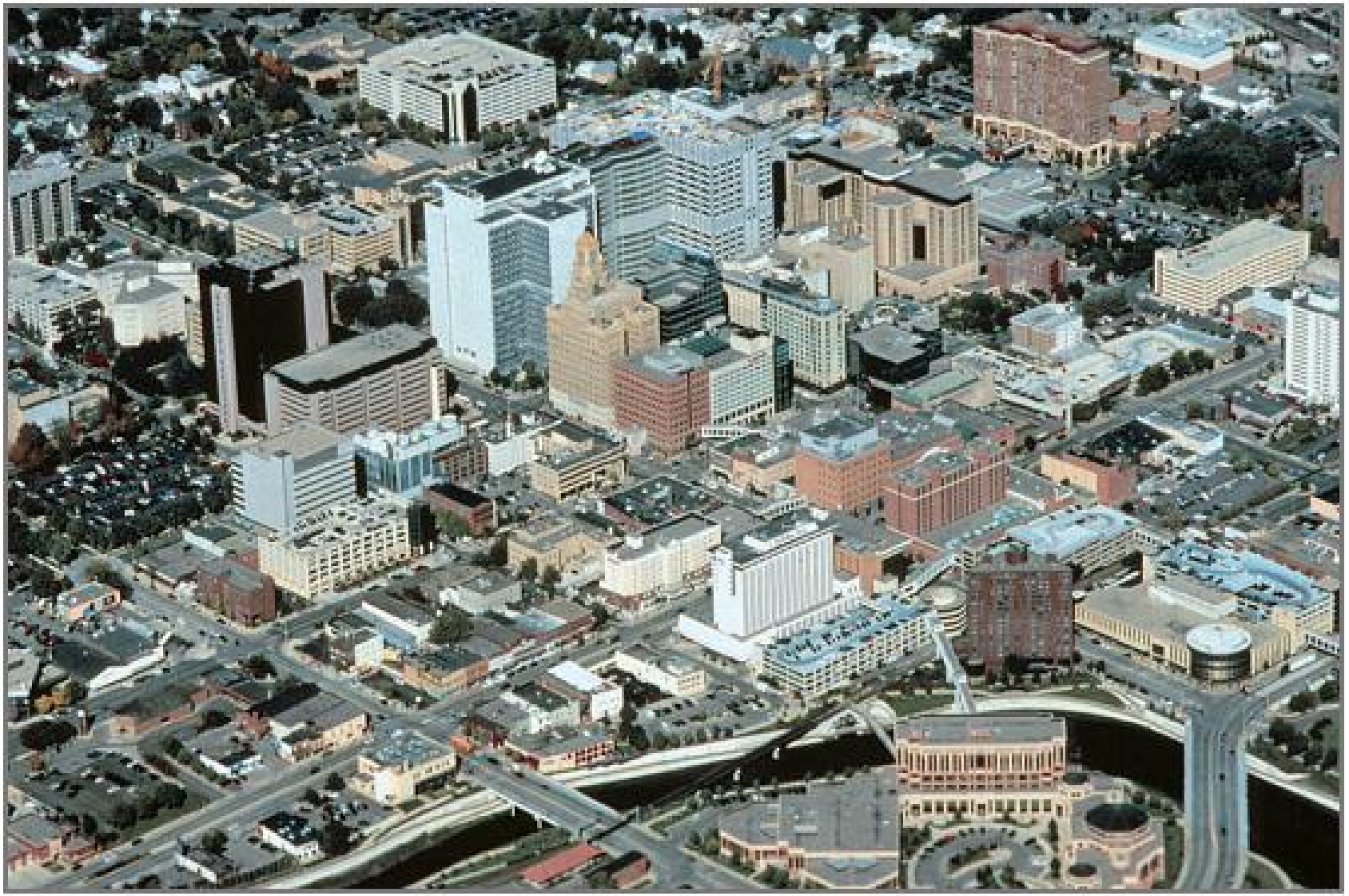
MAYO CLINIC

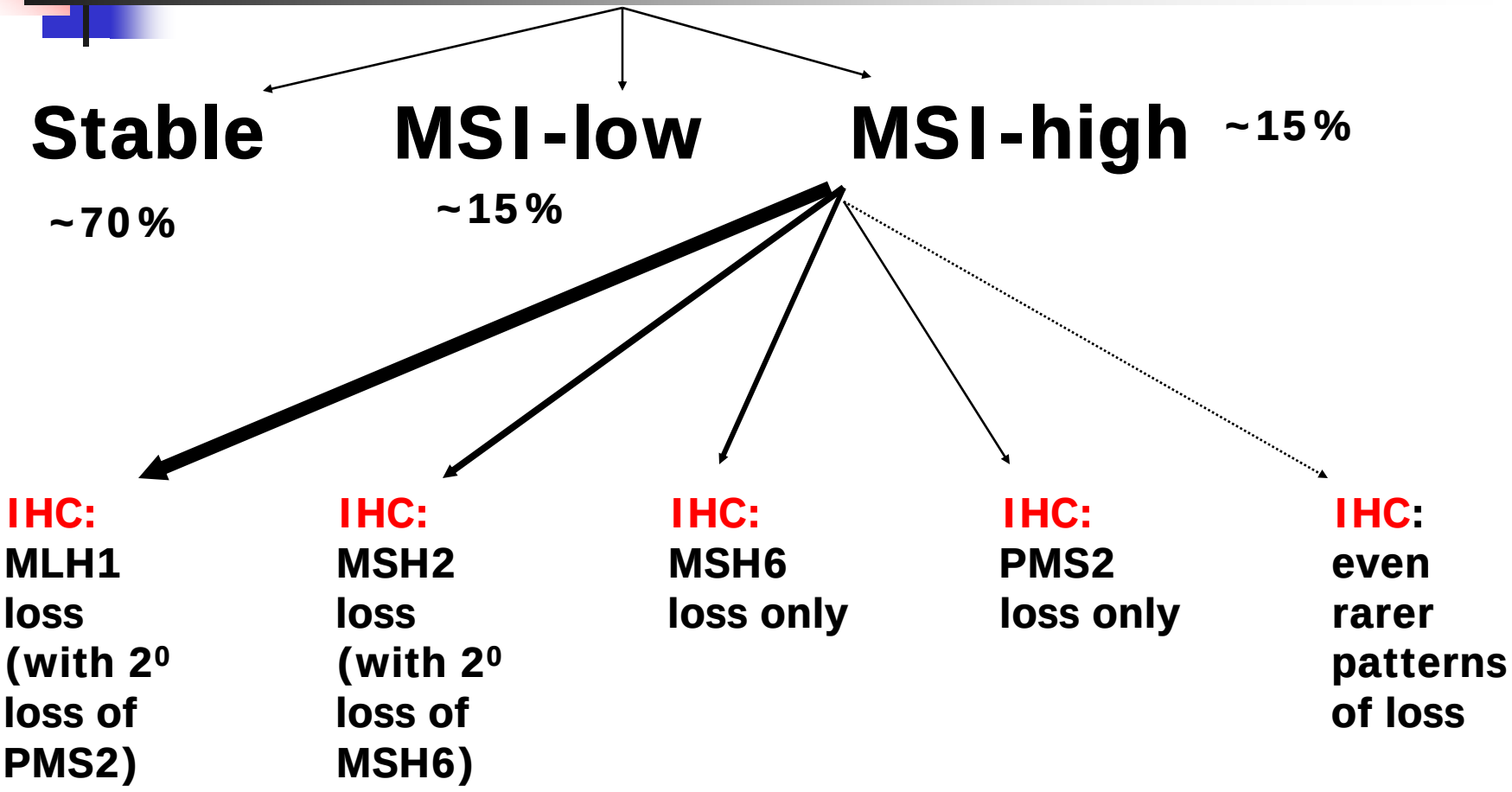
