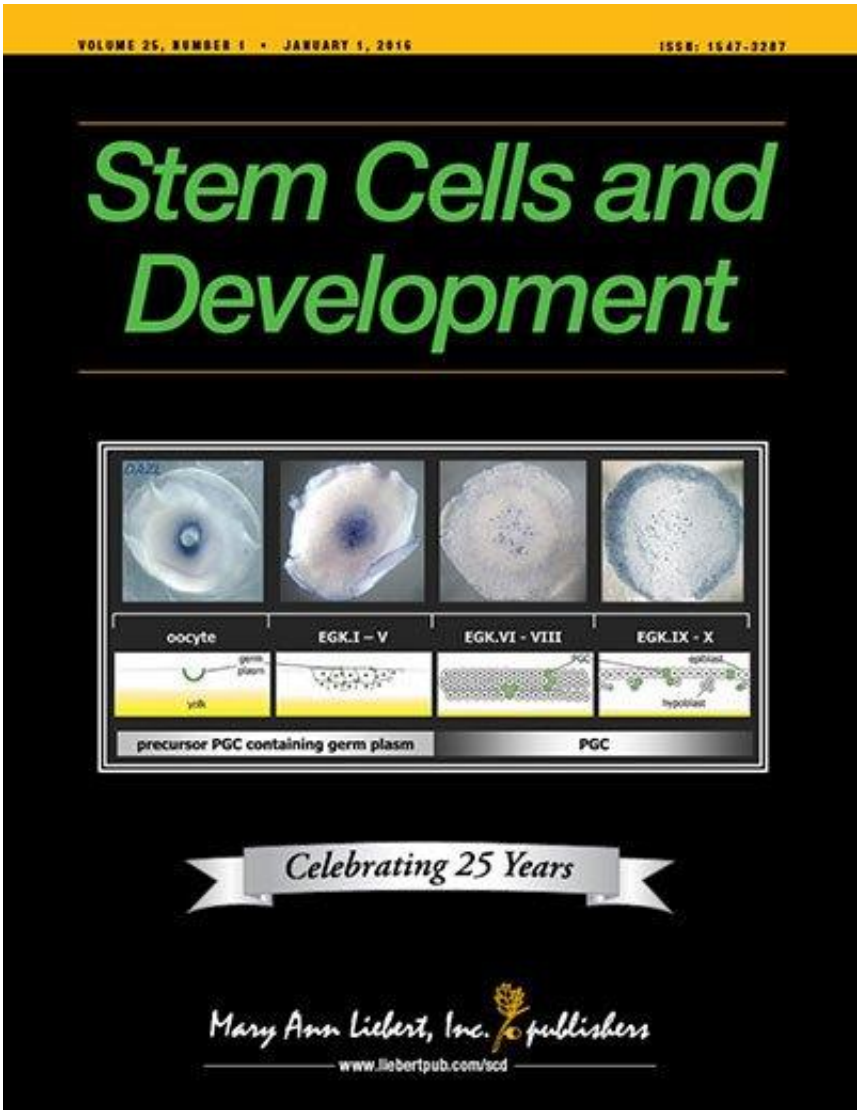
Medical Xpress

New stem cell model valuable tool for studying Andersen's syndrome

February 1 2016



Credit: Mary Ann Liebert, Inc., publishers

Successful reprogramming of muscle cells derived from biopsies of patients with Andersen's syndrome (AS) led to the formation of induced pluripotent stem (iPS) cells that can serve as a valuable model for understanding the cause of this rare disorder and discovering novel therapies. Details of the methods used to generate the AS-iPS cells and evidence of their capacity for self-renewal and pluripotency are presented in the study published in *Stem Cells and Development*. The article is available free on the *Stem Cells and Development* website until March 1, 2016.

In "Modeling Andersen's Syndrome in Human Induced Pluripotent Stem Cells," Jonathan Pini, and coauthors, University Sophia Antipolis, and the Faculté de Médecine, and Institute for Research on Cancer and Aging of Nice (INSERM and CNRS), Nice, France, used the four genes known as "Yamanaka's factors" to reprogram muscle cells from patients with AS and from healthy individuals. The researchers demonstrated that the reprogramming process did not affect the gene mutation known to cause AS, and that the two sets of iPS cells generated did not differ in terms of self-renewal, expression of pluripotent markers, or the ability to differentiate into all three germ layers.

"This is the first report of derivation and initial characterization of iPS cells from a patient with Andersen's syndrome, and as such will provide a boon to researchers of this condition and hopefully encourage derivation of further lines derived from different mutations," says Editor-in-Chief Graham C. Parker, PhD, The Carman and Ann Adams Department of Pediatrics, Wayne State University School of Medicine, Detroit, MI.

More information: Jonathan Pini et al. Modeling Andersen's Syndrome in Human Induced Pluripotent Stem Cells, *Stem Cells and Development* (2016). DOI: 10.1089/scd.2015.0258

Provided by Mary Ann Liebert, Inc