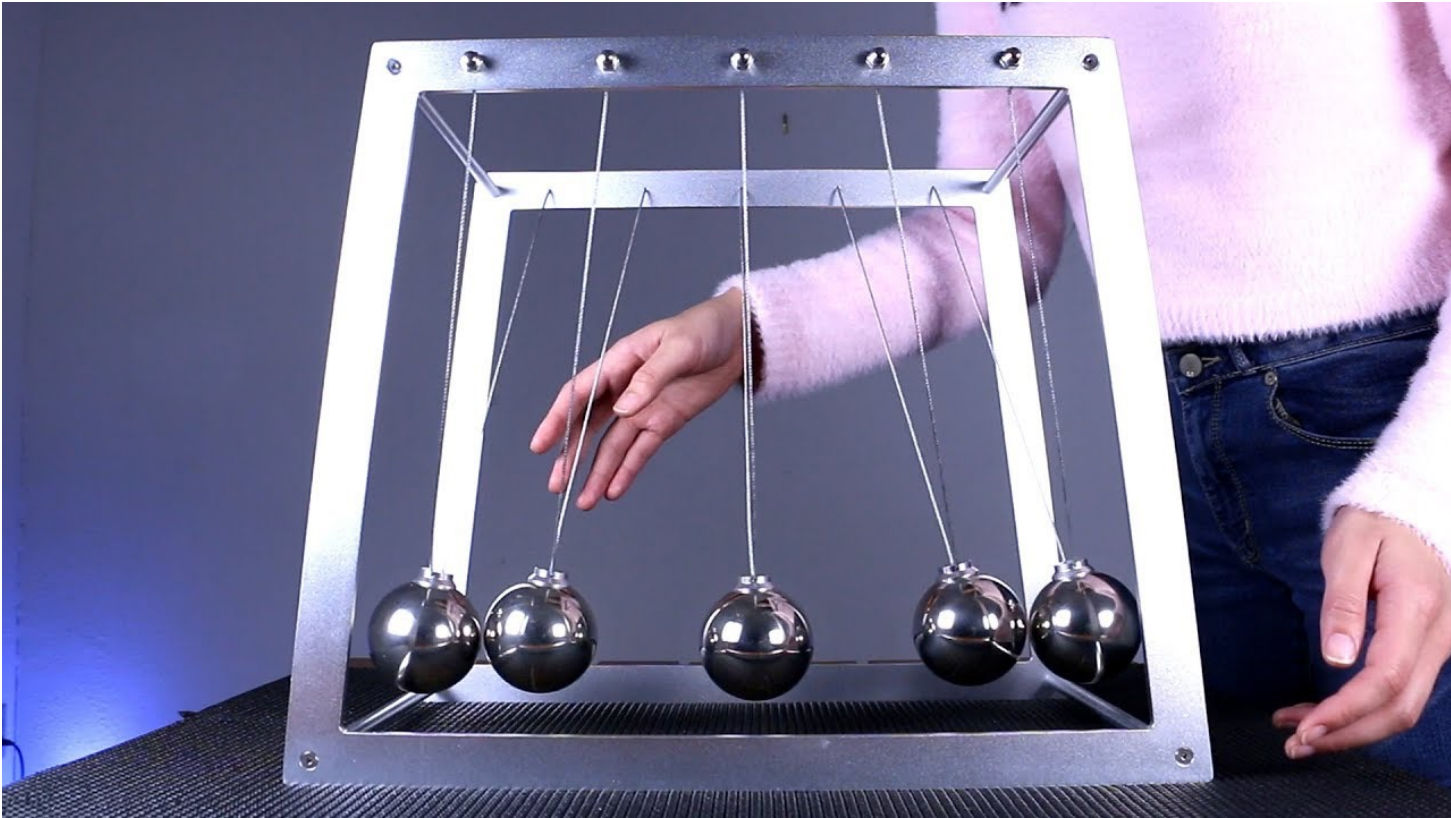
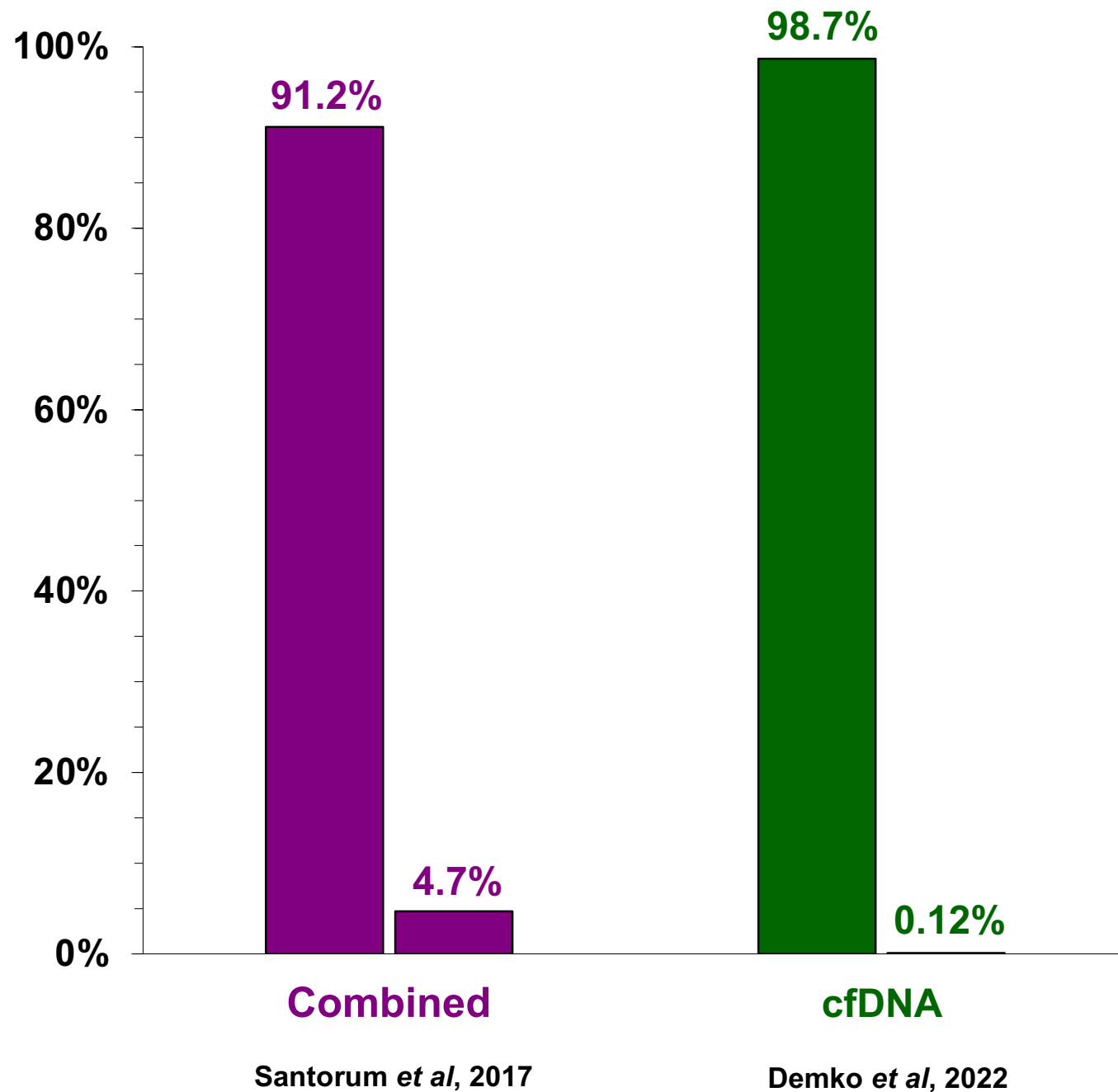


