Mouse models of Charcot-Marie-Tooth peripheral Neuropathy: towards disease mechanisms and therapeutic strategies

In 2006, the Burgess lab discovered mice with an autosomal dominant mutation in Gars (glycyl tRNA synthetase), resulting in peripheral axon degeneration. These mice create a model of the human disease Charcot-Marie-Tooth type 2D, and we have used a combination of genetic and biochemical approaches to understand the underlying disease mechanism and to pilot therapeutic approaches. These studies will have implications for other forms of Charcot-Marie-Tooth disease, and possibly other heritable neuromuscular disorders and motor neuron diseases.

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