A rare truncating BRCA2 variant and genetic susceptibility to upper aerodigestive tract cancer

An international collaborative study led by researchers in the Genetic Cancer Susceptibility Group of the International Agency for Research on Cancer (IARC) links a genetic variant (rs11571833, K3326X) with susceptibility to upper aerodigestive tract cancers. The results have now been published in the Journal of the National Cancer Institute. rs11571833 has been similarly associated with lung cancer, but only a modest risk of breast cancer. The more pronounced association in lung and upper aerodigestive tract cancers, together with the ambiguous functional consequence of this variant, suggests an alternate susceptibility mechanism for K3326X compared with the highly deleterious BRCA2 breast cancer susceptibility alleles. The new result highlights how large international genetic studies can provide novel insights into cancer susceptibility and cancer etiology.

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