

Mutations of Keratin Genes

Produced for the CRC for Premium Quality Wool undergraduate program by; **Dr. Barry Powell, The University of Adelaide.**



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for

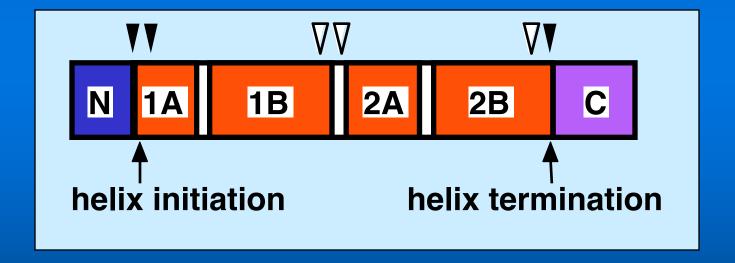
Premium

Quality

Wool

Genetic Mutations in the Epidermal Keratin IF cause Blistering Diseases

 Dowling-Meara and Weber-Cockayne syndromes



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300

CRC

for

Premium

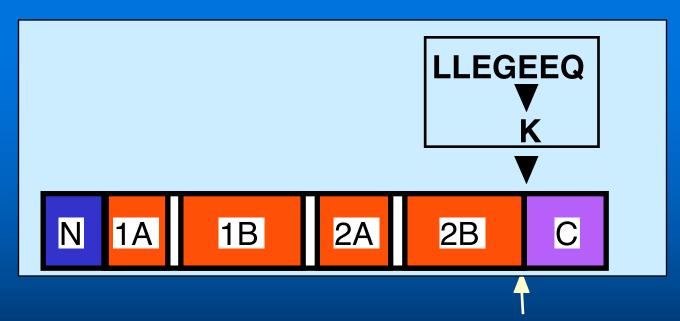
Quality

Wool

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Monilethrix: a Mutation in a Hair Type II IF Gene?

- Hair defect short, fragile, beaded hair
- Maps to human keratin type II locus
- Preliminary evidence for
 - glu (-) → lys (+) mutation



helix termination signal



Hair and Wool Mutations

- Over 50 mouse hair mutations known
 - (shaven, caracul, naked, velvet coat & hairy ears map to the type II gene locus)
- In sheep:
 - Felting lustre mutant
 - dominant gene
 - lustrous, felting fleece
 - reduced follicle density
 - no gly/tyr-rich KAPs
 - gene defect not known regulatory gene?

Barry Powell