Application: 4843

Baylor Genetics Genomic Technology Team

Page: General Information

Provide information about the company to be considered for the award. If you will be nominating an individual, specify the nominee's employer.

Name of Organization/Company

Baylor Genetics

Additional Contacts

I would also like to have others receive emails about the disposition of our entries.

Page: Entry Information

Entry Title

Baylor Genetics Genomic Technology Team

Category

G08. Technology Team of the Year - Biotechnology

Technology Team Submission Format

Written Answers

a. Briefly describe the nominated technology team: its history and past performance (up to 200 words). Required

For 45 years, Baylor Genetics has been at the forefront of genetic testing, empowering patients, providers and partners with answers that matter. Stemming from academic roots at the renowned Baylor College of Medicine, the company offers a spectrum of genetic testing, research and lab services to help patients and families make more informed decisions about their health, and help healthcare providers better identify, treat, and prevent disease.

Baylor Genetics' Genomic Technology team offers a variety of genetic testing products and services, with a particular expertise in Whole Exome Sequencing (WES) and Whole Genome Sequencing (WGS). These are single tests that can analyze a patient's genetic information and detect a wide spectrum of genetic abnormalities. They are among the fastest and most comprehensive precision diagnostics tools available today and are suited to patients who have conditions that are difficult to discern and require a broad view into potential underlying causes.

At its state-of-the-art 73,000-square-foot lab at the Houston Medical Center, the Genomic Technology team oversees each critical element of the diagnostic product development process, covering: R&D (analytical algorithms and best practices development); Diagnostics Digital Platform (technology development and transfer to enable large-scale production); and Quality Assurance and Live Production Support.

b. Outline the technology team's achievements since the beginning of 2022 that you wish to bring to the judges' attention (up to 250 words). Required

After extensive clinical validation, the Genomic Technology team adopted NovaSeq X – the latest and most powerful model sequencing system, which delivers extraordinary throughput and accuracy to perform data-intensive applications at scale – and is the first lab in TX to utilize the tool in a clinical setting.

The Genomic Technology team has created a workflow that ensures the highest quality and precision at every step. By integrating various patching points between instruments and the individuals running those instruments, the workflow is streamlined and interconnected, with event-driven, large computation of data to support data exchange and flow between different applications.

Additionally, the team has worked to vastly improve the analytical bandwidth, turnaround time and processing by 30-50%, and expanded data volume by 60%. Not long ago, Baylor Genetics processed about 12 thousand carrier screenings per month, but now the team processes over 20 thousand – a 60-70% gross improvement.

The team continues to streamline processes, further increase accuracy, and scale up tests to provide trustworthy results quickly. Their efforts include the incorporation of high-accuracy artificial intelligence, referred to as explainable artificial intelligence (XAI), to prioritize variants that are most likely to solve a case and to support its genetic findings. XAI and informatics automation are enabling Baylor Genetics to scale up its lab operations and clinical whole-genome sequencing (WGS).

c. Explain why the achievements you have highlighted are unique or significant. If possible compare the achievements to the performance of other players in your industry and/or to the team's past performance (up to 250 words). Required

Adoption of the cutting-edge sequencing platform, NovaSeq X, represents a significant leap forward and allows the team to take on increasingly complex and ambitious ultra-high-throughput projects. Baylor Genetics is one of the first few labs in the country to take this ambitious step.

Incorporation of XAI and informatics paves the way for significant streamlining without sacrificing accuracy. The previous workflow for whole exomes required manual evaluation of 400 to 700 variants per exome – a lengthy and burdensome process. Baylor Genetics recognized that variant interpretation for WGS posed a bottleneck and scalability challenge that would be unsustainable in the long term.

To scale up testing and improve turnaround time, the lab validated and implemented an automated workflow. Following sequencing, secondary analysis software makes variant calls, including complex structural variants, which are fed into tertiary analysis for interpretation. The XAI compares variants with a subject's phenotypic information to gene-disease data from public and internal databases and shortlists the top potential causal variants.

d. Reference any attachments of supporting materials throughout this nomination and how they provide evidence of the claims you have made in this nomination (up to 250 words). Optional

The Baylor Genetics lab validated its standardized XAI-driven bioinformatics process with a retrospective cohort of 180 rare genetic disease cases that were previously evaluated and solved with whole-exome sequencing and manual interpretation, publishing results in the June 2023 issue of Genetics in Medicine (see: https://www.gimjournal.org/article/S1098-3600(23)00843-2/abstract). In more than 93 percent of singleton cases, the top 10 suggestions generated by XAI included the causative variants previously identified by manual curation. When exome data from both parents was available, causative variants were included in the top recommendations 98 percent of the time, with 97 percent accuracy score overall.

In a prospective cohort of 334 new patients, the XAI-assisted process resolved 28.7 percent of cases, with an additional 12.6 percent having a possible diagnosis pending further evaluation. This diagnostic rate is comparable to that of Baylor Genetics' manual evaluation process for rare genetic diseases (25 to 36.7 percent) and average rates among other commercial labs in the US (26 to 30 percent).

Webpage Link

https://ai.nejm.org/doi/full/10.1056/AIoa2300009 (https://ai.nejm.org/doi/full/10.1056/AIoa2300009)

Would you like to add an additional webpage link?

Yes

Webpage Link 2

https://www.genomeweb.com/informatics/baylor-genetics-prepares-genome-world-explainable-ai-powered-variant-interpretation (https://www.genomeweb.com/informatics/baylor-genetics-prepares-genome-world-explainable-ai-powered-variant-interpretation)

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Webpage Link 3

https://www.genomeweb.com/clinical-sequencing/qa-improving-access-genomic-testing-baylor-genetics (https://www.genomeweb.com/clinical-sequencing/qa-improving-access-genomic-testing-baylor-genetics)

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Web Page Link 4

https://www.genomeweb.com/molecular-diagnostics/baylor-genetics-machine-learning-tool-shows-promise-clinical-rare-variant (https://www.genomeweb.com/molecular-diagnostics/baylor-genetics-machine-learning-tool-shows-promise-clinical-rare-variant)

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Web Page Link 5

https://www.baylorgenetics.com/news/using-ai-to-improve-diagnosis-of-rare-genetic-disorders/ (https://www.baylorgenetics.com/news/using-ai-to-improve-diagnosis-of-rare-genetic-disorders/)

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Web Page Link 6

https://www.baylorgenetics.com/news/baylor-genetics-to-share-scientific-insights-at-acmg-2024/ (https://www.baylorgenetics.com/news/baylor-genetics-to-share-scientific-insights-at-acmg-2024/)

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Web Page Link 7

https://www.baylorgenetics.com/news/baylor-genetics-accelerates-growth-combines-clinical-and-scientific-expertise-with-commercial-capabilities-to-meet-increased-needs-of-patients-providers-and-partners/ (https://www.baylorgenetics.com/news/baylor-genetics-accelerates-growth-combines-clinical-and-scientific-expertise-with-commercial-capabilities-to-meet-increased-needs-of-patients-providers-and-partners/)

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Supporting Document

No File Uploaded

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