**第２０８回　臨時　生物科学セミナー**

（第380回 Zoological Conference）

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**From zebrafish mutation to human disease: Identification of Stac3, a novel regulator of skeletal muscle**

**日時：　2014年3月4日（火）　15:00-16:30**

**場所：　東京大学　理学部２号館　　講堂 （4階）**

**Abstract:** Defects in excitation-contraction (EC) coupling, the process that regulates contractions by skeletal muscles, are associated with numerous muscle diseases. EC coupling transduces changes in membrane voltage by activating release of Ca2+ from internal stores to initiate contraction. A zebrafish genetic screen generated a locomotor mutation that was identified as one in stac3 that is selectively expressed by skeletal muscles. Our studies demonstrated that Stac3 was a novel and important component of the EC coupling complex in muscles. Furthermore, a mutation in human STAC3 was identified as the basis for the debilitating Native American myopathy (NAM). Analysis of NAM stac3 in zebrafish showed that the NAM mutation decreased EC coupling. The identification of Stac3 as a novel component of EC coupling and a causative gene for myopathy enhances our understanding of both EC coupling and the pathology of myopathies. Recent analysis of a related stac gene that is expressed in the CNS will also be discussed.

担当：　東京大学大学院理学系研究科・生物科学専攻・武田洋幸