

3/13/95

A Kamb - p16 & BRCA1

Myriad Genetics

MLM = malignant melanoma suscept. gene

- made physical map

- gene localization by homozygous deletion

- sequenced cosmid

MTS1 = p16 = MLM = CDKN2

MTS2 = p15

(no clear evidence that this is involved)

p16 - many deletions in this region

- also many pt. mutations (but not very high freq)

overexpression of p15 & p16 decrease

of clones/plane

alternatives to (E1 β vs E1 α)

BRCA1

- mapped by M. Claire King

- 1863 aa

- 100 kb

- v. hydrophilic

- Zn finger



BRCA1 mutations

most are frameshifts/nonsense

