Nový hereditární syndrom u karcinomu prsu

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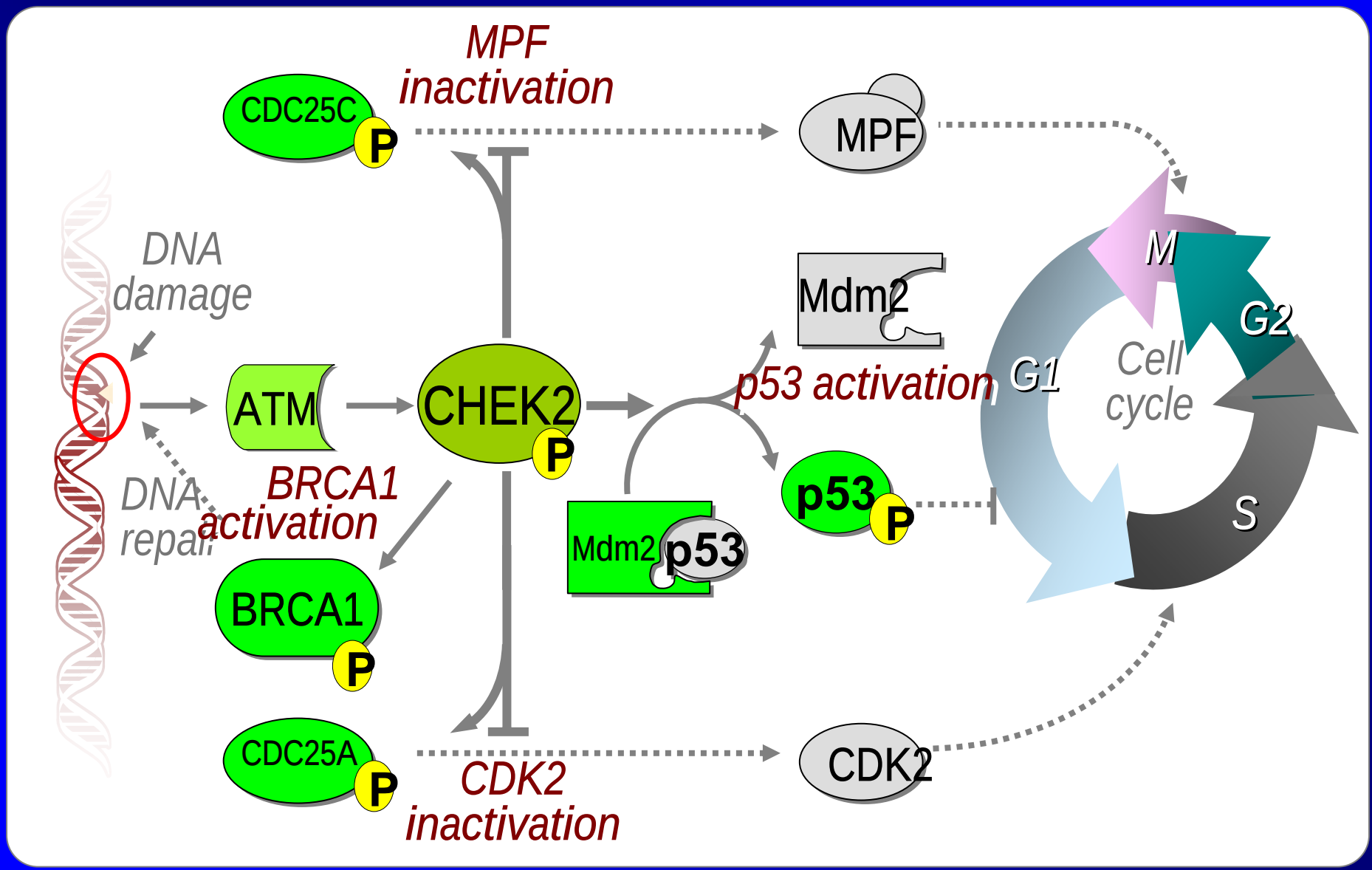
Geny zodpovědné za hereditární formy karcinomu prsu

