March 20, 2024

The Honorable Brett Guthrie
Chair, Health Subcommittee
House Committee on Energy and Commerce
2434 Rayburn House Office Building
Washington, DC 20515

The Honorable Anna Eshoo
Ranking Member, Health Subcommittee
House Committee on Energy and Commerce
272 Cannon House Office Building
Washington, DC 20515

The Honorable Cathy McMorris Rodgers
Chair
House Committee on Energy and Commerce
2125 Rayburn House Office Building
Washington, DC 20515

The Honorable Frank Pallone, Jr.
Ranking Member
House Committee on Energy and Commerce
2322A Rayburn House Office Building
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Dear Chair Guthrie, Ranking Member Eshoo, Chair McMorris Rodgers and Ranking Member Pallone,

On behalf of the more than 30 million Americans living with one of the over 10,000 known rare diseases, the National Organization for Rare Disorders (NORD) thanks the House Committee on Energy and Commerce's Health Subcommittee for holding a hearing focused on oversight of diagnostic testing that could have a profound impact on the rare disease community NORD so proudly represents.

NORD is a unique federation of non-profit and health organizations dedicated to improving the health and well-being of people with rare diseases by driving advances in care, research, and policy. NORD was founded over 40 years ago, after the passage of the Orphan Drug Act (ODA), to formalize the coalition of patient advocacy groups that were instrumental in passing this landmark law. Since that time, NORD has been advancing rare disease research and funding to support the development of effective treatments and cures; raising awareness and addressing key knowledge gaps; and advocating for policies that support the availability of affordable, comprehensive health care, including access to safe and effective therapies.

Many people living with a rare disease struggle to obtain an accurate diagnosis, have limited treatment options, and grapple with access to health care providers with expertise in their condition, and to afford the often high out-of-pocket costs associated with their treatment and care. The medical needs of those living with a rare disease are complex, as are the policies necessary to enable rare disease patients to thrive. NORD is grateful to the Subcommittee for holding today's hearing examining how best to regulate diagnostic tests without threatening access for patients with rare diseases.

Rare disease patients, families and health care providers need practical and feasible legislative solutions that are risk-based, data-driven, and that consider the unique challenges around rare diseases. The delay in reaching agreement on oversight of lab developed tests (LDTs) has diverted considerable resources and perpetuated substantial and inherently harmful uncertainty for rare disease patients that largely depend on LDTs. We urge Congress to find a solution now that prevents bad tests from harming patients while ensuring continued access to diagnostic testing for rare disease patients – our patients and the future of rare disease therapies depend on it.

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<u>The stakes are high for the roughly 1 in 10 Americans living with a rare disease</u>:

1. **LDTs are central to the medical care of rare disease patients – and the economics of developing diagnostic tests for rare diseases are fundamentally different from other disease areas**. As many as 80% of all rare diseases have a genetic component; many genetic tests in clinical use today are LDTs.[1] Similarly, most newborn screening tests administered by publicly funded and run newborn screening programs across the country are LDTs, as are many companion diagnostics such as biomarker tests central to the safe and effective use of many rare disease therapies, particularly in the oncology space.[2]

2. **Rare disease patients need and deserve accurate, reliable diagnostic tests <u>and</u> timely access to them.** In many cases, an incorrect diagnostic test result can cause more harm for the patient and family than no diagnostic test at all. Yet, treatment delays also harm patients; many rare disease patients already face a long 'diagnostic odyssey.'[3] Perhaps most importantly, an increasing number of innovative rare disease therapies have narrow treatment windows. Any delay in diagnosis can – and does – exclude rare disease patients from clinical trials, or from receiving FDA-approved therapies. Given the limited alternative treatment options for many rare diseases, such delays are often devastating for patients and families. Similarly, timely access to companion diagnostics such as biomarker tests,[4] which provide vital information about the safe and effective use of a corresponding drug or biologic, often determines if and when rare disease patients access life-altering therapies.

3. **Not all diagnostic tests are created equal or carry the same risks for patients.** It is not feasible – or desirable - to require the same level of oversight over all tests regardless of how they are used or what risks and benefits they pose to patients. Companion diagnostics such as biomarker tests in particular play an increasingly important role in rare disease drug development, and delays in the approval or clearance of a companion diagnostic can lead to devastating delays in drug approval or biologic licensure.

4. **The existing regulatory framework for devices was not made for LDTs and will not work well for rare disease LDT; careful, deliberate implementation of any sweeping changes to the LDT sector is vital for success.** Data on the use and performance of LDTs in rare diseases (and more broadly) to guide implementation are very scarce. This makes careful, deliberate implementation of

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[1] Richardson, L., Dobias, M., Akkas, F., Younoszai, Z., & McAndrew, E. (n.d.). *The Role of Lab-Developed Tests in then Vitro Diagnostics Market*. The Pew Charitable Trusts. https://www.pewtrusts.org/-/media/assets/2021/10/understanding-the-role-of-lab-developed-tests-in-vitro-diagnostics.pdf

[2] Id.