Mayo Clinic - Downtown



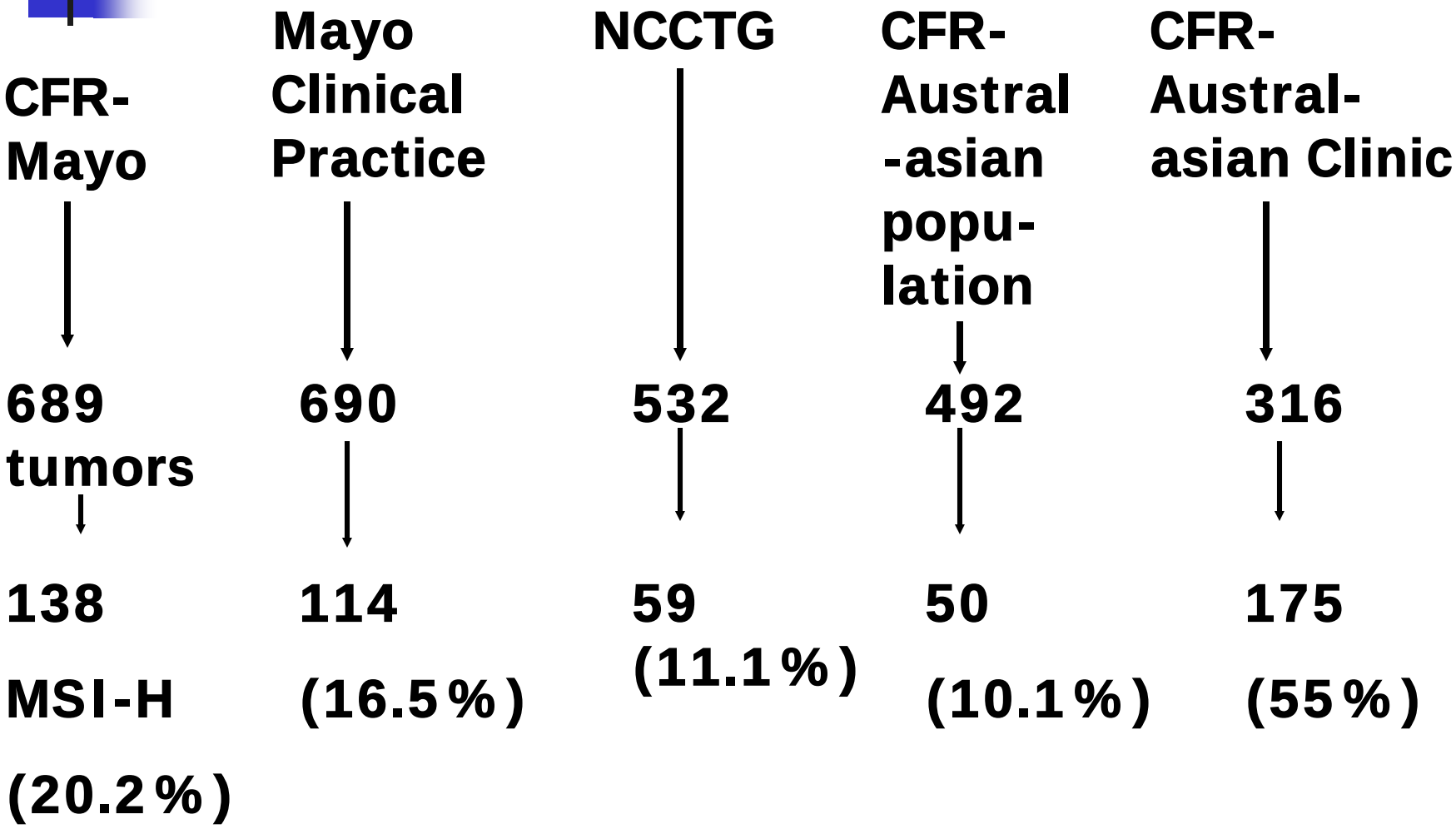
Isolated loss of *PMS2* expression in colorectal cancers: frequency, patient age, and familial aggregation

