

(A)



**Figure S1.** The location of the primers and CpGs investigated in this study for the five annotated regions: (A) CpGi 74, (B) CpGi 43, (C) CpGi 36, (D) CTCF1 and (E) CTCF2. Red letters indicate the external primers used (PCR1), blue highlight indicates the internal primers (PCR2), green letters are used for the CTG expansion and yellow highlight indicate the CpGs present. The numbers correspond to the CpGs analysed in this study. The other CpGs were not readable.

**Table S1.** Primer combinations and thermocycler settings

Primer Name	Sequence (5'-3')	Tm	Cycles
<b>Long PCR</b>			
DM-C	AACGGGGGCTCGAAGGGTCTCT	63.5	28
DM-DR	CAGGCCTGCAGTTTGCCCATC		
<b>Methylation</b>			
CpGi 74-F-1	GGTAAGTAATGGAGTTAGTTT	57	40
CpGi 74-R-1	ACTTCTCTATCTATACTACCA		
CpGi 74-F-2	CTGTAGGGGTAAAGGTTATAG	55	40
CpGi 74-R-2	CTATCTATACTACCAAAAACAA		
CpGi 43-F-1	GATAGTGAGATAGAGTGGAG	55	40
CpGi 43-R-1	AACATTTTTTAACCCCCAAAAC		
CpGi 43-F-2	AGTTTAGTTTTAGGTTTTAAGGT	55	40
CpGi 43-R-2	CTTATCTCCAATACCCCTTCTAA		
CpGi 36-F-1	GGGAATGAGTGATTTAGGATTT	57	40
CpGi 36-R-1	CTCTCCCTCTAAACAAAACACCT		
CpGi 36-F-2	AGTAGGGGTATAGGTATTTATTT	57	40
CpGi 36-R-2	TCTAAACAAAACACCTCTCTCTAC		
CTCF1-F-1	TGTYGTYGTTTTGGGTGTATTG	57	40
CTCF1-R-1	TTCCYGACTACAAAACCCCTTYG		
CTCF1-F-2	GTTGTATTGGGTGGTGGTTTA	57	40
CTCF1-R-2	CTACAAAACCCCTTYGAACCC		
CTCF2-F-1	TTYGGTTAGGTTTGGGTTT	57	40
CTCF2-R	TTAACAAAACAAATTTCCC		
CTCF2-F-2	TAAATTGTAGGTTTGGGAAG	57	40

1 and 2 in the primer name correspond to PCR 1 and 2 in the nested PCR. Tm= melting temperature

**Table S2.** DNA methylation analysis of CpGi 74 in blood samples across the clinical DM1 subtypes.

[illegible]

Late-onset	P38	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	P40	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	P41	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	P50	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	P51	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	P52	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	P53	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	P54	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	P55	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	P56	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	P57	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	P58	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
Asymptomatic Control	P59	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	P60	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	P61	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	P65	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	C1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	C2	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	C3	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	C4	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	0.75	1	1	1	1

**Table S3.** DNA methylation analysis of CpGi 43 in blood across the DM1 clinical subtypes

[illegible]

**Table S4.** DNA methylation analysis of CpGi 36 in blood across the DM1 clinical subtypes

<b>C2</b>	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
<b>C3</b>	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
<b>C4</b>	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	0.9	0.9	1

