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Supplemental Data

Modeling the Mutational and Phenotypic Landscapes of Pelizaeus-Merzbacher Disease with Human iPSC-Derived Oligodendrocytes

Zachary S. Nevin, Daniel C. Factor, Robert T. Karl, Panagiotis Douvaras, Jeremy Laukka, Martha S. Windrem, Steven A. Goldman, Valentina Fossati, Grace M. Hobson, and Paul J. Tesar

Figure S1.

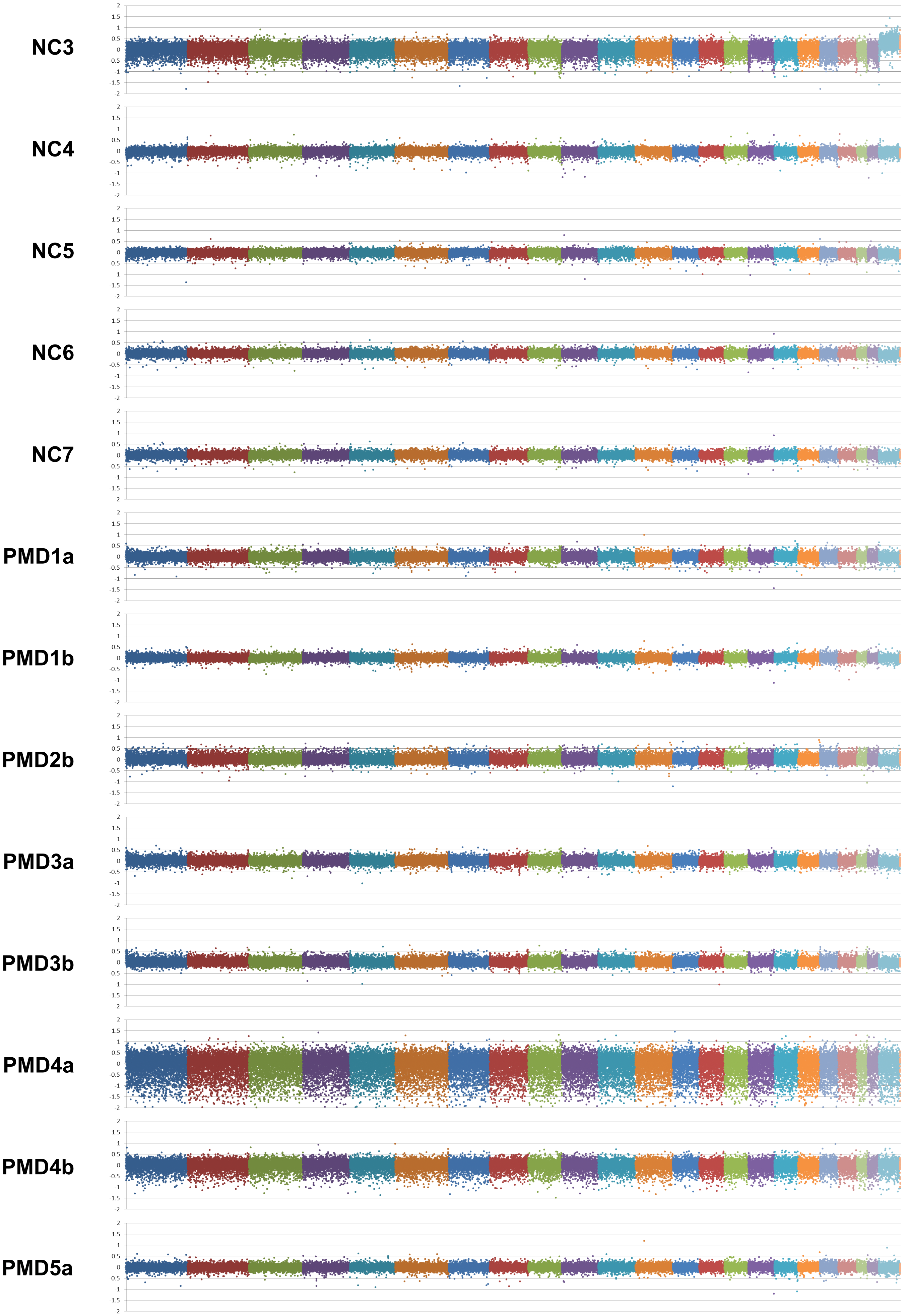
NC3
NC4
NC5
NC6
NC7
PMD1a
PMD1b
PMD2b
PMD3a
PMD3b
PMD4a
PMD4b
PMD5a
2
1.5
1
0.5
0
-0.5
-1
-1.5
-2

