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Summary

LOST EXPRESSION, FOUND FAMILIES: HOW IHC REVEALS LYNCH SYNDROME BEYOND AGE AND FAMILY HISTORY

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Colorectal

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A single-center study of 101 Lynch syndrome patients found that universal immunohistochemistry screening identifies affected families missed by traditional clinical criteria, particularly those with later-onset cancers and MSH6 or PMS2 mutations.

KEY POINTS

- Index cases identified through immunohistochemistry on tumor samples had a mean age of 57.93 years at genetic diagnosis compared to 46.35 years for those identified by Amsterdam II or revised Bethesda clinical criteria.
- Half of the index patients diagnosed through immunohistochemistry did not meet Amsterdam II criteria and would have been missed by clinical criteria alone.
- Patients with pathogenic variants in MSH6 and PMS2 genes, which have lower penetrance and later cancer onset, represented 50 percent of the immunohistochemistry group versus only 5 percent of the clinical criteria group.
- Universal immunohistochemistry screening detects mismatch repair-deficient tumors in patients diagnosed later in life and those without evident family history of cancer.
- The findings support applying immunohistochemistry screening for mismatch repair protein loss in both colorectal and endometrial cancers to identify Lynch syndrome families.

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