

Table S1: Overview of the iPSC lines used in this study

IPCS line	Gender	Genotype	Age of the donor	Derived from	Reprogramming method	Karyotype
NBS1	f	Homozygous <i>NBN</i> 657del5	18	Human dermal fibroblasts	Episomal reprogramming	46,XX
NBS8	m	Heterozygous <i>NBN</i> 657del5	7	Human dermal fibroblasts	Retroviral transduction	46,XX,add(5)(q22), der(11)t(5;11)(q14;q?23)[14]/46,idem, add(4)(q31)[10]
CTR1	m	-		newborn	Retroviral transduction	-
CTR2	m	-		51	Episomal reprogramming	46,XX

Table S5: Primer sequences used in this study

Gene	Forward	Reverse
<i>OTX2</i>	TGCAGGGGTTCTTCTGTGAT	AGGGTCAGAGCAATTGACCA
<i>ATM</i>	AGCTCGGATGCTTTCCTCAA	CTCCATCGAGAAGGTCCACG
<i>CDKN1A</i>	GGAAGACCATGTGGACCTGT	GCGTTTGGAGTGGTAGAAA
<i>DCX</i>	AAGACTTCAGCCAAGAGCCC	GCGTAGAGATGGGAGACTGC
<i>EMX1</i>	AGACGCAGGTGAAGGTGTGG	CAGGCAGGCAGGCTCTCC
<i>FOXA2</i>	CCACCACCAACCCACAAAATG	TGCAACACCGTCTCCCCAAAGT
<i>FOXB1</i>	CCCTCCCATITCTGTACGTTT	CTGGCGGCTCTTAGAGAT
<i>HOXA2</i>	TCAGCAAAATGCCCTCTCT	TAGGCCAGCTCCACAGTTCT
<i>HOXB2</i>	TITAGCCGTTTCGCTTAGAGG	CGGATAGCTGGAGACAGGAG
<i>HOXB4</i>	ACACCCGCTAACAAATGAGG	GCACGAAAGATGAGGGAGAG
<i>HOXB6</i>	GAAGTGAAGAGCGGACTCAC	CTGGGATCAGGGAGTCTTCA
<i>MDM2</i>	AAACTGGGGAGTCTTGAGGG	TGCACATTTGCCTGCTCCTC
<i>MK167</i>	TCGTCCCAGTGGAAGAGTTG	CGACCCCGCTCCTTTTGAT
<i>NNAT</i>	CGGCTGGTACATCTTCCGC	TGTCCCTGGAGGATTTGAAA
<i>NNAT (bissulfite-specific)</i>	GGGTTAGGTAGTTTGTGGA	CCCCCAAACCCTAATAAAT
<i>PAX5</i>	AGGATGCCGCTGATGGAGTAC	TGGAGGAGTGAATCAGCTTGG
<i>SOX2</i>	GAGCTTTGCAGGAAGTTTGC	GCAAGAAGCCTCTCCTTGAA
<i>TP53</i>	CAGGGCAGCTACGGTTTCC	CAGTTGGCAAAACATCTTGTTGAG