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Also, there are some great videos that can be found here: Singapore, 18 May, 2017 – Researchers from the National University of Singapore (NUS) have used a novel approach to identify a previously unknown genetic mutation that is responsible for infantile refractory epilepsy (i.e. seizures that do not respond to existing drugs). As a result, they have developed a potential therapy for this disease. This breakthrough is reported by Professor Julian Tang, Principal Investigator of the study and Associate Professor in NUS' Faculty of Science, in collaboration with Dr Christopher Tsai, NUS' Associate Professor, under the supervision of Associate Professor Eun-Sang Park, Director of the National Genomics Organisation (NUGO). The research was funded by the National Medical Research Council (NMRC), the Ministry of Education and NUS. "This study significantly contributes to our understanding of how genetic mutations can cause epileptic seizures, particularly infantile epileptic seizures, and hence has the potential to identify new therapies for this condition." explains Professor Julian Tang. The findings were presented today in the prestigious journal Neuron, an international journal for academic research, by Professor Julian Tang and Dr Eun-Sang Park. Children with infantile refractory epilepsy often develop cognitive impairment, mental retardation and other abnormalities as a result of the disorder. Despite this, the cause of most cases of infantile epilepsy remain unknown. So far, more than 600 gene mutations associated with infantile epilepsy have been identified. This new study shows that a single mutation in the KCNQ2 gene, also called Kv7.2, accounts for more than 50 percent of cases of infantile refractory epilepsy. "The new discovery has important implications for future therapies for children with infantile refractory epilepsy and other related seizure conditions," said Associate Professor Eun-Sang Park, who is also a group leader at the Singapore-MIT Alliance for Research and Technology (SMART). Researchers are optimistic that the new findings may lead to the development of more effective treatments for this condition. "The findings have highlighted the importance of the KCNQ2 gene in this condition and other forms of epilepsy," said Associate Professor Eun-Sang Park. "We also aim to extend our research to other related conditions including epilepsy and autism." The National University of Singapore (NUS 82157476af

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