Cystinosis nonsense mutation read-through mediated by ELX-02 restores protein function using in vitro and in vivo models

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Nephropathic Cystinosis is a recessive lysosomal storage disease that leads to Fanconi Syndrome and end stage renal disease

- Loss of function mutations in the Cystinosis Lysosomal Cystine Transporter (CYSNTS) leads to cystinosis
- ELX-02 is a Eukaryotic Ribosomal Selective Glicoside (ERSG) which can rescue full-length protein via ribosomal read-through

ELX-02 increases CTNS protein, function and mRNA in patient-derived fibroblasts with the W138X allele

ELX-02 reduces kidney half-cystine accumulation in mouse model of nonsense-mediated cystinosis

A Phase 2 study of ELX-02 in patients with Nephropathic Cystinosis

Conclusions
- ELX-02 read-through is sufficient to produce functional CTNS protein and increase CTNS mRNA
- Kidney exposure and demonstration of efficacy in vivo support dose-range selection for a Phase 2 clinical trial of ELX-02 in Nephropathic Cystinosis
- Completion of a Phase 1 study in renal insufficient patients provides modeling necessary for dose adjustments based on renal function

Acknowledgments
We thank the Cystinosis community and many contributors from McGill and of the CyNoMus Project that made this research possible. In addition, we thank the Eloxx Board members, past and present, for their efforts to advance therapeutic options for nonsense-mediated disorders.