

Protocol for surveillance in individuals with constitutional mismatch repair deficiency (CMMRD)

This guideline for the diagnosis, surveillance and management of people with constitutional mismatch repair deficiency has been drawn from the best available evidence and the consensus of experts in this area and it is regularly updated to reflect changes in evidence.

The expectation is that clinicians will follow this guideline unless there is a compelling clinical reason to undertake different management, specific to an individual patient.



**European
Reference
Network**

for rare or low prevalence
complex diseases

 **Network**
Genetic Tumour Risk
Syndromes (ERN GENTURIS)



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| Exam | | Frequency | Period | Strength* |
|---|---|---------------------------------|---|-----------|
| Clinical examination | | Every 6 months | From diagnosis | Strong |
| Brain MRI | | Every 6 months | 2 – 20 years | Strong |
| | | Annually | From 20 years | Moderate |
| Colonoscopy | | Annually** | From 6 years | Strong |
| Upper gastrointestinal endoscopy | | Annually** | Simultaneously with colonoscopy or at least from age 10 years | Weak |
| Video capsule endoscopy | | Annually | From 10 years | Moderate |
| Gynaecologic | Surveillance (clinical examination & transvaginal ultrasound) | Annually | From 20 years | Strong |
| | Prophylactic surgery | Not applicable | Discuss once family planning is completed | Moderate |
| Abdominopelvic ultrasound for gynaecological and urinary tract cancer screening | | Annually | From 20 years | Strong |
| Whole body MRI | | At least once | At diagnosis or when anaesthesia is no longer required | Strong |
| | | Discuss optional annual imaging | | Moderate |

* This grading is based on published articles and expert consensus: strong – expert consensus AND consistent evidence, moderate – expert consensus WITH inconsistent evidence AND/OR new evidence likely to support the recommendation, weak – expert majority decision WITHOUT consistent evidence.

**Interval should be increased to once every 6 months once polyps are detected.