Clin Cancer Res 2005;11(18) 6466-6471

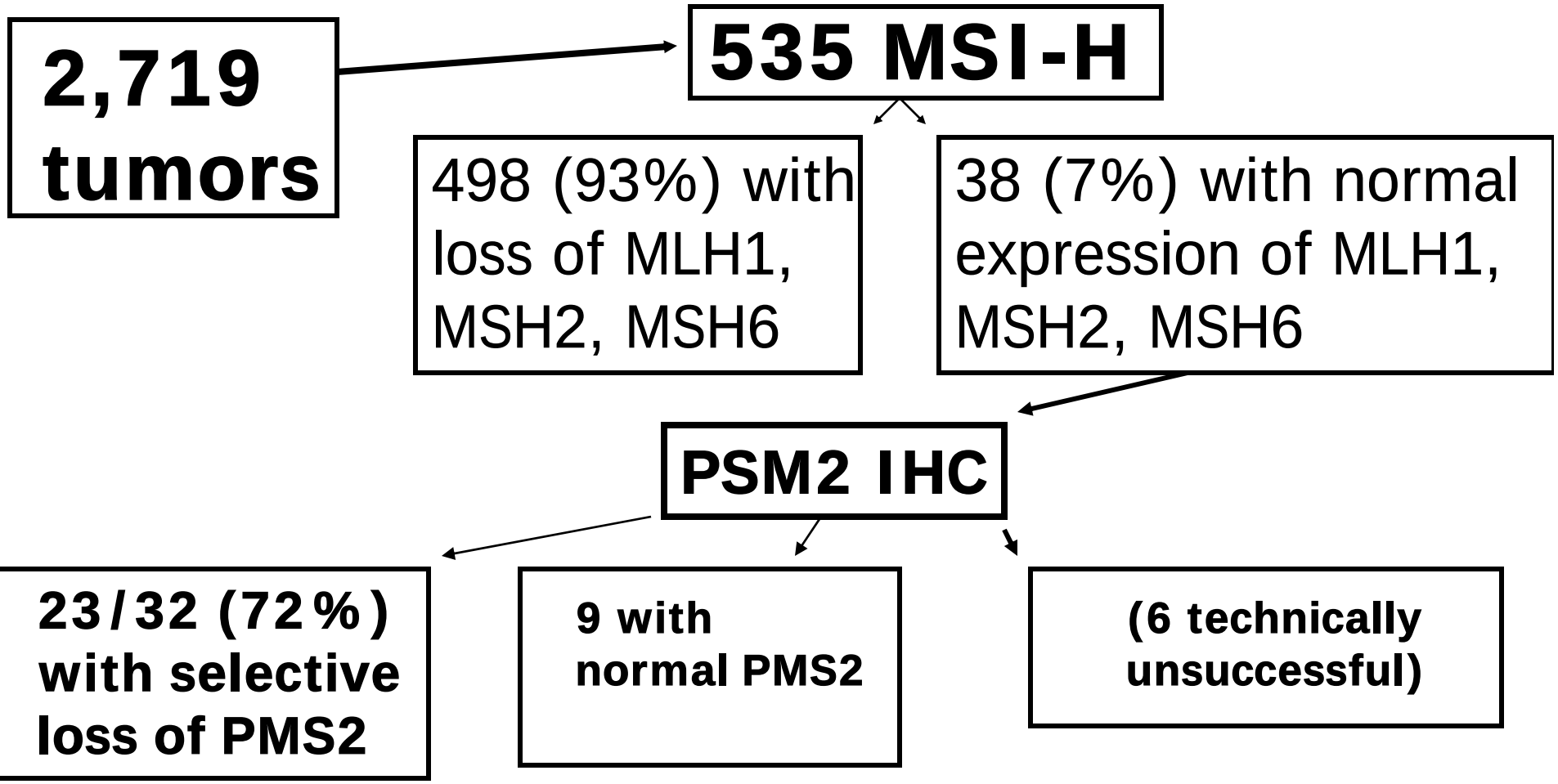
Microsatellite instability in colorectal cancer



