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Lab Resource: Stem Cell Line

Generation of induced pluripotent stem cells from a patient with spinocerebellar ataxia type 3

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ABSTRACT

Spinocerebellar ataxia type 3 (SCA3) is a dominantly inherited neurodegenerative disease caused by a trinucleotide repeat (CAG) expansion in the coding region of *ATXN3* gene resulting in production of ataxin-3 with an elongated polyglutamine tract. Here, we generated induced pluripotent stem cells (iPSCs) from the peripheral blood mononuclear cells of a male patient with SCA3 by using the Sendai-virus delivery system. The resulting iPSCs had a normal karyotype, retained the disease-causing *ATXN3* mutation, expressed pluripotent markers and could differentiate into the three germ layers. Potentially, the iPSCs could be a useful tool for the investigation of disease mechanisms of SCA3.

Resource table.

Name of Stem Cell line	TVGH-iPSC-SCA3-04
Institution	Department of Neurology, Taipei Veterans General Hospital
Person who created resource	Bing-Wen Soong, Huai-En Lu
Contact person and email	Bing-Wen Soong, bwsoong@vghtpe.org.tw
Date archived/stock date	July 11, 2016
Origin	Peripheral Blood Mononuclear Cells
Type of resource	induced pluripotent stem cell (iPSC)
Sub-type	cell line
Key transcription factors	Oct4, Sox2, cMyc, and Klf4
Authentication	identity and purity of stem cell line confirmed
Link to related literature	
Information in public databases	
Ethics	patient informed consent obtained/Ethics Review Board-competent authority approval obtained

1. Resource details

Spinocerebellar ataxia type 3 (SCA3) is a dominantly inherited neurodegenerative disease caused by trinucleotide repeat (CAG) expansion in the coding region of *ATXN3* gene on chromosome 14, which produces an elongated polyglutamine tract, leading to purkinje cell loss (Paulson, 2007). In this report, we successfully generated an iPS cell line, TVGH-iPSC-SCA3-04, from human peripheral blood mononuclear cells (PBMC) that were donated from a patient with SCA3. The PBMCs were reprogrammed by co-expressing Yamanaka factors, *OCT3/4*, *SOX2*, *KLF4*, and *cMYC* through the integration-free Sendai virus gene-delivery method (Takahashi et al., 2007; Takahashi and Yamanaka, 2006; Fusaki et al., 2009). Then, embryonic stem cell (ES)-like colonies were picked and cultured for characterization on day 21. The TVGH-iPSC-SCA3-04 at passage 10 featured a complete removal of all exogenous reprogramming factors (Fig. 1A). The resulting iPSCs had a normal karyotype and retained the disease-causing *ATXN3* mutation (Fig. 1B & C). The endogenous expression of the pluripotent markers, OCT4, SOX2 and NANOG was evaluated by RT-PCR (Fig. 1D). We also confirmed the protein expression of the pluripotent markers, OCT4, SOX2, NANOG, SSEA-3, SSEA-4, TRA-1-60 and TRA-1-81 by flow cytometry and immunocytochemistry staining (Fig. 1E & F). *In vitro* spontaneous differentiation potential towards the three-germ layers of the TVGH-iPSC-SCA3-04 cell line was demonstrated by the expression of

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[1] These authors contributed equally to this work.

