Giving hope to children with rare disorders

TGen helps researchers identify gene mutation with a fast, powerful Dell EMC HPC cluster

Life Sciences | United States

Business needs

TGen's Center for Rare Childhood Disorders sought to discover gene mutations by using a faster, more powerful technology platform for genetic sequencing.

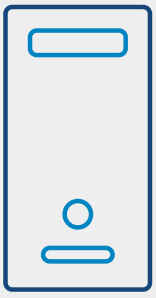
Solutions at a glance

- Data Center
- High-Performance Computing
  - Dell EMC Genomic Data Analysis Platform

Business results

- Accelerates genetic analysis
- Gives new hope to families suffering from symptoms related to the mutation
- Collaborates with other researchers to identify PYCR2 gene mutation

Supports
1 million
CPU hours monthly



Develops custom treatments
1 week
faster

For 18 years, Basil Pais and his wife struggled to find a diagnosis for their daughter. Born a healthy baby, she first started exhibiting problems at 3 months: she struggled to put on weight, she had a blank stare, and then she developed a staph infection. The infant was failing to thrive, but doctors couldn't figure out why.

Over time, because of progressive neuromuscular problems, she became unable to walk and talk and has suffered through an endless string of infections and surgeries. "We have worked with multiple specialists in different states, but tests always came back negative," says Pais. "It's been incredibly frustrating. Doctors just couldn't figure out what was going on with her. We are so committed to helping her, but this has been hard on all of us."

This is the type of challenge facing researchers at the Translational Genomics Research Institute's (TGen) Center for Rare Childhood Disorders. Every day, TGen bioinformaticians, geneticists, and clinicians perform genetic sequencing in an effort to identify diseases and help more people survive them. "We are committed to helping people like Basil's daughter. We never give up on trying to find a diagnosis," says Keri Ramsey, clinical co-director for the Center.

However, to accurately diagnose disorders, TGen needs the right high-performance computing (HPC) systems to quickly run extremely complex algorithms that analyze multiple terabytes of genetic and molecular data on a patient. "A single genome measures up to 4 terabytes, and sequencing that genome could take weeks to complete," says James Lowey, TGen's chief information officer. "It's also very difficult to try to determine the role of the thousands of genes we sequence." To effectively conduct its work, TGen requires an HPC cluster that provides the right amount of server performance, storage capacity, network bandwidth and availability.

> *"We waited 18 years to receive a diagnosis, so psychologically that makes a huge difference for us. Even though we don't have a cure yet, or even a treatment course, we feel like it's a first step toward getting our daughter the help she needs."*
>
> Basil Pais, Patient's Father

"By identifying this gene mutation, with help from our Dell EMC HPC cluster, TGen will potentially be able to identify other children who have the disorder."

Keri Ramsey,
Clinical Co-director, TGen Center
for Rare Childhood Disorders

Accelerating genetic analysis with a powerful Dell EMC HPC cluster

TGen researchers originally sequenced the Pais family's genes in 2012, but they could not find the genetic mutation responsible for the child's disorder. "We realized we needed to have more data available to us and better technology to facilitate the genetic sequencing," says Ramsey.

To address its needs, TGen implemented an HPC cluster based on the Dell EMC Genomic Data Analysis Platform. The cluster includes 96 Dell EMC PowerEdge M420 server blades with Intel® Xeon® Processor E5-2470 v2 processors, contained in three Dell EMC PowerEdge M1000e chassis. "We get 512 cores in 10U using the PowerEdge M420 servers," says Lowey. "That's a lot of power in a very compact footprint." The organization also relies on Dell EMC Networking S4810 switches at the top of the rack to facilitate cluster component communication.

The technology gives TGen a highly scalable cluster that helps the organization accelerate analysis and create personalized treatments faster. "Scientists and doctors can come to conclusions faster by processing more data in a shorter time frame, because Dell EMC builds systems to enable that," says Lowey.

Discovering the PYCR2 gene mutation

Utilizing its Dell EMC HPC cluster, in 2016 a team of TGen researchers led by Drs. Matt Huentelman and Vinodh Narayanan collected genetic sequencing data from the Pais family and were able to diagnose the daughter with a mutation in the PYCR2 gene. This occurred after a paper in the *American Journal of Human Genetics* had been published by researchers at Boston Children's Hospital on children with the same disorder. The team at TGen has since collaborated with the group at Boston Children's Hospital by sharing biospecimens in order to gain insight into this rare disorder.

"Because of our understanding of scientific advancements, as well as the capabilities we have with the Dell EMC HPC cluster, we were able to go back and analyze the sequence data for the family again," says Ramsey. "We finally made the connection between the gene mutation and the child's disorder. This was just the third family identified with the disorder at the time, which shows how rare it is."

Finding hope in a diagnosis

Although there is still no cure for his daughter's disease, Basil Pais still feels like a burden has been lifted. "We waited 18 years to receive a diagnosis, so psychologically that makes a huge difference for us," he says. "Even though we don't have a cure yet, or even a treatment course, we feel like it's a first step toward getting our daughter the help she needs."

The discovery also stands to benefit other families. "By identifying this gene mutation, with help from our Dell EMC HPC cluster, TGen will potentially be able to identify other children who have the disorder," Ramsey says. "We could possibly create a cohort of individuals who could participate in clinical trials for drugs that could help treat their symptoms." Adds Pais, "There are probably hundreds of cases like my daughter's, all of which might be able to be identified. The work TGen and Boston Children's Hospital have done is paving the way for more discoveries in the years ahead."

Supporting 1 million CPU hours each month

TGen researchers are taking advantage of the Dell EMC HPC cluster to develop custom treatments at least one week faster than before. "Our partnership with Dell EMC has enabled us to use the latest technologies to log 1 million CPU hours per month on our cluster, which has helped increase the volume and speed of our research," says Lowey. "Processing that amount of data was a massive challenge before we implemented this high-throughput, high-speed cluster."

Powering future research

TGen's Center for Rare Childhood Disorders is confident that it can use its Dell EMC HPC cluster to facilitate new research going forward. "We have the ability to more quickly perform genetic sequencing and meet the demand for processing increasing data volumes, because of the Dell EMC HPC cluster," Lowey says. "And this technology also helps us ask harder questions of the data, and hopefully answer those questions. We are committed to helping children with rare disorders, and we are better equipped to do that with this solution."

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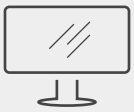

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