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Redefining Rare Disease Care in the Digital Age: Insights and Key Takeaways from a Digital Health Symposium Focused on Empowering Rare Disease Communities

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Abstract
At the Stanford-UCB Rare Disease Digital Health Symposium held in Stanford, California, on September 8, 2023, researchers, clinicians, payers, thought leaders, and rare disease caregivers and advocates discussed the current state of care delivery and future perspectives of digitally-enabled care for rare disease patient populations. Digital health aims to improve healthcare delivery through novel ways of providing access to more precise diagnosis, monitoring of disease progression, treatment, prognosis, and care management for rare disease patients. The meeting focused on highlighting challenges and unmet needs, data infrastructure and analytics, the need for targeted and effective personalized therapies, and the importance of digital care transformation. The meeting also covered the social and ethical impact of access to digitally delivered, patient-centered care, as well as views on implementation and patient autonomy and empowerment.

Introduction

UCB, a multinational biopharmaceutical company, and Stanford University have made significant investment and progress in digital healthcare initiatives ranging from solutions that help disease diagnosis, symptom tracking, and outcome measures to disease management optimization. Stanford is recognized as a leader in digital health and has focused on the design and implementation of care transformation solutions to ensure that they are viable and can be integrated seamlessly into the patient's care journey. This event advanced conversations around the translation of digital health solutions into the real world by informing how to best integrate DHTs into clinical workflows so that solutions fit more naturally into care delivery workflows and practices as care delivery models evolve.

Proceedings

The day began with an introduction by Dr. Russ Altman (Stanford University, Stanford, CA, USA), who provided background on the UCB and Stanford partnership, as well as the genesis of the symposia. The objective of the symposia was not just to discuss the

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scientific and clinical challenges of care delivery in rare disease but also to navigate a 360° view of how the ecosystem must better reach and serve these patients.

After introductions, Dr. Kimberly Moran (UCB Biopharma SRL, Atlanta, GA, USA) spoke about the importance of care transformation in the rare disease space, which is often overlooked and under-resourced. Rare disease, as a category, is encompassed by small but many uniquely identified disease states that, together, encapsulate a broad swath of the overall patient population and significant portion of unmet need. Dr. Moran emphasized that with the power of data, analytics, and technology, we have great potential to better understand the complexity and individual nature of these diseases and then better support a more personalized patient journey. Taking a "how might we" approach, Dr. Moran suggested that we can imagine many use cases in which we can harness the power of technology and data to allow patients to better navigate their everyday lives. The event co-chairs, Dr. Ron Li (Stanford Medicine, Stanford, CA, USA), Emily Lewis (UCB Biopharma SRL), and Dr. Anuradha Dayal (Stanford Medicine), emphasized that holding this event at an academic institution is a privilege because it allows for a diverse group of stakeholders to come together and share ideas and to critically reflect on the changes that need to be made in the healthcare system in order to better serve this special patient population.

In his keynote lecture, Dr. Daniel Kraft (NextMed Health, San Francisco, CA, USA) discussed the potential of digital health to radically transform healthcare. He envisions a future in which healthcare is more digital, connected, crowdsourced, intelligent, data-driven, precise, equitable, and democratized. Dr. Kraft discussed a paradigm in which healthcare data is more personalized and accessible to healthcare agents. The hallmark of this paradigm is the convergence of many technologies, including robotics, nanotechnology, genomics, artificial intelligence (AI), quantum computing, and connected health, in an environment where hardware is becoming faster, smaller, and cheaper. He also shared a paradigm where the confluence of these technologies allows sick care to become true health care, and the episodic care we have grown accustomed to can become more integrated and continuous. What was once a "one size fits all," fee-for-service, paternalistic healthcare system is now becoming a more personalized, precise, AI-enhanced, and value-based system which is much more proactively focused on wellness.