Figure S1 (continued).

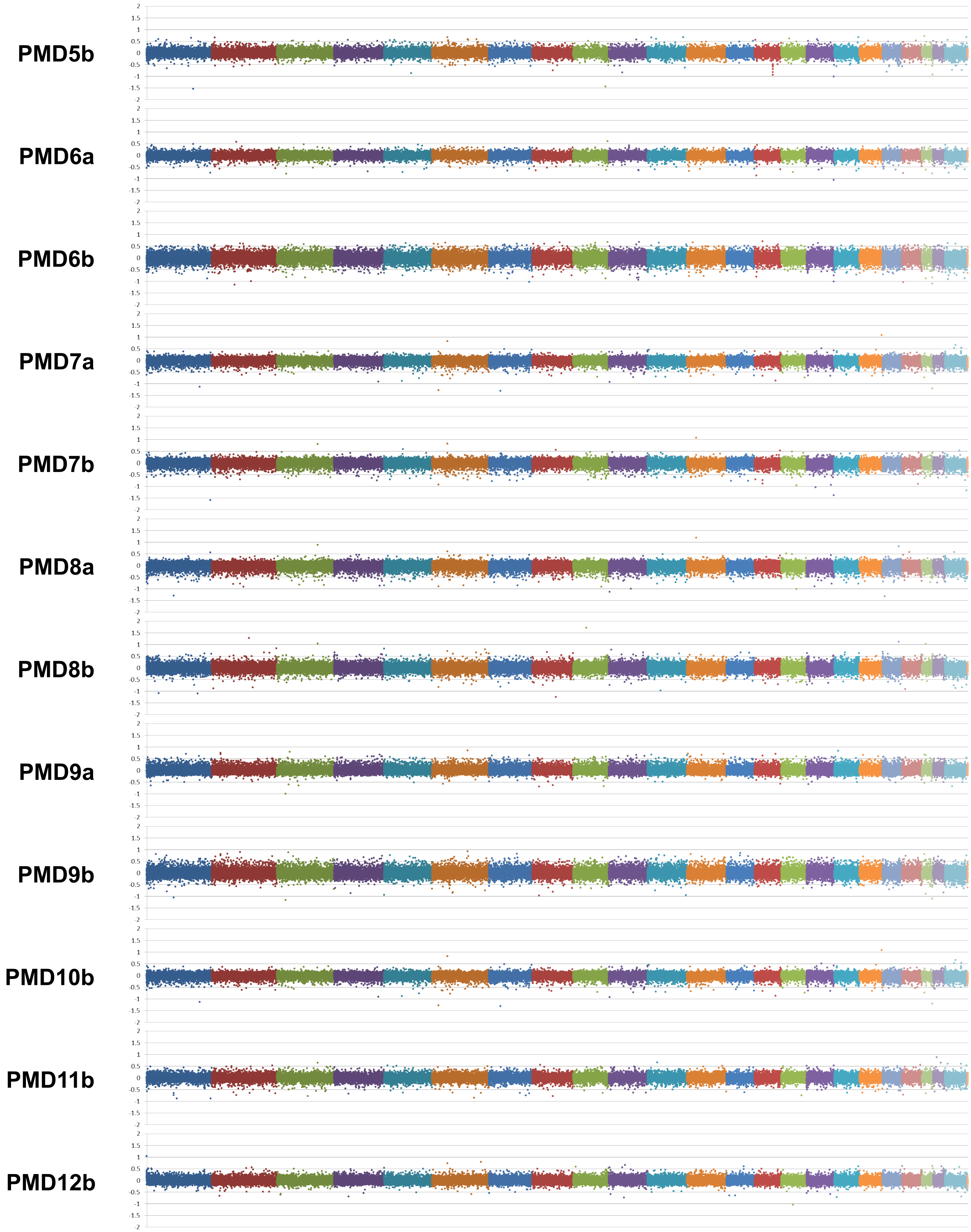

Figure S1. High Density SNP Microarray Analysis
The remainder of the lines not included in Main Figure 1E. Plots demonstrating the gross genomic integrity of derived pluripotent lines. Relative copy number was calculated for each SNP in a high density SNP microarray. Every 100th SNP, arranged by ranked genomic coordinate, was plotted as a Log R Ratio. SNPs are colored by chromosome.

