I would like to confirm that I had the support of the SSIEM by means of a Travel Bursary to attend the International Conference on Ureagenesis Defects and Allied Conditions – Novel Models and Treatment Options - on the 16th to 20th October 2022, in Valencia (Spain).

This is a very convenient and effective type of scientific meeting, with a limited number of attendees, most of them with an excellent expertise, where almost all bring some contribution in the form of long or short communication, enabling the interactivity and sharing experiences around a concise area of knowledge. The meeting, that was not supported by the Industry, was organized by Prof. Vicente Rubio and the local committee and was the second of this format after the first one that was organized by Johannes Häberle and Beat Thony on March 19-21, 2018, in Pontresina (Engadin, Switzerland). Among the International Scientific Committee there were some experts very well known to the members of SSIEM such as Carlo Dionisi-Vici and Johannes Häberle.
The meeting in Valencia consisted of 3 very intensive days of presentations, not only restricted to the Urea Cycle disorders with still some "unknowns" and enigmas about pathophysiology of ureagenesis and hyperammonaemia, and the experience with liver transplantation, but also allied conditions such as mutations in CAD gene, encoding a multifunctional enzyme involved in de novo pyrimidine biosynthesis that has been reported to be associated with early-onset epileptic encephalopathy, sometimes responsive to uridine treatment. There was also a special focus on citrin deficiency, with the presence of the laureate Prof. John E. Walker, Cambridge, UK (Nobel Prize in Chemistry in 1997) and the Citrin Foundation who was presented by the co-founder (Barbara Yu). There were also very interesting communications on experimental work on aralar – SLC25A12 and citrin – SLC25A13 (malate-aspartate shuttle). Promising results were presented about new treatment pathways, for example pharmacological chaperones and AAV gene therapy for OTCD, and liver-targeting mRNA therapy for OTC and ASA deficiencies. Very recent data were also showed on the clinical trial of enzyme replacement therapy for Arginase 1 deficiency, and, finally, I was able to present very good results of liver transplantation in two patients with arginase 1 deficiency which experienced improvement of the neurological symptoms.
I'm very grateful to the SSIEM initiative of a Travel Bursary facilitating the assistance to a very interesting scientific meeting and allowing also insight talks along the Botanical Gardens and short walks inside the Old City and Cultural Center of Valencia.

Best wishes,

Dr. Guillem Pintos-Morell, MD, PhD
Retired Member of SSIEM