Potential of cfDNA aneuploidy screening to reduce invasive prenatal testing



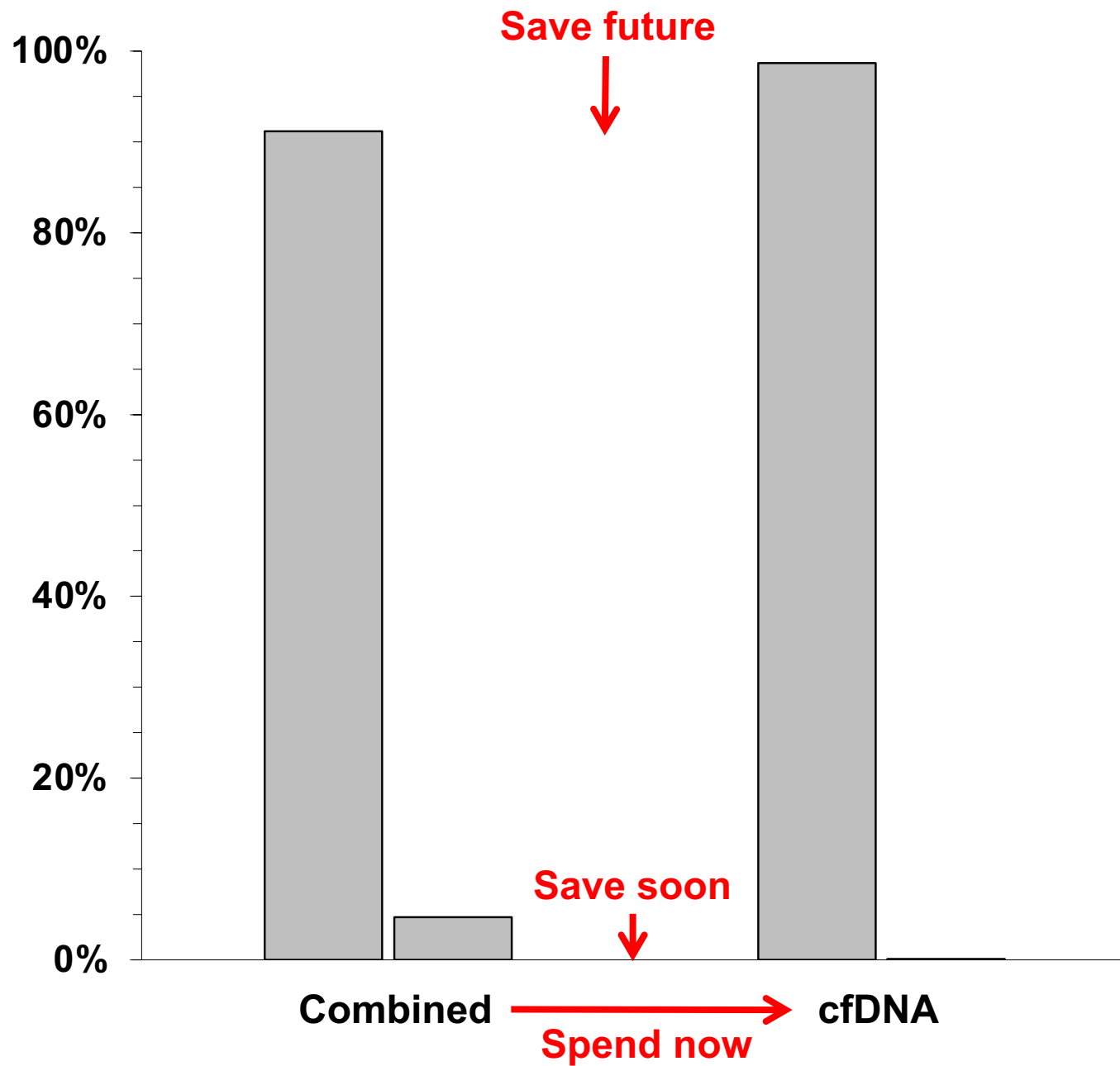
Howard Cuckle



WIN
 WIN







False-positive rate

6,0%

5,0%

4,0%

3,0%

2,0%

1,0%

0,0%

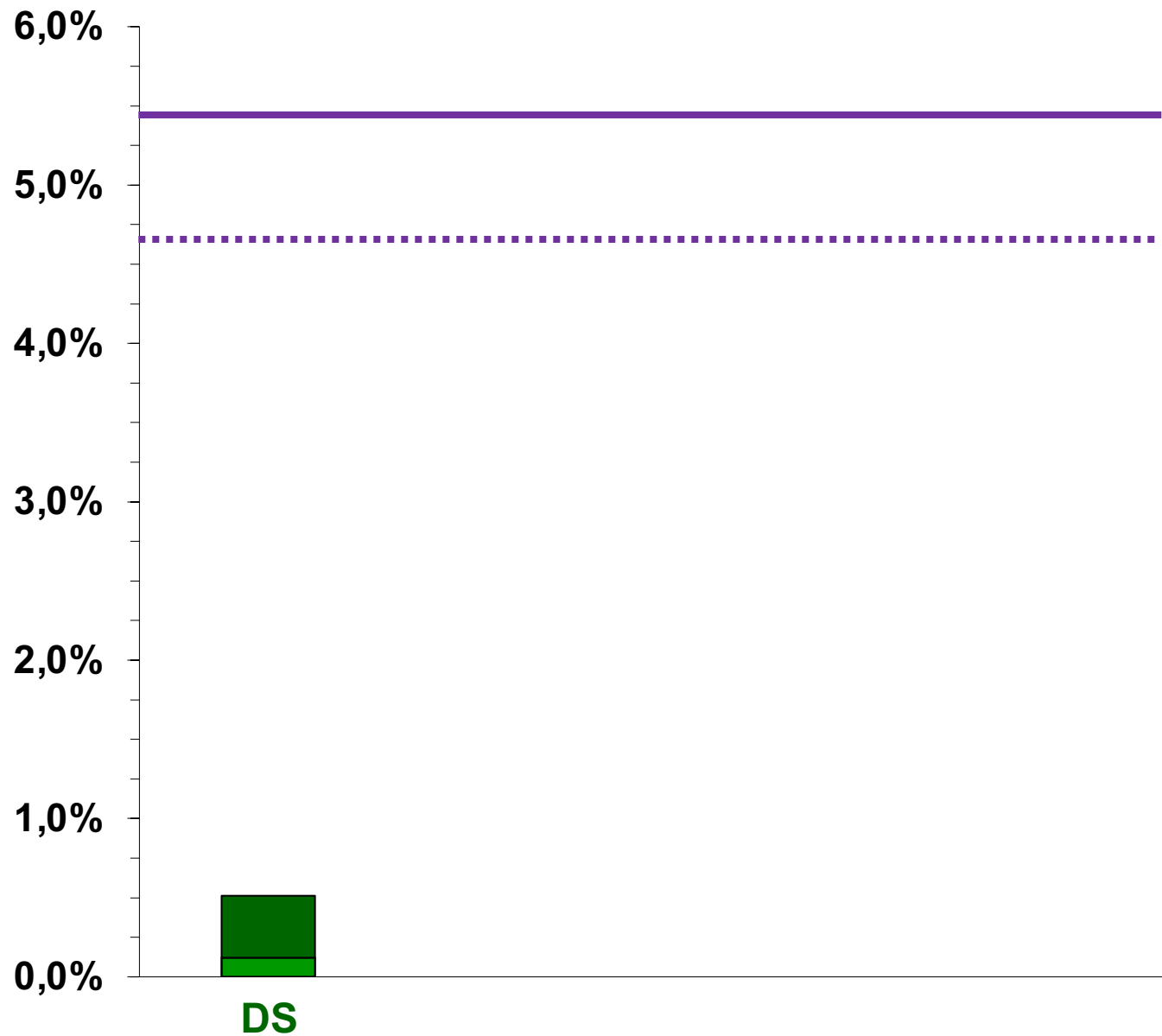
DS

Santorum *et al*, 2017

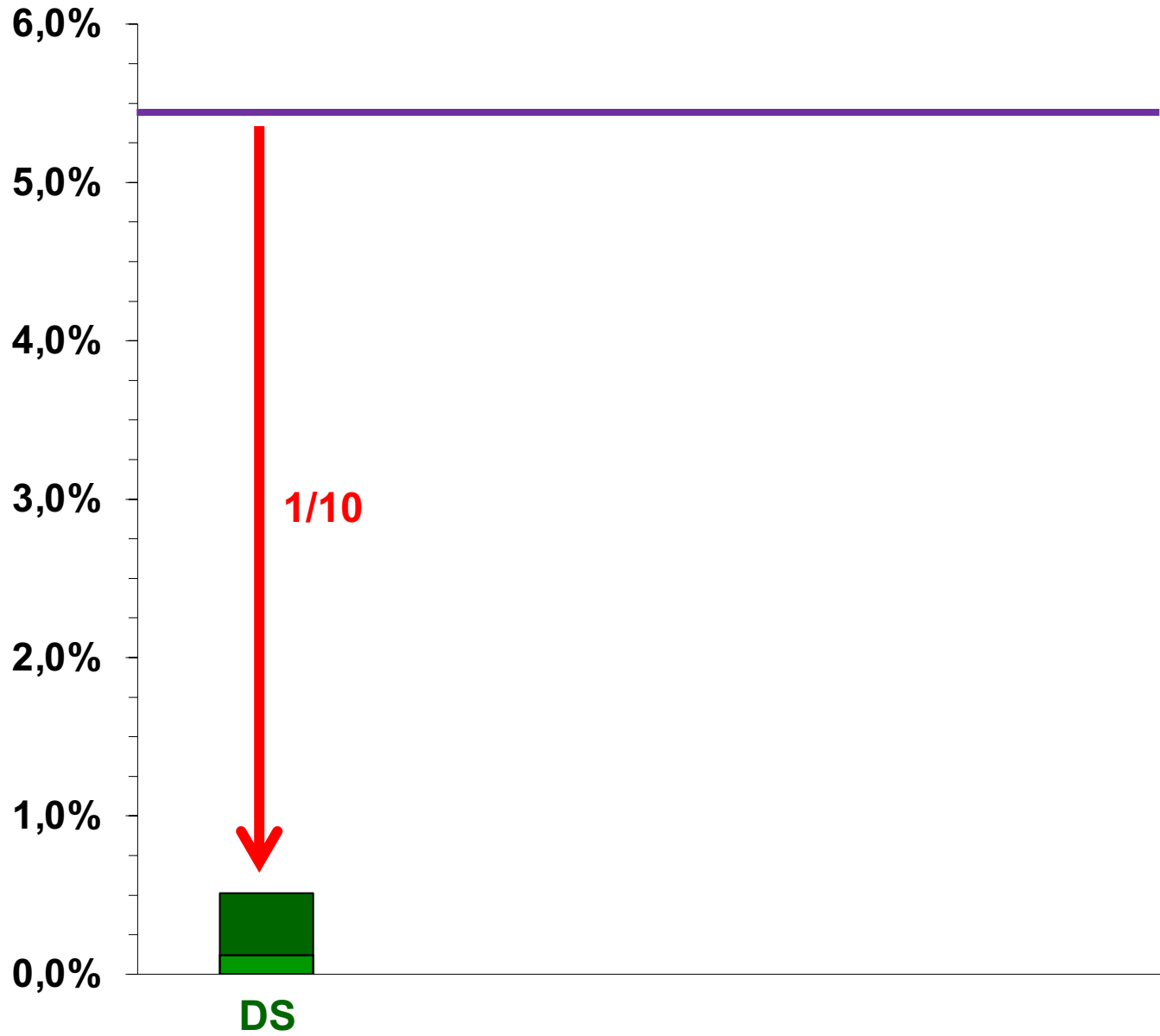
Demko *et al*, 2022



All positive rate



All positive rate



Detection rate

100%

80%

60%

40%

20%

0%

DS

ES

PS

