Short human genetics opinion for a health insurer

Lynch syndrome – surveillance colonoscopy interval

Human genetics opinion on reimbursement of annual colonoscopies in a 27-year-old carrier of a pathogenic MLH1 variant

Fictitious sample case – case, persons, events and instruction are simulated, not a real expert opinion. Cited sources are real.

Instructed by	Private health insurer (private Krankenversicherung) [fictitious], Outpatient Benefits Department	Reference	KV-2026-13075 [fictitious]
Insured person	J. S. [fictitious], born 1999	Date	8 November 2026

Result in brief

The German S3 guideline recommends colonoscopies in Lynch syndrome as a rule every 24 months. In the presence of risk factors, explicitly including a variant in *MLH1*, the interval can be shortened to a minimum of 12 months. For the insured person, an annual interval is therefore guideline-compliant and individually justifiable. There is no general standard of annual colonoscopy. The question of reimbursement is reserved to the insurer.

1. Instruction and question

The insurer requests an opinion on reimbursement of annual colonoscopies. It refers to recommendations that provide for an interval of two to three years. It asks whether the annual interval is medically justified and whether a longer interval is sufficient. This opinion distinguishes three levels: the guideline recommendation, the individual medical justification and the contractual question of reimbursement. The insurer decides on the third level.

2. Records

The following applies to this fictitious sample case: the 27-year-old insured man is asymptomatic. His mother developed colorectal cancer at the age of 38. Loss of MLH1 expression was noted in the tumour tissue, and a pathogenic germline variant in *MLH1* was subsequently detected in her. The insured man carries the same variant. The specific variant is not named in the fictitious files, and its classification as pathogenic is taken as an assumption of the sample case. A first colonoscopy at the age of 26, complete to the caecum and with good bowel preparation, revealed a small tubular adenomatous polyp with low-grade dysplasia, which was completely removed. The treating gastroenterologist has planned annual follow-up examinations.

3. Assessment

Risk of disease. Cancer risk in Lynch syndrome differs markedly according to gene, age and sex [1]. In the Prospective Lynch Syndrome Database, the cumulative colorectal cancer incidence under surveillance up to the age of 70 in *MLH1* carriers was 46%, the highest value of the four genes studied. First carcinomas occurred in *MLH1* and *MSH2* carriers from the age of 25 [2]. These are cohort figures, not an individual prediction.

Effectiveness of colonoscopy. In a controlled study over 15 years, colonoscopy at three-year intervals reduced the colorectal cancer rate in affected families by 62%. No death from colorectal cancer occurred in the surveillance group [3]. Carcinomas also occur under surveillance, but in the prospective database they rarely led to death [2].

Guidelines. The S3 guideline Colorectal Cancer (S3-Leitlinie Kolorektales Karzinom, version 3.2, March 2026, recommendation 5.13, grade of recommendation B, level of evidence 3) recommends starting colonoscopy between the ages of 20 and 25 in *MLH1* carriers. Colonoscopy should be repeated every 24 months. In the presence of risk factors such as previous colorectal cancer or a high-risk gene (*MLH1*, *MSH2*), the interval can be shortened to a minimum of 12 months. In the background text, the guideline additionally names male sex and age over 40 years [4]. The European guideline of the EHTG and ESCP

recommends colonoscopies for *MLH1* carriers from the age of 25 at intervals of two or three years [5]. A comparative study from three countries with different intervals found no difference in colorectal cancer incidence or tumour stage [6].

Individual justification. Two risk factors expressly named in the guideline are present in the insured man: a variant in the high-risk gene *MLH1* and male sex. In addition, there are the very early age of onset in his mother and an adenoma already removed at the age of 26. The previous examination was complete and of good quality, so the shortened interval does not serve to compensate for an inadequate examination. An annual interval thus lies within the guideline recommendation. However, there is no evidence of superiority over the 24-month interval [6].

4. Answers to the questions

Question 1: Is a colonoscopy interval of twelve months medically justified?

> **Answer:** Yes. It corresponds to the shortening for *MLH1* expressly provided for in the S3 guideline and is individually justified by the family and personal history. It is a discretionary medical decision within the guideline, not a general standard.

Question 2: Is an interval of 24 months sufficient?

> **Answer:** An interval of 24 months would correspond to the standard recommendation of the S3 guideline and would also be professionally justifiable. Both intervals lie within the professional standard. What consequence this has for reimbursement is a contractual question.

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