Mutations in COQ2 linked to multiple-system atrophy

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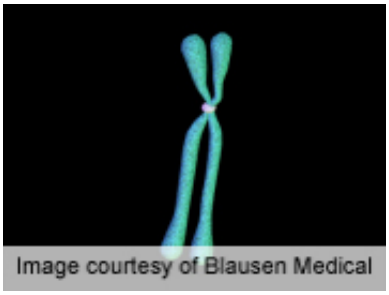

Functionally impaired variants in the *COQ2* gene, which encodes the enzyme parahydroxybenzoate-polyprenyl transferase, have been identified in multiplex families with multiple-system atrophy and in patients with sporadic disease, according to a study published online June 12 in the *New England Journal of Medicine*.

(HealthDay)—Functionally impaired variants in the *COQ2* gene, which encodes the enzyme parahydroxybenzoate-polyprenyl transferase, have been identified in multiplex families with multiple-system atrophy and in patients with sporadic disease, according to a study published online June 12 in the *New England Journal of Medicine*.

Jun Mitsui, M.D., Ph.D., from the University of Tokyo, and colleagues performed whole-genome sequencing of a sample obtained from a member of a multiplex family with multiple-system atrophy. Mutational analysis was also performed on samples from members of five other families, and from three patient series (Japanese, European, and North

American). Enzyme activity of parahydroxybenzoate-polyprenyl transferase was measured.

In two multiplex families, the researchers identified a homozygous mutation (M78V-V343A/M78V-V343A) and compound heterozygous mutations (R337X/V343A) in *COQ2*. A common variant (V343A) and multiple rare variants were also identified in *COQ2*, all of which were functionally impaired and were linked with sporadic multiple-system atrophy. The variant V343A was only seen in the Japanese series.

"Functionally impaired variants of *COQ2* were associated with an increased risk of multiple-system atrophy in multiplex families and patients with sporadic disease, providing evidence of a role [for] impaired COQ2 activities in the pathogenesis of this disease," the authors write.

More information: Abstract
Full Text