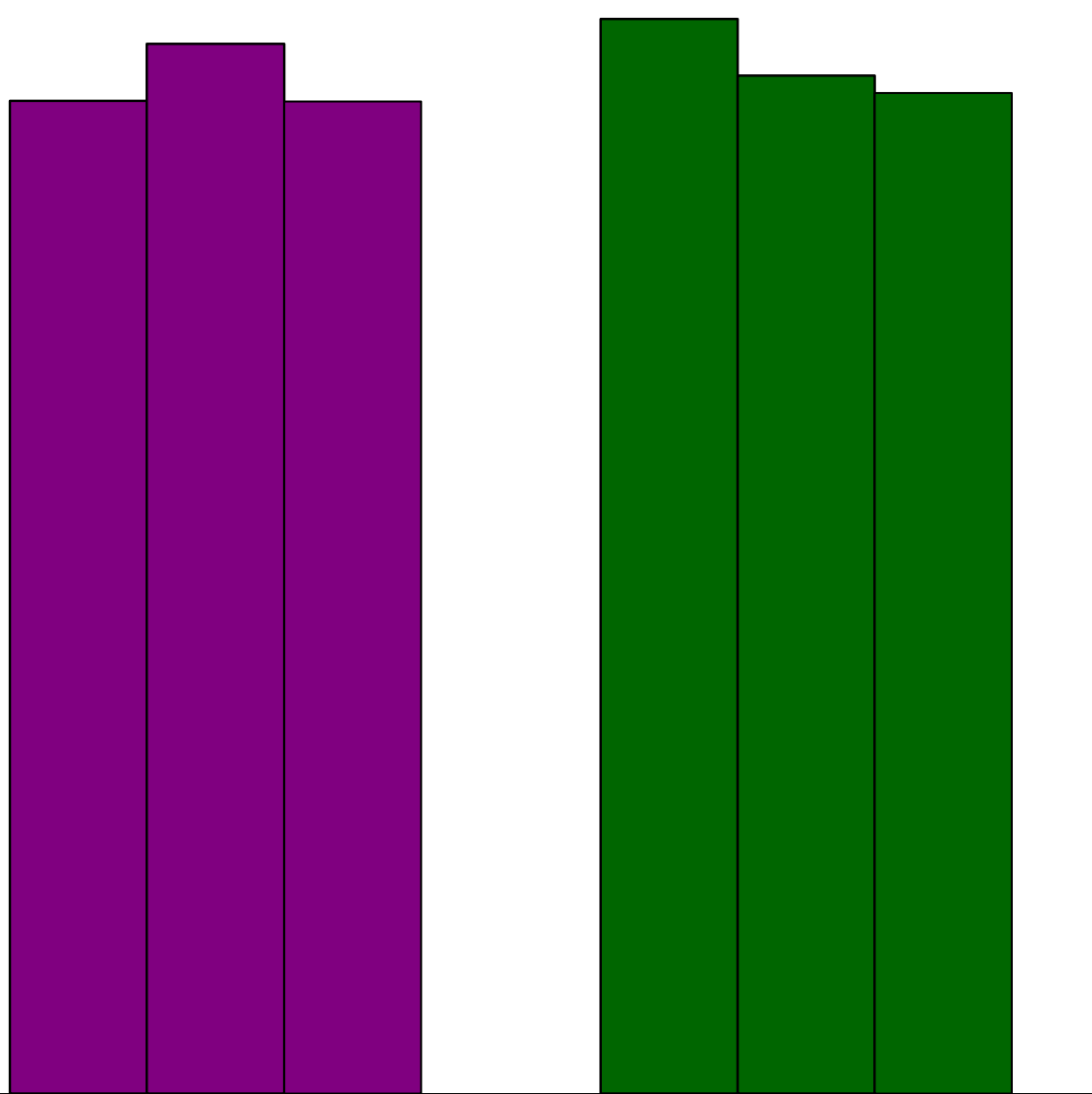
Santorum et al, 2017

DS

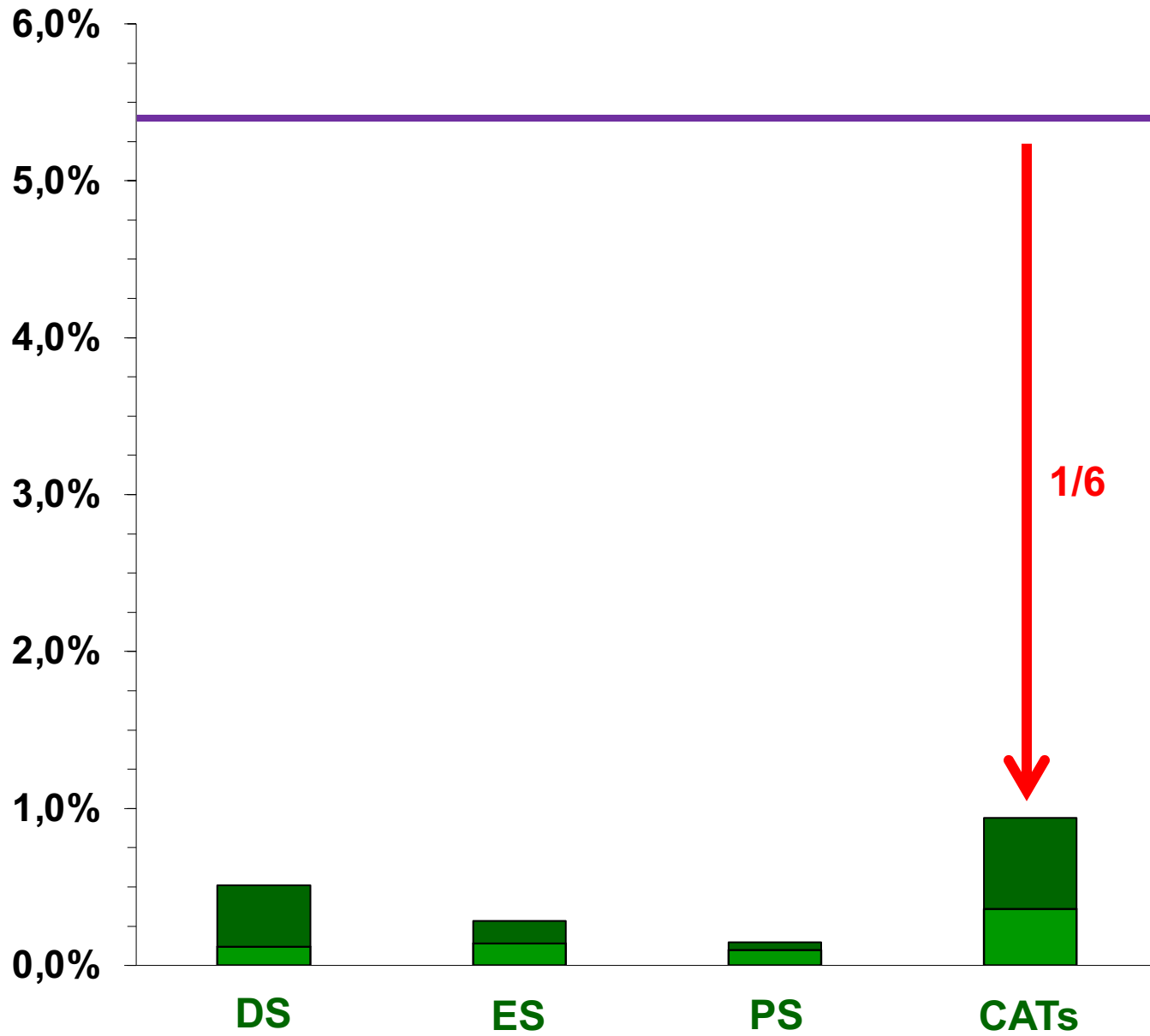
ES

PS

Demko et al, 2022



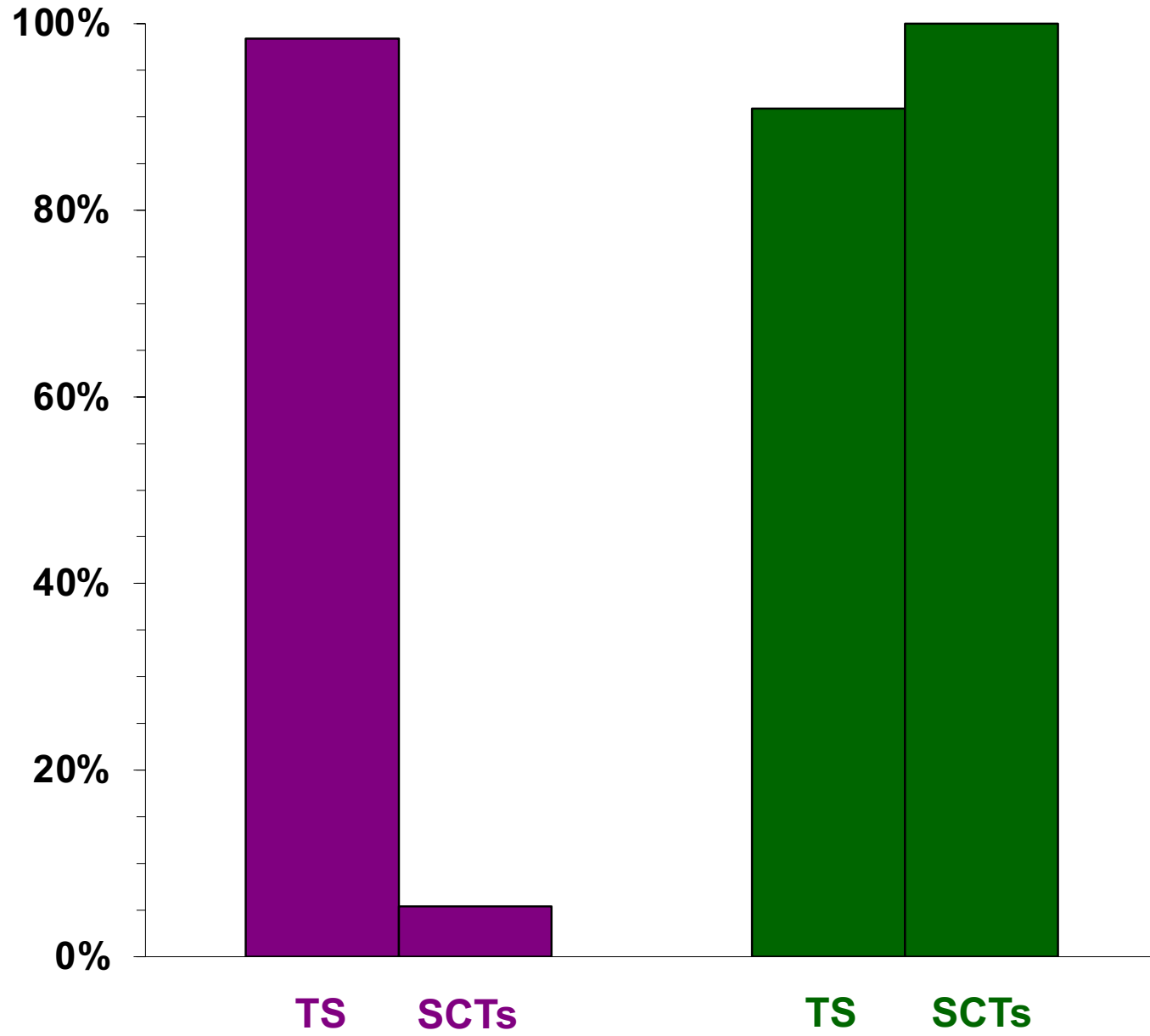
All positive rate





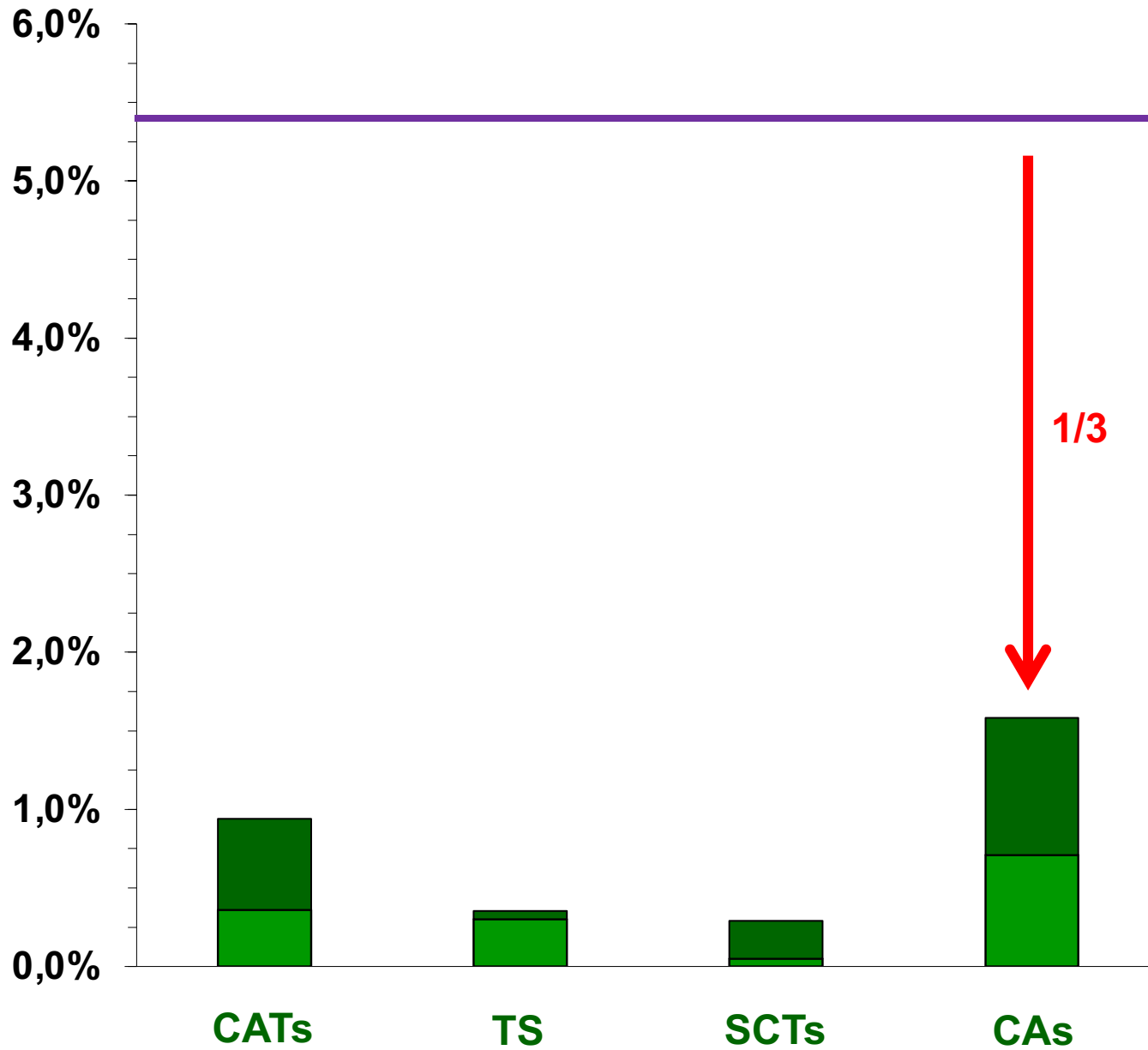
Sex Chromosome Aneuploidy

Detection rate



Santorum et al, 2017; Morris et al, 2008; Demko et al, 2022; Rose et al, 2022

All positive rate



coping is
each iteration can
classify features into
manageable.

