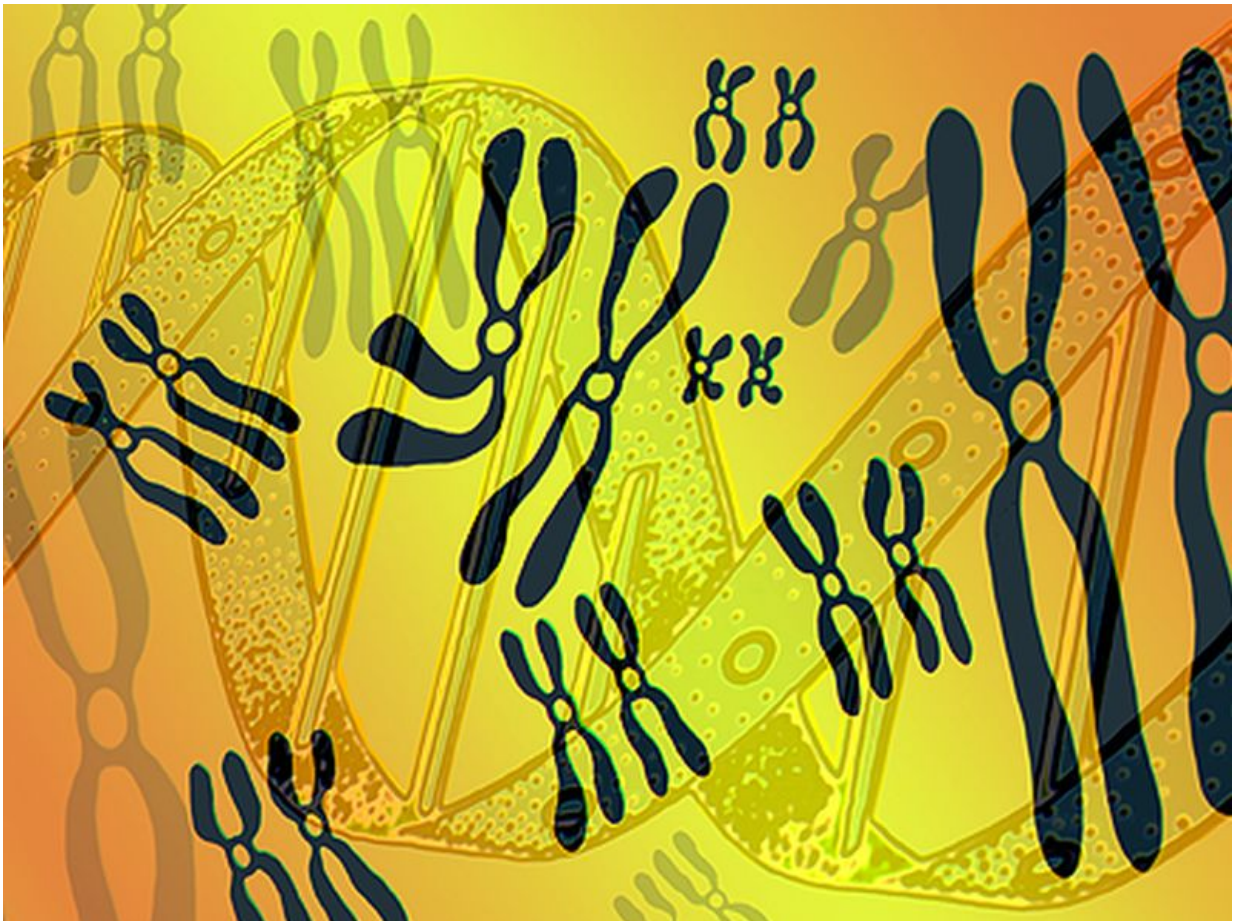


HTRA1 mutations tied to cerebral small vessel disease

August 11 2017



(HealthDay)—Heterozygous *HTRA1* mutations may play a role in

familial cerebral small vessel disease (SVD), according to a study published online Aug. 6 in *CNS Neuroscience & Therapeutics*.

Ilaria Di Donato, M.D., from the University of Siena in Italy, and colleagues performed mutational analysis of *HTRA1* gene in 142 *NOTCH3*-negative [patients](#) and 160 healthy age-matched controls.

The researchers found that five different *HTRA1* heterozygous [mutations](#) were detected in nine patients from five unrelated families. In these patients, the clinical phenotype was typical of SVD, and the onset was pre-senile. A subcortical leukoencephalopathy, with involvement of the external and internal capsule, [corpus callosum](#), and multiple lacunar infarcts, was seen on brain magnetic resonance imaging. Additionally, cerebral microbleeds were seen, but anterior temporal lobe involvement was not present.

"Our observation further supports the pathogenic role of the heterozygous *HTRA1* mutations in familial SVD," the authors write.

More information: [Abstract](#)
[Full Text \(subscription or payment may be required\)](#)

Copyright © 2017 [HealthDay](#). All rights reserved.

Citation: HTRA1 mutations tied to cerebral small vessel disease (2017, August 11) retrieved 20 April 2024 from
<https://medicalxpress.com/news/2017-08-htra1-mutations-tied-cerebral-small.html>

<p>This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.</p>
--