Mutation ID'd in Waldenstrom's macroglobulinemia

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(HealthDay)—MYD88 L265P is a common, recurring mutation in patients with Waldenström's macroglobulinemia, according to a study published in the Aug. 30 issue of the *New England Journal of Medicine*.

Steven P. Treon, M.D., Ph.D., from the Dana Farber Cancer Institute in Boston, and colleagues performed whole-genome sequencing of bone marrow lymphoplasmacytic lymphoma (LPL) cells in 30 patients with Waldenström's macroglobulinemia. The findings were validated using Sanger sequencing in an expanded cohort of patients with LPL, those with other B-cell disorders that have some of the same features as LPL, and healthy donors.

The researchers identified a somatic variant (T?C) in LPL cells at position 38182641 at 3p22.2 among the patients with Waldenström's macroglobulinemia. This variant was seen in all samples from 10 patients with paired tissue samples and in 17 of 20 samples from patients with unpaired samples. The variant predicted an alteration in an amino acid (L265P) in *MYD88*, which led to IRAK-mediated NF-?B signaling. MYD88 L265P was identified using Sanger sequencing in tumor samples from 49 of 54 patients with Waldenström's macroglobulinemia and in all three patients with non-immunoglobulin M (IgM)-secreting LPL. In paired normal tissue samples from patients with Waldenström's macroglobulinemia or non-IgM LPL and in B cells from healthy donors, MYD88 L265P was absent.

"MYD88 L265P is a commonly recurring mutation in patients with Waldenström's macroglobulinemia that can be useful in differentiating Waldenström's macroglobulinemia and non-IgM LPL from B-cell disorders that have some of the same features," Treon and colleagues conclude.

Several authors are employees of Complete Genomics.

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