Scope Creep

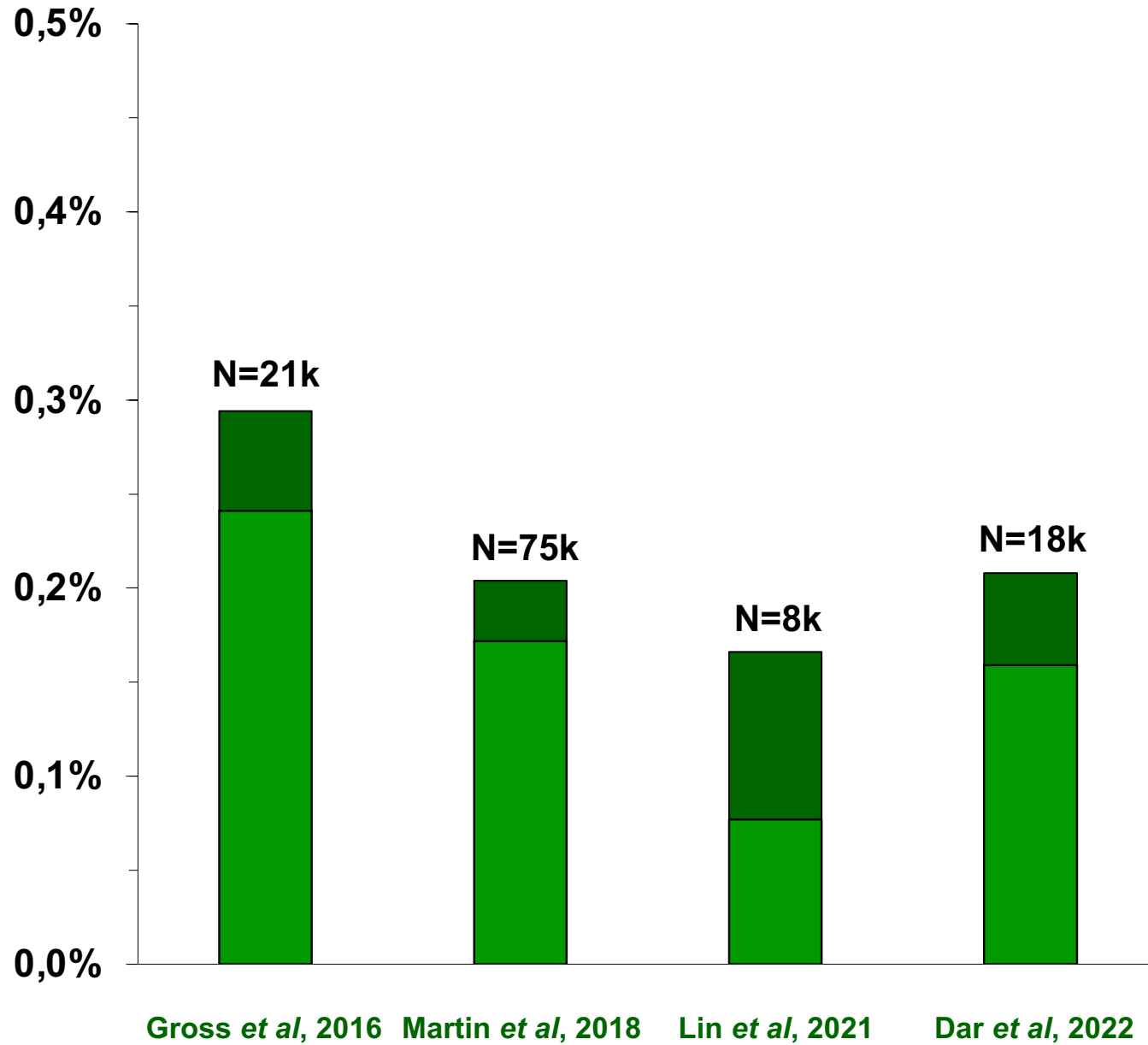
Scope creep is the gradual, progressive increase in the project's scope by the project team or the customer. Generally, when added together, these changes result in schedule overruns.

