

A black and white microscopic image of several chromosomes. An arrow on the left points to a specific region on one of the chromosomes. The chromosomes are dark, condensed structures against a light background.

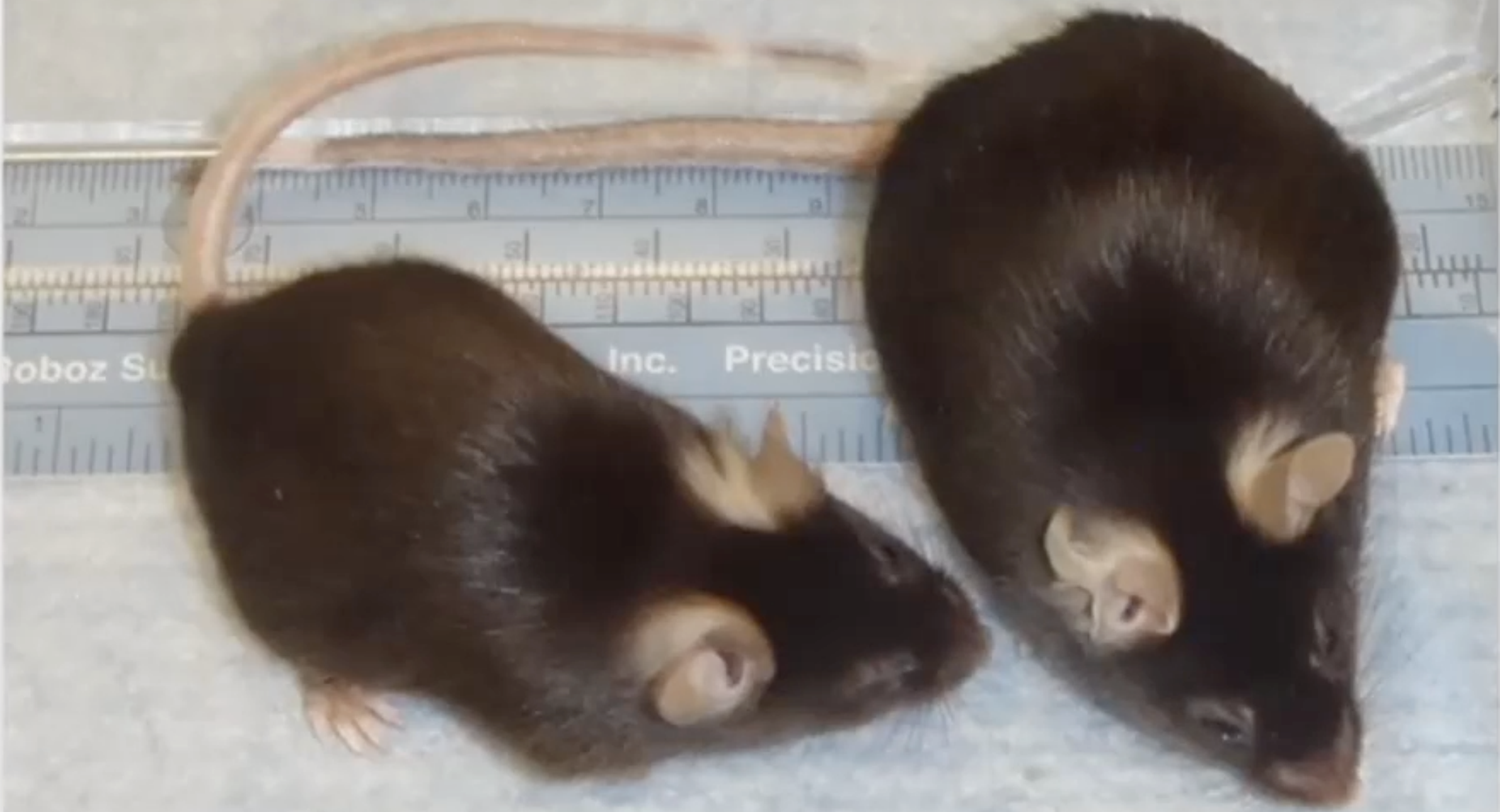
Animal Models of Bloom Syndrome: Understanding Human Health