**Table S5.** DNA methylation analysis of CTCF1 in blood across the DM1 clinical subtypes

CpG →		1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20	21	22	23	24	25	
Co	P1	NA	NA	0.15	0.25	0.25	0.25	0.25	0.5	0.5	0.6	0.65	1	0.65	0.65	0.5	0.75	0.75	NA	NA	NA	NA	NA	NA	NA	NA	
	P1-2	0.25	0.25	0.25	0.4	0.25	0.25	0.25	0.4	0.25	0.5	0.5	0.4	0.4	0.5	0.4	0.65	0.6	0.6	0.5	0.6	0.9	0.9	0.9	0.9	NA	
	P2	NA	0.15	0.15	0.25	0.25	0.25	0.25	0.4	0.4	0.5	0.5	0.5	0.5	0.5	0.5	1	0.8	1	0.5	0.75	0.9	0.9	0.75	0.75	NA	
	P3	0	0.15	0.25	0.15	0.15	0.25	0.25	0.25	0.35	0.4	0.4	0.4	0.4	0.4	0.4	0.55	0.6	0.6	0.5	0.5	0.75	0.75	0.75	0.75	0.75	0.75
	P4	NA	0.25	0.25	0.25	0.25	0.25	0.25	0.3	0.4	0.5	0.5	0.5	0.4	0.4	0.4	0.75	0.75	0.75	0.5	0.5	NA	NA	NA	NA	NA	NA
Ch	P5	1	1	1	1	1	1	1	1	0.45	1	1	1	1	1	1	1	1	1	0.6	0.75	1	0.75	1	1	1	1
	P6	0.25	0.25	0.25	0.4	0.25	0.25	0.25	0.4	0.25	0.25	0.25	0.25	0.4	0.4	0.25	0.5	0.5	0.6	0.5	0.6	0.8	0.9	1	1	NA	
	P7	1	1	1	0.6	0.75	0.75	0.75	0.75	0.75	0.75	0.8	0.75	0.85	0.85	0.75	0.85	0.85	0.85	0.9	0.9	0.9	0.9	0.9	0.9	0.9	
	P8	0	0	0	0	0	0	0	0	0	0	0	0	0.25	0	0	0	0	0	0	0	NA	NA	NA	NA	NA	
	P9	0	0	0	0	0	0	0	0	0	0	0	0	0.25	0	0	0	0	0	0	0	NA	NA	NA	NA	NA	
Juv	P10	NA	0.25	0.5	0.5	0.25	0.25	0.25	0.25	0.25	0.4	0.5	0.45	0.45	0.5	0.4	0.75	0.75	0.75	0.5	0.75	0.75	0.75	0.75	0.75	0.75	0.75
	P11	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	
	P12	NA	0.5	0.75	0.5	0.5	0.5	0.5	0.5	0.5	0.6	0.75	0.55	0.5	0.5	0.5	0.75	0.5	0.55	0.4	0.5	0.75	0.8	0.75	0.8	0.8	
	P13	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	
	P14	NA	0	0	0	0	0.25	0	0	0.25	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	
A	P15	NA	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	NA	NA	NA	NA	NA	NA	NA	NA	NA	
	P16	0	0	0	0	0	0	0	0	0	0	0	0.5	0	0	0	0	0	0	0	0	0	0	0	0	0	
	P17	0	0	0	0	0	0	0	0	0	0	0	0.25	0	0	0	0	0	0	0	0	0	0	0	0	0	
	P18	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	
	P19	NA	0.25	0.25	0.25	0.25	0.25	0.25	0.4	0.5	0.5	0.5	0.5	0.5	0.5	0.5	0.8	0.9	0.9	0.5	0.6	0.9	1	0.75	0.75	NA	
	P20	NA	0.15	0.15	0.25	0.25	0.25	0.25	0.4	0.4	0.5	0.5	0.5	0.5	0.5	0.5	0.75	0.75	0.75	0.75	0.5	0.4	0.9	0.9	0.5	0.9	
	P21	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	
	P22	NA	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	
	P23	NA	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	
	P24	NA	0.05	0.05	0.05	0.05	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	NA	NA	NA	NA	
	P25	NA	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	NA	NA	NA	NA	
	P26	NA	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	NA	NA	NA	NA	
	P27	NA	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	NA	NA	NA	NA	
	P28	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	
	P29	0.05	0.05	0.05	0.05	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
	P30	0.05	0.05	0.05	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
	P31	0.4	0.4	0.25	0.45	0.6	0.75	0.75	0.75	0.7	0.8	0.75	0.8	0.8	0.8	0.8	0.75	0.75	0.85	0.85	0.85	1	1	0.9	0.75	1	1
	P32	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
	P33	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
	P34	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	NA	NA	NA	NA	NA	NA
	P35	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	NA	NA	NA	NA	NA	NA
	P36	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
	P37	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	NA	NA	NA	NA	NA
	P38	0	0	0	0	0	0	0	0	0	0	0	0	0.25	0	0	0	0	0	0	0	0	NA	NA	NA	NA	NA
	P39	0	0	0	0	0	0	0	0	0	0	0	0	0.25	0	0	0	0	0	0	0	0	NA	NA	NA	NA	NA
P40	0	0	0	0	0	0	0	0	0	0	0	0	0.25	0	0	0	0	0	0	0	0	NA	NA	NA	NA	NA	
P41	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	NA	NA	NA	NA	
P42	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	
P43	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	
P45	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	
P46	0	0	0	0	0	0	0.25	0	0.25	0	0.25	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	
P47	NA	0	0	0	0	0	0	0.1	0	0	0	0	0	0	0	0	0	0	0	0	0	NA	NA	NA	NA	NA	
P48	NA	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	NA	NA	NA	NA	
P49	NA	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	NA	NA	NA	NA	NA	
P50	0.1	0.1	0.1	0.2	0.2	0.2	0.2	0.2	0.2	0.2	0.25	0.25	0.25	0.25	0.3	0.25	0.4	0.5	0.5	0.5	0.6	NA	NA	0.75	0.6	0.5	
P51	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	NA	NA	NA	NA	NA	NA	NA	
P52	NA	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	
P53	NA	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	

[illegible]

**Table S6.** DNA methylation analysis of CTCF2 in blood across the DM1 clinical subtypes

[illegible]