most important, c
scope creep. No
The pro
a mana

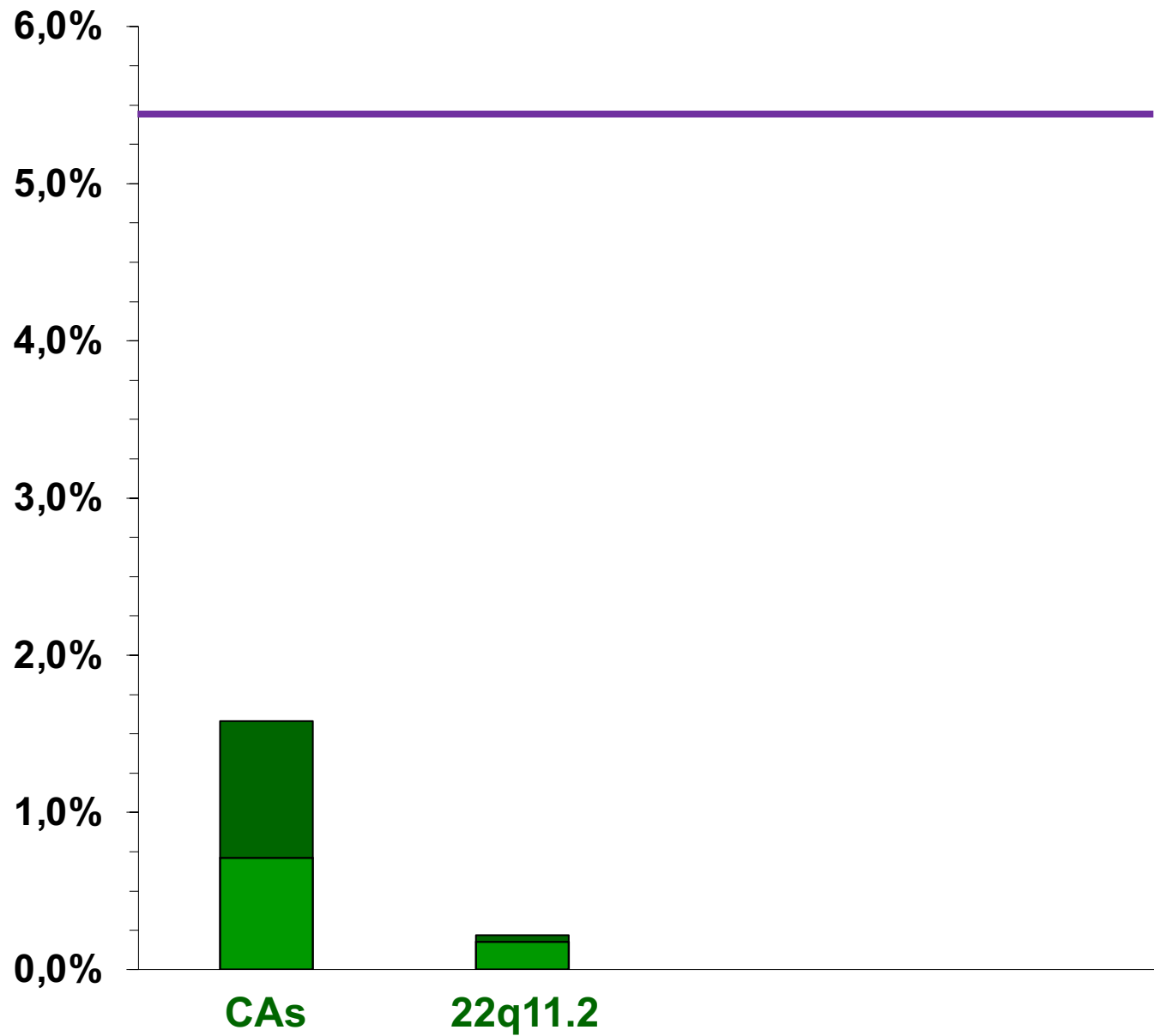
Micro-deletion syndromes

All positive rate

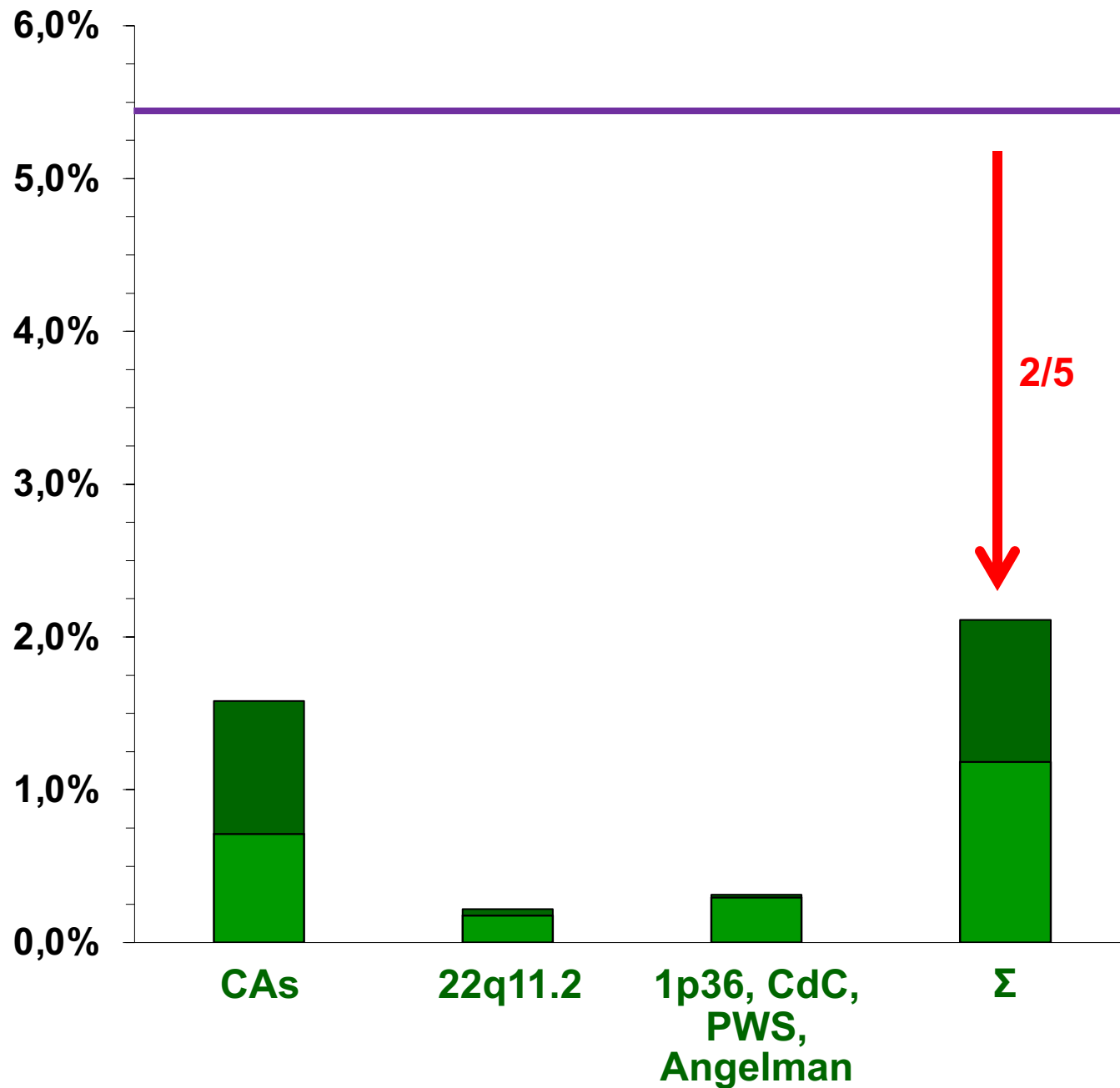
22q11.2 deletion



All positive rate



All positive rate





Rare Autosomal Trisomy & Segmental Aneuploidy

All positive rate

Rare Autosomal
Trisomy

Segmental
Aneuploidy

0,5%

0,4%

0,3%

0,2%

0,1%

0,0%

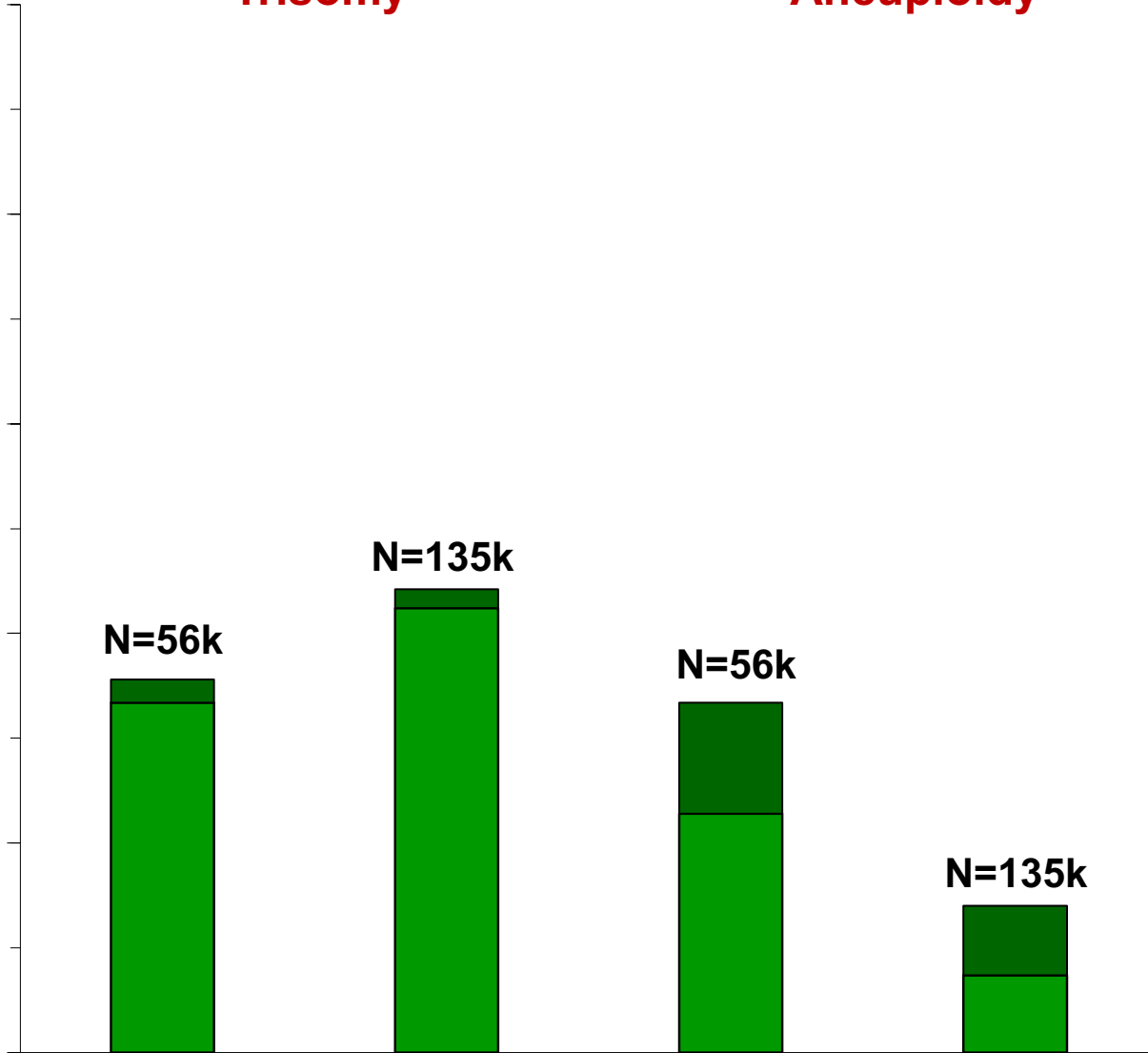
N=56k

N=135k

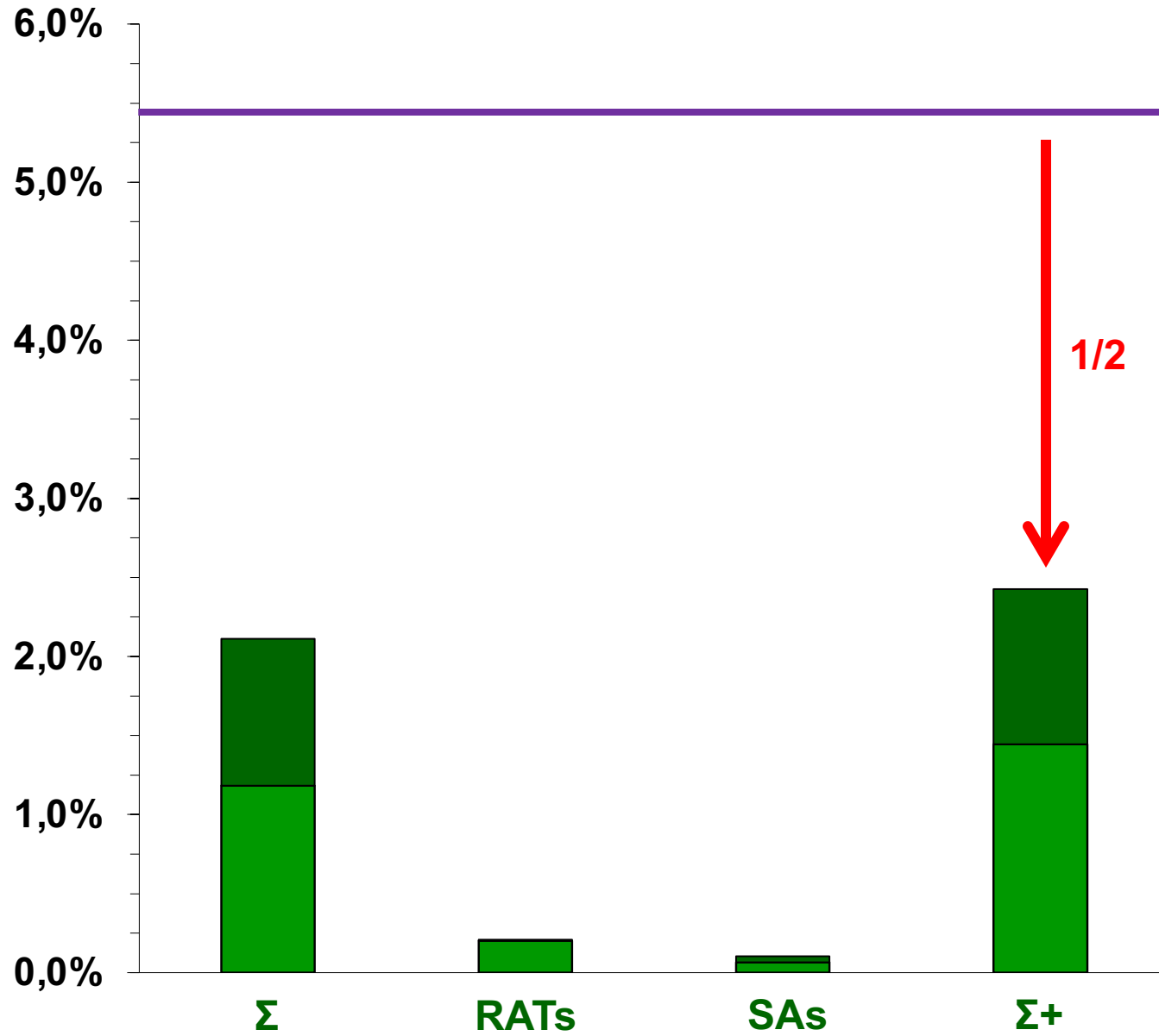
N=56k

N=135k

Meij *et al*, 2019 Bogaert *et al*, 2021 Meij *et al*, 2019 Bogaert *et al*, 2021



