

**B**

**C**

**H**

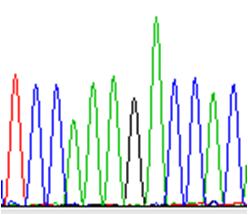
**E**

**G**

**D**

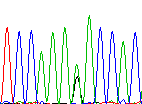
**F**

**I**



**A**

m.13513G



m.13513G>A

Control

Mutant

**Supplementary Figure S1.** Generation and characterization of N44SV.1. **A**) Electropherograms showing the presence of the m.13513G>A mutation in LND554SV.4 and the absence in N44SV.1. **B**) Typical ES-like colony morphology of the N44SV.1 iPSC line growing on feeder cells. **C**) Positive staining for alkaline phosphatase. **D**) qPCR showing the expression of the pluripotency associated genes *OCT4, SOX2, KLF4, CRIPTO, NANOG* and *REX1*. All data normalized with GAPDH. **E**) Immunofluorescence analysis showing expression of typical pluripotent ES cell markers: OCT4, NANOG, SOX2, SSEA3, SSEA4, TRA-1-60 and TRA-1-81; scale bars: 300 µm. **F**) Embryoid body based in vitro differentiation assays demonstrating the ability of N44SV.1 to differentiate into ectoderm (Tuj1+), mesoderm (SMA+), endoderm (AFP+). **G**) RT-PCR for detecting the clearance of the vectors used for reprogramming. **H**) Normal Karyotype (46, XY) in N44SV.1 iPSC line. **i**) DNA fingerprinting analysis from fibroblast and N44SV.1 revealing genetic identity.