The first *Clec16a* mutant mouse exhibits defects in digits and tail

Belinda S. Harris, Heather E. Fairfield, Laura G. Reinholdt, David E. Bergstrom, and Leah Rae Donahue

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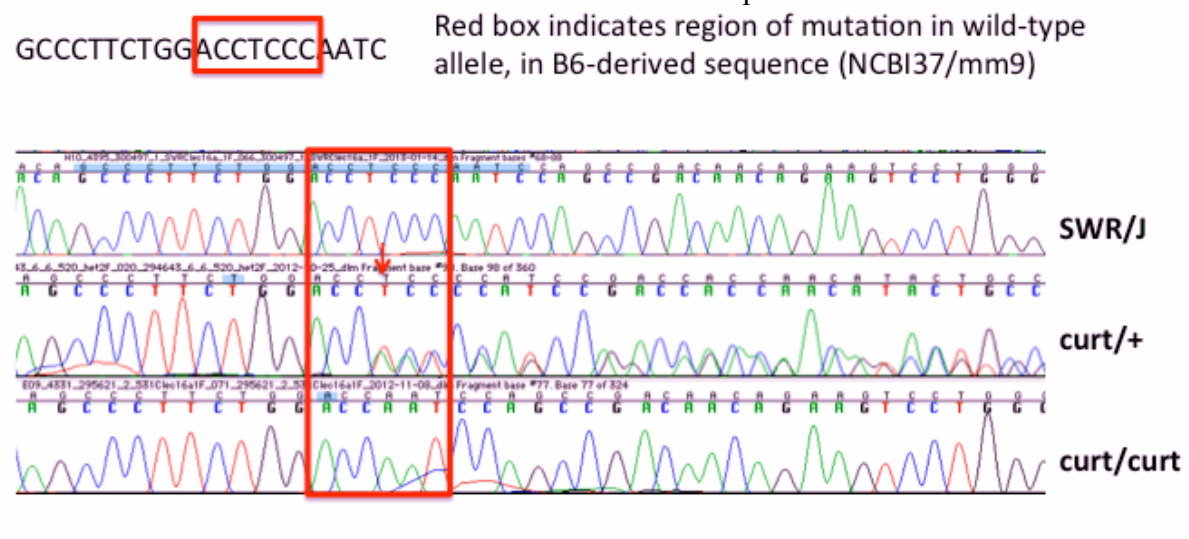
Mutation (allele) symbol: *Clec16a*^{curt}

The curvy tail mutation arose spontaneously at The Jackson Laboratory and was previously characterized with phenotypes such as small body size, squinting eyes, and crooked tail and digits. Mapping data demonstrated linkage to Chromosome 16 and whole exome sequencing was used to identify candidate mutations in the mapped region. Commercial alignment and SNP/INDEL calling tools as well as a second alignment tool (BWA) and SNP caller (SAM Tools) were used to identify candidate variants. A 4-nucleotide deletion allele was found beginning at position 10,694,722 on Chromosome 16 (NCBI37/mm9) in C-type lectin domain family 16, member a (*Clec16a*). Primers were generated that produce a 383 base pair product spanning the predicted mutation; *Clec16a* forward (CCCCAAGGGTCTTACTGTCA) and *Clec16a* reverse (GAAGCCAGAGCTGCCAATAG). Sequence analysis of additional mutant and wild type genomic DNA samples was used to confirm the presence of the 4-nucleotide deletion from ACCTCCC to ACC---- in mutant DNA samples.



Comparison of sequence chromatograms from SWR/J and heterozygous controls and homozygous mutant sample. Double chromatogram in curt/+ sample and single chromatogram in mutant sample indicates 4-bp deletion encompassing nucleotides 10,694,725-10,694,728 on chromosome 16 (NCBI37/mm9).

This small deletion in exon 21 is predicted to result in a frameshift mutation, leading to the introduction of novel amino acids and an early termination codon. The *Clec16a* gene is highly conserved, and is present in both vertebrates and invertebrates. C-type lectin-domain family members are typically extracellular proteins and have many diverse functions. This spontaneous mutation and the phenotypes associated with the curvy tail mouse present the first opportunity to characterize the function of *Clec16a* and its potential role in development.