All positive rate





Predictive value positive

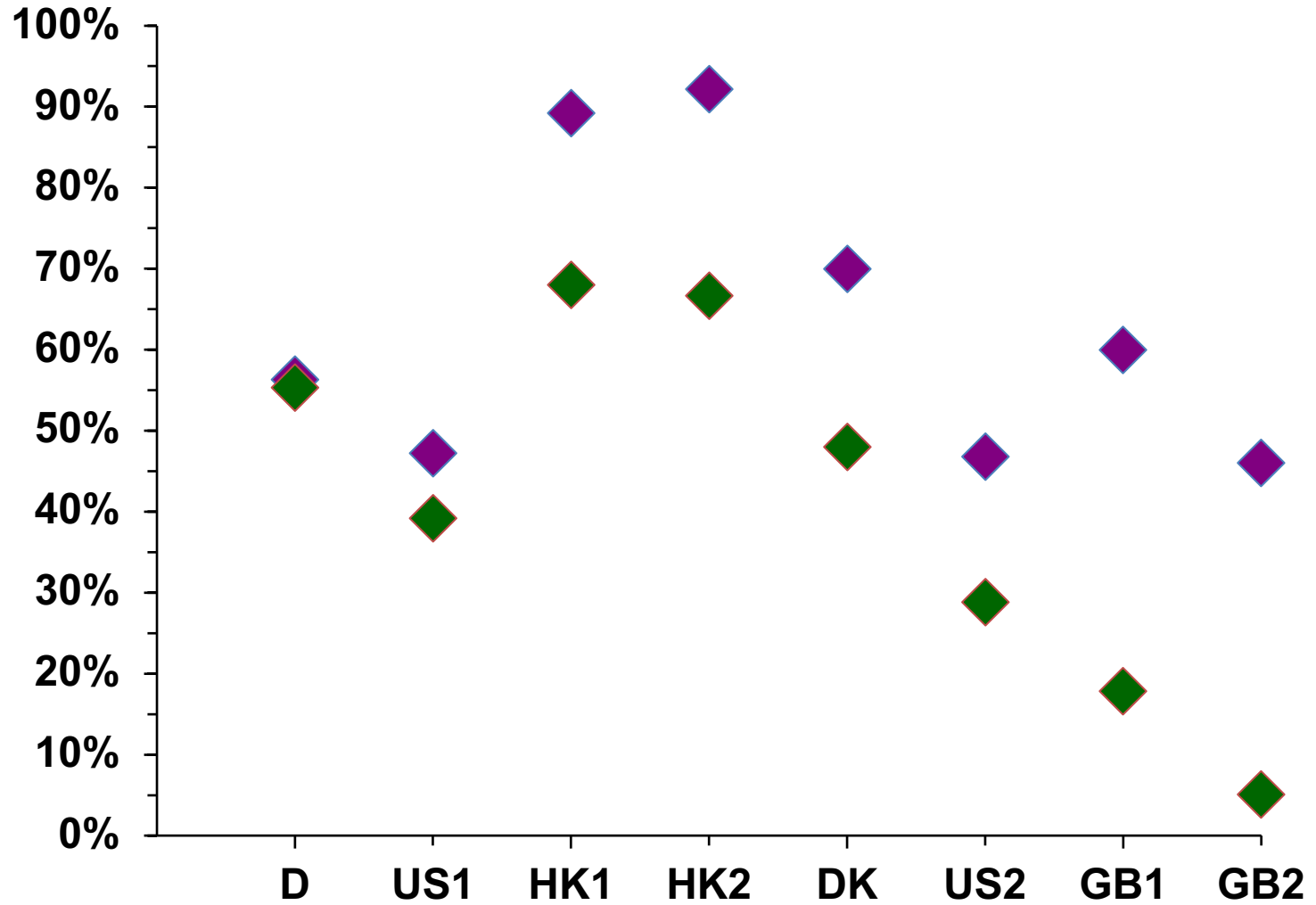
DS only=1 in 15

All=1 in 8

DS only=3 in 4

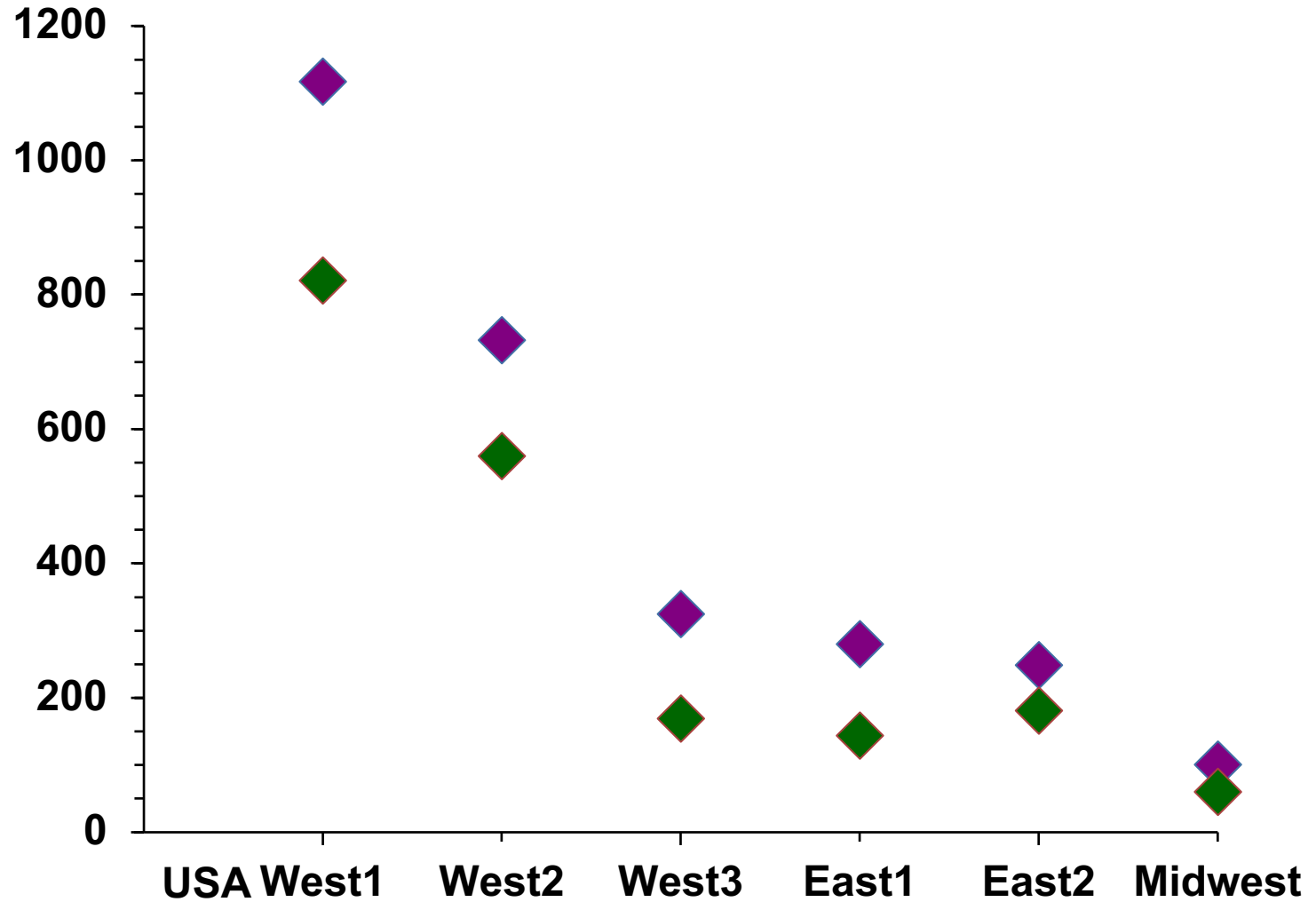
All=2 in 5

Invasive test uptake



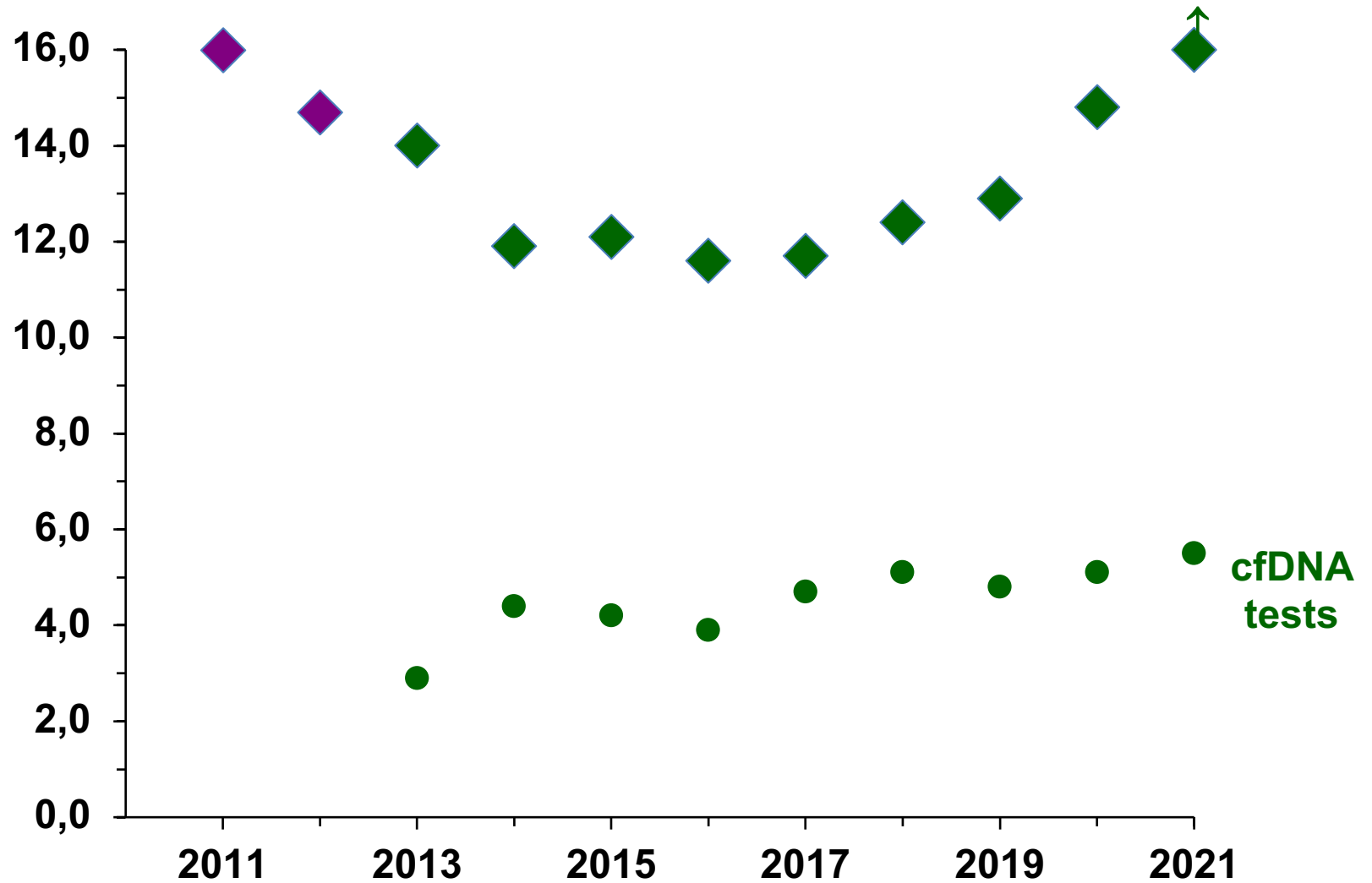
Secondary cfDNA studies worldwide

Invasive tests/year



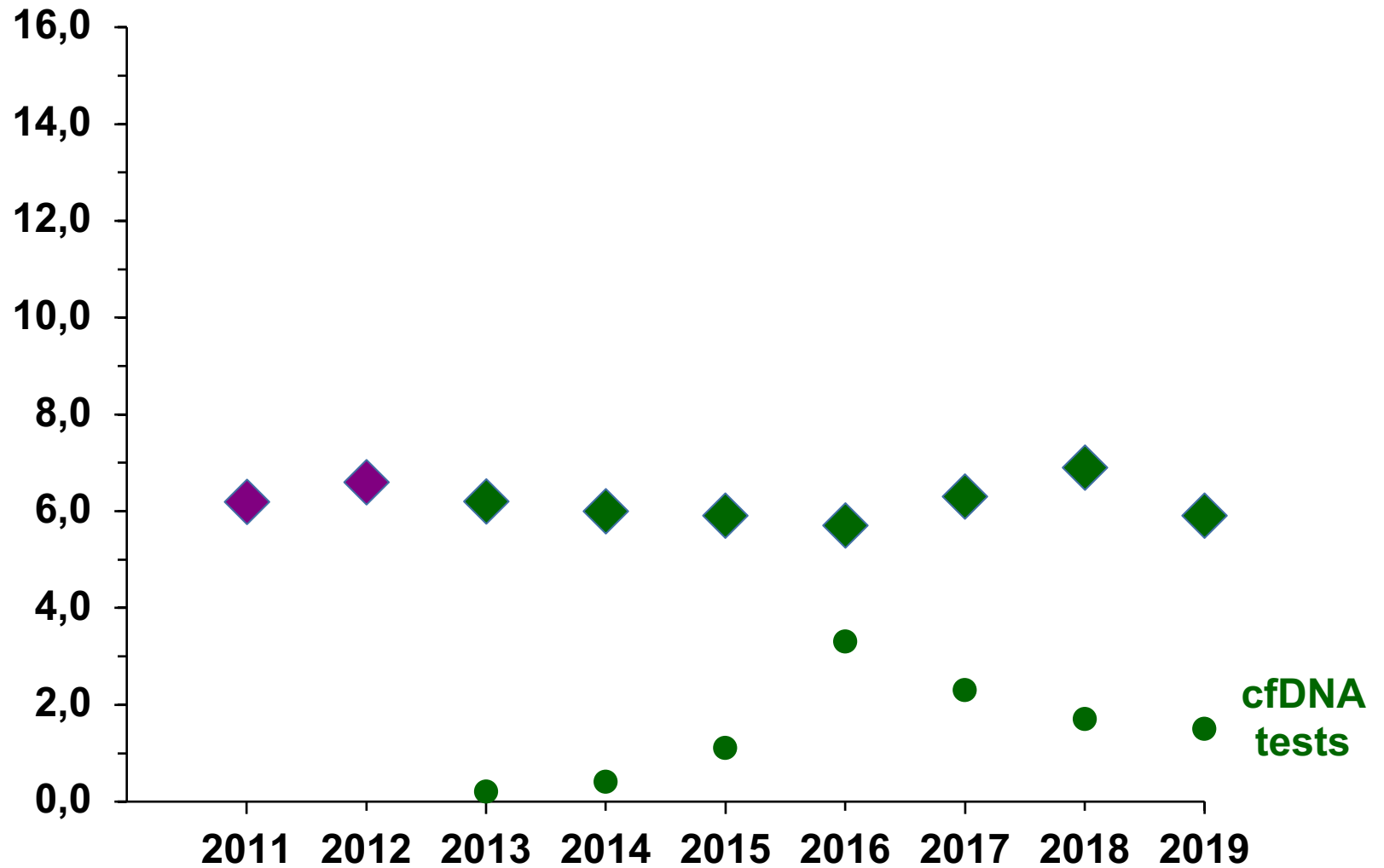
Selective cfDNA: AMA, U/S, Secondary

Invasive tests (%)



