

Unmet Medical Device Needs for Patients With Rare Diseases

Background

The impact of rare diseases is likely far greater than the term implies. The lives of nearly 30 million Americans, half of whom are children, are directly affected by approximately 7,000 rare diseases.¹ Statistics for the number of people seeking care with disorders of unknown or unclear etiology (i.e., undiagnosed rare disease patients) remain elusive. When these potential numbers are considered alongside known numbers, the probability that every health care professional in the United States cares for at least one patient with a rare disease—knowingly or unknowingly—becomes a relevant consideration for resource allocation and policy development within the U.S. health care ecosystem.

The U.S. Food and Drug Administration (FDA) and the National Center for Advancing Translational Sciences (NCATS)/Office of Rare Diseases Research (ORDR) at the National Institutes of Health (NIH) sought to better understand the medical device needs of patients with rare diseases. Medical devices represent a highly diverse spectrum of promising technologies for rare diseases, both in diagnostic testing options and in treatments. These technologies range anywhere from simple medical instruments to cutting-edge scientific advances in implants and nanotechnology.

The Orphan Drug Act generally defines a rare disease as one affecting fewer than 200,000 people in the United States, yet many rare diseases affect only tens to hundreds of people. This level of rarity adversely affects the potential for improving diagnostic and therapeutic options to better serve this population. In the past three decades, the Orphan Drug Act has stimulated a significant increase in the development of drugs and biologics for these diseases; however, development of devices for rare diseases has lagged behind.

From late 2015 to 2016, FDA and NCATS/ORDR at NIH conducted a needs assessment to better understand unmet medical device needs for rare diseases; generate meaningful data to inform patients, practitioners, policymakers, and device developers on the needs, barriers, and incentives related to medical device development for rare diseases; and increase public awareness of these needs. The assessment included a subfocus on pediatric rare disease patients. This report describes the results of that assessment, which offers key findings about device needs in adult and pediatric rare disease populations.

Methods

The agencies conducted an online survey of four clinician groups that advise or work with FDA concerning device development or with NCATS regarding clinical trials of rare diseases. Two of these groups consisted of clinicians focusing on pediatric product issues, which provided a better understanding of the unique needs of pediatric patients. The complete clinician groups included physicians and non-physicians with patient experience (e.g., dentists, optometrists, and therapists). In this report, those who responded to the survey from the clinician groups are referred to as respondents or clinicians.

The survey was designed to elicit information regarding (1) satisfaction with current diagnostic and therapeutic devices, (2) unmet diagnostic and therapeutic device needs for specific rare diseases identified by each respondent, (3) unmet diagnostic and therapeutic device needs for rare disease populations in general, (4) impediments to medical device development, and (5) familiarity and

¹ Global Genes. *Rare diseases: Facts and statistics*. 2015. <http://globalgenes.org/rare-diseases-facts-statistics/>. Accessed January 21, 2017.

experience with Humanitarian Use Devices (HUDs). Those with direct experience were also asked about current diagnostic and therapeutic practices for specific rare diseases they identified, including limitations in current practices. For a companion manuscript, a separate statistical analysis was performed on results solely from participating physicians who had direct experience with or knowledge of rare diseases and the results of that analysis will be published soon. There was no intent to prioritize needs by disease or to emphasize needs in one population over any other.

Survey Respondents

In total, 1,342 clinicians received the survey, including 1,154 physicians and 188 non-physicians (827 members of the FDA Center for Devices and Radiological Health Advisory Committee,² 26 members of the FDA Pediatric Advisory Committee,³ 63 members of the FDA Pediatric Device Consortia,⁴ and 426 members of the NCATS/ORDR Rare Diseases Clinical Research Network program).⁵ In total, 588 completed the survey, for a response rate of 44 percent. The respondents reported expertise covering many specialties, and 33 percent had a pediatric focus (a pediatric specialty or significant experience with pediatric patients). A large majority (90 percent) reported they had direct experience diagnosing or treating patients with rare diseases or had knowledge of rare diseases. Of those with direct experience, 93 percent had seen such patients in the past two years.

Findings

The survey results clearly documented that patients with rare diseases face numerous unmet needs related to diagnostic and therapeutic devices. In addition, device needs of pediatric patients sometimes differ from those of adults. For example, devices must be able to grow with a child, be modified to a smaller size, or be less invasive. Overall, respondents believed that creating entirely new devices is what is most needed, rather than modifying existing devices or repurposing devices for other indications. The limitations of existing diagnostic devices included their lack of sensitivity and specificity and their cumbersome and invasive nature. Respondents noted that meeting therapeutic device needs would improve care for patients across all types of rare diseases. However, the costs of research and development, lack of profitability for industry, and challenges of conducting trials in small, heterogeneous populations stand in the way of progress in this area. Notably, genetic tests are essential tools necessary for the diagnosis and treatment of many rare conditions, and the critical shortage of such tests was mentioned repeatedly by survey respondents. Overall findings from physician respondents were similar to those from non-physician respondents.

