



# 東北医学会 特別講演

日時:平成29年9月7日(木) 10:00~11:00

会場:東北大学 星陵会館2階大会議室

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(オランダ)



## 【演題】

Understanding the mechanisms  
underlying neurodevelopmental  
disorders and identifying treatments

## 【講演要旨】

Intellectual disability (ID) is present in 2% of all children. Recent evidence suggests that in the majority of the cases a genetic mutation is the primary cause of the intellectual disability. Knowledge of the role of mutated gene is an important step in trying to understand the molecular mechanisms leading to ID, and helps us to design therapeutic strategies in which we try to ameliorate the symptoms associated with these disorders.

In this presentation I will in particular focus on our research on Angelman Syndrome. Angelman syndrome (AS) is a severe neurological disorder, affecting 1:20,000. Children with AS present with severe intellectual disability, motor dysfunction and absence of speech. In addition, there is a high comorbidity with epilepsy and behavioral deficits. AS is caused by spontaneous mutations in the UBE3A gene. Currently, there are no effective treatments for this devastating disorder. Therefore, the primary aim of our research is to advance our knowledge of the underlying pathophysiology of AS with the ultimate aim to identify promising approaches for improving therapeutic treatment.

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この講演会は『国際交流セミナー(アドバンスド講義科目)を兼ねています。  
受講学生は、「国際交流セミナー履修簿(参加するセミナーの名称、月日等を事前に記入すること)を持参し、講演会終了後に”専用シール”を受け取ること。

後援:文部科学省科学研究費補助金新学術領域研究【領域略称「個性」創発脳】  
第60回日本神経化学会大会