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Mismatch repair deficiency in adult granulosa cell tumours

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Introduction and objective: Adult granulosa cell tumours (AGCTs) are rare ovarian malignant neoplasms. Their aetiopathogenetic mechanisms remain largely unelucidated. Lately, defects in mismatch repair (MMR) have been implicated in the pathogenesis of AGCTs. The demonstration of MMR deficiency in these tumours can help identify patients potentially eligible for immune checkpoint inhibition therapy. The present study was carried out to explore the role of MMR deficiency in the aetiopathogenesis of AGCTs.

Methodology: This was a retrospective study conducted on histopathologically confirmed AGCT cases. MMR protein expression was evaluated by immunohistochemistry (IHC) on tissue microarrays using an antibody panel of MSH2, MSH6, MLH1, and PMS2.

Results: A total of 40 ovarian AGCTs evaluated for MMR deficiency, none demonstrated loss of expression of any of the four MMR proteins.

Conclusion: The results of our preliminary study show that there is no association between MMR deficiency with AGCT. Nevertheless, larger multicentre studies are needed to confirm or refute this observation.

Keywords: adult granulosa cell tumour; mismatch repair; microsatellite instability; immunohistochemistry; tissue microarray

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