"Rotatin is a new causal gene for congenital dilated cardiomyopathy"

We have generated iPSC-CMs for a child with congenital dilated cardiomyopathy and found significant impairment in contractility and mitochondrial function, and recapitulated structural defects found in patient’s iPSC-CMs. CRISPR-Cas9-mediated knockout of RTTN, putative causal gene identified by next gen sequencing technology, recapitulated cardiomyocyte defects, and the correction of the missense mutation in the disease iPSCs restored cardiomyocyte structure and function, confirming causality. It suggests that RTTN may be important gene in cardiac regulation.

9:00 am – 9455 MRB IV

Refreshments provided