[3] *Barriers to Rare Disease Diagnosis, Care and Treatment in the US*. (2020, November 19) National Organization for Rare Disorders.. https://rarediseases.org/wp-content/uploads/2020/11/NRD-2088-Barriers-30-Yr-Survey-Report_FNL-2.pdf

[4] Center for Devices and Radiological Health. *Companion Diagnostics*. (2023). U.S. Food and Drug Administration. https://www.fda.gov/medical-devices/in-vitro-diagnostics/companion-diagnostics#:~:text=A%20companion%20diagnostic%20is%20a,corresponding%20drug%20or%20biological%20product.

any sweeping changes to the LDT sector particularly important. It also reinforces the need for ample input from ALL parts of the impacted communities.

The rare disease community needs Congressional leaders to come together in a bipartisan manner on this topic; we need legislative solutions that can adequately address the unique challenges and needs of all patients including the 30 million Americans living with rare diseases. We hope the upcoming hearing will build on the similar hearing almost 10 years ago.[5]

- We recognize a key question this week – as 10 years ago - will likely be whether FDA should oversee LDTs at all, or whether they are better regulated by modernizing regulations under the Centers for Medicare and Medicaid Services (CMS) through the Clinical Laboratory Improvement Amendments (CLIA). FDA and CMS issued a joint statement in January of this year stating "[…] CMS does not have the expertise to assure that tests work" and "[…] the complementary FDA and CMS frameworks are both critical to assuring patients can rely on the clinical accuracy of their test results."[6] Given CMS's strong and recent view on the issue, we worry further discussions of this issue will likely prove of limited practical utility.

- As in 2014, FDA sketched out a new proposal to regulate LDTs in the fall of 2023 – the latest chapter in a long string of unsuccessful regulatory attempts dating all the way back to 2006.[7,8,9] When the public comment period closed last December, the agency had received more than 6,700 public comments, many expressing concerns about the proposed rule.[10] The resulting final rule is currently under Office of Management and Budget (OMB) review. NORD, as well as many other stakeholders, have serious concerns about the proposed rule and the potential unintended consequences associated with its practical implementation, similar to concerns NORD raised almost 10 years ago.[11,12] The final

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[5] *21st Century Cures: Examining the Regulation of Laboratory-developed Tests*. (2024, March 20). https://www.congress.gov/event/113th-congress/house-event/LC39273/text

[6] Center for Devices and Radiological Health. (2024, January 18). *FDA and CMS: Americans Deserve Accurate and Reliable Diagnostic Tests, Wherever They Are Made.* U.S. Food and Drug Administration. https://www.fda.gov/medical-devices/medical-devices-news-and-events/fda-and-cms-americans-deserve-accurate-and-reliable-diagnostic-tests-wherever-they-are-made

[7] Congressional Research Service. *FDA Regulation of Laboratory-Developed tests (LDTs).* (2022, December 7). https://crsreports.congress.gov/product/pdf/IF/IF11389

[8] Center for Biologics Evaluation and Research (CBER). *Framework for Regulatory Oversight of Laboratory Developed Tests.* (2014, October 3). U.S. Food and Drug Administration. https://www.fda.gov/media/89841/download

[9] Covington and Burling, LLP. *HHS Issues New LDT Policy, Rescinding FDA Premarket Review Policies.* (2020, August 25). https://www.cov.com/en/news-and-insights/insights/2020/08/hhs-issues-new-ldt-policy-rescinding-fda-premarket-review-policies

[10] Federal Register. *Medical Devices; Laboratory Developed Tests.* 88 FR 68006. (proposed 2023, October 03). https://www.federalregister.gov/documents/2023/10/03/2023-21662/medical-devices-laboratory-developed-tests

[11] *NORD Supports Lifting the HDE Cap.* National Organization for Rare Disorders. (2015, September 15). https://rarediseases.org/assets/files/policy-statements/2015-09-21.NORD-Supports-Lifting-the-HDE-Cap.pdf

[12] *NORD Comments on Proposed LDT Rule.* National Organization for Rare Disorders. (2023, December 04). https://rarediseases.org/wp-content/uploads/2023/12/NORD_comments_LDT_proposed_rule_2023_final.pdf

rule is likely to raise many (or more) of the same concerns. Unfortunately, similar to 10 years ago, the forthcoming FDA regulatory proposal from FDA is unlikely to generate practical, tangible solutions.

We simply cannot continue this fruitless debate indefinitely. We urge Congress to come together in a bipartisan manner to find a practical solution now that prevents bad tests from harming patients while ensuring continued access to diagnostic testing for rare disease patients – our patients and the future of rare disease therapies depends on it.

NORD is grateful for the Subcommittee's attention to these critical issues and looks forward to working with the Subcommittee to better support the rare disease community. Please do not hesitate to reach out to Karin Hoelzer at KHoelzer@rarediseases.org when NORD can be of assistance to the Subcommittee's important work.

Sincerely,

Karin Hoelzer, DVM, PhD
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