A de novo germline MLH1 mutation in a Lynch syndrome patient with discordant immunohistochemical and molecular biology test results

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Résumé en anglais	We describe a patient with a Homo sapiens mutL homolog 1 (MLH1)-associated Lynch syndrome with previous diagnoses of two distinct primary cancers: a sigmoid colon cancer at the age of 39 years, and a right colon cancer at the age of 50 years. The mutation identified in his blood and buccal cells, c.1771delG, p.Asp591Ilefs*25, appears to be a de novo event, as it was not transmitted by either of his parents. This type of de novo event is rare in MLH1 as only three cases have been reported in the literature so far. Furthermore, the discordant results observed between replication error phenotyping and immunohistochemistry highlight the importance of the systematic use of both pre-screening tests in the molecular diagnosis of Lynch syndrome.
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