BRCA1/2	*60 – 80%*
P53	*40 – 60%*
STK11	*15,2x vyšší než průměr*
PTEN	*25 – 50%*
ATM	*20 – 40%*

Nově identifikován další gen

CHEK2	***40%***

CHEK2: funkce



CHEK2: alterace

Nejčastější:

1100delC

Dlouhé delece 9 a 10 exonu

CHEK2 a jiné malignity

- Karcinom m. měchýře 1,9x

CHEK2 a karcinom prsu

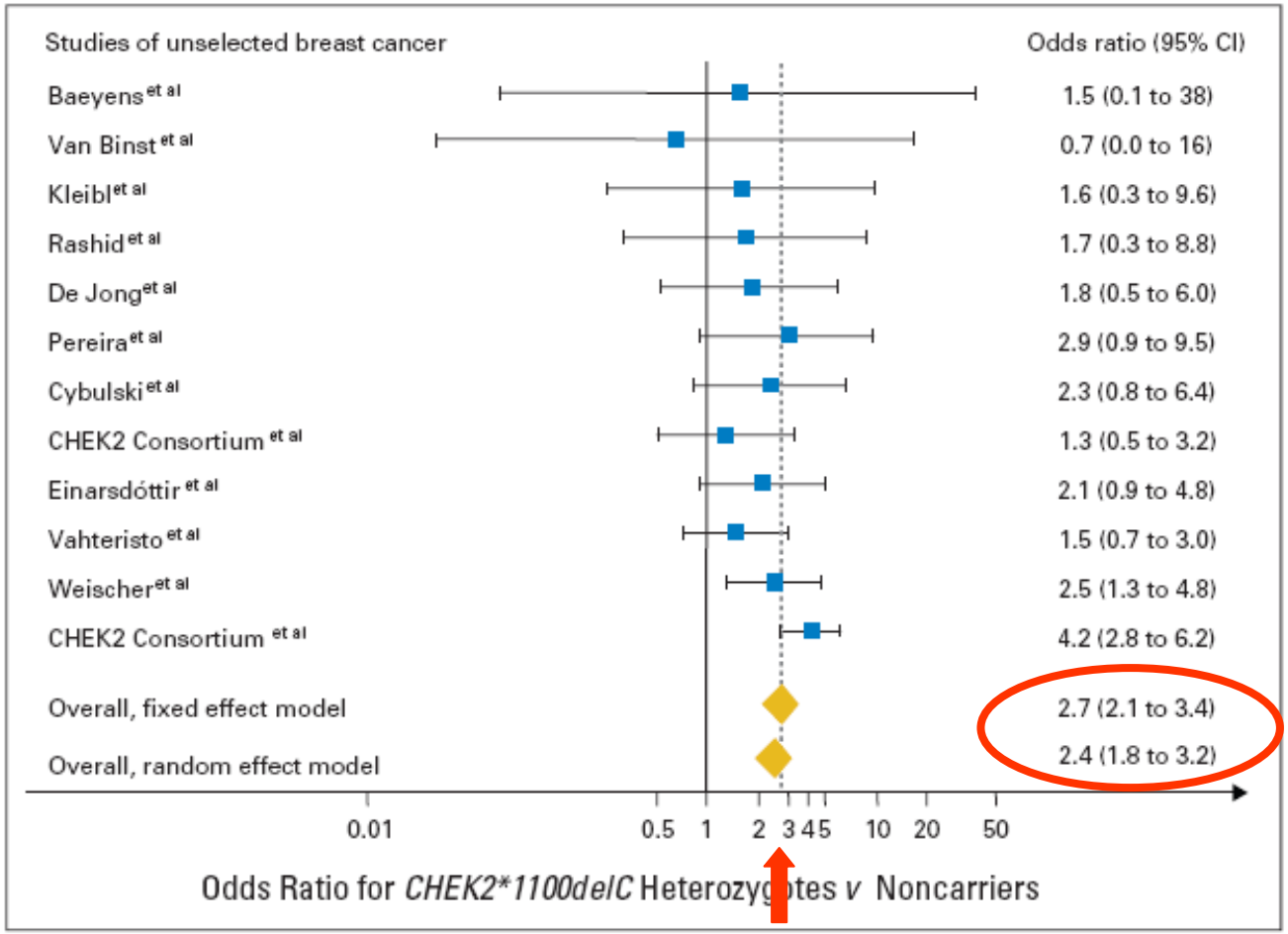


Fig 1. Meta-analysis of the risk of unselected breast cancer for *CHEK2**1100delC heterozygotes versus noncarriers. Studies appear in weighted order and start with the smallest contributor at the top.

