

Beta Cell Interest Group (BIG)

Current and ongoing beta cell research is presented in this weekly seminar by faculty, postdoctoral fellows and students. If you are interested in attending the Beta Cell Interest Group (BIG) seminars and joining the BIG community, please contact [David Jacobson](#).

Keywords: Beta cell

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Meeting Details

Start Date / Time	April 25, 2018 at 9:00 AM
End Date / Time	April 25, 2018 at 9:55 AM
Duration	55 minutes
Location	512 Light Hall
Presenter Name	Emily Walker (Stein lab)
Presentation Title	The S64F MAFA mutation causes strikingly different physiological phenotypes in males and females
Status	This meeting has already occurred

Meeting Agenda/Notes

Summary: Recently, a missense variant of human MAFA, Ser64Phe (S64F), was identified in patients with familial insulinomatosis and diabetes mellitus with diabetes more prevalent in males than females. The S64F mutation was found to impair phosphorylation within the MAFA transactivation domain, increasing protein stability and transactivation domain activity in beta cell lines. We hypothesized that islet beta cell function is negatively affected by elevated and dysregulated S64F MAFA levels. Therefore, we developed an endogenous S64F MAFA mouse model to examine the distinct regulatory mechanisms of this mutant. Here I will discuss the profound sexual dimorphism of the S64F MAFA mutant in regards to diabetes, and analogous to what occurs in human patients.