Dr. Kraft then underscored the importance of data integration to move from clinical knowledge to clinical utility more quickly. We are at a stage where we need to figure out how healthcare data that we collect from many sources, such as health IT, clinical decision support, software as a medical device, telemedicine, extended reality, remote monitoring, wearables, screening and diagnostics, and population health, can coalesce into unified, actionable insights (e.g., a "check engine" light) that can provide patients, providers, and caregivers with nudges when needed. This will require a proactive, systems approach to better identify and understand the point(s) when an individual might transition from health to disease so that intervention can occur.

In conclusion, Dr. Kraft drew attention to the larger picture, based on the fact that a shift is occurring from the "quantified self" to "quantified health." This implies that now that we can quantify many things about our personal health, we can work toward actually being healthier. While it would be impractical to have an app or wearable for every condition, there is a need to begin developing consolidated and integrated solutions that will appeal to a wide range of users. These solutions should be personalized and have engaging user interfaces that align the incentives of all stakeholders. If this era comes to pass, it will empower clinicians to practice at the highest level of their licensure, reduce medical errors, and cultivate greater digital empathy as part of their "webside manner." In this trajectory, where healthcare is shifting from the doctor's office to home, phone, and on and in our bodies, we hope to see this reflected in improved clinician and patient experiences, better patient outcomes, and lower overall costs.

Stanford Center for Digital Health

The morning continued with Dr. Eleni Linos (Stanford Medicine) introducing herself as the new Director for the Center for Digital Health (CDH). Dr. Linos affirmed that, although the CDH has been in existence for some time, it is re-launching with a renewed vision for digital health research. The center plans to focus on research in rare diseases, among other interests. In support of this work, the CDH aims to be the connector that brings experts from across the university together with entrepreneurs, tech companies, and pharmaceutical companies to explore diverse and global perspectives. The center has several new programs including pilot grants, leadership lunches, a seminar series, and an industry affiliates program in service of this goal.

Current State of Care within Rare Disease

Dr. Anuradha Dayal moderated the first panel of the day, with Dr. David Bergman (Stanford Medicine) and Dr. Jon Berstein (Stanford Medicine) discussing the

odyssey that rare disease patients go through to find a diagnosis. Dr. Bernstein expressed his enthusiasm for the rapid pace of change within the rare disease field, as facilitated by the emergence of AI which has enabled an exponential discovery of new rare diseases in the past decade. Digital health has also allowed him to better identify, understand, and communicate with patients in ways that were not previously possible through digital connections such as telemedicine, mobile health apps, and remote patient monitoring tools. In the same way that someone new to speaking a foreign language might not be able to distinguish between two similar sounds, clinicians need to be cross-trained in multiple domains and exposed to diverse experiences so that they do not interpret clinical cases in a narrow-minded way. Moreover, he spoke about how AI can seamlessly aggregate and sort many data sources, allowing clinicians to keep abreast of the latest advances in scientific discovery. Ultimately, the advancement of technologies like AI is moving disease states that were previously thought to be "impossible" to treat toward the realm of "possible."

Dr. Bergman, a complex care physician, underscored that the legacy paradigm in which medicine functioned as a paternalistic meritocracy built on knowledge (e.g., that the physician imparted to the patient) is being inverted. He conceded that, particularly in rare diseases, patients themselves are often the experts on their disease. This knowledge imbalance and re-thinking of the doctor-patient (and family) relationship have meant that Dr. Bergman has started to take on a different role in facilitating communication between families and the roughly 6 or 7 other providers the patient might also be seeing. This is an area where he suggests digital health can help ensure that all key stakeholders are on the same page, involved in shared decision-making, and able to reach a common understanding of the necessary path forward.

Dr. Bergman also highlighted the inherent and unique challenges within the physician-caregiver-patient relationship when taking care of rare disease patients. Oftentimes, rare disease patients have impaired communication, which in turn shifts conversations around state of disease and quality of life to discussions between the physician and parent – thus, the patient themselves cannot participate in meaningful discussions around their own care. In addition, physicians who take care of rare disease patients need to have difficult conversations with parents and families regarding a shared mental model for disease trajectory, potential for clinical improvement, and sometimes initiating discussions regarding palliative care. He then stressed that technology like AI can help healthcare providers take a more equitable look at their patients by standardizing treatment recommendations based on more diverse and representative datasets of specific rare disease patients, which reduces variability and increases equity in care delivery. As a physician taking care of a patient with a rare disease, they might only encounter 1 or 2 patients in their career lifetime with the same diagnosis. AI could help "re-calibrate" care delivery by collectively detecting patterns and progressions of specific rare diseases and presenting how to effectively address them in their clinical decision-making while providing explainability interfaces that allow clinicians to understand the reasons behind their recommendations and offer insights for continuous improvement.

