



Program Agenda

SLC13A5 Deficiency Annual Research Roundtable 2018

August 20, 2018

Stanford University
Li Ka Shing Center (LKSC), Room LK208
291 Campus Drive
Palo Alto, CA 94305

Time	Topic	Speaker/ Moderator	Institution
8:00-8:30	Check-In and Breakfast	--	--
8:30-8:40	Welcome	Kimberly Nye	TESS Research Foundation
8:40-8:50	Rapid Intros	All	All
8:50-9:00	Clinical Overview	Brenda Porter	Stanford University
9:00-9:10	Research Overview	Matthew Bainbridge	Rady Children's Hospital
9:10-9:20	Tooth abnormalities in SLC13A5 Deficiency (Skype)	Agnès Bloch- Zupan	University of Strasbourg
9:20-9:45	Using Drosophila to understand the biology of SLC13A5 Deficiency	Stephen Helfand	Brown University
9:45-10:10	Humanized SLC13A5 mouse to understand the role of the citrate transporter in humans	Vadivel Ganapathy	Texas Tech University Health Sciences

10:10-10:30	Break	--	--
10:30-10:55	Metabolomics of SLC13A5 Deficiency	Sarah Elsea	Baylor College of Medicine
10:55-11:20	The Na ⁺ /citrate transporter in human neurons	Ana Pajor/ Anne Murphy	UCSD
11:20-11:45	Biomarkers of SLC13A5 Deficiency: Blood and CSF Citrate levels	Andreas Birkenfeld	Technische Universität Dresden
11:45-12:10	3D cortical spheroids as disease model for SLC13A5 Deficiency	Toshihiko Ezashi	University of Missouri
12:10-1:10	Working Lunch	All	All
1:10-1:35	Gene therapy for SLC13A5 Deficiency	Rachel Bailey	UT Southwestern
1:35-2:00	How to get a therapy to market	Dawn Blessing	Rare Disease Drug Development and Corporate Strategy
2:00-2:20	Group Photo and Break	--	--
2:20-2:45	SLC13A5 Antibodies, patient derived iPSCs, and registry	Deepti Dubey	TESS Research Foundation
2:45-4:30	Brainstorm session	All	All
4:30-5:00	Meeting Wrap-up and Action Items	Deepti Dubey/Kimberly Nye	TESS Research Foundation
5:00-5:30	Carpool to MacArthur Park Restaurant	All	27 University Avenue Palo Alto, CA 94301 (650) 321-9990
5:30-8:00	Dinner at MacArthur Park Restaurant	All	All