**Supplementary Information**

# Faithful *SGCE* imprinting in iPSC-derived cortical neurons: an endogenous cellular model of myoclonus-dystonia

Karen Grütz, MSc; Philip Seibler, PhD; Anne Weissbach, MD; Katja Lohmann, PhD; Francesca A. Carlisle, BSc; Derek J. Blake, DPhil; Ana Westenberger, PhD; Christine Klein*, MD; Anne Grünewald, PhD

**Corresponding author:**

Christine Klein, MD

Christine.klein@neuro.uni-luebeck.de

Institute of Neurogenetics, University of Lübeck, Lübeck, Germany

**Supplementary Table S1** Primers for quantification of cDNA, PCR-amplification and sequencing of bisulfite-treated genomic DNA, and PCR-amplification and sequencing of *SGCE* cDNA

| Gene | Template | Application | Forward 5'-3' | Reverse 5'-3' |
|---|---|---|---|---|
| *ACTB* | cDNA | qPCR | TGAAGTGTGACGTGGACATC | GGAGGAGCAATGATCTTGAT |
| *HPRT1* | cDNA | qPCR | ATCAGACTGAAGAGCTATTGTAATGACCA | TGGCTTATATCCAACACTTCGTG |
| *OCT4* endo [1] | cDNA | qPCR | CCTCACTTCACTGCACTGTA | CAGGTTTTCTTTCCCTAGCT |
| *OCT4* trans [1] | cDNA | qPCR | CCTCACTTCACTGCACTGTA | CCTTGAGGTACCAGAGATCT |
| SOX2 endo [1] | cDNA | qPCR | CCCAGCAGACTTCACATGT | CCTCCCATTTCCCTCGTTTT |
| *SOX2* trans [1] | cDNA | qPCR | CCCAGCAGACTTCACATGT | CCTTGAGGTACCAGAGATCT |
| *KLF4* endo [1] | cDNA | qPCR | GATGAACTGACCAGGCACTA | GTGGGTCATATCCACTGTCT |
| *KLF4* trans [1] | cDNA | qPCR | GATGAACTGACCAGGCACTA | CCTTGAGGTACCAGAGATCT |
| *cMYC* endo [1] | cDNA | qPCR | TGCCTCAAATTGGACTTTGG | GATTGAAATTCTGTGTAACTGC |
| *cMYC* trans [1] | cDNA | qPCR | TGCCTCAAATTGGACTTTGG | CGCTCGAGGTTAACGAATT |
| *NANOG* [1] | cDNA | qPCR | TGAACCTCAGCTACAAACAG | TGGTGGTAGGAAGAGTAAAG |
| *GDF3* [1] | cDNA | qPCR | AAATGTTTGTGTTGCGGTCA | TCTGGCACAGGTGTCTTCAG |
| *GATA4 endoderm* [1] | cDNA | qPCR | CTAGACCGTGGGTTTTGCAT | TGGGTTAAGTGCCCCTGTAG |
| *AFP endoderm* [1] | cDNA | qPCR | AGCTTGGTGGTGGATGAAAC | CCCTCTTCAGCAAAGCAGAC |
| *SOX17 endoderm* | cDNA | qPCR | CTCTGCCTCCTCCACGAA | CAGAATCCAGACCTGCACAA |
| *RUNX1 mesoderm* [1] | cDNA | qPCR | CCCTAGGGGATGTTCCAGAT | TGAAGCTTTTCCCTCTTCCA |
| *MSX1 mesoderm* | cDNA | qPCR | CGAGAGGACCCCGTGGATGCAGAG | GGCGGCCATCTTCAGCTTCTCCAG |
| *MYH6 mesoderm* | cDNA | qPCR | TCAGCTGGAGGCCAAAGTAAAGGA | TTCTTGAGCTCTGAGCACTCGTCT |
| *NCAM ectoderm* [1] | cDNA | qPCR | ATGGAAACTCTATTAAAGTGAACCTG | TAGACCTCATACTCAGCATTCCAGT |
| *PAX6 ectoderm* | cDNA | qPCR | GTCCATCTTTGCTTGGGAAA | TAGCCAGGTTGCGAAGAACT |
| *NES ectoderm* [1] | cDNA | qPCR | GCGTTGGAACAGAGGTTGGA | TGGGAGCAAAGATCCAAGAC |
| *MAP2* | cDNA | qPCR | CAGGTGGCGGACGTGTGAAAATTGAGAGTG | CACGCTGGATCTGCCTGGGGACTGTG |
| *PEG10* | cDNA | qPCR | TGTCGTCCGGAGCCACTC | AGGGTGACGGTTGGGGTG |
| *Brain-specific SGCE* | cDNA | qPCR | GGAGATTTTCGTTTGACAAC | CAACATGCATAACATATGCCAG |
| *SGCE* PromM [2] | gDNA | PCR, Seq | GTGTTATGTTTTATAAATAGATAAG | CAACTCATATACCTCTACAATTC |
| *SGCE* Prom Un-methylated | gDNA | PCR, Seq | TTTTATAAATAGATAAGTGTTTTAAATGT | CAACTCATATACCTCTACAATTC |
| *SGCE* Prom Methylated | gDNA | PCR, Seq | TTTATAAATAGATAAGCGTTTTAAATGC | ACTACGACTACTTTATTTATAACGCAAA |
| *SGCE* whole transcript [3] | cDNA | PCR, Seq | CTGATGCTGAACTGGCCAAG | CAACATGCATAACATATGCCAG |
| *SGCE* Ex 3 internal [2] | cDNA | Seq | GCGAGATTAGTAATGATCCC | |

endo - endogenous, trans - transgene, qPCR - quantitative PCR, Seq – Sequencing


1 Park, I. H. *et al.* Reprogramming of human somatic cells to pluripotency with defined factors. *Nature* **451**, 141-146, doi:nature06534 [pii] 10.1038/nature06534 (2008).

2 Muller, B. *et al.* Evidence that paternal expression of the epsilon-sarcoglycan gene accounts for reduced penetrance in myoclonus-dystonia. *Am J Hum Genet* **71**, 1303-1311 (2002).

3 Xiao, J., Nance, M. A. & LeDoux, M. S. Incomplete nonsense-mediated decay facilitates detection of a multi-exonic deletion mutation in SGCE. *Clin Genet* **84**, 276-280, doi:10.1111/cge.12059 (2013).