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A case series of proven and likely de novo MMR mutations.

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If you have a possible case of de novo MMR mutation, please contact me. It will help if you can answer the questions below, but it is appreciated that it may not be possible to address all the points.

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1. Your name, institution and who else you would like credited on the paper.

2. The age, gender and brief description (e.g. occupation) of the patient. Do they have any unusual medical history or problems?

3. Who referred the patient to you and why? To whom did the patient present first, e.g. surgeon or geneticist?

4. Details of cancer/s, age of onset, histology, staging. Were any polyps present? Was polyposis present or excluded?

5. Treatment of cancers, outcome, survival, subsequent medical problems.

6. Family history, presence of Lynch Syndrome-associated cancers, other cancers/tumours/malignancies, parental age and health, number and gender of siblings. Suspicious deaths in family. Please note if cancers are confirmed or not. Did the proband have offspring?

7. Was MSI performed, how many/which markers showed instability? Was *BRAF* V600E testing carried out, what was the result?

8. Was Immunohistochemistry performed, what did it show?

9. Which genes were sequenced? Was MLPA performed?

10. What is the mutation? Has it been previously reported on the InSiGHT database?

11. Have the parents or other family members been tested for the mutation – what was/were the results?

12. Has non-paternity, non-maternity or adoption been ruled out? How was this tested?