Transforming Data into Breakthroughs: 5 Years of RDCA-DAP Journey and Impact

Founded in 2019, the Rare Disease Cures Accelerator-Data and Analytics Platform (RDCA-DAP) is an FDA-funded initiative that provides a centralized and standardized infrastructure to support and accelerate rare disease characterization with the goal of accelerating therapy development.

Alexandre Bétourné, Ph.D., Pharm.D., is Executive Director for the Rare Disease Cures Accelerator-Data and Analytics Platform initiative and previously served as its Scientific Director. Dr. Bétourné works with the RDCA-DAP team to expand its reach into new diseases areas, accessing new data and enhancing C-Path's relationships within the rare disease community. He holds both a PhD and a PharmD from the University of Toulouse in France, has three patents, and has written multiple papers that intersect with several different rare disease areas.

In honor of Rare Disease Month, Alex took some time to sit down and discuss the impact RDCA-DAP has made in the last five years, the collaborations it has fostered, the work of the Rare and Orphan Disease Program, and his biggest point of pride in the program.

In the five years since the RDCA-DAP initiative was launched, what do you think has been the platform's biggest impact for rare disease drug development?

RDCA-DAP currently hosts datasets for over 40 diseases and thrives to make it available on our public interface. As we continued to grow this database over the years, we've seen a growing number of accredited users actively exploring what is made available. This is a success by itself since making data more visible to scientists over the world is critical. Further, we have opened dozens of workspaces where our users are able to access fully anonymized and deidentified patient-level data to perform approved scientific projects. Industry, academics, healthcare companies, and regulators are actively leveraging the platform tools to inform their clinical activities, including trial design and endpoints development, and overall advance their understanding of diseases and disease progression. This is tremendous and will ultimately lead to more treatments for patients, as best exemplified by the first drug approval for Friedreich's Ataxia, which was supported by specific analyses done by the company on the largest database available for the disease; we made this available due to our collaborations with the Friedreich's Ataxia Research Alliance (FARA).

In the five years since RDCA-DAP was formed, what do you think have been the biggest signs of progress in your mission to break data silos?

I think rare disease patients and foundations, for the most part, have always been aligned with our mission to break silos in data sharing. Overall, patients want to share data and want that data to be used. From my direct perspective interacting with a broad array of rare registries and groups, advanced or very early in their efforts, I'm seeing a greater understanding of how this can be achieved. I see a lot of new groups implementing de-identification systems, working on consent language that will ensure data is shared, and thinking very prospectively about how they can maximize the utility of the data collected from patients. This, hopefully, will facilitate future data sharing to platforms like RDCA-DAP. We are also seeing a lot of our discussions and collaborations leading to the building of partnerships with other data platforms. This is critical going forward, as silos will continue to occur if we don't foster an ecosystem where rare disease data platforms collaborate and develop means to share data between platforms or support federated analytics.

What is something that someone living with a rare disease, is a care partner for someone living with a rare disease, or is an advocate for those living with rare diseases, should know about the work being done by C-Path's Rare and Orphan Disease program?

This could seem very obvious but the first thing that comes to mind is that we are a rare disease effort. What we do at C-Path is very unique and might sometimes be hard to understand for those that don't know us. We work for rare disease patients and everything we do is aimed at lifting roadblocks to drug development, so that more treatments will be available. Whatever we do is guided by this final goal, and in pursuit of that goal, we constantly engage and work with the entire rare disease space. We talk with patients and advocacy groups all the time, and it is key to our mission to involve them in our discussions and work. When it comes to sharing data to the RDCA-DAP, I would like to convey to patients that their data is safe. Of course, we provide a very secure place to store data and share it with scientists and only gather de-identified data to protect their anonymity. In addition, speaking as a member of our data use committee that reviews the users' requests for access to data, I have never seen any request that was ill-intended. All the approved work that is being conducted is to the benefit of science, drug development and, ultimately, patients. The platform is helping people, and I'm convinced that the output from our users will, in the long term, prove to be of major significance for rare and orphan diseases patients.

What is the biggest accomplishment or point of pride you have from working with the rare and orphan disease program?

We are here! The platform was opened to the public 3 years ago with a tremendous number of successes. We were able to build a very large database, and scientists are actively engaging with the platform. We have also built and expanded our activities in rare diseases to develop a very compelling program, with multiple small- or large-scale public-private partnerships working to facilitate drug development in Friedreich's Ataxia, Progressive Supranuclear Palsy, Spinocerebellar Ataxias primary mitochondrial diseases, Limbgirdle Muscular Dystrophy, Duchenne, Huntington disease, Polycystic Kidney Disease, Lysosomal Storage Diseases, Alpha-1 Antitrypsin Deficiency and ALS. Each of these efforts are advancing one or multiple solutions that will accelerate the development of drugs for patients. Above all, my biggest point of pride is that our program (and colleagues) is always looking onward. We want to diversify our activities to help as many rare diseases as we can. We want to continue to grow our databases to sustain the development of more solutions to help patients. We want to grow our collaborations and lead an ecosystem that facilitates conversations, partnerships and facilitates drug development.