Supplementary Online Content


eSupplement. Equation for Prediction of Pathogenic APC and Biallelic MUTYH Mutations

This supplementary material has been provided by the authors to give readers additional information about their work.
eSupplement. Equation for Prediction of Pathogenic APC and Biallelic MUTYH Mutations

Predicted probability of a pathogenic APC mutation = \( \frac{\exp(L_{APC})}{1 + \exp(L_{APC}) + \exp(L_{MUTYH})} \)

Predicted probability of pathogenic biallelic MUTYH mutations = \( \frac{\exp(L_{MUTYH})}{1 + \exp(L_{APC}) + \exp(L_{MUTYH})} \)

Where:

\[
L_{APC} = 1.7821 - 1.5525\times(V1) - 0.7148\times(V2) + 0.2452\times(V3) + 2.1359\times(V4) + 3.2522\times(V5) - 0.0724\times(V6) - 0.1620\times(V7) - 0.0224\times(V8) + 0.5489\times(V9)
\]

\[
L_{MUTYH} = -3.117 + 0.0817\times(V1) + 1.1556\times(V2) + 1.9642\times(V3) + 2.5692\times(V4) + 1.6877\times(V5) - 0.0129\times(V6) + 0.7661\times(V7) - 0.0173\times(V8) - 0.1965\times(V9)
\]

Definition of Variables

\( a, b \)

Variable | Definition
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V1 | Presence of 1-9 adenomas in the proband
Set to 0 if absent, 1 if present
V2 | Presence of 10-19 adenomas in the proband
Set to 0 if absent, 1 if present
V3 | Presence of 20-99 adenomas in the proband
Set to 0 if absent, 1 if present
V4 | Presence of 100-999 adenomas in the proband
Set to 0 if absent, 1 if present
V5 | Presence of \( \geq 1000 \) adenomas in the proband
Set to 0 if absent, 1 if present
V6 | Proband’s age (in years) when first diagnosed with an adenoma
(Age at adenoma diagnosis should be estimated if unknown)
V7 | Presence of colorectal cancer diagnosis in the proband
Set to 0 if absent, 1 if present
V8 | Proband’s age (in years) when first diagnosed with colorectal cancer
(Age at colorectal cancer diagnosis should be estimated if unknown; if V7=0, then V8 must be 0)
V9 | Presence of a first-degree relative with colorectal cancer
Set to 0 if absent, 1 if present

\( a \) “Proband” refers to the individual being evaluated

\( b \) Variables V1 - V5 are mutually exclusive

If any one of these variables = 1, the remaining four variables must be set to 0
For example, if a proband has 20-99 adenomas, then V3 = 1, and V1, V2, V4, and V5 must be set to 0

**NOTE:** The model was developed from a selected sample, where participants had undergone testing based on a personal or family history suggestive of a polyposis syndrome. Therefore, the prevalence estimates, particularly in the groups with few numbers of individuals in the development cohort must be interpreted with caution because of statistical uncertainty. The model should only be used to predict the probability of APC and MUTYH mutations in an individual with a history of \( \geq 10 \) adenomas.

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