2020 Bloom Syndrome Conference Webinar

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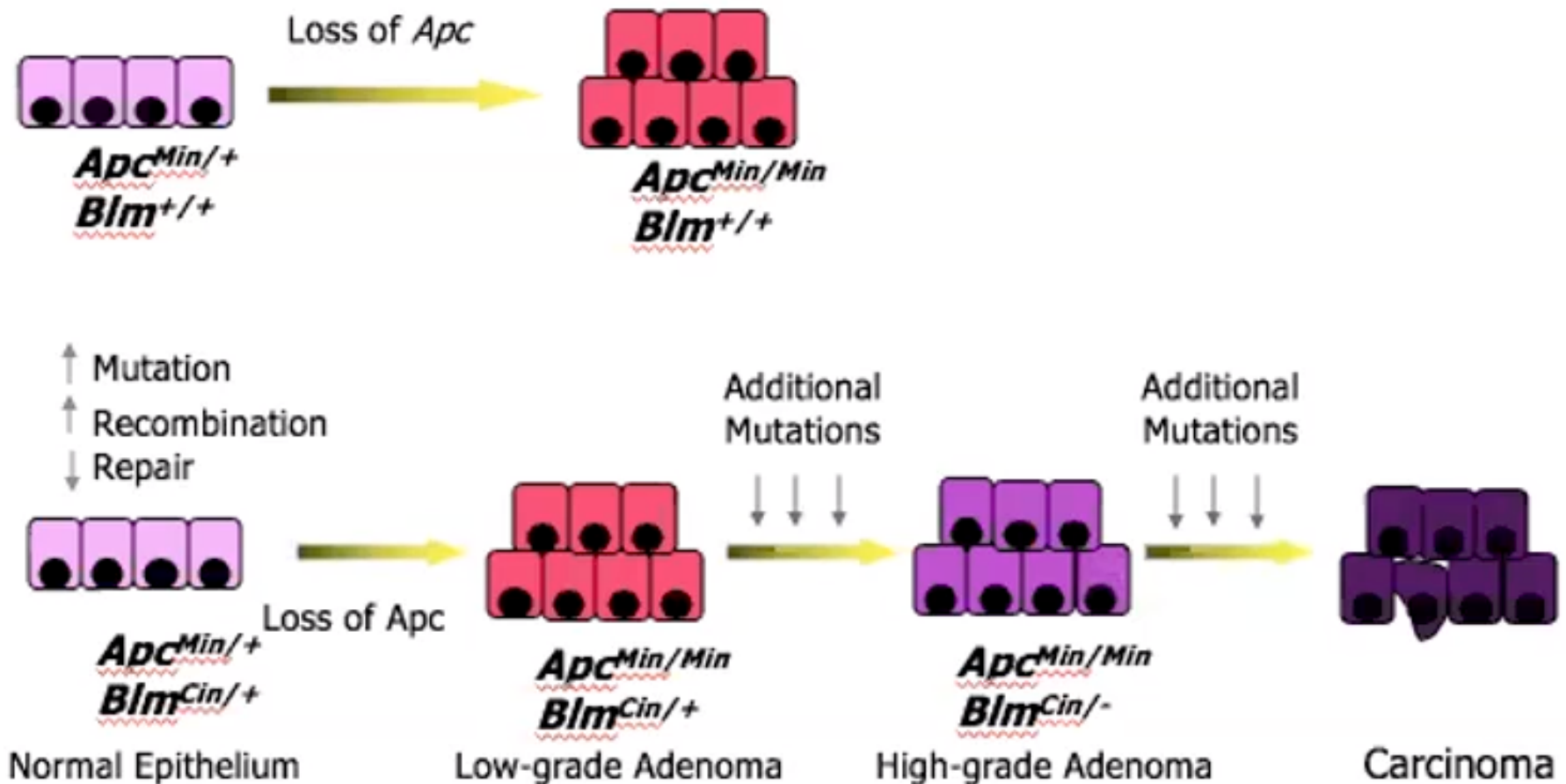
Mouse models of Bloom syndrome replicate some of the human characteristics of BLM mutation.