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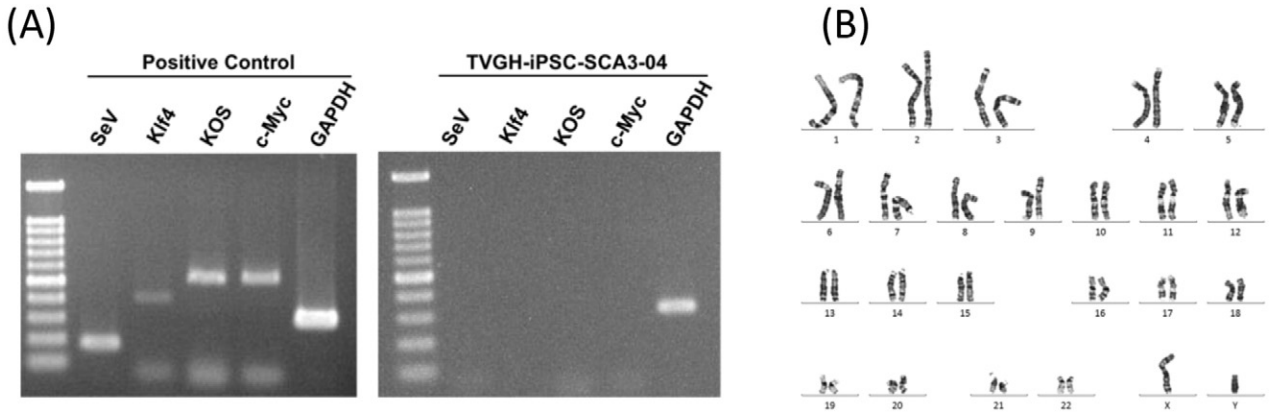
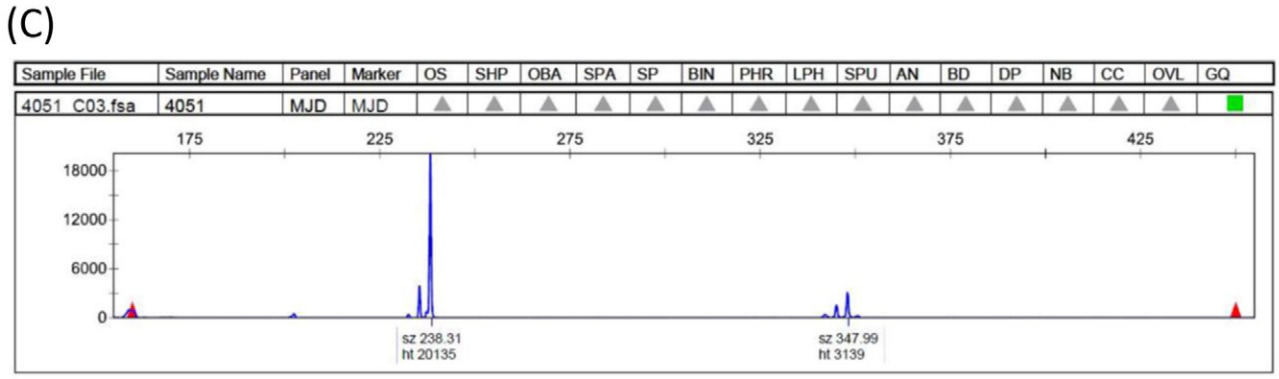

(A)
Positive Control
TVGH-iPSC-SCA3-04
SeV
Klf4
KOS
c-Myc
GAPDH
(B)