Figure S2.

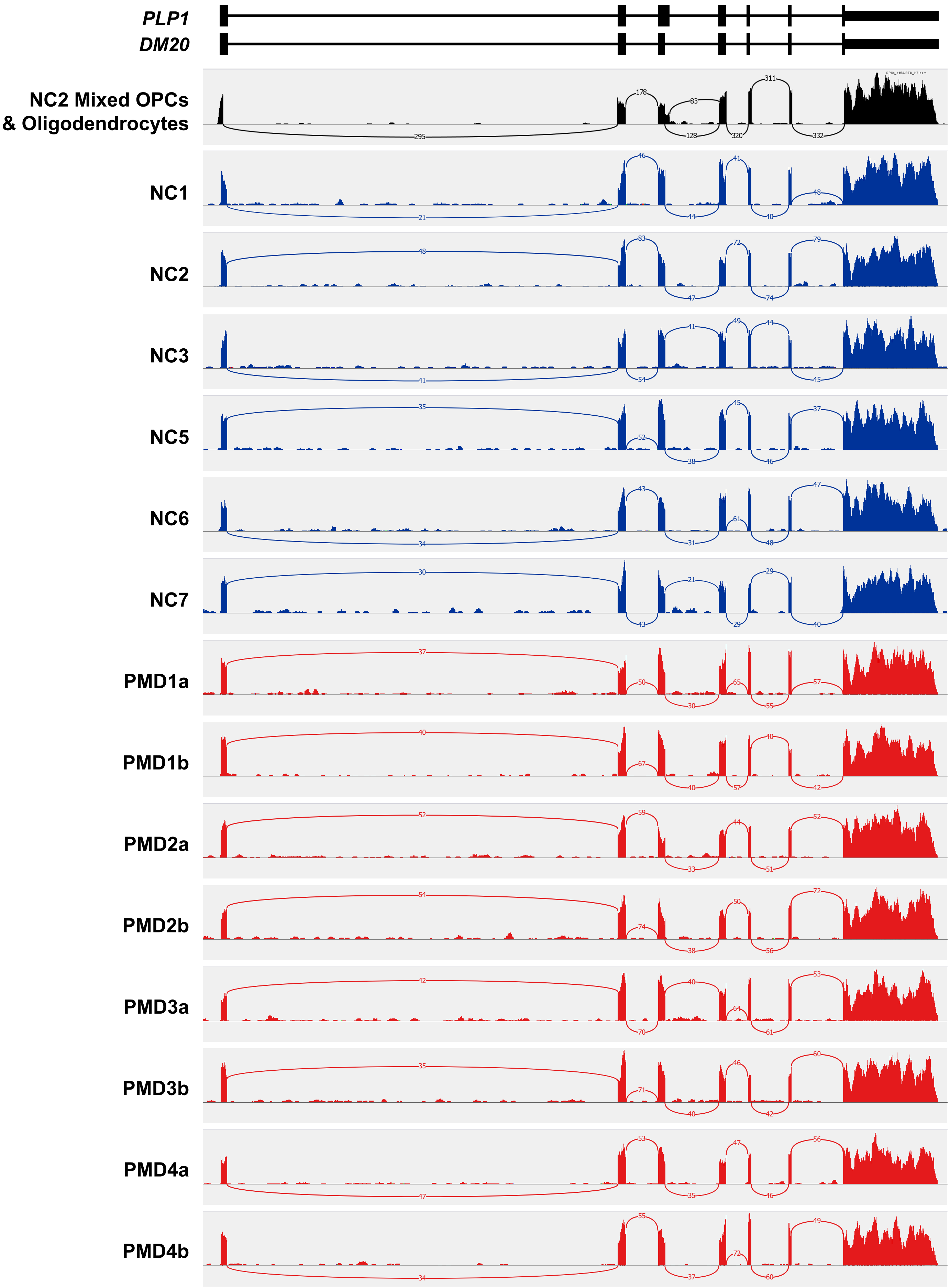
PLP1
DM20
NC2 Mixed OPCs & Oligodendrocytes
178
83
311
295
128
320
332
NC1
46
41
48
21
44
40
NC2
83
48
72
79
47
74
NC3
41
49
44
41
54
45
NC5
35
45
37
52
38
46
NC6
43
47
61
34
31
48
NC7
30
21
29
43
29
40
PMD1a
37
50
65
57
30
55
PMD1b
40
40
67
40
57
42
PMD2a
59
52
44
52
33
51
PMD2b
54
72
50
74
38
56
PMD3a
42
53
40
64
70
61
PMD3b
35
60
46
71
40
42
PMD4a
53
47
56
47
35
46
PMD4b
55
49
72
34
37
60