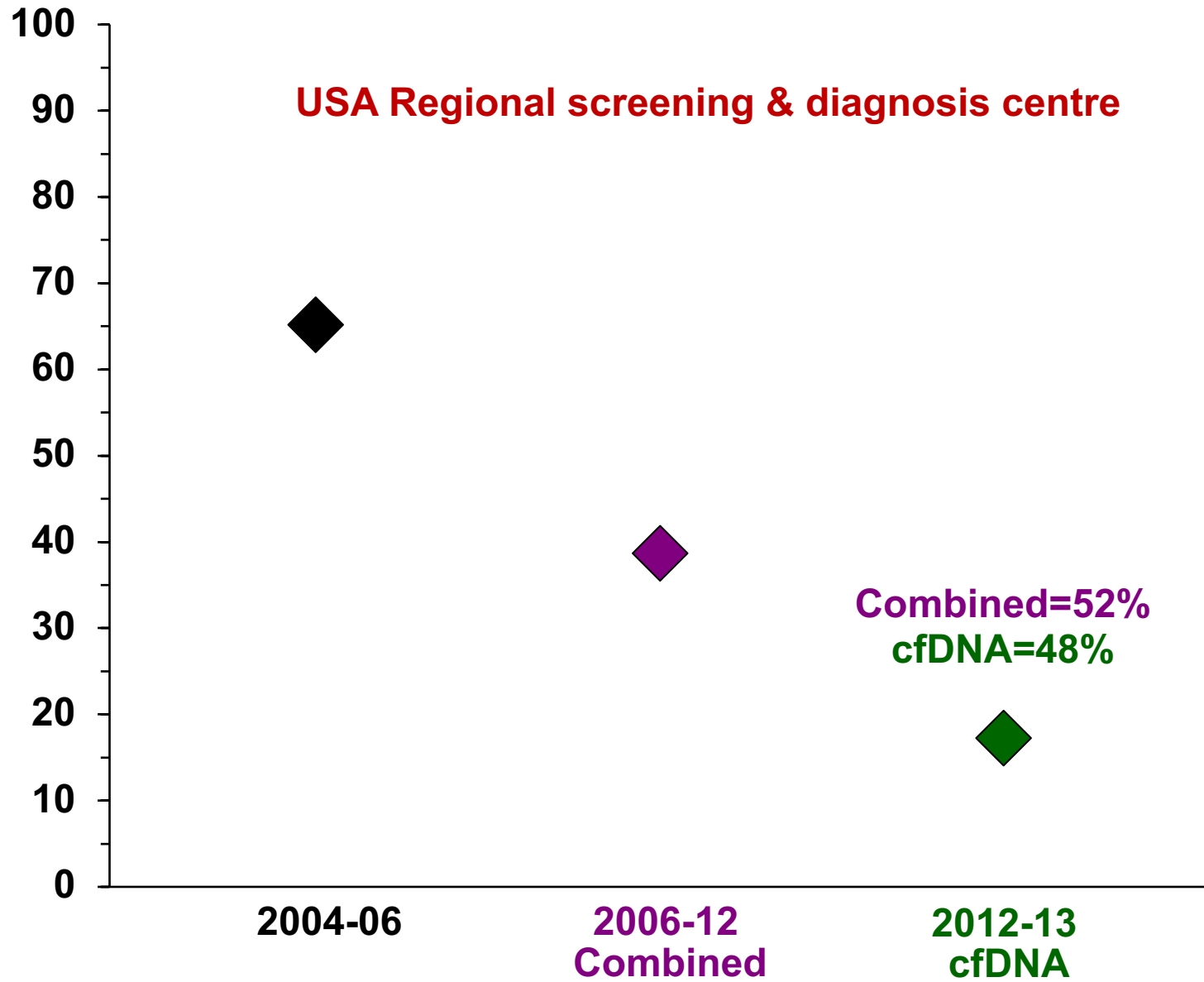
Israel (secondary cfDNA not recommended)

IPND rate (%)

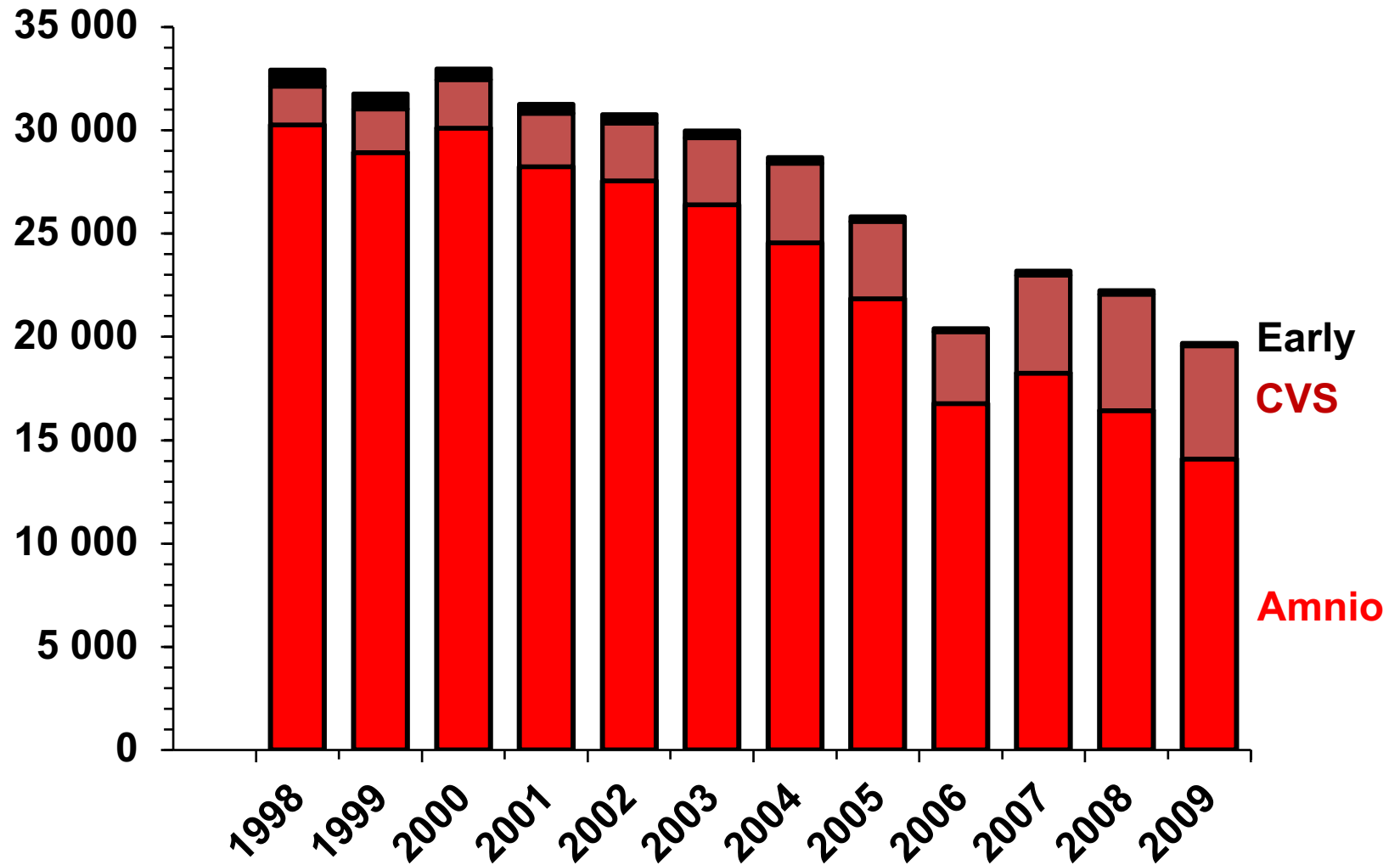


Denmark (2017-19: Secondary cfDNA option)