CHEK2 a karcinom prsu

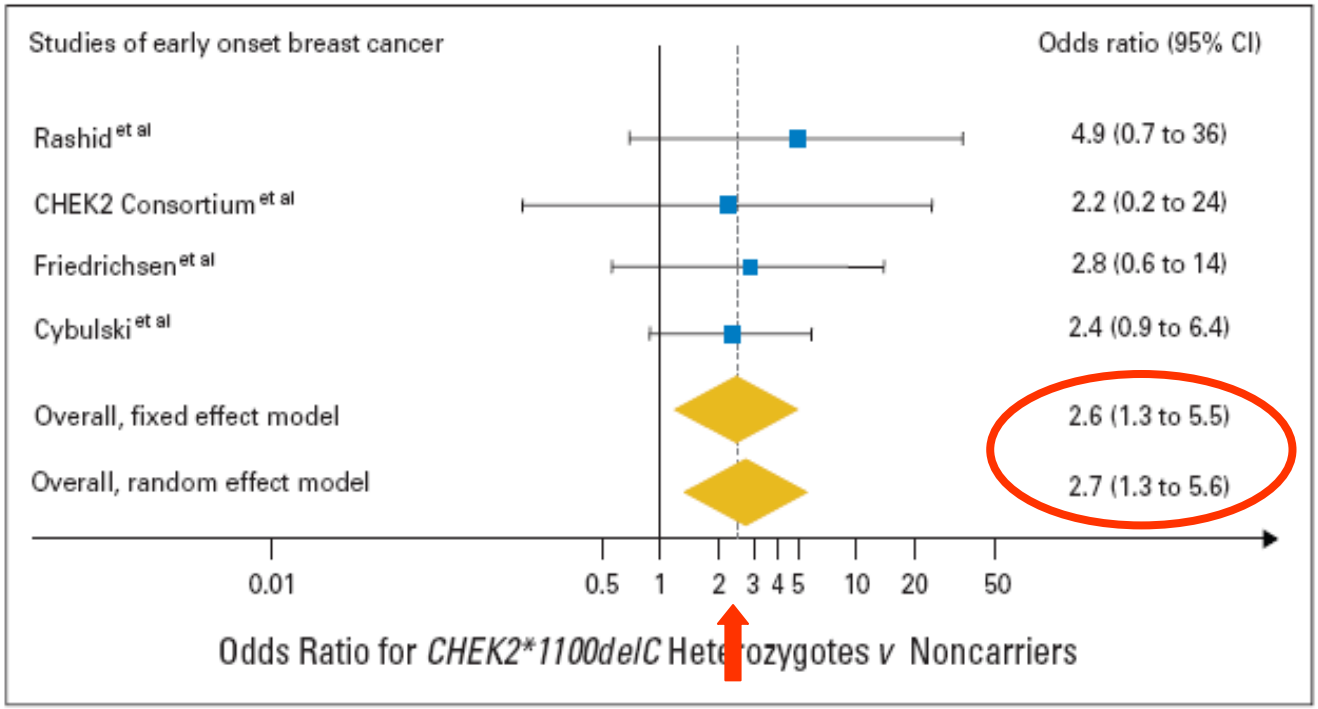


Fig 2. Meta-analysis of the risk of early-onset breast cancer for *CHEK2**1100delC heterozygotes versus noncarriers. Studies appear in weighted order and start with the smallest contributor at the top.

