

New autoinflammatory disease linked to NOD2 mutations ID'd

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A new autoinflammatory disease characterized by features including inflammatory polyarthritis/polyarthralgia and dermatitis, and involving mutations in the nucleotide-binding oligomerization domain 2 gene has been identified, according to research published online Oct. 26 in the *Journal of the American Academy of Dermatology*.

(HealthDay)—A new autoinflammatory disease characterized by features including inflammatory polyarthritis/polyarthralgia and dermatitis, and involving mutations in the nucleotide-binding oligomerization domain 2 (*NOD2*) gene has been identified, according to research published online Oct. 26 in the *Journal of the American Academy of Dermatology*.

Qingping Yao, M.D., Ph.D., of the Cleveland Clinic, and associates prospectively sought to characterize a new category of autoinflammatory disease in 22 non-Jewish, white patients (13 females, nine males; median age at diagnosis, 40.1 years), all of whom had autoinflammatory



phenotypes and NOD2 gene mutations.

The patients, who were studied between January 2009 and February 2012, had a mean disease duration of 4.7 years and included three females who were siblings. The researchers identified inflammatory polyarthritis/polyarthralgia (20/22), dermatitis (19/22), weight loss (13/22), and episodic self-limiting fever (13/22) as common clinical features. In 13 patients, gastrointestinal symptoms occurred; sicca-like symptoms were identified in nine patients; and recurrent chest pain was seen in five patients. All patients had mutations in the *NOD2* gene, with two variants identified.

"The characteristic clinical phenotype, notably dermatitis, coupled with certain *NOD2* variants constitutes a new autoinflammatory disease entity, which we have named as *NOD2*-associated autoinflammatory disease," the authors write.

More information: Abstract

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