Invasive tests/month



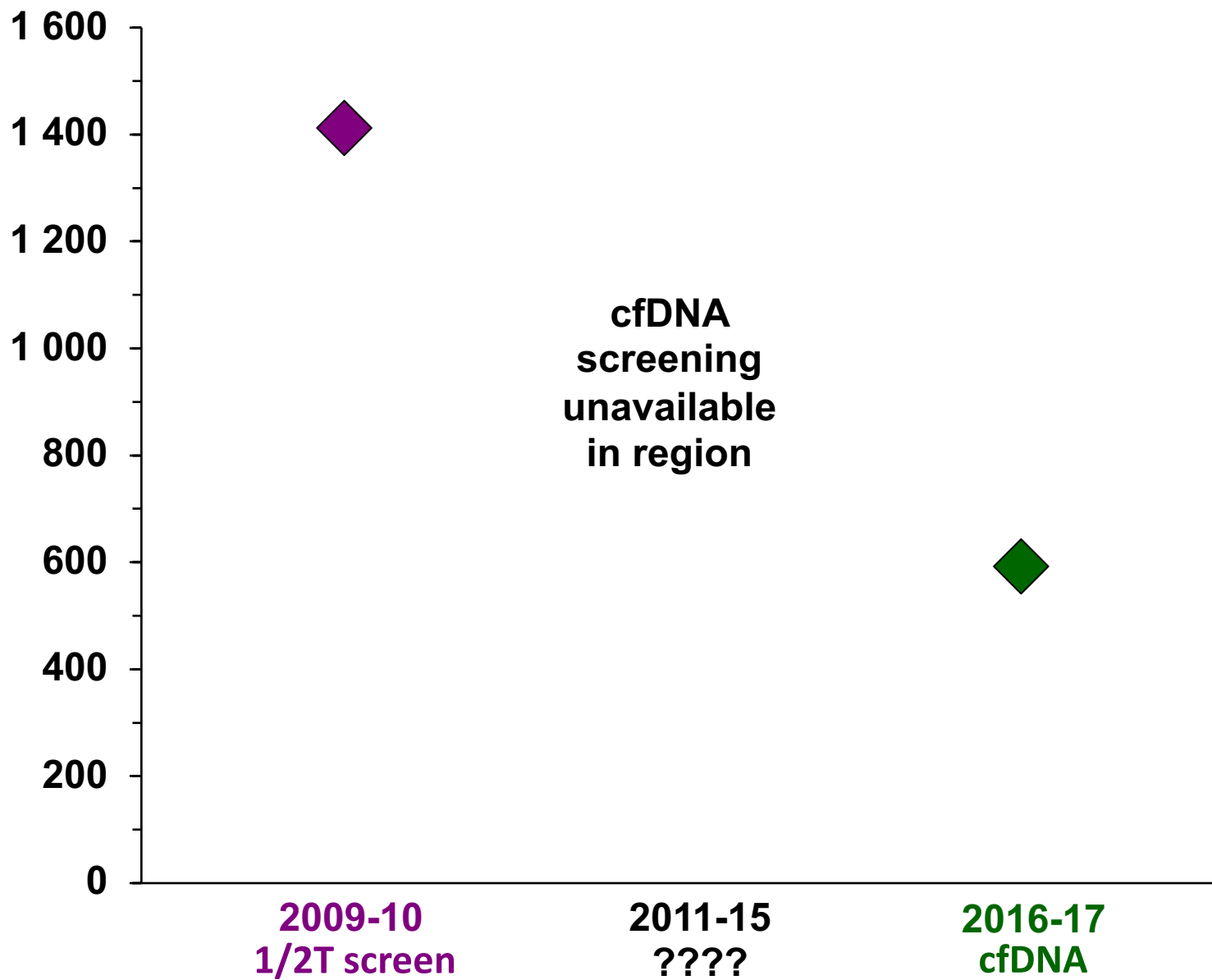
Invasive tests/year



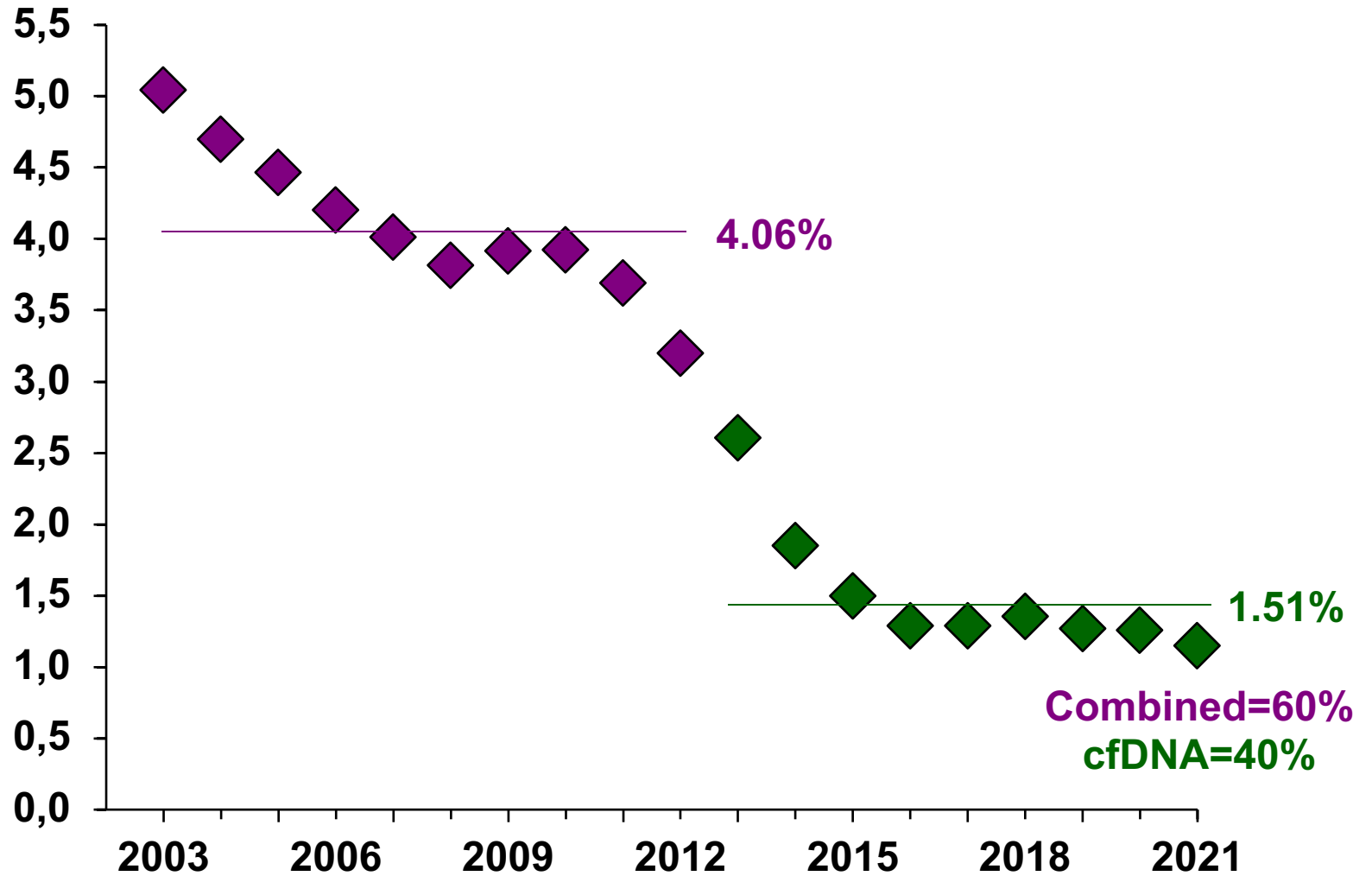
California State-wide Screening Programme

Invasive tests

Izmir Tertiary referral centre



Invasive tests (%)

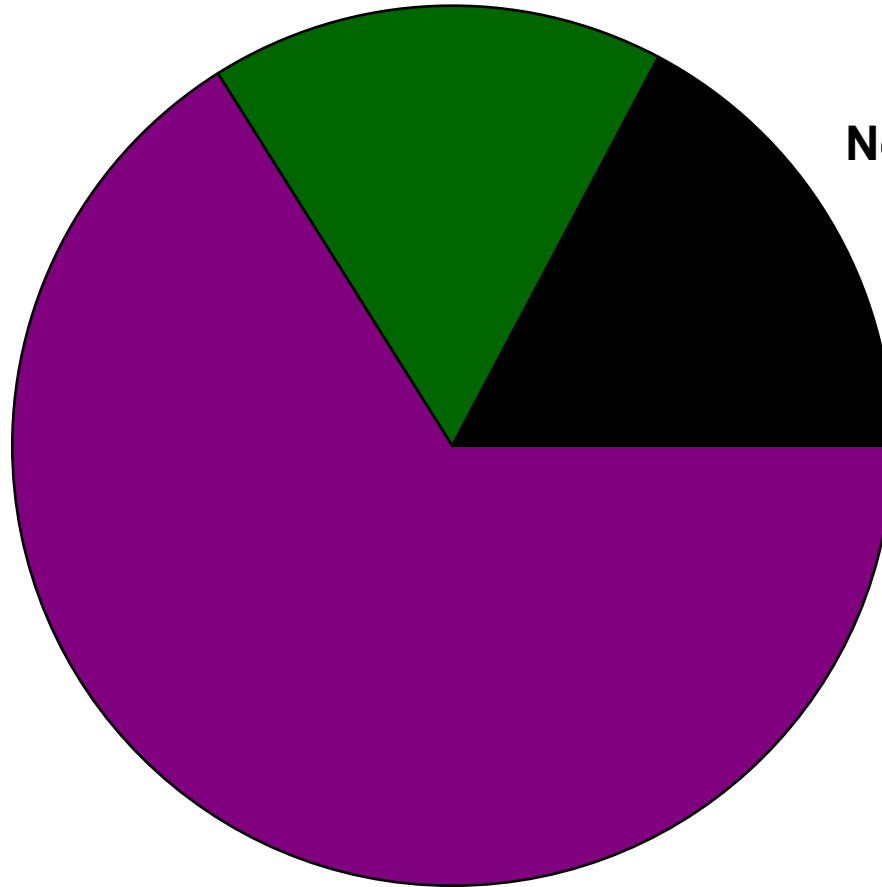


Australia, cfDNA self-funded since 2013 (AU\$450)

Victoria, Australia (2015)

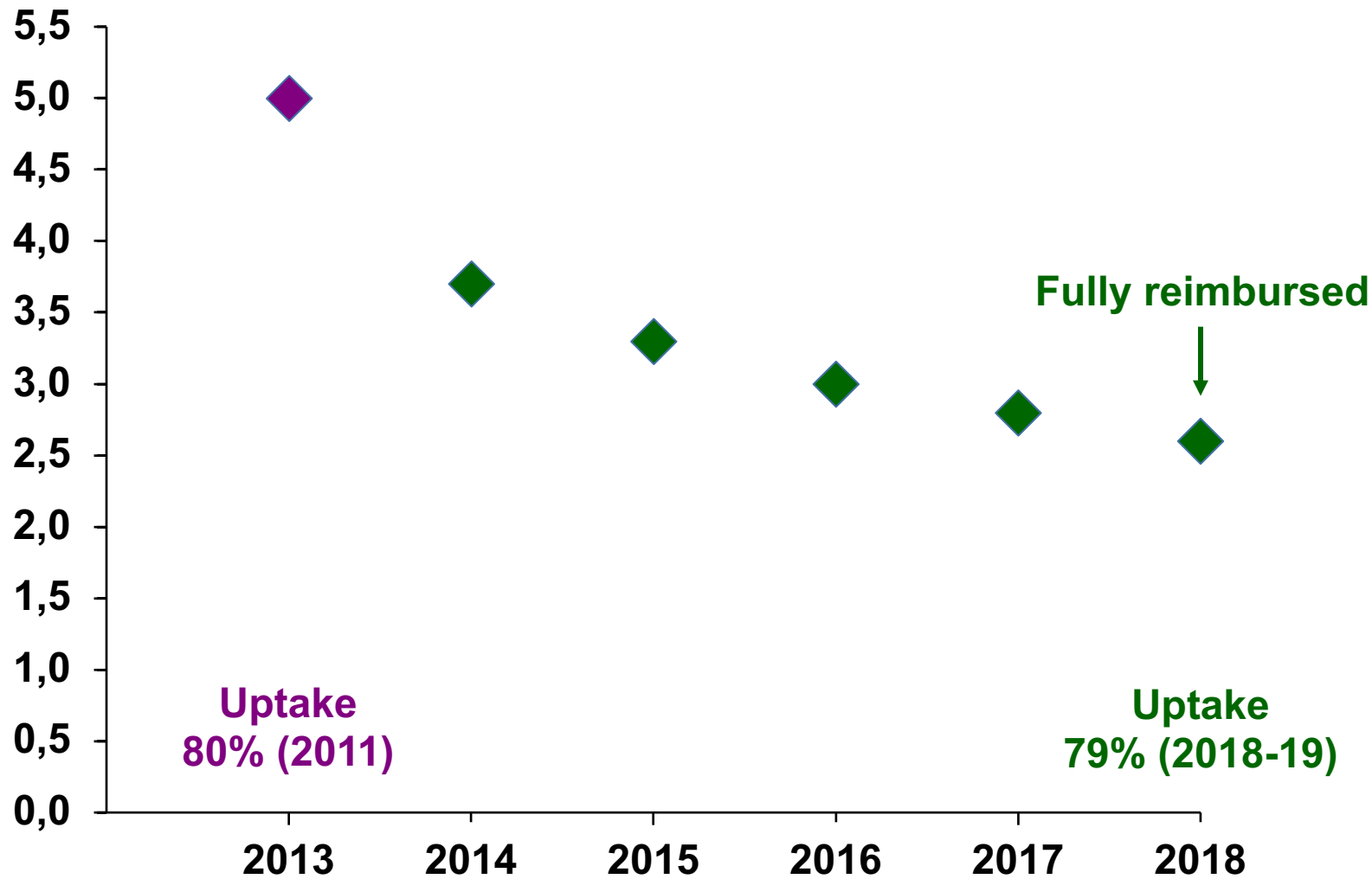
cfDNA screening (Invasive=1.9%)

Neither (Invasive=5.4%)

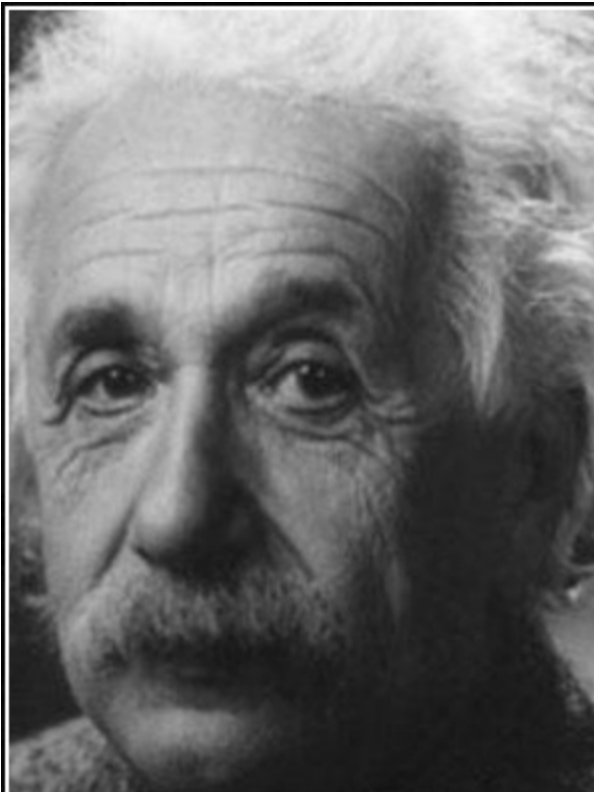


1T/2T screening (Invasive=2.0%)

Invasive tests (%)



Belgium



In theory, theory and practice are
the same. In practice, they are not.

— *Albert Einstein* —