CHEK2 a karcinom prsu

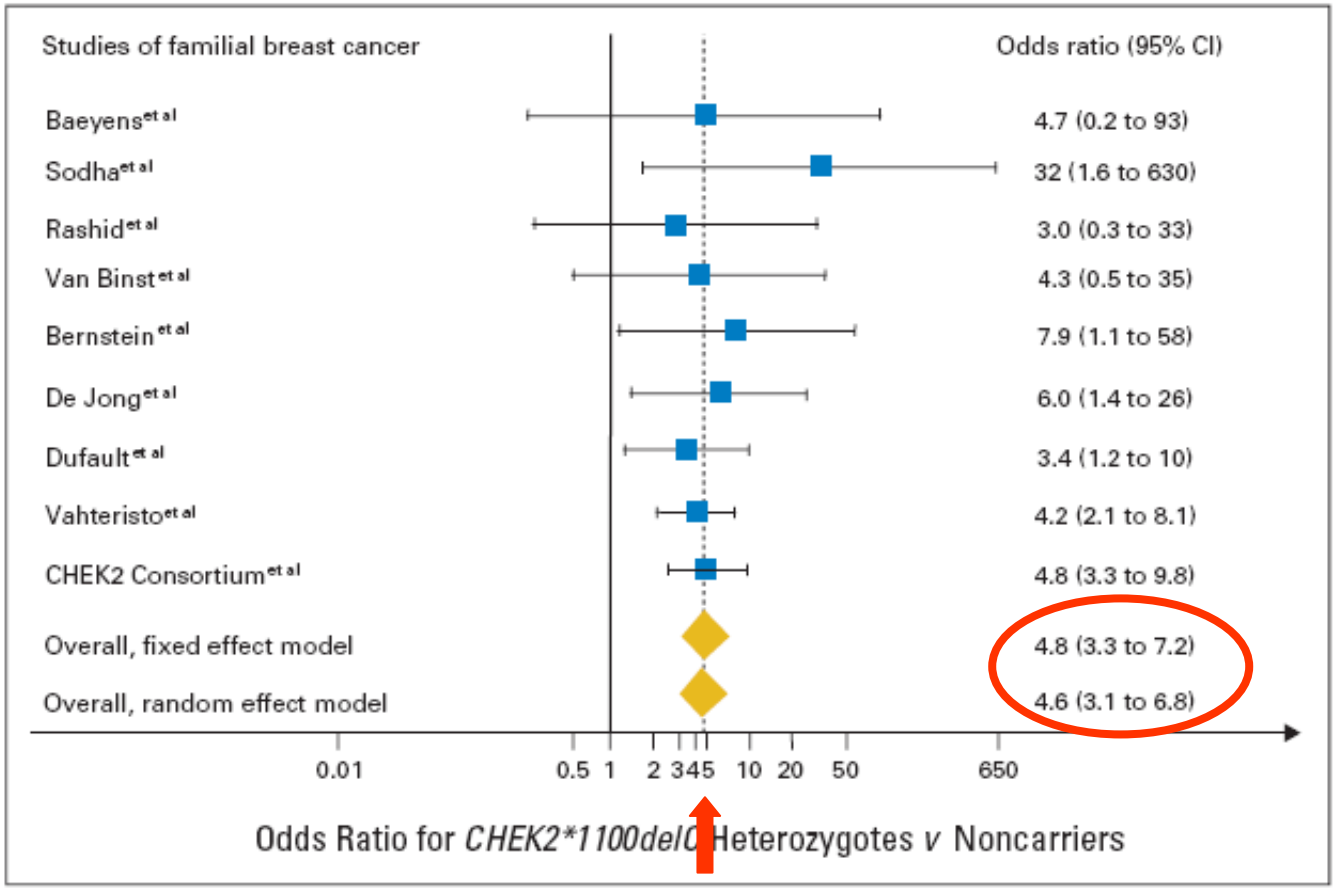
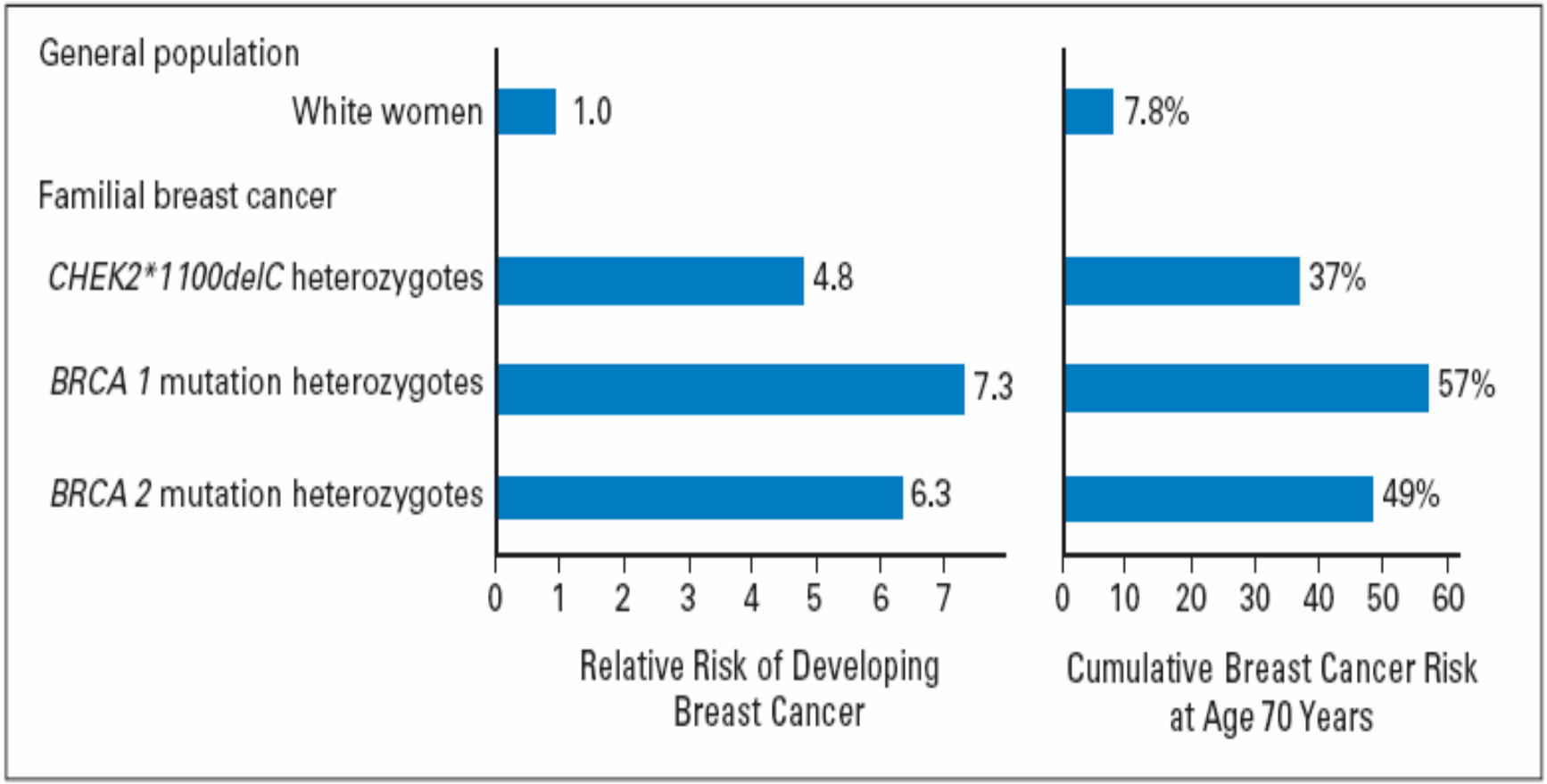


Fig 3. Meta-analysis of risk of familial breast cancer for *CHEK2**1100delC heterozygotes versus noncarriers. Studies appear in weighted order starting with the smallest contributor at the top.

CHEK2 a karcinom prsu



CHEK2 a karcinom prsu

- Výstupy pro praxi:
  - S poruchou genu *CHEK2* je spojeno vysoké riziko vzniku karcinomu prsu
  - Péče o osoby s mutací odpovídá péči o osoby s poruchou genů *BRCA1/2*
  - Testování genu *CHEK2* je rutinní součástí molekulárně genetické analýzy genů *BRCA1/2*

Děkuji za pozornost