	P27	0	0	0	0.05	0.05	0	0	0	0	0	0	0
	P28	NA	NA	NA	0	0	0	0	0	0	0	0	0
	P29	NA	NA	NA	0	0	0	0.2	0	0	0	0	0
	P30	0	0	0	0.05	0	0	0	0	0	0	0	0
	P31	NA	NA	NA	0.5	0.6	1	0.75	0.75	1	1	1	1
	P32	0	0	0	0	0	0	0	0	0	0	0	0
	P33	0	0	0	0	0	0	0	0	0	0	0	0
	P35	0	0	0	0	0	0	0	0	0	0	0	0
	P37	0	0	0	0	0	0	0	0	0	0	0	0
	P38	0	0	0	0	0	0	0	0	0	0	0	0
	P39	0	0	0	0	0	0	0	0	0	0	0.05	0
	P40	0	0	0	0	0	0	0	0	0	0	0	0
	P41	0	0	0	0	0	0	0	0	0	0	0	0
	P42	0	0	0	0	0	0	0	0	0	0	0	0
	P43	0	0	0	0	0	0	0	0	0	0	0	0
	P44	NA	NA	NA	0	0	0	0.5	0	0	0	0	0
	P45	0	0	0	0	0	0	0	0	0	0	0	0
	P48	0	0	0	0	0	0	0	0	0	0	0	0
	P49	0	0	0	0	0	0	0	0	0	0	0	0
	P50	NA	NA	NA	0	0	0	0	0	0	0	0	0
	P51	NA	NA	NA	0	0	0	0	0	0	0	0	0
	P52	0	0	0	0	0	0	0	0	0	0	0	0
	P53	0	0	0	0	0	0	0	0	0	0	0	0
	P54	0	0	0	0	0	0	0.1	0	0	0	0	0
Late-onset	P55	0	0	0	0	0	0	0	0	NA	NA	NA	NA
	P56	0	0	0	0	0	0	0	0	NA	NA	NA	NA
	P57	0	0	0	0	0	0	0	0	0	0	0	0
	P58	0	0	0	0	0	0	0	0	0	0	0	0
	P59	0	0	0	0	0	0	0	0	0	0	0	0
	P60	0	0	0	0	0	0	0.05	0	0	0	0	0
Asymptomatic	P63	0	0	0	0	0	0	0.1	0	0	0	0	0
	P64	0	0	0	0	0	0	0	0	NA	NA	NA	NA
	P65	0	0	0	0	0	0	0	0	NA	NA	NA	NA
Control	Ctrl 1	0	0	0	0	0	0	0	0	0	0	0	0
	Ctrl 2	0	0	0	0	0	0	0	0	0	0	0	0
	Ctrl 3	0	0	0	0	0	0	0	0	0	0	0	0
	Ctrl 4	0	0	0	0	0	0	0	0	0	0	0	0
	Ctrl 5	0	0	0	0	0	0	0	0	0	0	0	0
	Ctrl 6	0	0	0	0	0	0	0	0	0	0	0	0
	Ctrl 7	NA	0	0	0	0	0	0	0	0	0	0	0

Range 0 to 1 indicates 0 to 100% methylation. NA = not available. P1-2= five year follow-up P1

**Table S7.** Clinical characteristics of congenital cases

Patient	Methyl CTCF1/2	Age of onset (years)	Age at sampling (years)	Gender	ePAL (CTGs)	Myotonia	Facial weakness	Axial weakness	Limb weakness	MIRS	Cardiac involvement	NVM	Hyper-somnolence	Cataracts	mRS	DM1-ACTIV
P1	yes/no	birth	10	female	831	no	mild	moderate	moderate proximal	4	no	no	no	no	3	32
P2	yes/yes	birth	18	male	587	no	severe	mild	severe proximal	5	LAFB	no	U	no	5	1
P3	yes/no	birth	2	male	1011	no	moderate	moderate	NA	4	no	no	no	no	NA	NA
P4	yes/no	birth	16	male	730	yes	moderate	mild	moderate distal	3	no	yes	no	no	3	38

<b>P5</b>	yes/yes	birth	16	male	222	yes	unknown	none	moderate distal	3	no	no	no	no	2	40
<b>P6</b>	yes/yes	birth	15	female	280	no	mild	moderate	moderate proximal	4	no	no	yes	no	3	30

Methyl CTCF1/2= hypermethylated at the CTCF1 and CTCF2 site; ePAL= estimated progenitor allele size; NA = not applicable; U= unknown; AV = atrioventricular; LAFB = left anterior fascicular block; mRS = modified Rankin Scale; MIRS = Muscular Impairment Rating Scale; NMV = nocturnal mechanical ventilation.