(C)
Sample File
Sample Name
Panel
Marker
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SHP
OBA
SPA
SP
BIN
PHR
LPH
SPU
AN
BD
DP
NB
CC
OVL
GQ
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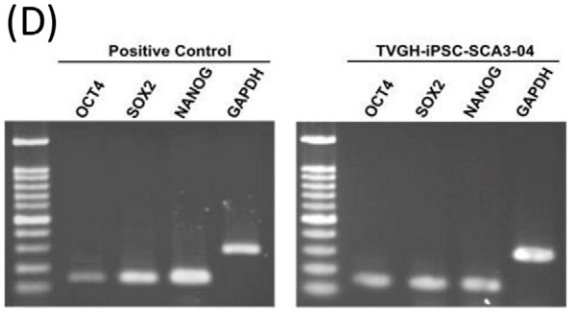
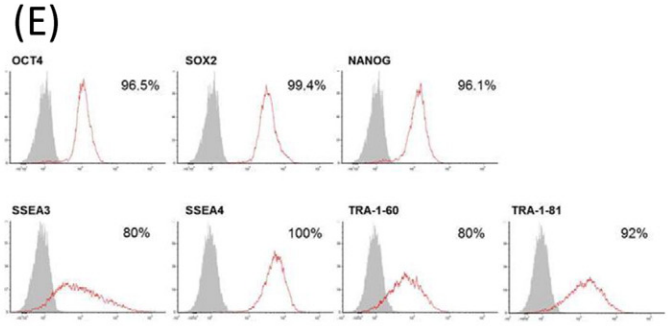
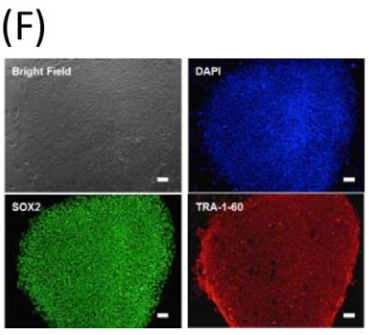
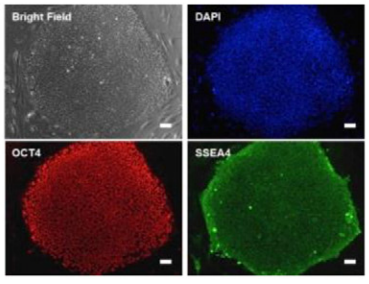
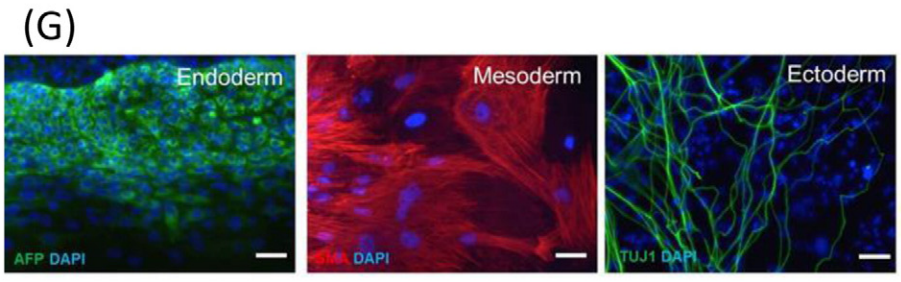
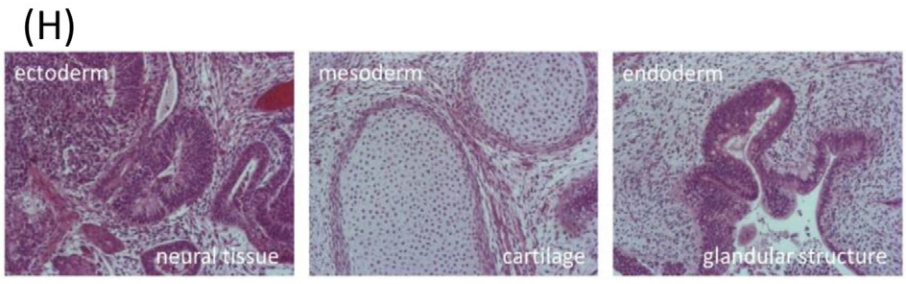

(D)
Positive Control
TVGH-iPSC-SCA3-04
OCT4
SOX2
NANOG
GAPDH


(E)
OCT4
96.5%
SOX2
99.4%
NANOG
96.1%
SSEA3
80%
SSEA4
100%
TRA-1-60
80%
TRA-1-81
92%


(F)
Bright Field
DAPI
SOX2
TRA-1-60


Bright Field
DAPI
OCT4
SSEA4


(G)
Endoderm
AFP DAPI
Mesoderm
SMA DAPI
Ectoderm
TUJ1 DAPI


(H)
ectoderm
neural tissue
mesoderm
cartilage
endoderm
glandular structure