Empowering patients and their caregivers was a consistent theme in this panel, and Dr. Bergman highlighted that we should seek opportunities to empower both groups to translate the knowledge they receive into personalized and actionable decision-making. Care coordination was also highlighted as an unmet need, especially when children age out of pediatric care. Digital health has the potential to close this gap by connecting patients to the right clinician within their care team at the right time or matched with a more suitable age-appropriate care team across the lifespan. Dr. Bergman also shared his hope that powerful analytics could lead to predictive algorithms that alert families to more actionable insights in real time, particularly in cases where there may be a high risk of a significant impending medical event occurring.

At this point in the panel, two mothers of rare disease patients, Nancy Buhr (Stanford Children's Health, Palo Alto, CA, USA) and Corina Provencher (Stanford Children's Health), were brought to the stage to discuss their personal experiences. They shared the challenges of raising and advocating for their children in various every day and emergent capacities. In articulating their needs, they mentioned they would love digital tools which could continuously monitor relevant symptomatology, streamline the collection and sharing of data across their healthcare providers, and simply connect them to other families to foster a sense of community.

While discussing the DHTs that are currently in use, such as wearable devices for remote patient monitoring, the panel questioned who within the care team should be responsible for monitoring the data. Dr. Bergman argued that, in an ideal scenario, it would not be the care team at all but rather the patient or caregiver's responsibility to monitor these data. We can leverage and develop digital health tools to empower individuals to know what to do,

who to call, and where to go when they are alerted to evolving data measurements which could indicate the need to escalate care.

Using Multi-Modal Data to Identify and Diagnose Rare Disease Patients

Danielle Hamel (Tempus Labs, Inc., Chicago, IL, USA) commenced by sharing a startling statistic from Global Genes that it can take the average rare disease patient 6 to 8 years to get an accurate diagnosis. We are now seeing this number come down to around 5 years [1]. Hamel then explained how a platform approach can help reduce this timeline by curating, de-identifying, structuring, and connecting multi-modal data from disparate sources and mapping it to clinical meaningfulness in terms of therapeutic response and patient outcomes. What is unique about Tempus' products is their ability to pair unimodal clinical tests and products with complementary information to provide a more comprehensive picture of the patient. A better understanding of the individual patient then begets a capacity for better detection of patterns across patients. By leveraging clinically redundant information, Tempus can prioritize more informative but potentially less expensive or invasive tests by using one modality to inform or predict the result of another. Going forward, Tempus intends to double down on AI-enabled comprehensive care by closing care gaps, augmenting workflows, and catalyzing multi-disciplinary care.

Current State of Digitally Enabled Care and Remote Patient Monitoring

Dr. Ben Vandendriessche (Byteflies, Antwerp, Belgium) opened by discussing the current challenges in care delivery, namely point-of-care solutions which are expensive and intermittent as well as patient self-reporting practices which are subjective and generally garner low adherence rates. The progression of care from fragmented to centralized has led to a focus on virtual, patient-centered care enabled by the home environment.

Although Byteflies has its roots in remote patient monitoring for clinical trial settings, they now have a "remote patient monitoring (RPM) as a service" offering called Care@Home, in which providers can prescribe days to weeks of at-home monitoring. Byteflies leases and ships monitoring equipment directly to patients' homes to collect medical grade data on demand. Cloud services enable them to annotate data and perform AI-enabled workflows to derive diagnostic insights. These insights are then made available to the prescribing provider in a dashboard for clinical decision-making in a user-friendly way. The offering also comprises EMR integration for clinicians and a companion app for patients to view the insights. What is special about Byteflies' program is that it was co-developed with healthcare providers (HCPs) and patients to meet the needs of specific care paths within a therapeutic area. More specifically, it was designed to be compatible with associated clinical workflows to improve important clinical metrics like time-to-diagnosis and time-to-treatment.