**Table S8.** Clinical characteristics of juvenile cases

<i>Patient</i>	<i>Methyl CTCF1/2</i>	<i>Age of onset (years)</i>	<i>Age at sampling (years)</i>	<i>Gender</i>	<i>ePAL (CTGs)</i>	<i>Myotonia</i>	<i>Facial weakness</i>	<i>Axial weakness</i>	<i>Limb weakness</i>	<i>MIRS</i>	<i>Cardiac involvement</i>	<i>NVM</i>	<i>Hyper-somnolence</i>	<i>Cataracts</i>	<i>mRS</i>	<i>DM1-ACTIV</i>
<b>P13</b>	no/no	15	39	female	286	yes	mild	none	mild/moderate proximal+distal	4	LAFB	no	yes	no	2	U
<b>P14</b>	no/no	12	22	male	NA	yes	none	none	none	2	NA	no	no	no	1	40
<b>P15</b>	no/no	18	54	female	392	yes	moderate	U	mild/moderate proximal+distal	4	1° AV block	no	no	yes	3	13
<b>P16</b>	no/no	11	12	male	517	yes	U	U	none	2	no	no	U	no	1	36
<b>P17</b>	no/no	16	19	female	273	yes	mild	U	mild distal	1	no	no	no	no	1	40
<b>P18</b>	no/no	11	16	female	222	yes	none	none	mild distal	2	no	no	no	no	1	40
<b>P19</b>	yes/yes	15	26	female	U	yes	none	none	none	2	no	no	yes	no	1	39
<b>P20</b>	yes/yes	15	24	male	189	yes	none	none	none	2	no	no	no	no	3	37
<b>P21</b>	no/no	12	40	female	428	yes	mild	moderate	mild distal	3	no	yes	yes	no	2	18
<b>P22</b>	no/no	19	37	female	251	yes	mild	none	mild distal	3	no	no	no	no	1	29
<b>P23</b>	no/no	12	14	female	642	yes	mild	none	none	2	no	no	yes	no	1	40
<b>P24</b>	no/no	14	16	female	229	yes	mild	none	none	2	no	no	no	no	1	40
<b>P25</b>	no/no		15	male	235	yes	none	none	none	1	1°AV block	no	no	no	0	40
<b>P26</b>	no/no	15	16	male	323	yes	mild	none	none	2	no	no	no	no	1	40
<b>P27</b>	no/no	16	37	female	330	yes	mild	mild	mild/moderate proximal+distal	4	1°AV block	no	yes	yes	2	29
<b>P28</b>	no/no	17	45	male	236	yes	mild	U	mild/moderate proximal+distal	3	1°AV block + LAFB	yes	no	no	3	24
<b>P29</b>	no/no	18	26	female	254	yes	none	none	none	1	no	no	no	no	1	40
<b>P30</b>	no/no	18	24	male	274	yes	mild	none	mild distal	2	no	no	yes	no	1	33
<b>P31</b>	yes/yes	16	53	female	420	yes	mild	none	mild/moderate proximal+distal	3	no	no	no	yes	2	27
<b>P32</b>	no/no	15	29	male	237	yes	moderate	none	none	2	pacemaker	yes	no	no	3	19
<b>P33</b>	no/no	19	24	male	228	yes	mild	none	mild distal	2	no	no	yes	no	1	37
<b>P34</b>	no/no	13	20	female	368	yes	mild	none	mild distal	2	no	no	yes	no	1	39
<b>P35</b>	no/no	14	15	male	318	yes	mild	none	none	2	no	no	yes	no	1	39

Methyl CTCF1/2= hypermethylated at the CTCF1 and CTCF2 site; ePAL= estimated progenitor allele size; U= unknown; AV = atrioventricular; LAFB = left anterior fascicular block; mRS = modified Rankin Scale; MIRS = Muscular Impairment Rating Scale; NMV = nocturnal mechanical ventilation

**Table S9.** Logistic regression model ePAL/modal vs. Methylation status

<i>Model</i>	<i>Variables</i>	<i>OR</i>	<i>CI</i>	<i>p-value</i>	<i>N</i>
<b>1</b>	ePAL versus MetS	1.005	1.002-1.008	0.004	53
<b>2</b>	Modal versus MetS	1.004	1.002-1.007	0.001	46

Significance is set at  $p \leq 0.05$ . ePAL= estimated progenitor allele size; MetS= methylation status



**Table S10.** DNA methylation analysis of CpGi 74 in lymphoblastoids

CpG →		1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19
<i>Congenital</i>	P1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	P2	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	P4	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
<i>Childhood</i>	P7	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	P8	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	P9	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	P10	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	P11	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
<i>Juvenile</i>	P13	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	P20	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
<i>Adult</i>	P36	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	P38	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
<i>Late-onset Control</i>	P56	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	C1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	C4	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	C5	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	C6	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1

Range 0 to 1 indicates 0 to 100% methylation.

**Table S11.** DNA methylation analysis of CpGi 43 in lymphoblastoids

CpG →		1	2	3	4	5	6	7	8	9	10	11	12	13	14	15
<i>Congenital</i>	P1	0	0	0	0	0	0	0	0	0	0	0	0	0	0.35	0.3
	P2	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.25
	P4	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.15
<i>Childhood</i>	P7	0	0	0	0	0	0	0	0	0	0	0	0	0.1	0	0.2
	P8	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
	P9	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.3
	P10	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
	P11	0	0	0	0	0	0	0	0	0	0	0	0	0	0.3	0.6
<i>Juvenile</i>	P13	0	0	0	0	0	0	0	0	0	0	0	0	0	0.2	0.2
	P20	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
<i>Adult</i>	P36	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
	P38	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
<i>Late-onset Control</i>	P56	0	0	0	0	0.2	0	0	0	0	0	0	0	0	0.4	0.3
	C1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
	C4	0	0	0	0	0	0	0	0	0	0	0	0	0	0.45	0.45
	C5	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
	C6	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0

Range 0 to 1 indicates 0 to 100% methylation.

