

NIGMS Science Advance

Project Title: Human Induced Pluripotent Stem Cell (IPSC) applied to brain disease

Institution and State: Rhode Island Hospital, Providence, Rhode Island

PI Name: Eric M. Morrow

Background: Developed expertise in reprogramming oral mucosa peripheral blood lymphocytes into IPSC and then differentiating this into neurons to study the abnormalities seen in different forms of autism. They have recruited 6 families with NHE6 gene mutations and developed neuronal cell lines from them

Advance: Defined defects in neurite branching in neurons derived from these patients. A unique investigation into the defects underlying one serious form of autism

How NIGMS Grant Enabled Advance: Funded the actual studies

Public Health Impact Statement: Advance in possible therapies for neurologic diseases and an important advance toward possible treatment for selected forms of autism

NIH Director's theme(s) relevance*: Advances in understanding autism which might lead to therapies for selected forms of autism

Grant Support: COBRE grant

Publication Citation and Link (if applicable):

[Genetic and phenotypic diversity of NHE6 mutations in Christianson syndrome.](#)
Pescosolido MF, Stein DM, Schmidt M, El Achkar CM, Sabbagh M, Rogg JM, Tantravahi U, McLean RL, Liu JS, Poduri A, Morrow EM. Ann Neurol. 2014 Oct;76(4):581-93. doi: 10.1002/ana.24225. Epub 2014 Sep 19; PMID:25044251, PMCID: PMC4304796.

Key Words: Autism, Induced pluripotent stem cells and NHE6 mutations

NIGMS Point of Contact:

**NIH Director's Themes: Genomics, Translational Research, Health Care Reform, Global Health, Reinvigorating the Biomedical Community.*

<http://nexus.od.nih.gov/all/2009/09/01/five-themes-for-the-nih-3/>

<http://news.sciencemag.org/funding/2010/02/nih-director-bends-budget-fit-five-themes>