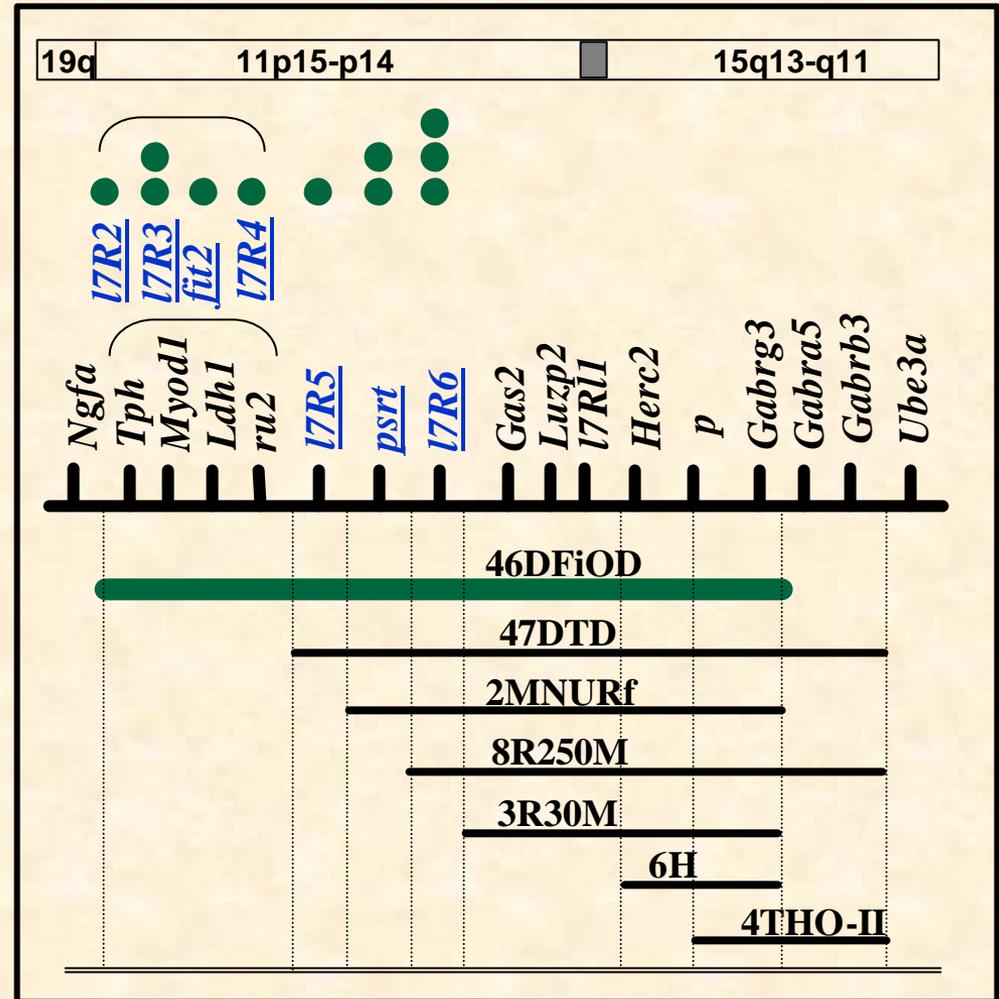


Mammalian Genetics and Genomics Program–December, 2001 (Rinchik et al., *PNAS*, 2002, in press) Contact: E. M. Rinchik (865) 241-2158

- Eleven recessive ENU-induced mutations (filled symbols at right) recovered in a small, 1- to 2-centiMorgan region of mouse Chromosome 7 that is homologous to Human Chr 11p14-p15
- Wide variety of recessive phenotypes (continual seizures, postnatal failure-to-thrive, failure of prenatal development)
- Mutations each genetically finely mapped and ordered by crosses with radiation-induced deletions (extents shown by lines below chromosome map at right)
- Ready for gene identification and correlation with DNA sequence and transcription maps

