Researchers discover genetic mutation causing excessive hair growth

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(Medical Xpress) -- Researchers in the Keck School of Medicine of the University of Southern California (USC), together with scientists in Beijing, China, have discovered a chromosomal mutation responsible for a very rare condition in which people grow excess hair all over their bodies. Investigators hope the finding ultimately will lead to new treatments for this and less severe forms of excessive hair growth as well as baldness.

The study, "X-linked congenital hypertrichosis syndrome is associated with interchromosomal insertions mediated by a human-specific palindrome near SOX3,"
will appear in the June 2, 2011 issue of the *American Journal of Human Genetics*.

Pragna Patel, professor in the Keck School, the USC Institute for Genetic Medicine, and the Herman Ostrow School of Dentistry of USC, is a co-investigator of the study, and Sunju Choi, a research associate in Patel's lab, is a co-first author. The principal investigator is Xue Zhang, professor and chair of medical genetics at the Peking Union Medical College.

The initial discovery of the mutation came from Zhang's lab, which examined the condition, known as CGH, in a Chinese family. Researchers there worked with Choi to confirm the finding in a Mexican family that Patel first began studying in 1993. Males with this disorder have hair covering their entire face including their eyelids and their

upper body, while females have thick patches of hair on their bodies.

"In 1995, we traced the approximate location of the mutation responsible for CGH to a section of the X chromosome in the Mexican family," said Patel, whose laboratory focuses on understanding the genetic basis of inherited diseases. "We sequenced nearly 100 genes but could not find any mutations. Then recently, Xue Zhang and his colleagues in Beijing were able to pinpoint the exact location, discovering that there was an insertion of chromosome 5 into the X chromosome in the Chinese family. When the Mexican family was examined, a piece of chromosome 4 was found to be inserted into the same part of the X chromosome, thus confirming that these extremely rare events caused the disorder."

The investigators suggest that insertion of the "extra" DNA sequences into the X chromosome apparently turns on a gene, likely SOX3, located near the insertion site. SOX3 is a strong candidate because other members of this gene family have been shown to play a role in hair growth.

In addition, the insertion has occurred within a block of DNA sequence called a "palindrome," in which the sequence of the four building blocks of DNA (akin to letters of the alphabet) read exactly the same as their complementary sequences, but in the reverse direction. The particular palindrome at the site that the researchers studied is only found in humans.

"We don't yet know the significance of the palindromic sequence in this case," Patel said. "But it appears to be unstable, and can be entirely absent in many individuals with normal hair growth. It's only when there is insertion of certain chromosome segments at this site that people have extra hair."

Earlier, researchers had theorized that the CGH mutation is "atavistic" – a trait that reappears after being absent for a long time. One example of an atavistic trait is extra nipples in both men and women.

"It's like the information is there in the genome, but is silenced," Patel said. "Then somehow it's reactivated, and can manifest as the trait. We don't know yet if this is the case with CGH."

Further studies will test if this is indeed true. "If in fact the inserted sequences turn on a gene that can trigger hair growth, it may hold promise for treating baldness or hirsutism [excessive hair growth] in the future, especially if we could engineer ways to achieve this with drugs or other means," Patel said.

Provided by University of Southern California

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