Figure S2 (continued).

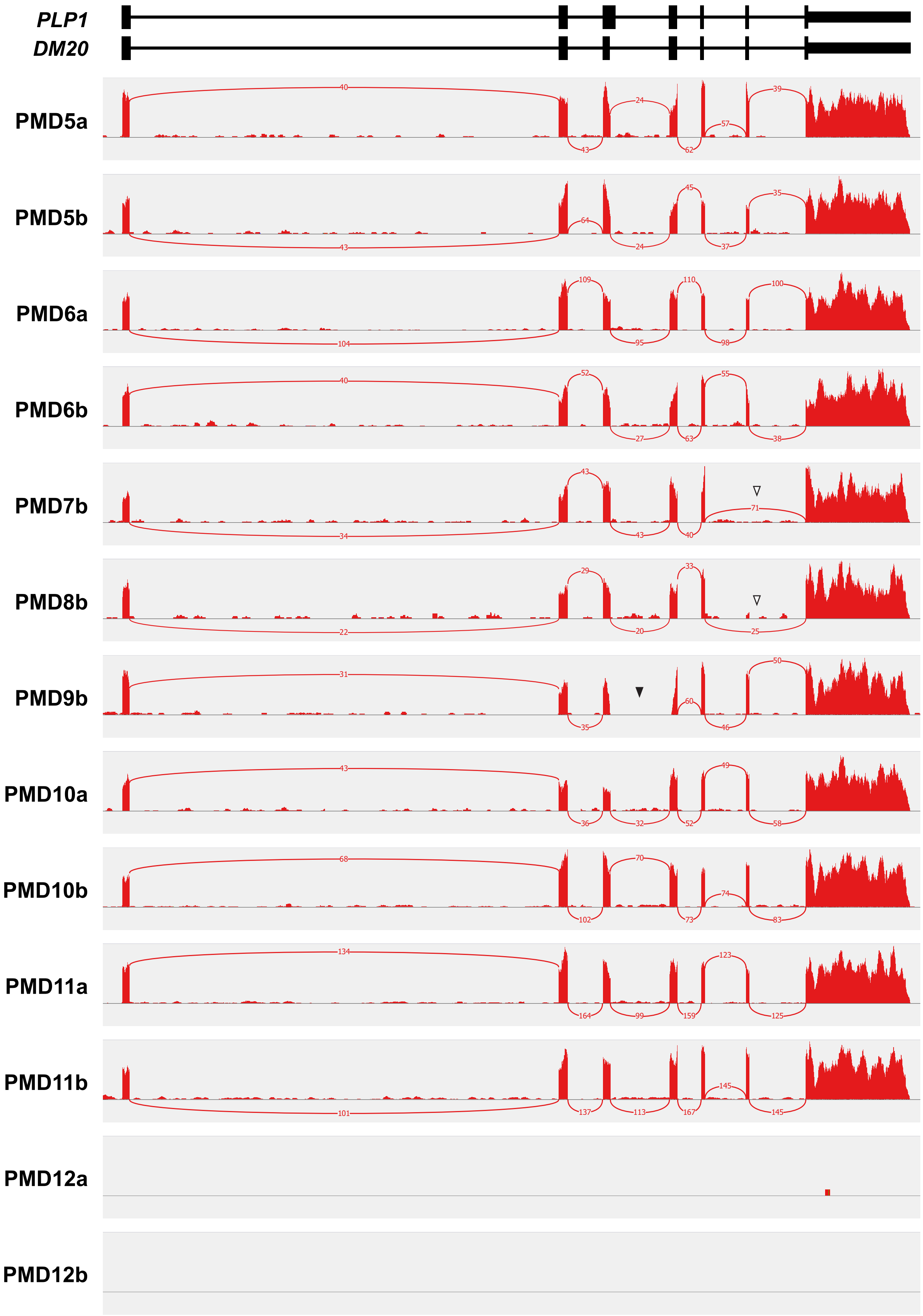

Figure S2. mRNA Splicing Analysis

The remainder of the lines not included in Main Figure 2E. PLP1 mRNA splicing was quantified in PMD hiPSCs and controls using Integrated Genome Viewer's Sashimi plot function. PMD7 and 8 demonstrate skipping of exon 6 (white arrowheads). The exon 3-4 junction cannot be annotated in PMD9 (black arrowhead) due to its partial deletion. For comparison, splicing analysis of day 154 differentiated NC2 mixed OPCs and oligodendrocytes is also provided, demonstrating the presence of both DM20 and PLP1 transcripts.

Table S1. Primers for *PLP1* Exon Sequencing.

PLP1 Exon	Forward Primer	Reverse Primer
1	AAAGCGAAATTCCAGGCAAGC	GATAGAGGGAAGTGAGGGGGT
2	AAGGATTCTGGGTCAATCTCACA	CACAGAGGGAAGACTCGGGA
3	TGGCGGGAGGGGCATATGTTTC	AGACTCGCGCCCAATTTTCCCC
