Environmental human DNA offers new opportunities for public good

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Scientists collected high-quality DNA from footprints made by one of the researchers on an uninhabited island. Sequencing the DNA revealed identifiable information about the participant's genome. Credit: David Duffy

In May, University of Florida scientists announced that they had unearthed high-quality, information-rich human DNA from nearly every spot they could think of. Rivers, beaches, oceans—even vacuumed up from the air.

The era of human environmental DNA, or eDNA, has arrived.

That discovery means huge changes are on their way across science—and our lives—says David Duffy, the UF researcher who led the recent project unveiling how widespread our genetic information really is in the environment.

There are countless exciting new opportunities, from protecting public health to identifying violent criminals. We could uncover the secrets of the past with DNA from hidden gravesites or protect the environment; all with nothing more than a cup of water at a sewage plant or an air filter hooked up in a park.

But that's just the tip of the iceberg. "This can potentially be useful for things we haven't even thought about yet," said Duffy, a professor of wildlife disease genomics at UF's Whitney Laboratory for Marine Bioscience. "We're only at the beginning of understanding the possibilities of this technology."

Nabbing criminals

Straight out of TV procedurals, one obvious application of this new kind of DNA analysis is upgrading our ability to identify criminals. "We potentially already have the technology to do forensics using environmental DNA samples," Duffy said. But eDNA forensics will look different than today's crime lab.

U.S. police departments today rely on matching about 20 individual portions of the human genome spread across the 23 pairs of chromosomes. With enough matches, it's almost certain that the DNA comes from a unique individual.

But eDNA is too fragmented and mixed up, making it impossible to tell if different chromosomes in a sample come from one person or many. So eDNA forensics would need to focus on genetic variation that is on individual chromosomes, in hopes that a long stretch of sequenced DNA captures multiple regions that vary between people.

In the future, eDNA analysis could also look for missing or duplicated chunks of genetic code, which are known as deletions and insertions, which eDNA tech can readily identify. But more work is necessary to understand how these errant marks in our DNA vary between people before it can be used to identify criminals.

"We have to catch up with our knowledge of these genetic differences to build the medical and forensics databases to better understand the diversity of human insertions and deletions," Duffy said.

Improving forensics with eDNA relies on the idea that finding someone's DNA in a room, or even on a murder weapon, means they were at the crime scene or pulled the trigger. But that might not be the case.

"You cannot assume that because you found someone's eDNA in a location, that means that person was there. All you can say is their DNA was there," said Connie Mulligan, a professor of anthropology at UF who researches human genetics and forensics. "If you're sampling the air outdoors, you may not even be able to say the person was nearby once you consider air currents."

With her graduate student Samantha McCrane, Mulligan recently showed that DNA can easily be transferred from one person to another and then to a gun through a simple handshake. These transfers can even happen multiple times, making a DNA identification on a weapon murky at best. So the criminologists of the future must be cautious in how they interpret eDNA signals.

"Our imaginations are probably not wild enough to conceive of all the scenarios in which a person's DNA can be moved to places that that person never was, whether through touch, or coughing, or sneezing, or air currents," Mulligan said.

Sewage plant health boost

During the COVID-19 pandemic, public health officers were able to see viral surges weeks in advance by spotting signs of the virus at sewage plants. This wastewater monitoring was most people's first taste of the power of eDNA, and its success during the pandemic means this kind of pathogen monitoring will likely continue. The technology has already been applied to many other human pathogens.

"It totally makes sense to monitor wastewater for any kind of human pathogen," said Mulligan. "It's easy. It's cheap.

Although the largest benefit to population health likely comes from pathogen surveillance like this, it's possible that signals of genetic susceptibility to diseases in humans could provide a way to protect the health of whole communities.

"If you're a public health specialist, it might be very useful to have an anonymized readout of what sort of susceptibility any given population has to particular diseases," Duffy said. One application could be linking chemical spills or radioactive waste to cancer mutations in a town by collecting eDNA from wastewater. Identifying a definitive link between contaminants and disease could help people demand cleanup or medical care.

Protecting the environment

Wastewater monitoring could also help scientists protect vital waterways. Human waste is a powerful contaminant for rivers and lakes, but it can be difficult to prove the source of contamination. If a lot of human DNA is in a water sample—especially if it's matched to the population of a nearby town—scientists could pinpoint faulty treatment plants or broken sewer lines as likely culprits and remedy the problem.

And eDNA will continue to benefit wildlife scientists, who—like Duffy—developed many of today's eDNA techniques to study and protect animals. Duffy has already used eDNA to research the viral cancers to which sea turtles are susceptible. With the ability to easily sequence the DNA of every species in a sample, scientists could identify key habitats to conserve, study hidden migrations, or look for species that are presumed extinct.

"Our human eDNA discoveries are showing us what it will be possible to achieve with wildlife and biodiversity eDNA in the very near future," Duffy said.

What's next

These are just some of the potential benefits we will draw from everimproving eDNA science. And the underlying technology is improving all the time. Sequencing eDNA is getting more reliable and cheaper every year.

"This is why I'm convinced we are only scratching the surface," Duffy said. "While this is a major advancement, there are many more on the horizon."

More information: Samantha M. McCrane et al, An innovative transfer DNA experimental design and qPCR assay: Protocol and pilot study, *Journal of Forensic Sciences* (2023). DOI: 10.1111/1556-4029.15243

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