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Lab resource: Stem cell line

Human embryonic stem cells derived from abnormal blastocyst donated by glucose-6-phosphate dehydrogenase deficiency patient

Hao Zhou [a,1], Xiaoying Zhou [b,1], Qingqing Yang [a], Long Li [a], Juan Du [a,c,d], Guangxiu Lu [b,c,d], Ge Lin [a,b,c,d], Qi Ouyang [a,b,c,*]

[a] Institute of Reproductive and Stem Cell Engineering, School of Basic Medical Science, Central South University, Changsha, China
[b] National Engineering and Research Center of Human Stem Cell, Changsha, China
[c] Key Laboratory of Stem Cells and Reproductive Engineering, Ministry of Health, Changsha, China
[d] Reproductive & Genetic Hospital of CITIC-Xiangya, Changsha, China

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ABSTRACT

A human embryonic stem cell (hESC) line was derived from abnormal embryo donated by Glucose-6-phosphate dehydrogenase (G6PD) deficiency patient. Sequencing analysis confirmed that the hESC line possessed the mutant contributing to abnormal expression of G6PD. Further characteristic analysis demonstrated that the favism hESC line maintained stable and normal karyotype, expressed pluripotent markers and had the capacity of generating the derivatives from all three germ layers.

Resource table:

Name of stem cell construct	*ch*HES-415
Institution	National Engineering and Research Center of Human Stem Cell
Person who created resource	Xiaoying Zhou, Qi Ouyang
Contact person and email	Qi Ouyang: loretta0730@163.com
Date archived/stock date	Sep 25, 2014
Origin	abnormal blastocyst from Glucose-6-phosphate dehydrogenase deficiency patient
Type of resource	human embryonic stem cells derived from PGD–analyzed abnormally embryo
Sub-type	cell line
Key transcription factors	NANOG, POU5F1, SOX2
Authentication	Identity and purity of cell line confirmed (Fig. 1)
Link to related literature (direct URL links and full references)	N/A
Information in public databases	N/A

Resource details

Human embryonic stem cell (hESC) line *ch*HES-415 was derived from abnormal blastocyst donated by G6PD deficiency patient after informed consent. Sequencing analysis confirmed a heterozygous missense mutation c.1376G > T (p.R459L) of *G6PD* in the cells(Fig.1A). This mutation was already documented in the main databases such as HGMD (http://www.hgmd.cf.ac.uk/ac/search.php) and ClinVar (http://www.ncbi.nlm.nih.gov/clinvar), indicating a pathogenic effect of this nucleotide change. The result is consistent with that of the proband. These cells expressed pluripotency-related genes NANOG, POU5F1, SOX2 (Fig.1B) and were positive for pluripotent markers POU5F1, NANOG, TRA-1-81 and TRA-1-60 as well as alkaline phosphatase (Fig.1C). During long-term culture on the mitotically inactivated mouse embryonic fibroblasts (MEFs), this cell line maintained a stable karyotype 46, XX (Fig.1D). The differentiation capacity of this cell line was confirmed through *in vitro* and *in vivo* assays. The cells from embryoid bodies expressed the key genes related with the development of main organs from all three germ layers, such as ectoderm markers (PAX6, SOX1, KRT17), mesoderm markers (T, FLK1, RUNX1) and endoderm markers (FOXA2, CXCR4, GATA4) (Fig.1B), while the teratoma contained a complex pattern of differentiation to three germ layers was detected in teratoma (Fig.1E).

Materials and methods

Source of embryo

The ethical committee of Reproductive and Genetic Hospital of CITIC-Xiangya approved this experiment. Abnormal embryo with

* Corresponding author at: Institute of Reproductive and Stem Cell Engineering, School of Basic Medical Science, Central South University, Changsha, China.
E-mail address: loretta0730@163.com (Q. Ouyang).
[1] These authors contribute equally to this study.

