Workshop: Translational Research in Ribosome-Based Diseases

Chairs: Jeffrey M. Lipton and Kathleen Sakamoto
Speakers: Jeffrey M. Lipton, Steve Ellis, Akiko Shimamura, Kathleen Sakamoto, Adrianna Vlachos

Several bone marrow failure syndromes are associated with mutations in genes that function in ribosome biogenesis, including dyskeratosis congenita (DC), Shwachman Diamond syndrome (SDS), and Diamond Blackfan Anemia (DBA). In addition to defective hematopoiesis, these diseases share the presence of congenital anomalies and predisposition to cancer. Moreover, deficiency of RPS14 was described in myelodysplastic patients with 5q- syndrome further connecting ribosomal deficiency and bone marrow failure. In this workshop, we propose to explore aspects of basic and clinical translational research on diseases associated with defective ribosomal synthesis. The basic biology of ribosome synthesis, alternative models of disease, controversies in current treatment, and future directions will be presented.

Jeffrey M. Lipton: Introduction to Diamond Blackfan Anemia (10 min)

Steve Ellis: Basic biology of ribosomes and defects in human diseases (20 min)

Kathy Sakamoto: Alternative models of Diamond Blackfan Anemia (10 min)

Akiko Shimamura: Shwachman Diamond Syndrome and Translation (20 min)

Adrianna Vlachos: Patient Cases/Controversies in DBA Treatment (20 min)

Discussion and Questions

References