Using Electronic Medical Record Data for Robust Rare Disease Research and Drug Development

Nasha Fitter (Ciitizen, Palo Alto, CA, USA) and Dr. Justin West (KCNT1 Epilepsy Foundation, Contoocook, NH, USA) began by sharing their stories as caregivers of children with FOXG1 and KCNT1 epilepsy, respectively, and the tremendous challenges they face in their children's activities of daily living such as bathing, feeding, and sleeping. In addition to daily care-taking, rare disease families are also starting advocacy groups, forming scientific advisory boards, creating natural history registries and biobanks, speaking with the pharmaceutical industry, raising funds for drug development, and learning the regulatory process behind research and development.

In discussing the particularly high mortality rate for diseases such as KCNT1 epilepsy, Dr. West emphasized the urgent need for life science companies to bring therapies to market. Invitae is a company which aims to re-imagine how to obtain data for 5- to 10-year natural history studies and eliminate phase III clinical trials for ultra-rare diseases. By utilizing innovative, cost-effective platforms to collect data and track trial endpoints at clinic visits their Ciitizen platform can help accelerate approvals with regulatory bodies like the FDA. Built with advocacy groups, the platform follows the patient and requests medical records from all providers the patient sees. Raw records are processed by a machine-learning engine to extract, normalize, and codify defined data elements from which de-identified data output is structured and ready to share for research extraction based upon ontologies to create natural history studies. Caregivers can also access these data to download and use for second opinions.

Fitter urged both clinicians and pharmaceutical companies to create working groups with patient advocacy groups to come to a consensus on data standards across standard of care visits so that these data can also be used in a research context. More specifically, there is also

a particular need for standardized questionnaires that are asked across all related rare diseases as well as disease-specific questions which can map to clinical endpoints.

Lightning Round: Solution Showcase

Dr. Charles Fisher (Unlearn.ai, San Francisco, CA, USA) started the showcase outlining the difficulties that various life sciences can have when many of them are enrolling clinical trials in the same patient population. He also reminded the audience that not only is it difficult to enroll trials in a timely manner, but in most cases, half of those enrolled participants will be randomized to a placebo group and never receive the experimental treatment. Thus, there is a tremendous interest from life science companies in reducing the number of participants on placebo or replacing the need for participants on placebo by using natural history studies as external controls, synthetic data, etc. Dr. Fischer asserted that while we cannot run single-arm studies, we can create digital twins of clinical trial participants which allows companies like Unlearn.ai to focus on maintaining the power of clinical studies while shrinking the size of the control group or increasing the power of clinical studies without increasing the sample size in phase II and III trials.

Indu Navar (EverythingALS, Los Altos, CA, USA) spoke about her technology non-profit, EverythingALS, created in honor of her husband Peter Cohen. EverythingALS aims to foster collaboration across stakeholders (e.g., namely the patient community, researchers, technologists, and pharmaceutical companies) in a meaningful way at a time in which we are reaching an inflection point for digital biomarkers. The network includes a pharma consortium and partners like academic medical centers, technology companies, and startups to harness the power of digital biomarkers. It is her mission to more accurately identify appropriate subsets of patients for research, clinical diagnosis, and tracking of disease progression. In their current work, EverythingALS is coupling digital breathing, movement, and speech biomarkers with analytics using data science and artificial intelligence to accelerate finding treatments and a cure for amyotrophic lateral sclerosis (ALS).

Focused on applying digital measurements early, EverythingALS is ensuring that diagnosis can happen faster so that patients are not too sick to qualify for clinical trials. The spirit of EverythingALS is citizen-driven research with an open innovation approach, enabled by a platform approach for promoting data sharing and analytics. With an ambition to partner with many sensor companies, they aim to coalesce sensor data with many other modalities including voice and video. They have built an app which provides a dashboard for patients, physicians, and pharmaceutical companies to access the data in formats best suited for them. For patients, there is a daily journal and progression tracker, as well as a conversational large language model agent which can be used to query their own data.