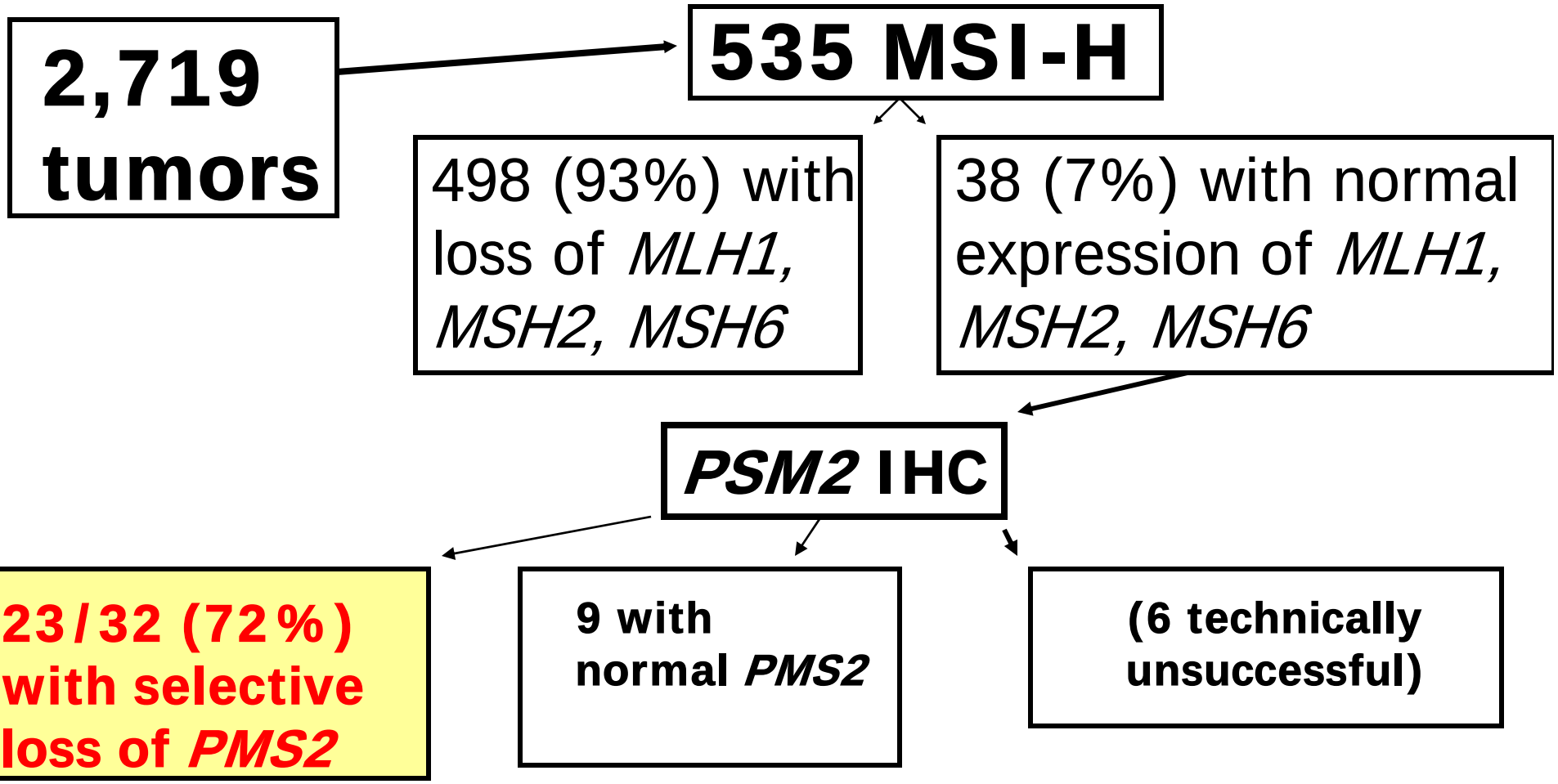
Collaborative study of PMS2



Collaborative study of PMS2



Collaborative study of *PMS2*



22/32 with selective loss of *PMS2*

- Median age 47.5 (28-67)
- Male 64%
- Proximal colon 52%
- Poor differentiation 50%
- More frequent in series selected for young age or +fam hx than in unselected group (NCCTG)

Fam hx on 22/23 cases with selective *PMS2* loss

- Only 2 met ACI or ACII criteria
- Most did not suggest family cancer syndrome
- No family in this study had the syndrome associated with homozygous MMR deficiency

Conclusions: Selective *PMS2* loss in CRC

- Rare event– seen in 23/535 MSI-H tumors (0.4%)
- Most cases not associated with Lynch-like family history
- Additional studies required to define relative contribution of germline *PMS2* mutations and the mechanisms of *PMS2* inactivation in tumor