**Table S12.** DNA methylation analysis of CpGi 36 in lymphoblastoids

CpG →	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17
<b>Congenital</b>	P2	1	1	1	1	1	1	1	1	1	1	1	1	1	0.8	1	1
	P4	1	1	1	1	1	1	1	1	1	1	1	1	1	0.9	0.9	1
<b>Childhood</b>	P7	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	P8	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	P9	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	P10	1	1	1	1	1	1	1	1	1	1	1	1	1	0.9	0.9	1
<b>Juvenile</b>	P13	1	1	1	1	1	1	1	1	1	1	1	1	1	0.9	0.9	1
<b>Adult</b>	P36	1	1	1	1	1	1	1	1	1	1	1	1	1	1	0.9	1
	P38	1	1	1	1	1	1	1	1	1	1	1	1	1	0.9	0.9	1
<b>Late-onset</b>	P56	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
<b>Control</b>	C4	1	1	1	1	1	1	1	1	1	1	1	1	1	0.9	0.8	1
	C5	1	1	1	1	1	1	1	1	0.8	1	1	1	1	0.9	0.9	1
	C6	1	1	1	1	1	1	0.8	1	1	1	1	1	1	0.8	0.8	1

Range 0 to 1 indicates 0 to 100% methylation.

**Table S13.** DNA methylation analysis of CTCF1 in lymphoblastoids

CpG →	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20	21	22	23	24	25
<b>Congenital</b>	P1	0.25	0.25	0.25	0.4	0.5	0.5	0.5	0.5	0.5	0.5	0.5	0.5	0.5	0.5	0.45	0.5	0.5	0.6	0.4	0.6	0.75	0.75	0.75	0.75
	P2	0.2	0.2	0.2	0.3	0.3	0.45	0.4	0.4	0.45	0.45	0.5	0.4	0.4	0.5	0.4	0.6	0.55	0.55	0.45	0.6	0.75	0.75	0.75	0.8
<b>Childhood</b>	P4	0.1	0.1	0.1	0.1	0.1	0.1	0.2	0.1	0.1	0.1	0.1	0.1	0.1	0.15	0.1	0.1	0.1	0.1	0.2	0.25	0.25	0.2	0.3	0.25
	P7	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
	P8	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
	P9	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
	P10	0.25	0.25	0.25	0.5	0.4	0.4	0.45	0.5	0.5	0.5	0.5	0.5	0.65	0.5	0.75	0.5	0.55	0.45	0.55	0.75	0.8	0.8	0.8	0.75
<b>Juvenile</b>	P11	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
	P13	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
<b>Adult</b>	P20	0.2	0.2	0.2	0.4	0.3	0.3	0.3	0.4	0.4	0.35	0.4	0.4	0.4	0.45	0.5	0.5	0.55	0.5	0.75	0.75	0.75	0.75	0.7	0.7
	P36	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
	P38	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
<b>Late-onset</b>	P56	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
<b>Control</b>	C1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
	C4	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
	C5	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
	C6	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0

Range 0 to 1 indicates 0 to 100% methylation.

**Table S14.** DNA methylation analysis of CTCF2 in lymphoblastoids

CpG →	1	2	3	4	5	6	7	8	9	10	11
<b>Congenital</b>	P2	NA	0	0	0.1	0.1	0.1	0.15	0.1	0.15	0.2
	P4	NA	0	0	0.1	0.1	0.1	0.1	0.1	0.15	0.15
<b>Childhood</b>	P7	NA	0	0	0	0.15	0	0	0.05	0.05	0.05
	P8	0	0	0	0	0	0	0	0	0	0
	P9	NA	0	0	0	0	0	0.05	0.05	0.05	0.05
	P10	NA	0	0	0.05	0.1	0.1	0.1	0.1	0.15	0.1
	P11	NA	0	0	0	0.05	0	0	0	0.5	0.1

Juvenile	P13	0	0	0	0	0	0	0	0	0	0	0
	P20	NA	0	0	0.1	0.1	0.2	0.05	0.1	0.1	0.1	0.15
	P36	0	0	0	0	0	0	0	0	0	0	0
Adult	P38	0	0	0	0	0	0	0	0	0	0	0
	P56	0	0	0	0	0	0	0	0	0	0	0
Late-onset	C1	0	0	0	0	0	0	0	0	0	0	0
Control	C4	0	0	0	0	0	0	0	0	0	0	0
	C5	0	0	0	0	0	0	0	0	0	0	0
	C6	0	0	0	0	0	0	0	0	0	0	0