Onno Faber (Rarebase, Palo Alto, CA, USA) then spoke from his personal experience as both a patient and an entrepreneur, catalyzing the rare disease community to action utilizing a "turning many small problems into a big one" strategy. He began by outlining many problems that he sees requiring better solutions. These included diagnosis, finding the right clinic or doctor for immediate care, connecting to the community, identifying potential therapies, enabling clinical research, and finding ways for patients to live with the consequences of their disease by better adapting to their circumstances. Faber voiced a fundamental need for more ideas to explore in the research space as a starting point, believing that expanding the amount of ideas might expand the likelihood of finding a solution. He also voiced a need for a shared infrastructure which pools resources and tools across rare diseases and allows key stakeholders to learn from each other.

Stefanie Eichner (FDNA, Sunrise, FL, USA) introduced FDNA as the world leader in AI-powered image analysis to aid in diagnosing developmental and genetic disorders. Their mission is to enable pediatricians, school nurses, and even parents to perform some of the time-consuming data collection tasks, freeing the specialists to focus on relevant clinical diagnostics and therapy. With a data-driven, mobile-first solution, they empower parents to be better advocates for their children and, in doing so, get better access to care in a more timely manner.

Their app, called the Child Development Checker, aims to shorten the diagnostic odyssey for rare disease patients significantly. The app asks about each child's behaviors and symptoms, then allows the addition of a "selfie picture" which is then analyzed with proprietary AI algorithms to identify diagnostic findings. The app also provides helpful content about the potential health conditions that may be associated with the information provided and suggests referrals to specialists. FDNA also boasts a clinician-facing platform called Face2Gene which is a clinical decision support tool incorporating data from the Child Development Checker app. In Face2Gene, the clinician can include additional clinical data, make assessments, collaborate with other specialists, order targeted testing, and compare diagnostic options to confirm a differential diagnosis. With the synergy of these two tools,

FDNA hopes to bring value to the healthcare ecosystem with a return on investment for providers, pharmaceutical companies, and contract research organizations alike.

Enabling Inclusive Care Models and Adoption at Scale

The day concluded with a panel moderated by Dr. Ron Li and Dr. Anuradha Dayal. The panel featured Rob Sederman (Ambit, Inc., Morristown, NJ, USA), Dr. Marc Overhage (Elevance Health, Indianapolis, IN, USA), Effie Parks (Once Upon A Gene, Mercer Island, WA, USA), Dr. Topher Sharp (Stanford University), Isil Arican (Stanford Children's Health), and Michael Graglia (SynGAP Research Fund, Inc., San Diego, CA, USA).

The panel began with a deeply human perspective, asking for those involved in the ecosystem to truly enter the rare disease world by connecting with a person who has a rare disease and listening to their story. These patients and families want to be seen and heard not as a monolith or a single data point, but as a whole person. This sentiment has also been shared by industry as they begin shifting to use more tools like "whole health indices" which take into account additional factors like social needs, communication, and engagement, but there are still many more factors that need to be considered to be representative of the whole patient (and their caregivers) and their lived experience. To this end, it was also mentioned that designing products for caregivers, in addition to patients, is an often overlooked unmet need.

Continuing with the theme of patient empowerment, it was discussed that, even with HIPAA right of access, patients are sometimes still at the mercy of their clinicians to obtain access to their own health data in a digestible and understandable format. Beyond information and resource sharing, there is a need to educate patients and their caregivers on how to speak with clinicians more effectively with these data and determine which pieces of information are most relevant. There was an assertion that the healthcare system is collectively still doing medicine "to" patients rather than "with" patients, and digital tools can facilitate the connection needed to empower those on the patient and caregiver end. In cases where patients and caregivers do have access to some electronic health records, it is impossible for them to find what they need in the sea of documents. As a proposition, Gragglia suggested that greater weight should be given to more consequential documents within patients' medical records (e.g., a causative genetic mutation leading to a life-changing diagnosis), triggering them to rise to the top of the documentation for ease of access.

