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Who do you think you are? Conference puts Russian genome research in spotlight



Shura Collinson
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National genome projects that study the gene pool of a specific country can not only reveal much about a country's history and its inhabitants' roots, they can also bring about advances in medical treatment. But while some countries have poured millions of dollars into national programmes, the Russian genome remains largely unexplored territory. An upcoming conference at the Skolkovo Foundation, [Genome Russia](#), will bring together experts from around the world to explore genome projects in other countries and exchange ideas for Russia's own fledgling programme.

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Russia is home to nearly 200 different peoples or ethnicities, including the Tyvans (pictured).

Photo: Kremlin.ru.

The study of population genetics has two main aims: to improve medical treatment and establish the history of a country's people and their origins.

"It's important to know the distribution and frequency of various genes in order to know what varieties of illnesses are widespread among the population, how to treat them, how they will react to various medicines: that's the most important thing," says Yuri Nikolsky, the science director of the Skolkovo Foundation's biomed cluster who is co-chairing the Genome Russia conference at the foundation on April 14.

"It's also an important academic task to know who's descended from whom, to establish the history of the development of various peoples, especially in multiethnic countries like Russia," he said. Russia is home to nearly 200 different peoples or ethnicities, according to the 2002 and 2010 censuses.

Since the first human genome sequence was drafted in 2001, the benefits of national genome programmes have been demonstrated in other countries.

In Saudi Arabia, for example, scientists discovered varieties of diseases specific to the region, and now health care and diagnoses have been adapted accordingly, said Nikolsky.

A late start

Russia, however, is only now working on a nationwide programme: the [Genome Russia](#) project coordinated

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by the [Theodosius Dobzhansky Center](#) for Genome Bioinformatics at St. Petersburg State University. The centre's chief scientific officer, Stephen O'Brien, and his colleague Vladimir Brukhin will open the conference with talks on the project's aims and progress, and many of the other speakers are part of the Genome Russia Consortium carrying out the research.

"Russia - a country with 1/10th of Earth's landmass, 1/50th of the world's people, and descendants from critical crossroads across human migration history - has a relatively unexplored human genome landscape," the project's authors wrote in a letter published in Science magazine last year. The Genome Russia project will "close the largest gap in the genome map of the world," they say.



Yuri Nikolsky, the conference's co-chair, pictured in Almaty with Kazakhs wearing traditional dress at the Kazakhstan stage of the Skolkovo Foundation's Startup Tour last week. Photo: Sk.ru.

The project, which was launched in January 2015, aims to develop a database of anonymous information about the whole-genome sequences of more than 2,000 people from different regions of Russia. Blood samples will be taken from those whose ancestors have lived in the region for at least several generations.

"We have recruited about 900 volunteers or study participants in our database and we are anticipating another 2,000 in the coming year or more from several ethnic groups," O'Brien told Sk.ru.

The reasons for Russia's lateness - other countries began their quest for the national genome in the early 2000s - are complex, Nikolsky and O'Brien agree.

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“Sequencing technology in Russia is impeded by an outdated tender, purchasing and reagent broker system which jacks up the cost considerably,” said O’Brien.

“Add to that the ruble devaluation and the fragile economy in Russia, and the resources are scarce,” he said.

“Money is certainly a factor, because these programmes are quite expensive,” agrees Nikolsky.

Currently, the Genome Russia project is financed by St. Petersburg State University, but O’Brien hopes for increased federal funding in the future.

The Genome Russia project will close the largest gap in the genome map of the world, its coordinators say.

There are also logistical problems in mapping the Russian genome.

“It’s forbidden to take biological materials out of Russia, and it’s hard to do genotyping here on a large scale because we don’t have much equipment,” said Nikolsky. “It would be easier to do the genotyping by taking the samples abroad, where they’d do all the sequencing, but that can’t be done legally.”

A lack of qualified scientists is, however, not a problem.

“There is certainly a dedicated and fully capable Russian genetics community ready to work together on the project,” said O’Brien.

Back to the roots

To understand where we come from, the first task is to establish the founder populations – modern groups descended from a few individuals who settled in one area, explains Nikolsky. Founder populations have already been identified in much of Western Europe, he said.

