

Researchers study gene mutations in children with 'uncombable hair syndrome'

2 September 2022, by Bob Yirka



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A large international team of researchers has conducted a genotypic spectrum analysis for uncombable hair syndrome (UHS) and in so doing has found two pathogenic missense variants in PADI3 that account for the majority of cases. In their paper published in *JAMA Dermatology*, the group describes how they studied the genes of 107 children with the syndrome to identify its genetic roots.

UHS is a rare condition where a person's hair grows in a way that prevents it from lying down when combed—the result is a wild patch of unruly hair. Prior research has shown that the syndrome is almost always in [children](#) who grow out of it as they move into their teens. Children with the syndrome have also been found to have inherited the syndrome from both parents, though neither of them has to have it for the child to be affected. It has also been noted that the vast majority of those affected are Caucasian and have blond hair, leading to its nickname of spun-glass hair syndrome. Prior research has also shown that the unruliness is due to hair growing at right angles to the scalp and this is due to misshapen hair

follicles. In 2016, some of the members of the current team were able to identify gene variants in 11 children they tested.

In this new effort, the researchers recruited a much larger group of volunteers for testing—107 children with the syndrome. In analyzing their [genes](#), the researchers found a genetic reason for the [syndrome](#) in 80 of the volunteers. And in 76 of those people, they found mutations in the PADI3 gene. They note that prior research has shown that the PADI3 gene is responsible for encoding an enzyme that is involved in the production of trichohyalin—one of the main proteins found in hair.

The researchers suggest their findings may prove useful to parents of children who develop unruly [hair](#), by helping rule out other more serious illnesses. Such a finding may also reassure both the child and his or her [parents](#) that the condition is likely only temporary.

More information: F. Buket Basmanav et al, Assessment of the Genetic Spectrum of Uncombable Hair Syndrome in a Cohort of 107 Individuals, *JAMA Dermatology* (2022). [DOI: 10.1001/jamadermatol.2022.2319](https://doi.org/10.1001/jamadermatol.2022.2319)

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APA citation: Researchers study gene mutations in children with 'uncombable hair syndrome' (2022, September 2) retrieved 22 September 2022 from <https://medicalxpress.com/news/2022-09-gene-mutations-children-uncombable-hair.html>

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