RAREBASE

Unleashing the power of AI on drug hunting for rare diseases

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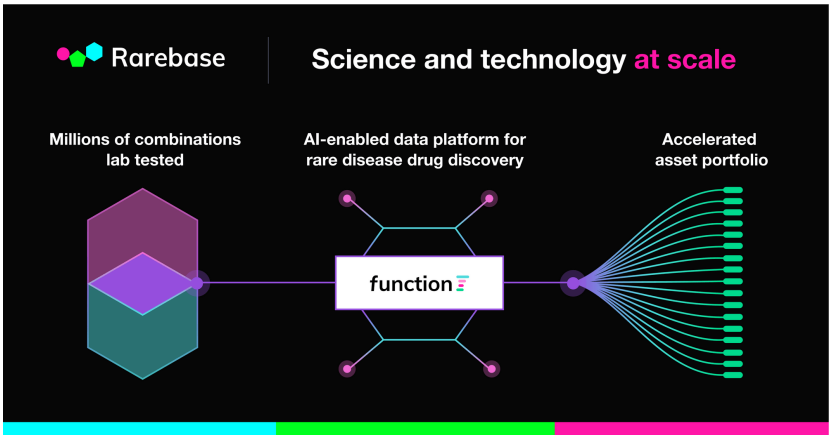

When Rarebase Director of Software Engineering Clayton Mellina first met Co-founder Onno Faber, he had just started a new engineering job at Google. He saw an upcoming genomics and AI event happening in San Francisco — an open genome hackathon for a patient with a rare auditory tumor disorder called neurofibromatosis

four or five years earlier.

Meeting rare disease researchers and integrating into the patient community sparked Mellina's journey to join Rarebase in the fight against rare diseases. He believes a patient-centric approach, coupled with breakthrough technologies in machine learning and high-throughput biology, will drive a revolution in human health.

Read on to discover Mellina's insights into the future of AI and how Rarebase scientists use data engineering to reimagine rare disease drug discovery.

Optimized data interpretation with machine learning

Machine learning helps interpret the wealth of biological data that comes out of the Rarebase laboratory, says Mellina, like analyzing cell images from a microscope.

"Images themselves are an important type of biological data. We have a high-content microscope in the lab that produces images that need interpretation, and we apply computer vision algorithms to extract features that are helping us interpret the underlying biology."

> **bring [experimental] modalities together, allowing us to jointly interpret all the types of data we're observing from biological systems," says Mellina.**

While images taken by a microscope help visualize limited aspects of cell biology, next-generation sequencing techniques can take a much deeper and broader dive into cell biology and drug response that are more translatable. The Rarebase Function™ product engine utilizes high-throughput drug screening and gene expression sequencing to examine thousands of drug candidates against virtually every gene expressed in the cell.

These experiments have generated a premiere drug-hunting dataset — and it takes AI applications to harness the full potential of that dataset towards rare disease drug discovery. "Machine learning is well-poised at this moment to bring [experimental] modalities together, allowing us to jointly interpret all the types of data we're observing from biological systems," says Mellina.

Drug discovery has always been resource-intensive, with billions of dollars invested and many years of work involved. The traditional approach has been a "jackpot model", explains Mellina, where researchers search for a molecule that can treat a specific patient population.

“The whole business model is based around this idea of you're going to end up with a lottery ticket at the end of clinical trials and see if your number comes up,” says Mellina. “Usually, it takes a lot of resources, billions of dollars, and many years — it's an expensive lottery ticket because at the end of clinical trials, you might come up empty, and that frequently happens.”

This model is not only expensive but also inefficient as it does not make use of the wealth of data available. By harnessing the power of data, drug discovery can be made more efficient and scalable. Data engineering enables drug hunters to identify promising compounds while minimizing the cost and time required for drug discovery.

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> **definitely is not in any way captured in this traditional model."**

The enormous cost of drug development can leave some patients behind. Drug developers typically fund the discovery process by “targeting a reasonably large patient population to [generate] something worth a jackpot — worth the effort and the cost,” explains Mellina. “In that story, where's the patient?”

Bringing innovation to rare diseases requires a strategy that pools resources to maximize efficiency in the discovery process. We must glean the most information from the data generated through these efforts and think on a larger scale for smaller patient populations.

“I think that there is an opportunity to extract signals from the full life cycle of human health in a way that's not done yet and definitely is not in any way captured in this traditional model,” says Mellina.

AI applications allow Rarebase scientists to pull data insights with greater efficiency, a synergistic partner to the scaleable experiments performed in our laboratory. Function utilizes these tools to

aligns a lead compound with diseases that may benefit most from its mechanism of action, driving research forward across an entire landscape of rare diseases.

The future of AI and data engineering for rare disease research

Data engineering has the potential to revolutionize drug discovery by making it more efficient, scalable, and patient-centric. AI facilitates the interpretation of large datasets cheaply and at scale, maximizing the knowledge gained from research efforts. Future applications may see AI incorporated as a virtual research assistant in laboratories, says Mellina.

“It's not at all too far to imagine a 5-10 year timescale where there are research assistant AIs that are reflecting synthesized knowledge about the experiments that we've run in the lab, pulling information from the body of scientific literature that might be relevant in aiding the scientific process.”

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> **same direction.”**

Rarebase will continue to leverage this revolutionary technology to accelerate drug discovery with our innovative AI-enabled product engine. This practical approach provides scalable, efficient drug discovery focused on rare disease-modifying therapies with a vision to help humanity through constant and impactful innovation.

As AI continues to grow and empower our research, the future of rare disease drug discovery looks brighter than ever before. “We have a lab, we have scientists, we have a need, and we have the opportunity to point those things in the same direction.”

Contact us to learn more.