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Two missense *STK11* gene variations impaired LKB1/AMPK signaling in Peutz–Jeghers syndrome

BACKGROUND
Peutz–Jeghers syndrome (PJS) is a rare hereditary neoplastic disorder mainly associated with serine/threonine kinase 11 (STK11/LKB1) gene mutations. Preimplantation genetic testing can protect a patient's offspring from mutated genes; however, some variations in this gene have been interpreted as variants of uncertain significance (VUS), which complicate reproductive decision-making in genetic counseling.
AIMS
To identify the pathogenicity of two missense variants and provide clinical guidance.
METHODS
Whole exome gene sequencing and Sanger sequencing were performed on the peripheral …

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December 28, 2023

Date of Revision

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