Wednesday, September 22, 2021
4:00 P.M. By Zoom

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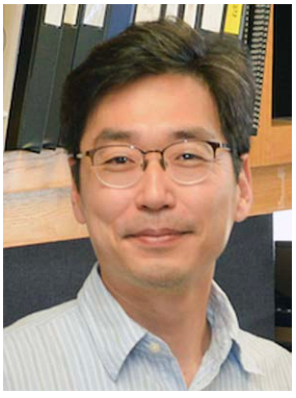
Biochemistry & Molecular Biophysics **Seminar**

Study of Mendelian Mitochondrial diseases: An interdisciplinary approach for disease discovery and mechanism studies

Wan Hee Yoon

Aging & Metabolism Research Program
Oklahoma Medical Research Foundation



1000 to 4000 children are born with a mitochondrial disease in the United States each year. Unfortunately, the genetic causes of the mitochondrial diseases of numerous children are not known. These patients suffer from a diagnostic odyssey lasting many years before they know the cause of their disease and potential treatments. The recent advent of genomic technologies including whole genome sequencing is enabling efficient diagnosis for known mitochondrial diseases and discovering new candidate genetic variants. The discovery of numerous disease-candidate variants from patients has created a pressing need for systematic approaches to determine if/how the variants affect mitochondrial function and human health. To meet this challenge, our group has developed an independent research program that leverages the combinational power of clinical data, genomics analysis, and studies in Drosophila and patient-derived cells. In this talk, I will present our recent discovery of two Mendelian mitochondrial diseases using state-of-the-art of Drosophila genetic technologies.