have noted that pediatricians vary with respect to instructions provided to families, with some families noting that their child's primary care physician suggested that a condition detected in an infant was likely a "false positive."

- Require that states disclose to new parents any tests listed on the RUSP but not included in the state NBS program, as well as testing and treatments available for those conditions.

- For states that do not include the full set of RUSP tests, require state investigation and disclosure of any newborn screening variability, including existence of differential testing for newborns covered by Medicaid, employer-sponsored coverage, ACA plans, and other payers.

- Convene an HHS Health Equity Task Force to examine and make recommendations regarding the conditions included on the RUSP list, state NBS panel variability on included tests, specimen collection, patient/payer costs, and follow-up. A task force modeled on the COVID-19 Health Equity Task Force within the HHS Office of Minority Health would be well-positioned to provide recommendations on refinements to NBS panels and the RUSP that would both reduce health inequities and improve health outcomes for all U.S. children with treatable rare diseases and conditions.

Substitution of EHB

Haystack Project has supported CMS' reversal of the 2019 Payment Notice provision amending the ACA regulations giving states the flexibility to permit issuer substitution of benefits within EHB categories. We remain convinced that state use of this "flexibility" would have a high

potential for harmful impacts on individuals with rare diseases and chronic conditions. While we appreciate that CMS is open to considering state flexibilities to address changing public policy focuses, the potential harm to particularly vulnerable patients continues to outweigh any benefit to plans and the relatively healthy individuals most likely to benefit from EHB substitution.

Conclusion

Once again, Haystack Project appreciates the opportunity to respond to the RFI with recommendations to refine the essential health benefits under the ACA in a manner that reflects current scientific evidence and reduces health inequities. We look forward to a continuing dialogue to increase the effectiveness of ACA coverage in facilitating timely, equitable diagnosis and treatment access for our patients.

If you have any questions or would like to discuss our response, please contact M Kay Scanlan, JD, at 410-504-2324.

Best regards,


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