Range 0 to 1 indicates 0 to 100% methylation. NA = not available

**Table S15.** DNA methylation analysis of CpGi 74 in skin and skin-derived cells

		CpG →	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	
Skin	Adult	P37	1	0.85	1	1	0.7	1	0.75	1	1	1	1	1	1	1	1	0.7	1	1	1	
		P68	1	0.75	1	1	1	1	1	1	1	1	1	1	1	1	1	1	0.5	1	1	1
		P69	0.75	1	1	1	1	1	1	0.95	1	1	1	1	1	1	1	1	0.7	1	1	1
	Control	C2	1	0.75	1	1	1	0.9	0.75	0.9	1	1	1	1	1	1	1	1	0.6	1	0.8	0.8
		C10	1	0.8	1	1	1	0.75	0.8	0.8	1	1	1	1	1	1	1	1	0.75	1	0.95	0.95
Skin fibroblasts																						
Skin fibroblasts	Juvenile	P13	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	0.5	1	1	1	
	Adult	P36	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	
		P38	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	
	Control	C2	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	
		C6	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	0.75	1	1	
		C9	1	1	1	1	1	1	1	1	0.75	1	1	1	1	0.75	1	1	0.75	1	1	1

Range 0 to 1 indicates 0 to 100% methylation.

**Table S16.** DNA methylation analysis of CpGi 74 in muscle and muscle-derived cells

	CpG →	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19
Muscle																				
Juvenile	P13	1	0.75	1	1	0.75	1	0.65	1	0.85	0.9	0.9	1	1	1	1	0.5	1	1	1
Adult	P37	1	0.75	1	1	0.73	0.9	0.55	0.9	1	1	1	1	1	1	1	0.85	1	1	1
	P68	0	0.5	1	0.6	0.75	0.75	0.4	0.83	0.83	0.83	1	0.9	1	1	1	0.6	1	1	1
Late-onset	P56	0.68	0.75	1	0.95	0.73	0.9	0.55	0.85	1	0.9	1	1	1	1	1	0.75	1	1	1
	P67	0.4	0.75	0.75	1	0.95	1	0.75	1	1	1	1	1	1	1	0.9	0.2	1	1	1
Control	C6	1	1	1	1	1	1	1	0.85	1	1	0.5	1	1	1	1	0.5	1	0.7	1
	C9	1	0.67	1	1	0.8	1	0.6	0.9	1	1	1	1	1	1	1	0.8	1	0.9	0.9
Myoblasts																				
Adult	P36	1	1	1	NA	NA	NA	NA	1	1	NA	NA	1	1	1	1	1	1	1	1
	P37	1	1	1	1	1	1	0.75	0.75	1	0.9	0.8	0.9	1	1	0.87	0.8	1	1	1
	P38	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
Late-onset	P56	1	1	1	NA	NA	1	0.95	1	1	1	1	1	0.9	0.9	1	1	1	0.9	0.8
Control	C2	1	0.75	1	NA	NA	1	0.8	1	1	1	1	1	1	1	1	0.8	1	1	1
	C3	NA	0.73	1	NA	NA	NA	NA	1	1	1	1	1	1	1	1	1	1	1	1
	C9	1	1	1	NA	NA	NA	0.9	1	1	1	1	1	1	1	1	0.8	1	1	1
	C10	1	0.75	1	NA	NA	NA	0.8	1	0.8	0.9	1	1	0.9	0.9	1	1	1	0.87	0.87
Myotubes																				
Juvenile	P13	NA	0.4	1	NA	NA	1	0.75	0.65	1	1	1	1	1	1	1	0.5	1	1	1

Adult	P36	1	1	1	1	0.9	1	1	0.8	1	1	1	1	1	1	1	1	1	1	1	1
	P37	1	0.5	1	1	0.75	1	0.75	0.75	0.75	1	1	1	1	1	1	1	0.6	1	1	1
	P38	1	1	1	NA	NA	NA	0.5	0.9	1	1	1	1	1	1	1	0.9	1	NA	1	1
	P56	1	1	1	NA	NA	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
	C2	1	1	1	NA	NA	1	0.75	1	1	0.9	1	1	1	1	1	1	1	1	1	1
	C4	1	0.9	1	NA	NA	1	0.9	0.8	1	1	1	1	1	1	1	0.75	1	1	1	1
	C6	NA	1	1	1	1	1	1	0.9	1	0.95	0.95	1	1	1	0.9	0.75	0.8	0.85	0.85	1
	C9	NA	1	1	NA	NA	NA	0.75	0.75	1	1	1	1	1	1	0.9	0.9	0.7	1	0.8	0.9

**Table S17.** DNA methylation analysis of CpGi 36 in skin and skin-derived cells

[illegible]

**Table S18.** DNA methylation analysis of CpGi 36 in muscle and muscle-derived cells

[illegible]

