

## Supplementary Information

### **Timing and localization of human dystrophin isoform expression provide insights into the cognitive phenotype of Duchenne muscular dystrophy**

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#### **This PDF file includes:**

Supplementary Figure 1

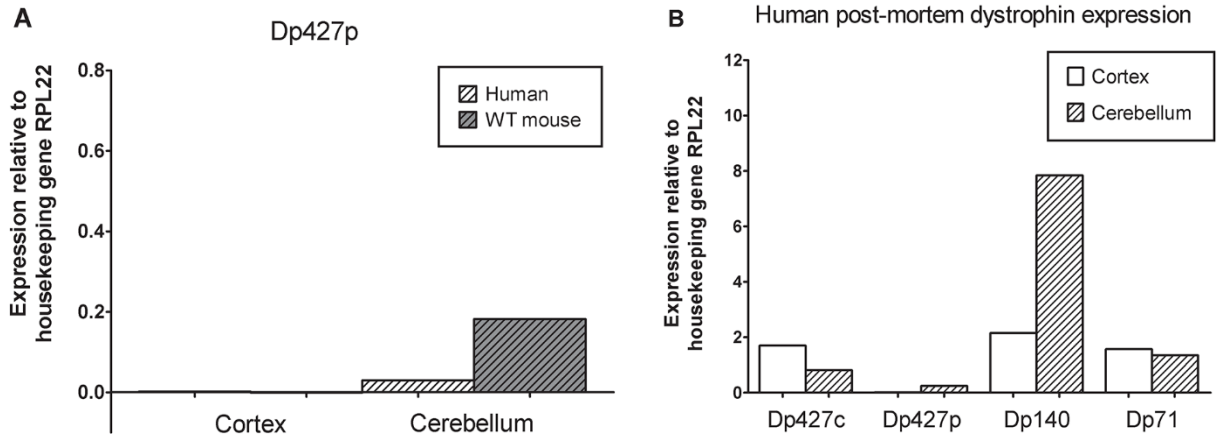
Supplementary Table 1

Supplementary Table 7

Supplementary Table 8

#### **Other Supplementary Material for this manuscript**

Tables S2 – S6 (as Excel files)



**Supplementary Figure 1. Ex-vivo qPCR isoform expression.** Dystrophin isoform expression qPCR results are depicted as a ratio with respect to housekeeping gene RPL22 to allow for comparison. Figure A shows expression of isoform Dp427p in the human and wild type (WT) mouse cortex and cerebellum. Figure B shows the expression of the isoforms Dp427c, Dp427p, Dp140 and Dp71 in the human cortex and cerebellum.

**Supplementary Table 1. Overview of the donor brains included, per age and sex, divided into developmental stages**

Developmental stage			Donors
1	8-9 pcw	Early fetal	8 pcw 1♂, 9 pcw 1♂
2	12-19 pcw	Early-mid fetal	12 pcw 3♀, 13 pcw 2♂ & 1♀, 16 pcw 3♂, 17 pcw 1♀, 19 pcw 1♀
3	21-37 pcw	Mid-late fetal	21 pcw 1♂ & 1♀, 24 pcw 1♂, 25 pcw 1♀, 26 pcw 1♀, 35 pcw 1♀, 37 pcw 1♂
4	4-12 mon	Infancy	4 mon 3♂, 10 mon 1♂, 12 mon 1♀
5	2-4 yrs	Early childhood	2 yrs 1♀, 3 yrs 1♂ & 1♀, 4 yrs 1♂
6	8-11 yrs	Late childhood	8 yrs 2♂, 11 yrs 1♀
7	13-15 yrs	Adolescence	13 yrs 1♀, 15 yrs 1♂
8	18-19 yrs	Early adulthood	18 yrs 1♂, 19 yrs 1♀
9	21-30 yrs	Young adulthood	21 yrs 1♀, 23 yrs 1♂, 30 yrs 1♂
10	36-40 yrs	Middle	36 yrs 1♂, 37 yrs 1♂, 40 yrs 1♀

**Supplementary Table 7. Roadmap Epigenomics samples.**

Sample number	Sample name	Sample annotation
1	Fetal Brain	Fetal Brain
2	Fetal Brain	
3	Neurospheres Cortex Derived	Adult Brain
4	Brain Hippocampus Middle	
5	Brain Substantia Nigra	
6	Brain Anterior Caudate	
7	Brain Cingulate Gyrus	
8	Brain Inferior Temporal Lobe	
9	Brain Angular Gyrus	
10	Brain Germinal Matrix	
11	Brain Mid Frontal Lobe	
12	NH-A Astrocyte Primary Cells	
13	Fetal_Muscle_Trunk	Fetal Muscle
14	Fetal_Muscle_Leg	
15	Fetal Heart	Fetal Heart
16	Skeletal Muscle	Adult Muscle
17	Skeletal Muscle	
18	Left_Ventricle	Adult Heart
19	Right_Ventricle	
20	Right_Atrium	
21	Aorta	
22	Adult Liver	Adult Liver
23	Fetal Kidney	Fetal Kidney

**Supplementary Table 8. qPCR primer sequences**

<b>Target</b>	<b>Species</b>	<b>Forward primer (5'..3')</b>	<b>Reverse primer (5'..3')</b>
<i>RPL22</i>	Human	tcgctcacctcccttctaa	tcacggtgatcttgctcttg
<i>Dp427p</i>	Human	cagcaaaaagctttcctatgaagg	aaattgtgcattaccattttgtg
<i>Dp427c</i>	Human	aggagaaagatgctgttttgca	aaattgtgcattaccattttgtg
<i>Dp140</i>	Human	tgctggctgctctgaactaa	gttctgacaacagtttgccg
<i>Dp71</i>	Human	ctgggaagctcactcctcca	tccagcaagttggttgagtc
<i>Rpl22</i>	Mouse	aggagtcgtgacctcgaac	ttggagaaaggcacctctg
<i>Dp427p</i>	Mouse	tcacagcaactggagtggaa	aattgtgcattatccattttgtga