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Title:	Author Correction: A Genetic Screen for Investigating the Human Lysosomal CystineTransporter, Cystinosis
Authors:	Deshpande, A.A. (/jspui/browse?type=author&value=Deshpande%2C+A.A.) Shukla, Anuj (/jspui/browse?type=author&value=Shukla%2C+Anuj) Bachhawat, A.K. (/jspui/browse?type=author&value=Bachhawat%2C+A.K.)
Keywords:	Cystinosis Genetic Screen CystineTransporter
Issue Date:	2019
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Abstract:	Cystinosis, a lysosomal transporter is involved in the efflux of cystine from the lysosome to the cytosol. Mutations in the human cystinosis gene (CTNS) cause cystinosis, a recessive autosomal disorder. Studies on cystinosis have been limited by the absence of a robust genetic screen. In the present study we have developed a dual strategy for evaluating cystinosis function that is amenable to rapid genetic analysis. We show that human cystinosis expressed in this yeast confers growth on cystine when the protein is mistargeted to the plasma membrane by the deletion of the C-terminal targeting signal, GYQDL. We also screened a vacuolar protein sorting deletion library, and subsequently created multiple vps deletion mutants for kinetic studies. The double deletion, vps1Δvps17Δ, greatly enhanced uptake. This enabled validation by kinetic studies, including first studies on the WT CTNS protein (that contained the GYQDL motif). Using this screen we isolated several gain of function mutants, G131S/D, G309S/D, A137V, G197R, S270T, L274F and S312N showing enhanced growth on low concentrations of cystine. Kinetic analysis yielded insights into the role of the residues (including one of the patient mutations, G197R). The results indicate that the screen could be effectively used for interrogating and understanding the CTNS protein.
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