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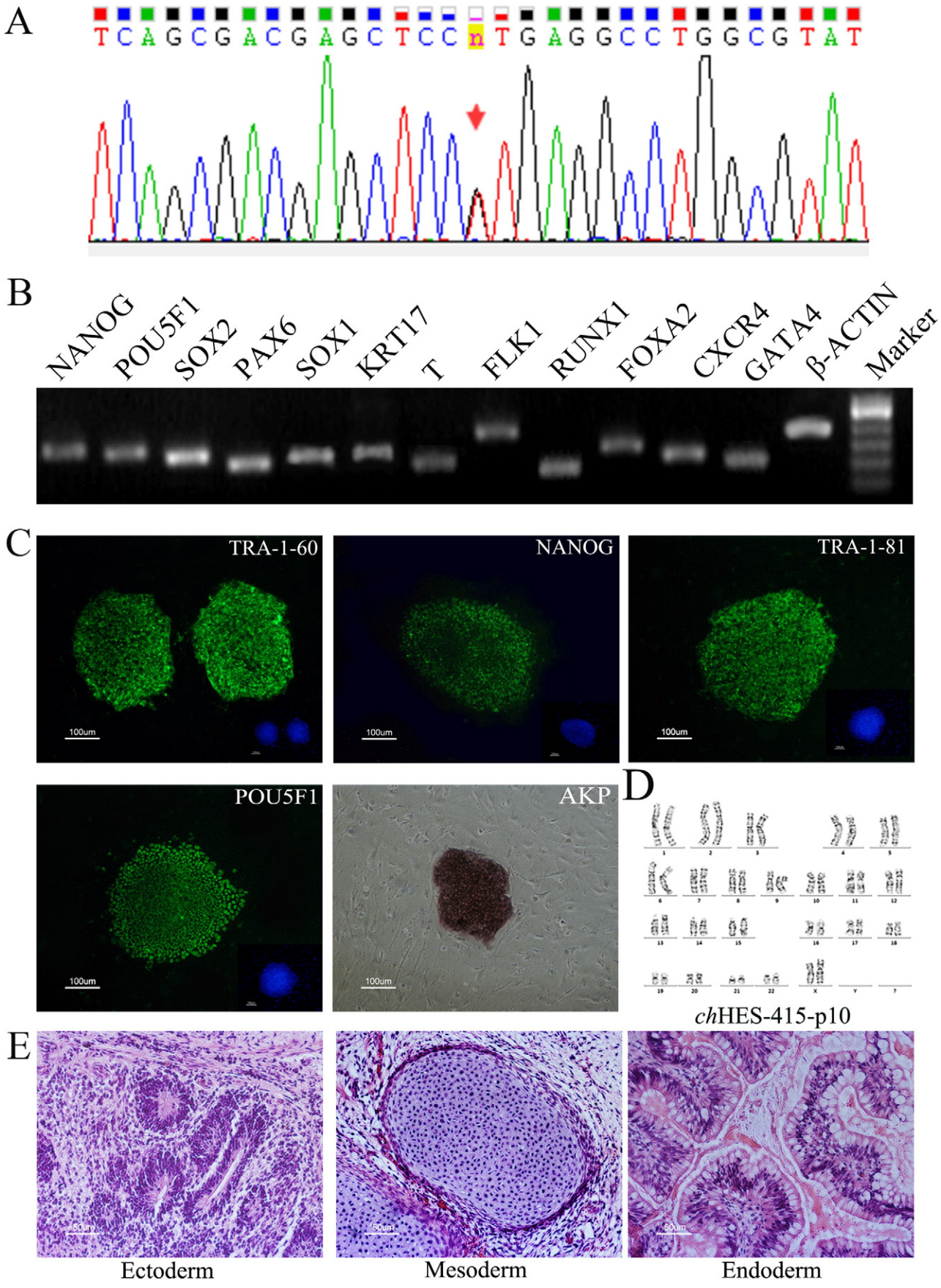
A
T C A G C G A C G A G C T C C n T G A G G C C T G G C G T A T
B
NANOG
POU5F1
SOX2
PAX6
SOX1
KRT17
T
FLK1
RUNX1
FOXA2
CXCR4
GATA4
β-ACTIN
Marker
C
TRA-1-60
NANOG
TRA-1-81
POU5F1
AKP
100um
D
chHES-415-p10
E
Ectoderm
Mesoderm
Endoderm