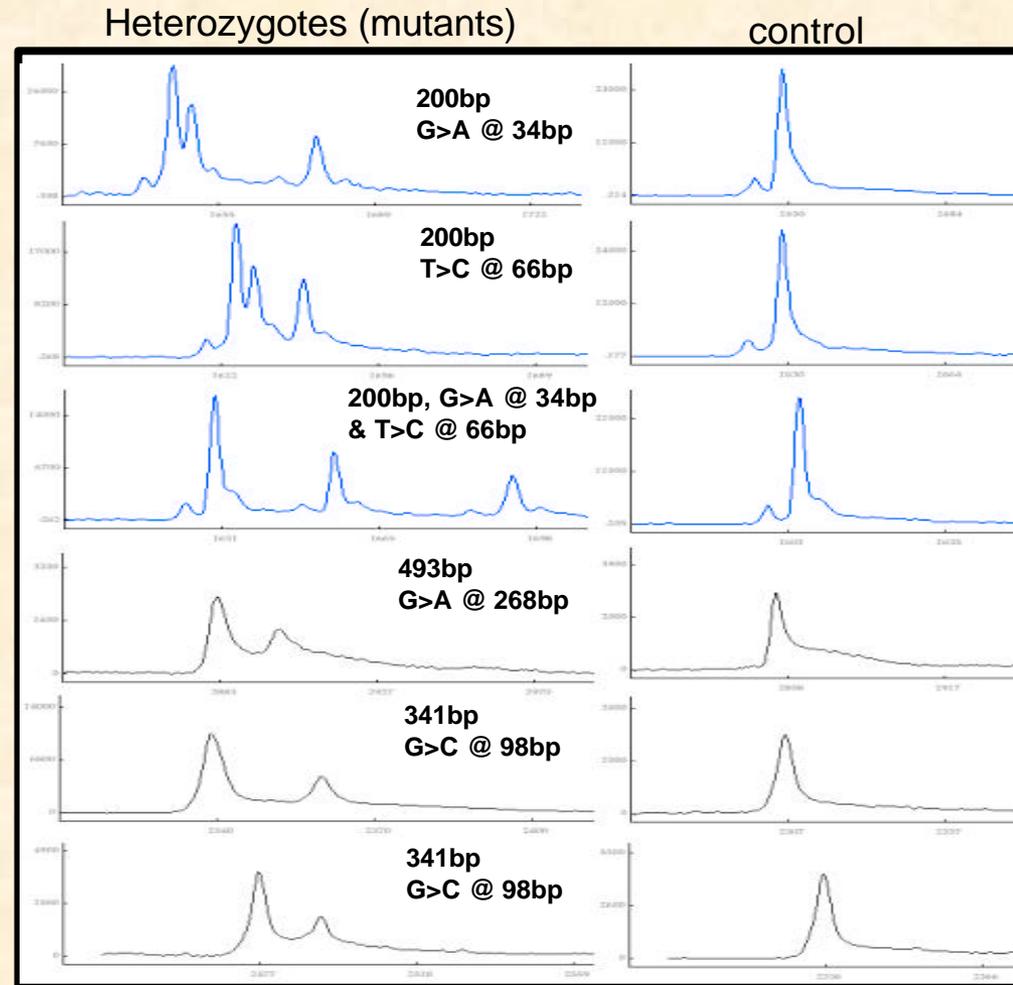


Mammalian Genetics and Genomics Program–December, 2001 (Li et al., *Electrophoresis*, 2002, in press) Contact: C. T. Cuiat (865) 241-0672

- Temperature Gradient Capillary Electrophoresis (TGCE) detects the presence of heteroduplex molecules formed by the normal and corresponding mutant DNA segments due to their differential mobilities from the homoduplexes during capillary electrophoresis.

- Detection of different mutations in a single temperature gradient run (60-70 degrees). TGCE allows rapid simultaneous scanning for point mutations or SNPs in different gene segments (e.g., various exons or conserved elements).

- TGCE protocols were developed to allow mutation scanning using crude DNA template preparations (mouse tail DNA preps) and unpurified PCR reactions. One 96-well plate is analyzed in ~45 minutes.



Mammalian Genetics and Genomics Program–December, 2001

(Miltenberger et al., *Genetics*, 2002, in press) Contact: E. J. Michaud (865) 574-0797

- The *agouti* gene regulates hair pigmentation in mice and many other mammals. Recessive loss-of-function mutations result in black hair color, whereas dominant gain-of-function mutations result in yellow hair color, obesity, diabetes, and cancer.
- Molecular and phenotypic analysis of 25 recessive mutations at the *agouti* locus, ranging from mild reduction in gene function to complete loss of gene function, reveals important *agouti* gene and protein structure-function relationships.