***Blm*^{DD/DD} ♀ at 12 weeks**

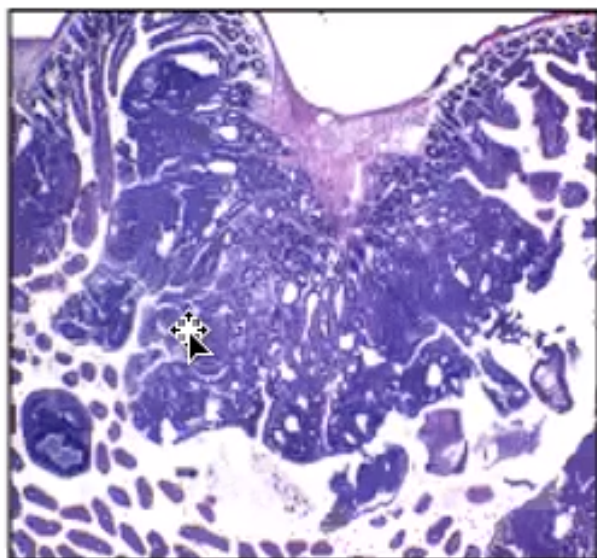
***Blm*^{+/+} ♀ at 12 weeks**

Heterozygosity for *Blm* mutation in a mouse model of intestinal cancer increases tumor number and tumor invasion.

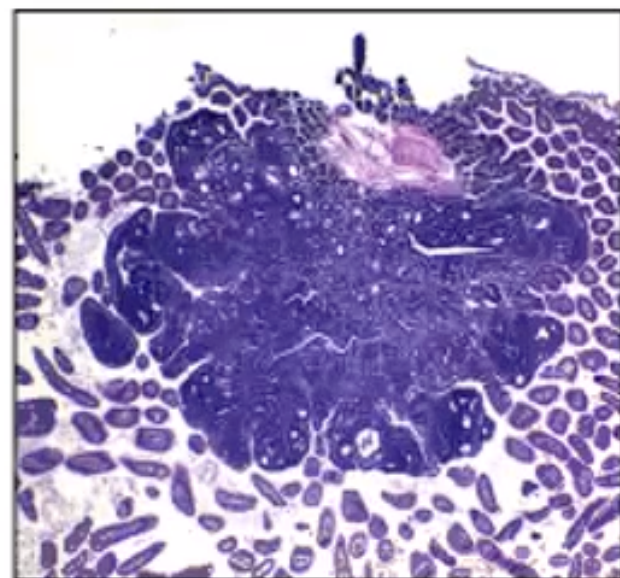


DNA repair deficiency (*Blm*^{+/-}) facilitates intestinal tumor progression in the *Apc*^{Min/+} mouse.

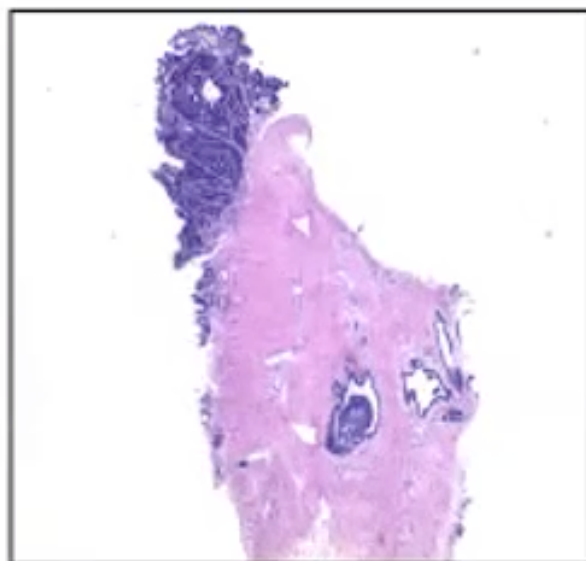
(Goss et al., 2001)



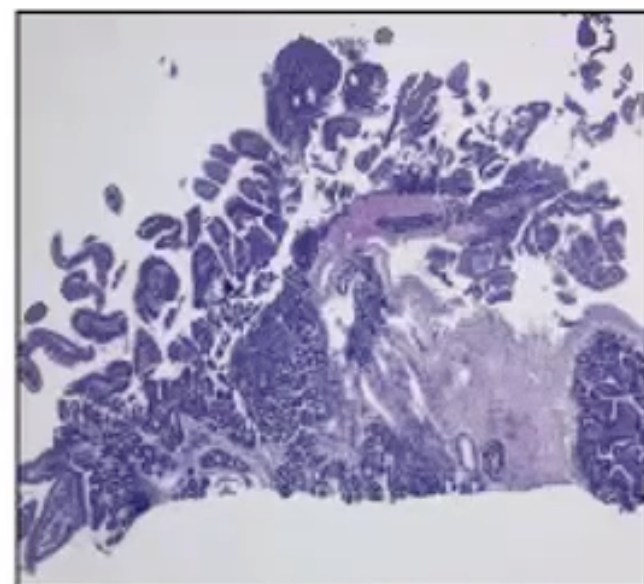
adenoma with low grade dysplasia at 4.5 months



adenoma with high grade dysplasia at 6.75 months



adenoma with herniation at 8 months



adenocarcinoma at 10 months

Heterozygosity for *hBLM^{ASH}* is associated with a 2-3-fold increase in risk of colorectal cancer.