² For more information about the Center for Devices and Radiological Health, visit <https://www.fda.gov/AboutFDA/CentersOffices/OfficeofMedicalProductsandTobacco/CDRH>

³ For more information about the Pediatric Advisory Committee, visit <https://www.fda.gov/AdvisoryCommittees/CommitteesMeetingMaterials/PediatricAdvisoryCommittee/>

⁴ For more information about the Pediatric Device Consortia grant program, visit <https://www.fda.gov/ForIndustry/DevelopingProductsforRareDiseasesConditions/PediatricDeviceConsortiaGrantsProgram/>

⁵ For more information about the Rare Diseases Clinical Research Network, visit <https://ncats.nih.gov/rdcern>

Key Findings

- **Clinicians overwhelmingly cited multiple needs for new or improved medical devices for diagnosing and treating rare diseases**



461 unique rare diseases were cited with 917 specifying unmet device needs
91% believed a new or improved device is needed
64% were dissatisfied with existing diagnostic and/or therapeutic devices

- **There is a critical need for entirely new devices rather than modifying or repurposing devices, which are often inadequate**



77% cited a need for an entirely new diagnostic and/or therapeutic device
23% cited a need for only modified or repurposed diagnostic and/or therapeutic devices

- **Existing devices have several limitations in diagnosing or treating rare diseases**



79% reported diagnostic devices for genetic disorders as an unmet need
37% currently repurpose an FDA-approved therapeutic device

- **Several impediments to developing new devices for rare diseases were mentioned**



74% saw the lack of profitability to industry as a large impediment
67% saw the cost of development as a large impediment

- **The Humanitarian Device Exemption (HDE) provides a helpful pathway for bringing devices to market, but there are obstacles to its use.**



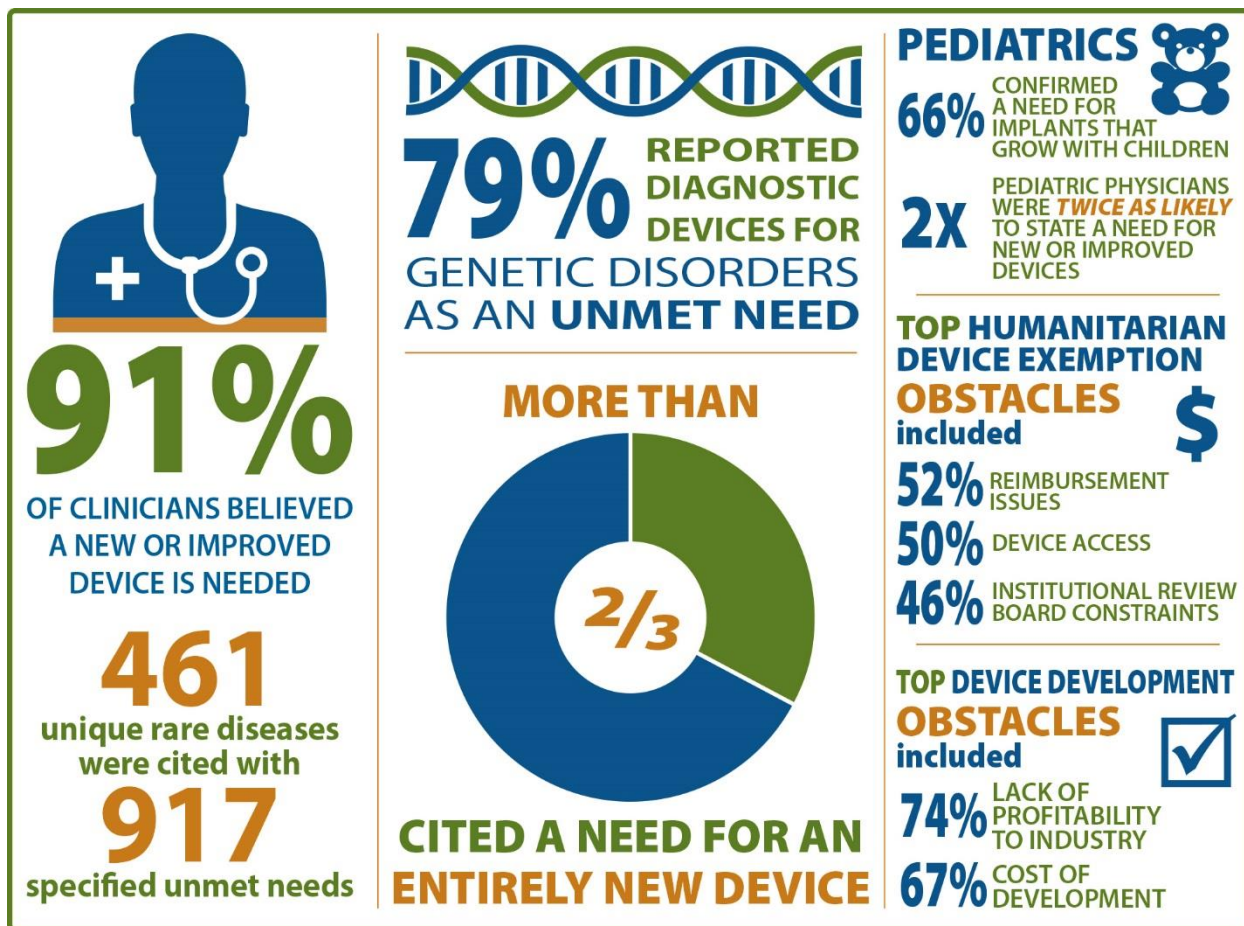
Top challenges cited by the 51% of respondents reporting familiarity with HUD/HDEs include the following:

52% said reimbursement
50% reported gaining access to HDE devices
46% indicated institutional review board constraints

- **While there are unique pediatric challenges, respondents with pediatric experience reported high levels of dissatisfaction similar to those without pediatric experience**



33% of clinicians had a pediatric focus
66% believed there is a pediatric need for implants that grow along with the child
44% confirmed intrathecal ports for drug delivery as a pediatric need



In summary, this national survey of government-associated clinicians verifies the need to develop devices for rare diseases and highlights the uniqueness of subpopulations. As described in the conclusion of this report, FDA and NIH provide programs to address these issues that encourage the development of devices for unmet medical device needs, as well as incentive programs that provide funding for the clinical development of products. Sustained support of the medical device ecosystem will accelerate the development of critically needed devices for rare diseases, thereby enhancing care options for these vulnerable patients.