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Lab Resource: Stem Cell Line

Generation of a human iPSC line from a patient with Leigh syndrome

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ABSTRACT

Human iPSC line LND554SV.3 was generated from heteroplasmic fibroblasts of a patient with Leigh syndrome carrying a mutation in the *MT-ND5* gene (m.13513G>A; p.D393N). Reprogramming factors Oct3/4, Sox2, Klf4, and cMyc were delivered using a non-integrative methodology that involves the use of Sendai virus.

Resource table

Name of stem cell line	LND554SV.3
Institution	Departamento de Bioquímica, Instituto de Investigaciones Biomédicas "Alberto Sols", Facultad de Medicina (UAM-CSIC) and Centro de Investigación Biomédica en Red en Enfermedades Raras (CIBERER) Madrid, Spain. Instituto de Investigación Hospital 12 de Octubre ("i + 12"), Madrid, Spain.
Person who created resource	Teresa Galera
Contact person and email	M. Esther Gallardo, egallardo@iib.uam.es
Date archived/stock date	June 20, 2013
Origin	Human skin cells
Type of resource	Biological reagent: induced pluripotent stem cells (iPSC) from a patient with Leigh syndrome
Sub-type	Cell line
Key transcription factors	Oct3/4, Sox2, cMyc, Klf4
Authentication	Identity and purity of cell line confirmed (Fig. 1)
Link to related literature	http://www.ncbi.nlm.nih.gov/pubmed/23034978
Information in public databases	http://www.omim.org/entry/256000
Ethics	Patient informed consent obtained/Ethics Review Board-competent authority approval obtained

Resource details

The generation of the human iPSC line, LND554SV.3, was carried out using non-integrative Sendai viruses containing the reprogramming factors, *OCT3/4*, *SOX2*, *cMYC*, *KLF4* (Takahashi et al., 2007). For this purpose, fibroblasts from a described patient with Leigh syndrome, an inherited devastating neurodegenerative disorder, were employed (Monlleo-Neila et al., 2013). The patient's fibroblasts carried a heteroplasmic mitochondrial DNA (mtDNA) mutation in the *MT-ND5* gene (m.13513G>A; p.D393N) with a mutant mtDNA load of 55%. The presence of this mutation in the iPSCs was confirmed (Fig. 1A). Interestingly, the percentage of mutant mtDNA in the LND554SV.3 line was only 32% due to spontaneous segregation of the heteroplasmic mtDNA content (Fig. 1A). LND554SV.3 iPSC colonies displayed a typical ES-like colony morphology and growth behavior (Fig. 1B) and they stained positive for alkaline phosphatase activity (Fig. 1C). We confirmed the clearance of the vectors and the exogenous reprogramming factor genes by RT-PCR after eight culture passages (Fig. 1D). The endogenous expression of the pluripotency associated transcription factors *OCT4*, *SOX2*, *KLF4*, *NANOG*, *CRIPTO* and *REX1* was evaluated by quantitative

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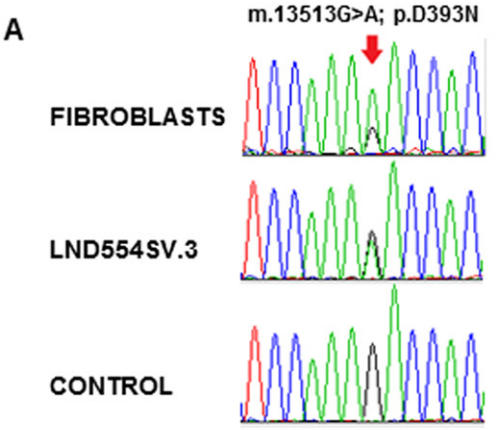

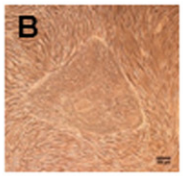

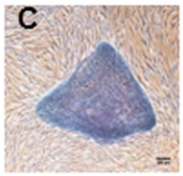

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Marker	Fibroblasts		LND554SV.3	
D2S1338	180	181	180	181
D7S820	209	220	209	220
D8S1179	167	172	167	172
D13S317	174	182	174	182
D19S433	194	198	194	198
D21S11	226	236	226	236
VWA	145	146	145	146
Amelogenin	X	Y	X	Y