4	GGCTTTGTTCAATGGCTAGGG	GTGGGTAGGAGAGCCAAAGC
5	GGCCATTCACATTGGCCTAC	TCTATGCTCATTGGCTCAGGC
6	CTGGGCACAACTGTAGGGAAC	GCCAATGCAAGTAGAAGTACGG
7	TCCCTGAGGAAAACTCAGTGC	GCAGGAACCAGCTATGAAGCA
3-4 Fusion in PMD9	TTCTCCAGGTCCCAGGGTAAG	AGTGCTTCCATAGTGGGTAGGA

Table S2. Prior Publications Involving PMD Samples in this Study.

Sample	Citation(s)
PMD1	[1]
PMD2	[1]
PMD3	
PMD4	[2]
PMD5	[3]
PMD6	[4; 5]
PMD7/8 (siblings)	[1; 6-8]
PMD9	[9]
PMD10	[1]
PMD11	
PMD12	[1; 10]

Table S3. Details on Control Pluripotent Cell Lines.

Panel ID	Cell Type	NIH Registry ID	Reprogramming Method	Gender
NC1	hESC	H1	--	Male
NC2	hESC	H7	--	Female
NC3	hESC	H9	--	Female
NC4	hiPSC	--	Episomal	Male
NC5	hiPSC	--	Lentiviral	Male
NC6	hiPSC	--	Lentiviral	Female
NC7	hiPSC	--	Lentiviral	Male

Supplemental References

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