<b>Controls</b>	<b>C3</b>	1	0.7	1	1	1	1	1	0.7	1	1	1	1	1	1	1	1	0.9
	<b>C9</b>	1	0.7	1	1	1	1	1	1	1	1	1	1	1	1	1	0.9	1
	<b>C10</b>	1	0.8	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1
<b>Myotubes</b>																		
<b>Juvenile</b>	<b>P13</b>	1	1	1	1	1	1	1	1	1	1	0.8	1	1	1	0.6	0.9	0.8
	<b>P36</b>	1	1	1	1	1	1	1	0.7	1	1	1	1	1	0.7	0.7	0.8	0.9
<b>Adult</b>	<b>P37</b>	1	0.5	0.75	0.75	1	0.55	1	0.55	1	1	0.8	1	1	1	0.7	1	1
	<b>P38</b>	1	0.6	1	1	1	1	1	0.75	1	1	1	1	1	0.8	1	0.9	1
<b>Late-onset</b>	<b>P56</b>	1	1	1	1	1	1	0.7	1	1	1	1	1	1	1	1	1	0.6
	<b>C2</b>	1	1	1	1	0.9	0.8	1	0.7	1	1	1	0.9	1	1	1	0.91	
<b>Control</b>	<b>C4</b>	1	0.6	1	1	1	1	1	0.9	1	1	1	1	1	0.8	0.9	1	0.9
	<b>C6</b>	1	0.7	1	1	1	1	1	1	1	1	1	1	1	1	1	1	0.6
	<b>C9</b>	1	0.7	1	1	1	0.7	1	0.9	1	1	1	1	1	1	1	0.8	0.8

Range 0 to 1 indicates 0 to 100% methylation. NA = not available

**Table S19.** DNA methylation analysis of CpGi 43 in skin and skin-derived cells

		CpG →	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15
Skin	Adult	P37	0	0	0	0	0	0.15	0	0.05	0.05	NA	NA	0	0	0	0
		P68	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
		P69	0	0	0	0	0	0.7	0	0	0	0	0	0	0	0	0
	Control	C2	0	0	0	0	0.05	0.1	0.05	0	0.05	0	0	0	0.1	0.1	0.1
		C10	0	0	0	0.05	0	0.1	0.05	0.05	0.05	0.1	0.1	0.1	0.1	0	0.1
Skin fibroblasts																	
	Juvenile	P13	0	0	0	0	0	0	0	0	0.05	0.4	0	0	NA	NA	0.4
	Adult	P36	0.2	0.25	0.5	0	0.25	0	0	0	0.4	0.5	0	0	0.5	0.05	0.8
		P37	0	0	0.05	0	0	0.05	0.05	0.05	0.1	0.1	0.05	0.1	0.1	NA	NA
	Control	P38	0	0	0	0	0	0	0	0	0.5	NA	0	0	0	0	0
		C2	0	0	0.1	0	0.1	0	0.1	0.1	0.1	0.1	0.1	0.1	0.05	0.1	0.3
		C5	1	0	0	0	0	0	0.6	0	0.75	0	0	0	0	0	0
		C6	0	0	0.4	0	0.3	0	0	NA	0.4	0.4	0.2	NA	0	0.5	0.75
		C9	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0

Range 0 to 1 indicates 0 to 100% methylation. NA = not available

**Table S20.** DNA methylation analysis of CpGi 43 in muscle and muscle-derived cells

		CpG →	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15
<b>Muscle</b>																	
<b>Juvenile</b>	<b>Adult</b>	<b>P13</b>	0.1	0	0.35	0	0.2	0.2	0.4	0.4	0.55	0.25	0	0.25	0.25	0.4	0.75
		<b>P36</b>	0	0	0.4	0	0.4	1	0.75	0.5	0.5	1	0.6	0	1	0.5	1
	<b>Control</b>	<b>P37</b>	0.25	0	0.5	0	0.4	0.4	0.5	0.5	0.75	0.75	0.5	0	0.75	0.75	0.8
		<b>P38</b>	1	0	0.4	0.35	0.4	0.4	0.8	0.45	0.5	0.5	0.5	0.25	0	0.75	1
	<b>Late-onset</b>	<b>P68</b>	0	0	0	0	0	0.15	0	0	0.2	0	0.25	0.35	0	0.5	
		<b>P56</b>	0.2	0	0.25	0.25	0.25	0.25	0.4	0.4	0.55	0.5	0.5	0.4	0.4	0.4	0.65
	<b>Control</b>	<b>P67</b>	0.35	0.25	0.25	0	0.05	0.35	0.35	1	0.75	1	0.45	0.45	0.75	0.1	0.8
		<b>C6</b>	0.85	0	0.25	0	0.2	0.2	0.75	0.4	0.75	0.55	0.45	0.45	0.45	0	0.85
	<b>Myoblasts</b>	<b>C9</b>	0.85	0.25	0.2	0	0.2	0.1	0.55	1	0.6	1	0.65	0.25	0.65	0.25	1
		<b>P36</b>	0	0.2	0	0.5	0.5	0.5	0.5	0.25	0.25	0.25	0.15	0.15	0.75	0.75	0.75
	<b>Adult</b>	<b>P37</b>	0.15	0	0	0.6	0	0	0	0	0	0	0.5	0	0	0.5	1

