Establishing a large genomic database in Israel

Illumina technology is helping Psifas capture unique genomic aspects of the Israeli population

PROF GABI BARBASH, CHIEF EXECUTIVE OFFICER OF PSIFAS

With 60 distinct subpopulations, including Arab Christians, Druze, and a diaspora of Jews originating from all over the world, Israel's population represents a global view of genomics. To take advantage of this diversity and promote better health across the country, Israel formed Psifas, a nationwide program designed to collect and analyze genomic data across the Israeli population. Psifas will sequence hundreds of thousands of individuals and create a publicly shared database for medical research purposes. The ultimate goals are to treat diseases more effectively, develop new therapeutics, promote personalized medicine, and enable earlier and more precise diagnosis. We interviewed Prof. Gabi Barbash, Chief Executive Officer of Psifas, about how Psifas works, what sets it apart from other biobanks, current projects, and future directions.

Why was Psifas created?

Gabi Barbash (GB): Psifas was established to create a big genomic database for clinical research. A main goal of Psifas is to serve as a national program to overcome Israel's fragmented genomic research infrastructure. Until now, sequencing projects were expensive, uncoordinated, and time-consuming, especially when it came to obtaining regulatory approvals and participant recruitment. We provide a unified platform that accelerates research by integrating sequencing, clinical data, and regulatory readiness into one national-scale infrastructure.

Also, in Israel, there is nothing like the National Institutes of Health in the US. However, we are in a position, despite the lack of these organizations, to allow researchers from the healthcare

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system to do research more easily and with some subsidy from the government because we are subsidizing them for the germline genetic sequencing in a limited capacity.

How does Psifas function as a national contract research organization (CRO) for genomic research?

GB: Psifas serves as Israel's one-stop shop for launching complex clinical genetic studies. By combining the advanced technology of the Illumina NovaSeq™ X System with nationwide volunteer recruitment and secure informatics infrastructure, Psifas demonstrates how a coordinated genomic infrastructure can shorten timelines, reduce costs, and ensure diversity across subpopulations.

Our research participants consent to providing clinical data from the community and hospitals. They provide blood for whole-genome sequencing, and they consent that we can come back to them for recall studies if we identify something for more in-depth research.

This enables us to accumulate a retrospective of data in our database that can be accessed by the Israeli medical research community.

What are the major advantages you have seen using the NovaSeq X System?

GB: We selected NovaSeq X System because it is the only platform capable of meeting our national-scale needs. The sequencing quality, ability to rapidly sequence large cohorts in a single run, and the competitive costs of sequencing on the NovaSeq X System have been transformative for Psifas and eliminated bottlenecks. These benefits allow us to make the Psifas database accessible at relatively low costs compared to the market, enabling research to advance without financial barriers.

How do you analyze your sequencing data?

GB: We perform almost all of our analysis using Illumina software, including the Illumina Connected Analytics platform on the web, which connects directly with the NovaSeq X Systems themselves, and then we use DRAGEN™ secondary analysis for variant calling in each sample. DRAGEN secondary analysis offers the necessary callers, including simple variations, expansion hunter, copy number, and structural variations for the full package.

Can you walk us through a recent research initiative that benefited from the NovaSeq X System and connected software capabilities?

GB: Yes. The Israeli Reference Genome Study (IRGS) is a prime example of our research. Within a short timeframe, thousands of samples from around 60 distinct subpopulations were collected and sequenced.

In fact, we will complete sequencing of 60K genomes collected through unselected population recruitment in December 2025 using the capacity of five NovaSeq X Systems in Israel. The capabilities of the NovaSeq X Systems are allowing us to sequence faster and produce the results, ready for researchers in the public domain, by early 2026.

What types of insights have emerged from your WGS studies so far?

GB: Psifas has developed a national genomic reference for Israel, significantly improving the accuracy of variant detection. Early insights are already helping to map rare variants and better understand disease risks within diverse subpopulations. By now Psifas has analyzed actionable genes among the first 56K volunteers and found that 2.2% of individuals carried at least one of these variants, which is an extremely important finding for future clinical and genetic studies. Unlike other biobanks, Psifas has an agreement to return information about actionable variants to the research participants, which we have already begun to do.

By integrating clinical and genomic data within secure, cloud-based trusted research environments (TRE), Psifas enables researchers to identify risk patterns, treatment responses, and population-level insights. Researchers in hospitals and industry are also using Psifas for studies on conditions such as myelodysplastic syndromes, schizophrenia, eye diseases, cancer, and more. Additionally, local and international biotech companies leverage the Psifas clinical genetic database for a range of purposes, much like they do with other leading biobanks around the world.

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What are the next steps for Psifas in terms of new cohorts or integrating other omics?

GB: Psifas is constantly looking to expand and evolve. We are very interested in integrating additional omics such as proteomics and transcriptomics. We are developing artificial intelligence (AI)–driven projects focused on genetics and other multimodal sources of data. We also plan to establish a dedicated center for longevity research and recruit new disease-specific cohorts in areas such as mental health, cardiology, and rheumatology.

How do you see Psifas evolving over the next 3–5 years?

GB: In the next 3–5 years, Psifas is on track to become Israel's central national infrastructure for genomic and clinical research, serving both academia and industry. In addition, Psifas will serve as the Israel's central CRO for conducting large-scale research, with biosample collection capabilities expanding well beyond what we are capable of today.

Our vision includes making Psifas one of the most significant and diverse genomic resources worldwide. We aim to be recognized internationally as a unique "virtual biobank," combining genetic, clinical, and longitudinal data under one secure platform.

We also see Psifas enabling advanced AI-driven research, where large-scale data is used to generate new insights into complex diseases, drug development, and personalized medicine. Looking ahead, Psifas could serve as the foundation for nationwide programs such as population-based prevention strategies, newborn screening at scale, and collaborations with global pharmaceutical companies seeking to leverage Israel's unique genetic diversity.

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