“In Russia, it’s a big grey area,” he said. “There could be 500,000 or a million genetic variations. And we will be able to work out the distribution of these variations.”

Once we have the genotypes of the founder populations, there are reliable mathematical methods that will allow each one of us to calculate the relative proportion of these populations in us, he said.

“Then we’ll know where we came from,” he said.

One such system – the Geographic Population Structure tool – that allows people to trace their DNA up to 1,000 years back to their region of origin was co-authored by the U.S.-based scientist Tatiana Tatarinova, who is chairing the conference together with Nikolsky.

"I believe that anything that helps to treat people is ethical, and knowing the national gene pool definitely helps a lot." - Yuri Nikolsky.

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In some countries that pioneered national genome projects like Iceland, high-profile debate raged over ethical issues, such as what consent is needed from individuals, the need for confidentiality and people's access to the information obtained.

In Russia, where the Orthodox Church has prevented the burial of the last two members of the murdered imperial family to be discovered, refusing to accept DNA evidence that shows the remains to be those of the two missing Romanov children, genome programmes also look set to raise questions.

Nikolsky's position on the ethics debate is unambiguous.

"I believe that anything that helps to treat people is ethical, and knowing the national gene pool definitely helps a lot," he says.

While there has been no national or large-scale programme until now, individual groups have carried out their own separate studies into Russian population genetics. The results have not only been fascinating, but have also seen practical benefits, Nikolsky said, citing a project carried out in a Russian Railways clinic that identified markers of predisposition to coronary heart disease.

"No Western models that had been applied had worked on Russian populations before they did that," he said.

Another Russian study was devoted to the Kets, a dwindling people that live in and around the basin of the Yenisei River in Siberia. A study of the gene pool of the Kets, of whom less than 2,000 remain, revealed that they have a direct genetic link to several groups of Native Americans.

"That research essentially proved that there was migration that began here in Siberia and ended in America," said Nikolsky. "The work couldn't have been done without genotyping, and it couldn't have been done in another 15 years' time, because there would simply not be enough Kets left, they are dying out," he added.



A 1914 photo of Kets. Research showed that the Kets have a direct genetic link to some Native Americans, revealing a migration route that began in Siberia and ended in America. Photo: Wikimedia Commons.

Russia and beyond

One of Russia's best known geneticists, Oleg Balanovsky of the [Vavilov Institute of General Genetics](#), will present a talk at the conference on studies of Russian ethnic genomes via mitochondrial, Y-chromosomal and full-genome markers.

It was the Vavilov Institute's laboratory of evolutionary genetics headed by Yevgeny Rogayev that identified the remains of Grand Duchess Maria and the Tsarevich Alexei – the last two murdered Romanovs whose identity the Orthodox Church disputes.

The same laboratory proved what historians had long assumed: that the disease from which Alexei suffered – along with many other members of European royal families descended from Britain's Queen Victoria – was haemophilia. Rogayev and his colleagues identified the type of haemophilia as the rarer form of the disease known as haemophilia B.

Other talks at the conference will be devoted to genomics in Qatar and Japan, and to practical medical applications of population genomics, such as its role in patient response to chemotherapy and possible use in

cardiovascular disease risk assessment.

Also speaking at the conference is Eran Elhaik, a lecturer at the U.K.'s University of Sheffield who created Geographic Population Structure together with Tatarinova. Elhaik's conclusion that Ashkinazi Jews trace their ancestry not to the Middle East, but to the Caucasus and Eastern Europe, sparked debate when his research was published several years ago. Elhaik will present his findings to the conference in a talk titled "The missing link of Jewish European Ancestry."

Some of Russia's leading minds in the field of genomics are pursuing their work abroad, such as Tatarinova, who is based at the University of Southern California.

"Most good biomedicine scientists from Russia left long ago," said Nikolsky, adding that there were advantages to be gained from this. "Now they have something to tell us [about research in the West], something to teach us," he said.

The Genome Russia conference will be held at the Hypercube at the Skolkovo innovation centre on April 14 from 9 am to 6.20 pm. Attendance of the English-language event is free of charge. To see the conference programme and register for the event, click [here](#).

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