The Molecular Mechanism of Barth Syndrome

We are studying tafazzin, a mitochondrial enzyme that is important for the function of the human heart. Tafazzin re-distributes fatty acids between phospholipid species and by doing so creates unique patterns of molecular species. Mutations in tafazzin cause Barth syndrome, a cardiomyopathy associated with muscle weakness, neutropenia, and abnormal growth. Our work relates to topics such as the structure and dynamics of mitochondria, the specific role of lipids in mitochondrial membranes, and the embryologic development of the heart.

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