Late-onset Control	P38	0	0	0.25	0.15	0.25	0.15	0.45	0.4	0.5	0.6	0.4	0.5	0.4	0.5	0.75
	P56	0	0	0.85	NA	NA	0.2	0.75	NA	0	0.2	NA	0	NA	0	0.2
	C2	0.25	0	0.6	0.25	0.65	0.65	0.78	0.85	0.9	1	0.5	0.75	0.85	0.6	0.9
	C3	0.35	0	0.55	0.4	0.55	NA	0.65	0.6	0.6	0.9	0.9	0.6	0.65	0.5	0.6
	C9	0.25	0	0.6	0.4	0.5	NA	0.75	0.85	0.85	1	0.75	0.95	0.75	0.65	0.75
	C10	0.4	0	0.67	0.25	0.67	0.5	0.75	1	1	1	0.75	0.75	0.6	0.75	0.75
Myotubes																
Juvenile																
Adult																
Late-onset Control	P13	NA	NA	0	0	0	0	0.5	0.2	0.35	0.35	0.4	0.1	0.25	0.25	0.75
	P36	0.4	NA	0.45	0.1	0.45	0.5	0.6	0.55	0.65	0.8	0.27	0.25	0.75	0.45	0.8
	P37	0.6	0	0.25	0.20	0.5	NA	0.78	1	0.8	1	0.70	0.85	0.35	0.65	0.9
	P38	0.05	0	0.15	0.1	0.15	0.2	0.25	0.25	0.5	0.3	0.2	0.25	0.6	0.5	0.75
	P56	NA	0	0	0.55	0	0.55	0.2	0.6	0.25	0.5	0.6	0.5	0.1	0.6	0.35
	C2	0.3	0	0.45	0.1	0.75	NA	0.6	0.6	0.75	0.75	0.55	0.7	0.75	0.6	0.8
	C4	0.5	0	0.7	0	0.75	0.5	0.8	0.75	1	1	0.75	0.8	0.5	0.75	0.75
	C6	0	0	0	0	0.25	0	0	0	0	0	0	0	0	NA	0.3
	C9	0.5	0	0.75	0.4	0.55	NA	NA	0.85	1	1	NA	0.75	0.85	1	

**Table S21.** DNA methylation analysis of CTCF1 in skin and skin-derived cells

Range 0 to 1 indicates 0 to 100% methylation. NA = not available, Skin fibros= skin fibroblasts

[illegible]

[illegible]

**Table S23.** CTG expansion sizes in blood and muscle of the patients providing a muscle biopsy

	Blood progenitor	Blood modal	Muscle progenitor	Muscle modal
Juvenile P13	286	445	691	736
Adult P36	180	338	208	335
Adult P37	199	374	469	788
Adult P38	115	130	135	207
Adult P68	322	619	351	758
Late-onset P56	300	381	NA	NA
Late-onset P67	108	246	128	512

**Table S24.** DNA methylation analysis of CTCF2 in skin and skin-derived cells

[illegible]

**Table S25.** DNA methylation analysis of CTCF2 in muscle and muscle-derived cells

		CpG →	1	2	3	4	5	6	7	8	9	10	11
<b>Muscle</b>	<b>Juvenile</b>	P13	NA	NA	NA	0	0	0	0	0	0	0	0
		P36	NA	NA	NA	0	0	0	0	0	0	0	0
	<b>Adult</b>	P37	NA	NA	NA	0	0	0	0	0	0	0	0
		P38	NA	NA	NA	0	0	0	0	0	0	0	0
	<b>Late-onset</b>	P68	NA	NA	NA	0	0	0	0	0	0	0	0
		P56	NA	NA	NA	0	0	0	0	0	0	0	0
	<b>Control</b>	P67	NA	NA	NA	0	0	0	0	0	0	0	0
		C4	NA	NA	NA	0	0	0	0	0	0	0	0
		C6	NA	NA	NA	0	0	0	0	0	0	0	0
		C9	NA	NA	NA	0	0	0	0	0	0	0	0
<b>Myoblasts</b>	<b>Juvenile</b>	P13	NA	0	0	0	0	0	0	0	0	0	0
		P36	NA	0	0	0	0	0	0	0	0	0	0
	<b>Adult</b>	P37	NA	0	0	0	0	0	0	0	0	0	0
		P38	NA	0	0	0	0	0	0	0	0	0	0
	<b>Late-onset</b>	P56	NA	0	0	0	0	0	0	0	0	0	0
		C2	0	0	0	0	0	0	0	0	0	0	0
	<b>Control</b>	C3	0	0	0	0	0	0	0	0	0	0	0
		C9	0	0	0	0	0	0	0	0	0	0	0
		C10	0	0	0	0	0	0	0	0	0	0	0
<b>Myotubes</b>	<b>Juvenile</b>	P13	NA	0	0	0	0	0	0	0	0	0	0
		P36	NA	0	0	0	0	0	0	0	0	0	0
	<b>Adult</b>	P37	NA	0	0	0	0	0	0	0	0	0	0
		P38	NA	0	0	0	0	0	0	0	0	0	0
	<b>Late-onset</b>	P56	NA	0	0	0	0	0	0	0	0	0	0
		C2	0	0	0	0	0	0	0	0	0	0	0
	<b>Control</b>	C4	0	0	0	0	0	0	0	0	0	0	0
		C9	0	0	0	0	0	0	0	0	0	0	0
		C6	0	0	0	0	0	0	0	0	0	0	0

Range 0 to 1 indicates 0 to 100% methylation. NA = not available