UCSC researchers trace the roots of a type of muscular dystrophy

By JANE PALMER

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SANTA CRUZ -- A team of scientists at UC Santa Cruz has uncovered how a genetic defect leads to the devastating disease myotonic muscular dystrophy.

Myotonic dystrophy, one of the nine types of muscular dystrophy, is a hereditary disease that causes weakness and muscle wasting in both children and adults. The most common type of myotonic dystrophy is caused by changes in a gene that has a repeating sequence of three DNA building blocks. In a normal gene, this sequence is repeated up to 35 times, but when it repeats more than 50 times, it can lead to myotonic dystrophy.

"The longer it becomes, the worse the disease gets," said Manuel Ares, a professor of molecular, cell and developmental biology at UCSC.

Ares and his collaborators at the University of Florida and at the University of Rochester, New York, have been able to identify the pathways by which such mutations wreak havoc on the human body, using research grants from the National Institutes of Health.

"We have been able to identify a pattern or fingerprint' of gene expression in cells that are starting to experience the effects of this mutation," Ares said.

Ultimately, Ares hopes that by analyzing the elements of this gene expression fingerprint, scientists will be able to trace where the symptoms of the muscle failure are coming from. Knowing the origins of the disease is one step toward knowing how to treat it, he said.

The devastating gene sequence acts like a sponge soaking up key molecules required to express many other genes properly, Ares said.

"You are not simply messing up the expression of one gene," Ares said. "All these other genes go haywire too."

Ares also hopes that the technology can be used to tell if proposed treatments are effective.

"We would want to know that the complex effects that we are observing are being reversed," he said.

Ares also hopes that the findings will be useful in identifying why the disease affects families differently.

"It is a progressive disease that gets much worse in some families than others, so we are hoping to capture some of the family differences in the fingerprint," Ares said.

"It is a very high resolution way of monitoring a very complex disease," he said.

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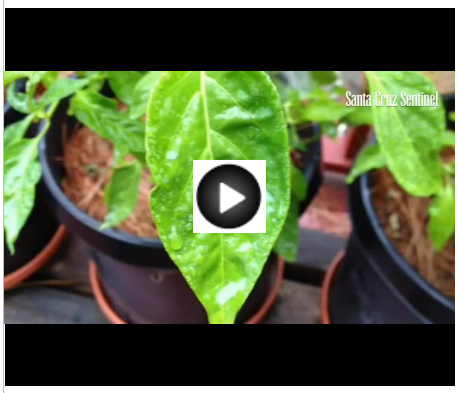

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