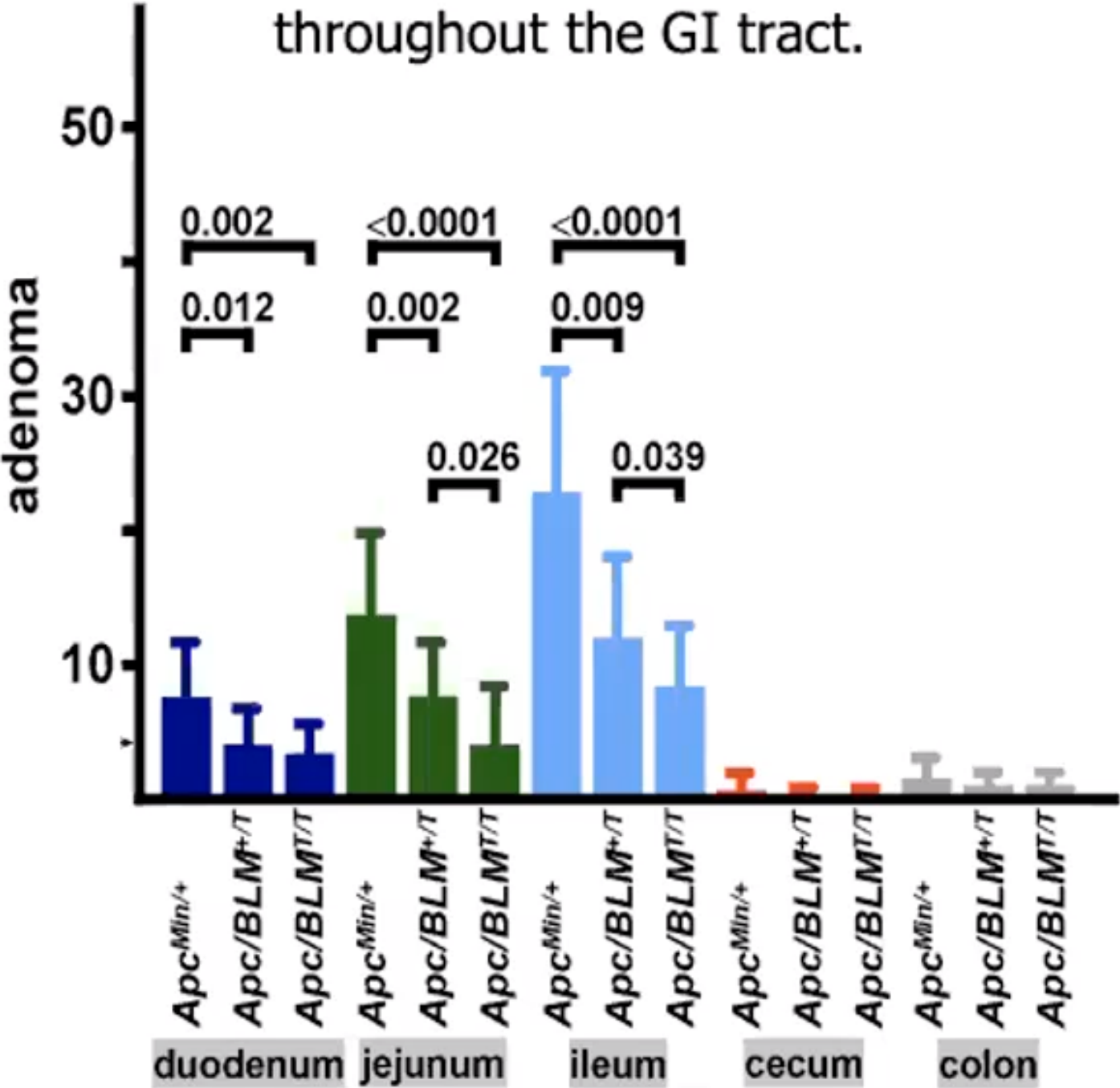
Heterozygosity for *hBLM^{ASH}* is not associated with any increase in risk of breast cancer.

