Genetisk udredning og mutationsscreening ved arvelige disposition til kolorektal cancer

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Hvem bør der tilbydes mutationsscreening?

1 tilfælde af CRC <50 år

Familier der opfylder FCC kriterier

Ved mistanke om polypose (5-10 adenomer afh. af alder, morfologi, dysplasigrad og familieanamnese)

Patienter med 1 eller flere juvenile polypper

I øvrigt påfaldende familieanamnese

Eksisterende gener inkluderet i CRC genpakke

MLH1, MSH2, MSH6, PMS2, EPCAM, APC, MUTYH, PTEN, STK11, BMPR1A, SMAD4

Nye gener og kontrolprogram ved fund af mutation[1]

Gen	Arvegang	Kontrolprogram
AXIN2[2-7]	AD	AFAP-progr. (koloskopi start senest 25 + øvre skopi)
GREM1[8-10]	AD	AFAP-progr. (Koloskopi start senest 25 + øvre skopi)
MSH3[11]	AR	AFAP-progr. (Koloskopi start senest 25 + øvre skopi)+ evt. "bred opmærksomhed"
NTHL1[12-14]	AR	*MUTYH*-progr. (koloskopi start senest 25 + øvre skopi). Endometriekontrol (UL) hvert 2. år fra 40 år. Opmærksomhed på øget risiko for bryst- og hudcancer.
POLD1[15-18]	AD	Koloskopi hvert 2 år fra 20 år. Endometriekontrol (UL) hvert 2. år fra 40 år
POLE [15-21]	AD	Koloskopi hvert 2. år fra 20 år. Overvej gastroskopi måske hvert 3 år fra ca 25 år
RNF43[22-25]	AD	Koloskopi hvert 5 år fra 40 år eller 10 år før yngste i familier, ikke øvre skopier

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