Studying Codon Bias and Kidney Dysfunction in a Mouse Model for *Familial Dysautonomia*

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Familial dysautonomia (FD) is a debilitating disease primarily known for its damage to the peripheral nervous system. However, kidney failure is the most common cause of death in FD patients. FD results from a mutation in the ELP1 gene, which is part of the 6-subunit complex, Elongator. It is known that some genes preferentially use specific synonymous codons, and Elongator is essential for translating genes that are enriched in either AA- or AG-ending synonymous codons. It has been assumed that kidney disease in FD results from irregular blood pressure and compromised innervation of kidney vasculature. However, here we show that ELP1 is robustly expressed in the kidney collecting duct, suggesting that it may play a direct role in kidney function and therefore the dysfunction in FD. To investigate this hypothesis, we made a mouse model where ELP1 is selectively ablated in the kidney collecting duct. Our data indicate an essential role for ELP1 in normal kidney function. These data suggest that compromised ELP1 levels in the kidneys of FD patients may be a contributing factor to chronic kidney disease. Our ongoing work focuses on identifying kidney-specific, codon-biased genes that are misregulated in the absence of Elongator and thus contribute to kidney dysfunction.