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Supplemental Data

Disease-Specific Induced

Pluripotent Stem Cells

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Figure S1.

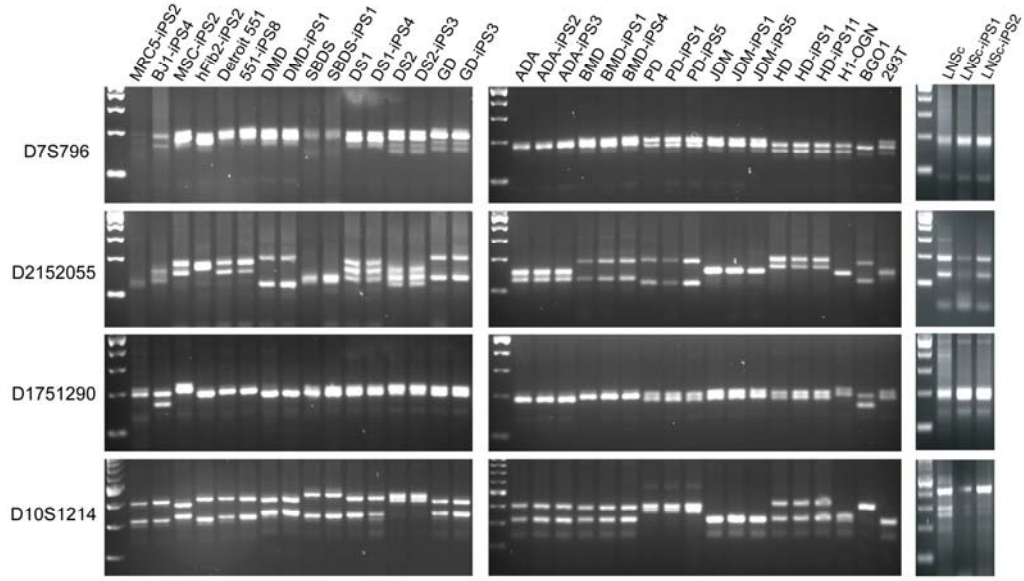

Figure S1. Qualitative DNA fingerprint analysis indicates that each line is derivative of its indicated parental fibroblast source.
PCR-based DNA fingerprint analysis using primer sets spanning highly variable tetra-nucleotide repeats are shown for four different loci: D7S796, repeat (GATA)n, average heterozygosity 0.95; D21S2055, repeat (GATA)n, average heterozygosity 0.88; D17S1290, repeat (GATA)n, average heterozygosity 0.84; and D10S1214, repeat (GGAA)n, average heterozygosity 0.97. Of note, the Down syndrome derived iPS lines (DS1-iPS4 and DS2-iPS3) as well as their respective parent fibroblasts (DS1 and DS2) each show three alleles at D21S2055 in keeping with the observation that most cases of DS derive from errors occurring within meiosis I of female germ cell development, where the two maternal amplicons represent alleles from each maternal grandparent with the third allele originating from within the paternal genome. From left to right at top are six lines of previously described (Park et al., 2008) human iPS cells: MRC5-iPS2 is a normal iPS cell line from fetal lung fibroblasts, BJ1-iPS4 is a

normal iPS cell line from neonatal foreskin fibroblasts, MSC-iPS2 is a normal iPS cell line from mesenchymal fibroblasts, hFib2-iPS2 is a normal iPS cell line from adult fibroblasts, and 551-iPS8 is a normal fibroblast iPS cell line. These are followed (from left to right) by patient-specific iPS lines as well as their parental fibroblast controls: DMD = Duchenne muscular dystrophy, SBDS = Shwachman-Bodian-Diamond syndrome, DS = Down syndrome, GD = Gaucher disease type III, ADA = adenosine deaminase deficiency-associated severe combined immunodeficiency, BMD = Becker type muscular dystrophy, PD = Parkinson disease, JDM = juvenile-onset type one diabetes mellitus, HD = Huntington disease, LNSc = Lesch-Nyhan syndrome carrier, H1-OGN and BG01 are human embryo-derived hES cells, and 293T is an immortalized human embryonic kidney-derived cell line used in the creation of the viral supernatants for reprogramming.

Figure S2.

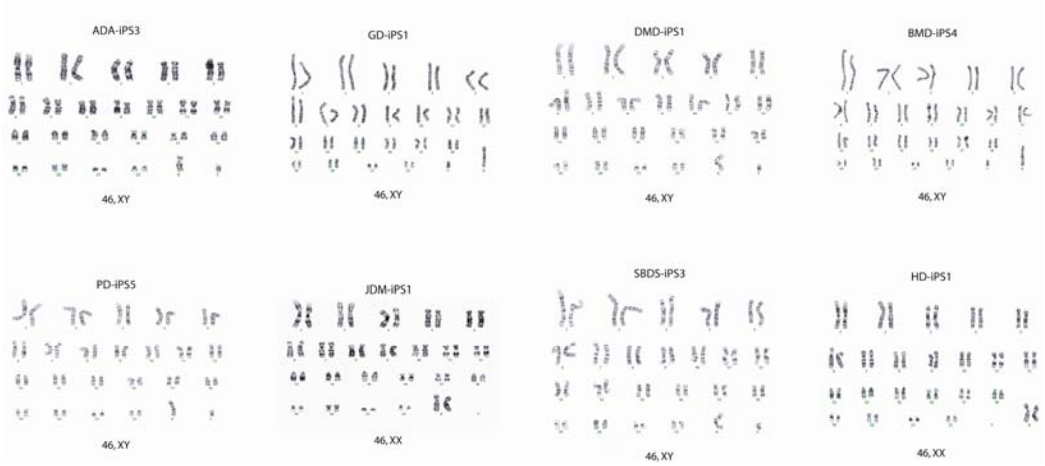

Figure S2. Disease-specific iPS cell lines maintain normal karyotypes.
When chromosomal contents were analyzed with high resolution G-banding karyotypes, ADA-iPS3, GD-iPS1, DMD-iPS1, BMD-iPS4, PD-iPS5, JDM-iPS1, SBDS-iPS3, and HD-iPS1 indicated normal, diploid chromosomal contents.

Supplementary Table 1. Primer sequences used to verify the genetic lesions in patient-derived iPS cells and transgenes in LNSc-iPS cells

Gene	Forward primer	Reverse primer	Sequencing primer
SBDS	GCAAATGGTAA GGCAAATACGG	AAGAAAATATCTGAC GTTTACAACATCTAA	AAAGACCTCGATGAAGTT
HD	AGGTTCTGCTTTTACCTG	CGGCTGAGGAAGCTGAGGA	AGGTTCTGCTTTTACCTG
ADA	CATGACTAGGATGGTTCA	CCTGTTATAAAGGGC CTG	CATGACTAGGATGGTTCA
GBA	TGTGTGCAAGGTCCAGGATCAG	ACCACCTAGAGGGGAAAGTG	TAGCTACTAAGGAATGTG
Lentiviral OCT4	CCCCTGTCTCTGTCACCACT	CCACATAGCGTAAAAGGAGCA	N/A
Lentiviral SOX2	ACACTGCCCCTCTCACACAT	CAT AGC GTA AAA GGA GCA ACA	N/A
Lentiviral cMYC	AAGAGGACTTGTTGCGGAAA	TTGTAATCCAGAGGTTGATTATCG	N/A
Lentiviral KLF4	GACCACCTCGCCTTACACAT	CATAGCGTAAAAGGAGCAACA	N/A
Lentiviral NANOG	ACATGCAACCTGAAGACGTG	CACATAGCGTAAAAGGAGCAA	N/A