Those present reflected on the fact that EHRs were developed for billing purposes but are not necessarily designed for clinical workflow, patient communication, or general usability across many stakeholders. Changing clinician workflows alone can change care delivery processes in the clinic entirely, so it is critical to be thoughtful about introducing change in the existing systems. There is heterogeneity in the way care is delivered across different practices depending on their size and location, which adds an additional layer of complexity.

For those looking to share EHR data, there is still data standardization and integration problem as data is coming into medical record systems from many different places, and each institution may have their own way of structuring it. Foundationally, the digitization, cleaning, and mapping of data within and outside EHR systems will be critical to being able to extract timely, meaningful insights as state-of-the-art machine learning algorithms come online. While decision-making around the improvement of EHR systems can be very challenging, sentiment coalesced around the central principles of making the system easier, putting the patient at the center, and ensuring data sharing is fluid while recognizing safety, privacy, and other common sense bounds.

Reflections in the latter half of the day centered around how the complexity of rare disease can bring societal problems, business problems, and process problems to the fore in a way that transcends the clinical realm and enters financial and operational dimensions. In terms of operational hurdles, it was raised that some monogenic diseases do not even have ICD diagnostic codes for which potential therapies could be billed against. While there are several programs providing free genetic testing for patients, we still do not have the diagnosis codes associated with all pathogenic variants for billing purposes or to identify individuals who might qualify for a clinical trial. Having codes for all disease states, and especially those in rare cases, is important because it will allow health systems to have better counts of incidence and prevalence of disease. These accurate counts can change the calculus in boardrooms where decisions on company pipeline are made. Interestingly, in rare conditions which recently received diagnosis codes, counts for those diseases skyrocketed which speaks to the need to identify the ground truth of rare disease epidemiology.

In addition to coding challenges, the panel raised the issue of the administrative burden that EHRs have put on physicians as the systems have been opened up for patient sharing. On average, doctors spend around 2 h in administrative documentation and patient questions for every 1 h of face-to-face care delivery. Generative AI in

particular was called out as a technology that could help reduce this burden by starting tasks like drafting prior authorization letters or response messages to patients. While all tasks would still have a human-in-the-loop to vet what the assistant generated, AI could allow clinicians to have a head start in tackling some of the more banal day-to-day tasks. While automation bias is a real concern, user-centered design, education, and training can bring awareness to these risks to mitigate them.

The general consensus, as the symposium began to conclude, was that we must begin by driving change where it is feasible. Strategically, we can take a common-sense approach to focusing on solving unmet needs within the rare disease population that also generalize to broader patient populations. Moreover, it was suggested that even taking small steps like feasibility trials where digital health tools are deployed in small cohorts of rare disease patients could be meaningful in these populations where digital data may not even exist. The panel acknowledged that in areas where data and technology do exist, there is actually a need for more collaborative efforts using them. Needs also differ depending on which rare disease is in question.

In closing, the greatest rate-limiting factor in servicing the needs identified was time to scale. We must find diseases that have overlapping symptoms, similar treating specialists, and common referral pathways and begin there to impact the largest number of rare patients. We also need to re-examine incentives and increase practices around data sharing and transparency. Ensuring that every stakeholder has a more complete picture of what is occurring in the health system will allow for a greater sense of ownership and accountability in the ecosystem.

Summary and Conclusion

Bringing together leading experts in the digital health space to share their views and future outlook on emerging trends, systemic impacts, and alternative futures has been invaluable. Events such as these underscore that digital strategy and vision must be aligned across and co-created with the greater ecosystem. With foresight, we can proactively inform next-best actions and shape the development and governance of advances in the space of innovative medicine in rare disease.

We believe you cannot reinvent until you reimagine, so we started there. Today's world can be uncertain, complex, and ambiguous as the pace of technological innovation and scientific advances hastens. Innovators must practice flexibility and adaptability to keep pace and make meaningful contributions. We recognize that a multi-stage, multi-layered strategic planning approach that is pressure-tested and refined by a diverse group of experts is essential to advancing flexible, participatory, and patient-centered models of digitally enabled care for rare disease patients.

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