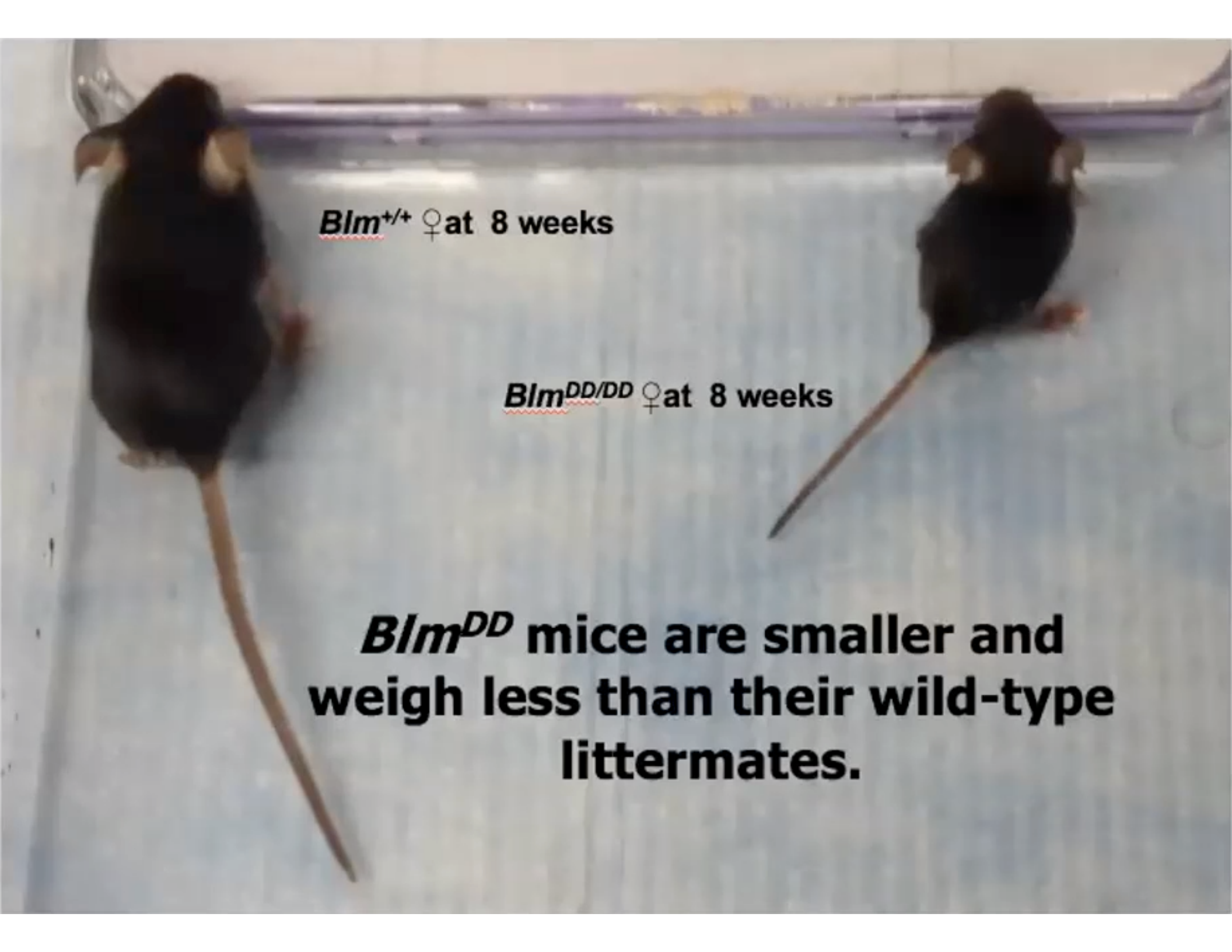
Heterozygosity for *mBlm* is associated with increase in virus-induced lymphoma.

Mutation of *BLM* in human is found at a high frequency in familial and sporadic mesothelioma.

Heterozygosity for *mBlm* is associated with increase in asbestos-induced mesothelioma.

Increasing *BLM* gene copy number using a human transgene in a mouse model of intestinal cancer reduces tumor number throughout the GI tract.






Blm^{+/+} ♀ at 8 weeks

Blm^{DD/DD} ♀ at 8 weeks

***Blm*^{DD} mice are smaller and weigh less than their wild-type littermates.**



Blm^{DD/DD} ♀ at 12 months

Blm^{DD/DD} mice
display cataracts
and graying fur by
one year of
age.

At one year, *Blm*^{DD/DD} mice demonstrate greater 18S *rDNA* copy number variation with age than wild-type littermates.

