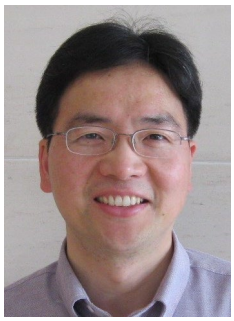
令和5年1月30日 January 30, 2023

# 基盤医学特論
## Tokuron Special Lecture



# Diverse pathophysiologic mechanisms of congenital defects of neuromuscular signal transmission


## Xin-Ming Shen, Ph.D.
### Associate Professor of Neurology
### Mayo Clinic College of Medicine & Science
### Rochester, Minnesota, USA


Shen博士は、神経筋接合部の分子病態解析と電気生理病態解析で世界をリードする研究者です。


Congenital myasthenic syndromes (CMS) are a diverse array of disorders characterized by fatigable muscle weakness due to abnormal neuromuscular transmission caused by variants of more than 30 genes, which encode proteins in presynaptic, synaptic, and/or postsynaptic regions of the neuromuscular junction. The treatment of CMS solely depends on the distinct functional characterization of disease-causing variants as some drugs that are beneficial in one syndrome can be detrimental in another despite similar clinical symptoms. The lecture will discuss how to characterize the pathophysiologic mechanisms of CMS by employing multiple tools, such as muscle biopsy and lab tests, genetic analysis, and molecular and electrophysiologic expression studies. These characterizations guide diagnosis and appropriate treatment for individual patients. The speaker will also discuss how the investigation of pathogenic variants provides opportunities to explore new attributes in normal physiologic mechanisms of the mutant proteins.


| | |
|---|---|
| 日時： | 2023 年 1 月 30 日（月）　午後 5 時から午後 6 時半 |
| Date: | January 30, 2023 (Mon), 17:00 – 18:30 |
| 場所： | Zoom 会議 |
| Venue: | Zoom meeting |
| 言語： | 英語 (パワーポイント・発表) |
| Language： | English (Both PowerPoint and Presentation) |
| 連絡先： | 神経遺伝情報学　大野欽司 (秘書内線 2447) |
| Contact: | Kinji Ohno, Neurogenetics (ext. 2447 for secretary) |
| 注意： | 事前連絡は不要です。基盤医学特論単位認定の対象となる講義です。 |
| Note： | No registration is required. Apply for a Tokuron lecture credit by filing a keyword and a report after the seminar through Google Form. |