

# Phenotyping Webtool Developed by Academics, Who Want 'Full Disclosure' of Limitations

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DNA



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Phenotyping—the prediction of physical characteristics

from DNA—has caused a minor sensation in the forensic sciences over the last three years. Some police departments and companies have put great stock in generating the predicted faces of unknown murderers and rapists from the genetic trail they left behind at crime scenes. Some critics at the same time have been skeptical, claiming that such images and traits may offer likelihoods of eye or skin color—but are otherwise not specific about nose structure or other major telltale features.

But an international team of academics have pushed out a new webtool for predicting eye color, hair color and skin tone that they say is pushing new boundaries for the science—especially since it is free of charge and available to all law enforcement agencies who want to use it.

The team, led by Susan Walsh at Indiana University—Purdue University Indianapolis School of Science, presents the “HIrisPlex-S” system in the journal *Forensic Science International: Genetics*.

Their main interest is in full disclosure of where the accuracy, and the possibility of error, are, Walsh said.

“What we do is totally out there,” said Walsh, in a *Forensic Magazine* interview this week. “We’ve said how we made it, we’ve said our caveats, we’ve said we’re good at doing this, and we’re bad at doing this ... this is where it’s at right now. We’re pretty open, and we’re free—and that’s

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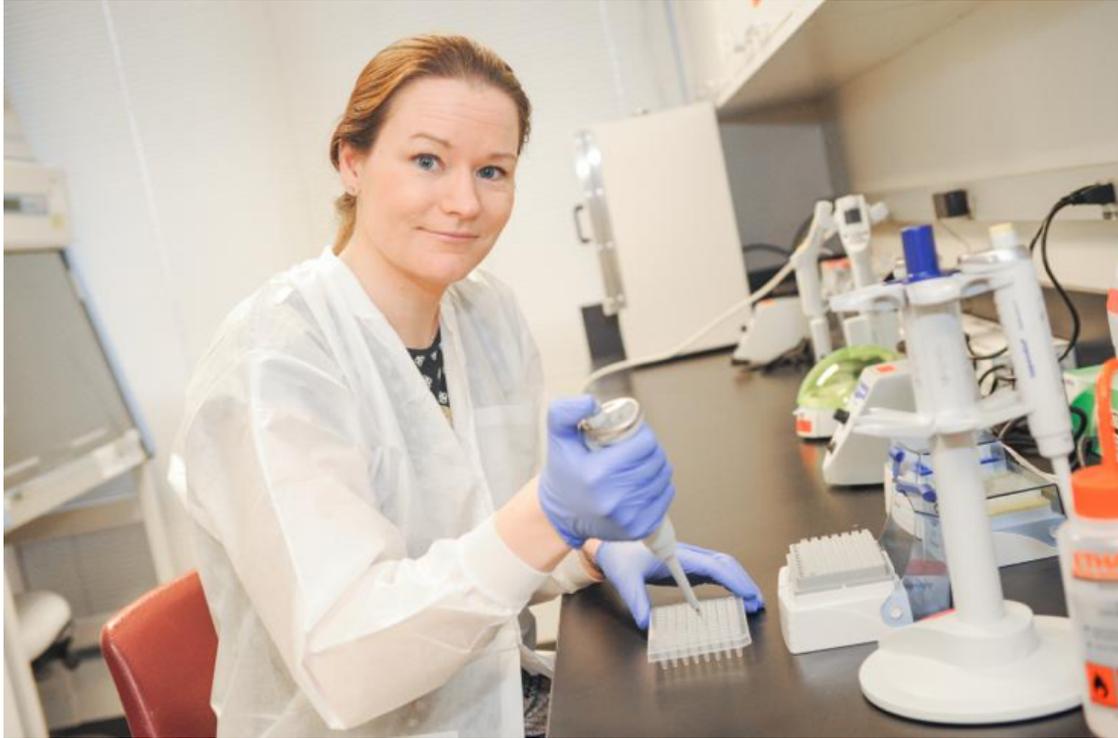


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how we like to work.”

Walsh and her colleagues believe they have put together the best system for phenotyping yet.



Their prediction system currently relies on 36 SNPs for skin tone, 6 SNPs for eye color and 22 SNPs for hair colors.

That means parsing out five different skin tones, three different eye colors and four hues of hair.

The system is also built for predicting based on limited or degraded DNA. In the *FSI: Genetics* paper, they have demonstrated that they could accurately make predictions from just 63 picograms of genetic material.

Their accuracy fluctuates depending on genetic nuances. For instance, they are as much as 97 percent accurate for predicting the darkest skin tones, but 74 percent for the lightest and fairest skin colors. The same goes with the different colors of hair and eyes, they present in a series of recent papers on the system beginning last May.

Already, the team has worked with the Indiana State

Police on some pending cases. Walsh would not elaborate on those cases—but she said she would want detectives worldwide to know that the HIrisPlex-S system is available for them, and for free. It doesn't use much DNA, and the assay (if agencies need to rely on Indiana University to do the lab work) costs just \$20.

"I really want investigators, the forensic scientists, the police, to see this," said Walsh. "This is doable themselves on cases that are stuck, and on cold cases which are just sitting around."

Walsh, who is partly funded by the National Institutes of Justice, said her work has been in parallel to those done by a Spanish phenotyping team at the University of Santiago de Compostela led by Christopher Phillips. That team had made some breakthroughs in skin tone, which furthered the overall science, Walsh said.

But the Indiana University academic said she is skeptical of the facial reconstructions done by private companies for law enforcement.

"We are nowhere near understanding facial prediction," said Walsh. "At the end of the day, there's no genetics behind it ... Pigmentation—absolutely."

Parabon NanoLabs, headquartered in Virginia, has become what appears to be the industry leader in phenotyping for law enforcement agencies across the country. They also recently announced a "genetic genealogy" division to their company earlier this month, in the wake of the sensational arrest of the alleged Golden State Killer in California. They tout some breakthroughs, including one in which an accused killer in Texas turned himself in after seeing the estimated recreation of his face—and another in which North Carolina investigators were re-directed by the genetic facial "sketch" because of unknown parentage of the true murderer.

Steven Armentrout, the Parabon CEO, said their patented Snapshot technology uses hundreds of thousands of SNPs for a “one-stop shop” including genealogy, ancestry prediction, phenotyping and kinship inference. Using an assay for pigmentation alone can limit the important inferences, Armentrout said.

The Snapshot technology has been scientifically tested, with some of the methodology published in the journal *BioData Mining* last year by the company, Armentrout added. A blind evaluation of the tool was conducted by the United of North Texas Health Science Center in 2016 using 24 sample subjects, and the results were presented at that year’s International Symposium on Human Identification. To date, that evaluation has not been published, but others are in the works, Armentrout said.

“We are planning to publish a developmental validation of Snapshot’s kinship inference algorithm later this year,” the CEO added.

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