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Lab resource: Stem cell line

Generation of spinocerebellar ataxia type 2 patient-derived iPSC line H266

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ABSTRACT

Spinocerebellar ataxia type 2 (SCA2) is a neurodegenerative disease primarily affecting the cerebellum. Very little is known about the molecular mechanisms underlying the disease and, to date, no cure or treatment is available. Here, we demonstrate the generation of an induced pluripotent stem cell (iPSC) line of a SCA2 patient. The selected clone has been proven to be a *bona fide* iPSC line, which retains a normal karyotype. Due to its differentiation potential into neurons, this iPSC line will be a valuable tool in studying a disease-specific phenotype of SCA2.

1. Resource table

Name of Stem Cell construct	H266 clone10
Institution	University of Copenhagen and Bioneer A/S
Person who created resource	Adele G. Marthaler, Benjamin Schmid
Contact person and email	Adele G. Marthaler, adele.marthaler@sund.ku.dk
Date archived/stock date	July 2014
Origin	Human skin fibroblasts
Type of resource	Induced pluripotent stem cells; derived from skin fibroblasts of patient with spinocerebellar ataxia type 2
Sub-type	Cell line
Key transcription factors	Episomal plasmids containing *hOCT4*, *hSOX2*, *hL-MYC*, *hKLF4*, *hLIN28*, and *shP53* (Addgene plasmids 27077, 27078 and 27080; Okita et al., 2011)
Authentication	Identity and purity of stem cell line confirmed (Fig. 1)
Link to related literature (direct URL links and full references)	
Information in public databases	

2. Resource details

Human skin fibroblasts, obtained by skin biopsy of a symptomatic, female 25-year-old spinocerebellar type 2 (SCA2) patient (anonymized as H266), were reprogrammed using episomal vectors carrying transcripts for human *OCT4*, *SOX2*, *KLF4*, *L-MYC*, *LIN28*, and small hairpin RNA for *TP53* (Okita et al., 2011). The clone described in this publication was termed H266 clone (c) 10. The absence of the reprogramming plasmids was confirmed by quantitative PCR (qPCR) on genomic DNA (Fig. 1A).

SCA2 is a dominantly inherited neurodegenerative disorder caused by a mutation in the *ATXN2* gene. Normal alleles contain 22 CAG repeats with CAA interruptions (also coding for glutamine), whereas disease causing alleles contain trinucleotide repeats of 33 or more CAGs (usually without any CAA interruptions) (Pulst et al., 1996). The repeat lengths for patient H266 were determined to be 22 and 44 by fragment length analysis (data not shown) and confirmed to be present also in iPSC line H266 c10 by sequencing both alleles individually (Fig. 1B).

Furthermore, the expression of key pluripotency genes was observed both on RNA, as well as protein level, as demonstrated by qRTPCR analysis and immunocytochemistry, respectively (Fig. 1C and D). Additionally, the cells had the capacity to form derivatives of all three germ layers upon embryoid body differentiation (Fig. 1E). Taken together, this validates the true pluripotent potential of the generated iPCS line.

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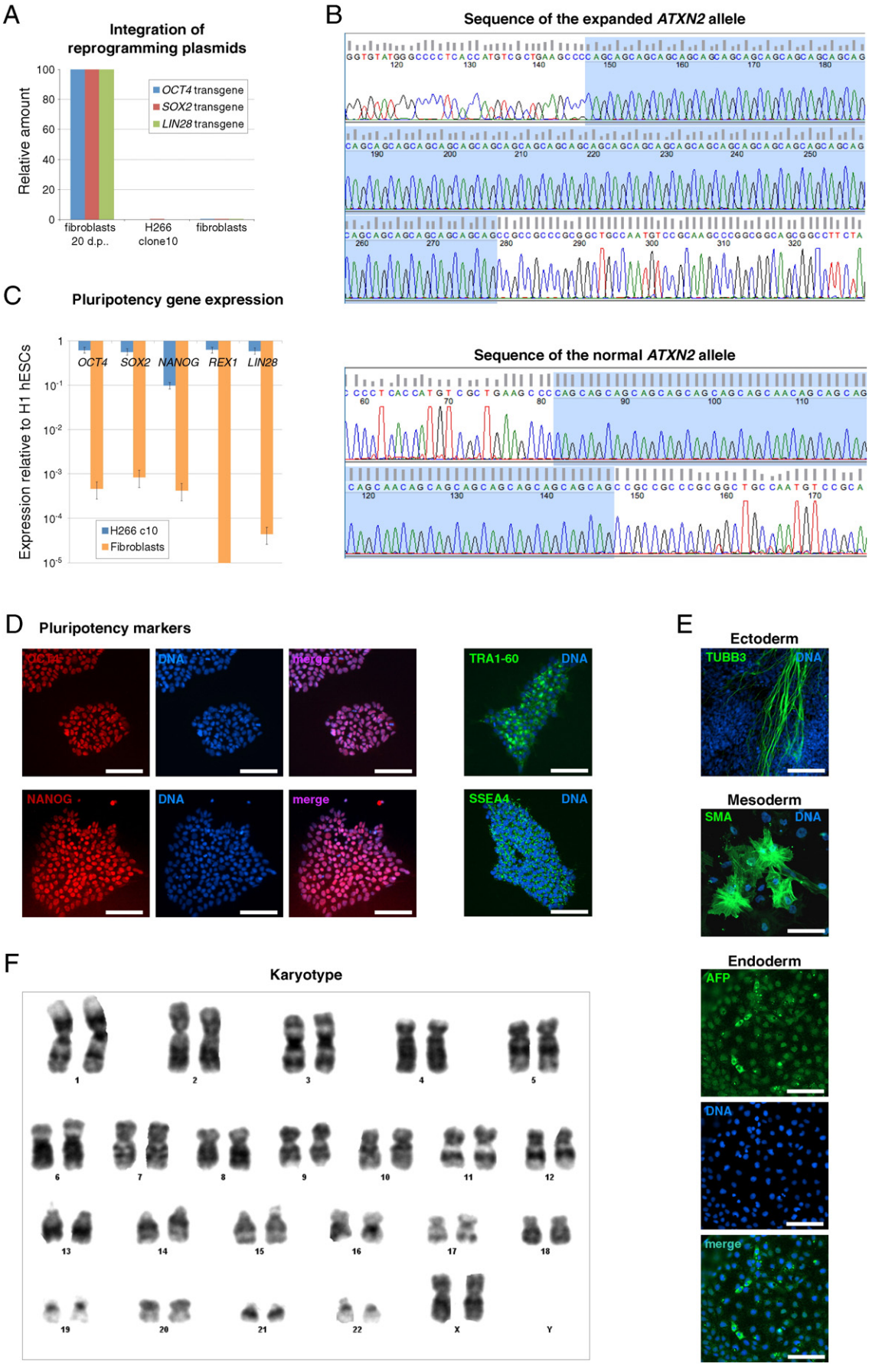

Fig. 1. (*caption on page 168*).