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Supplemental Data

Molecular Defects in the Motor Adaptor BICD2 Cause Proximal Spinal Muscular Atrophy with Autosomal-Dominant Inheritance

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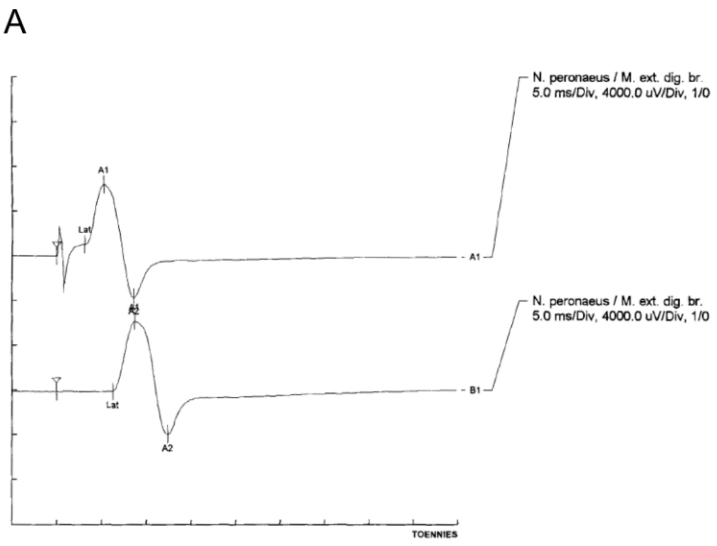
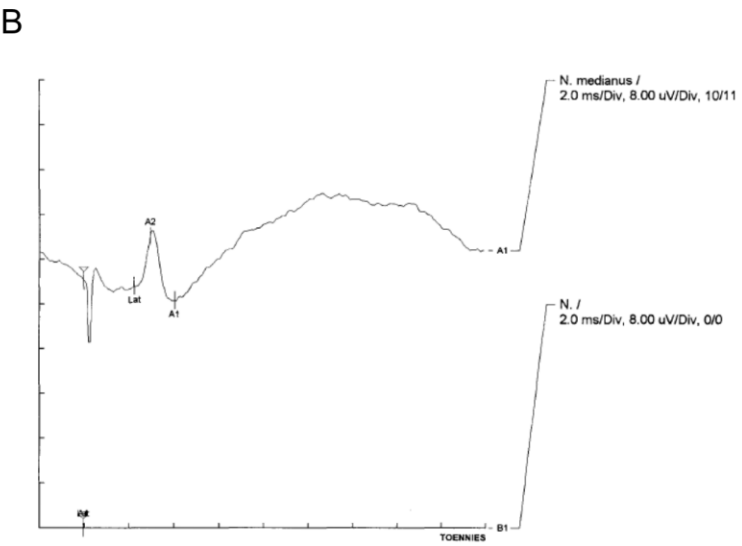


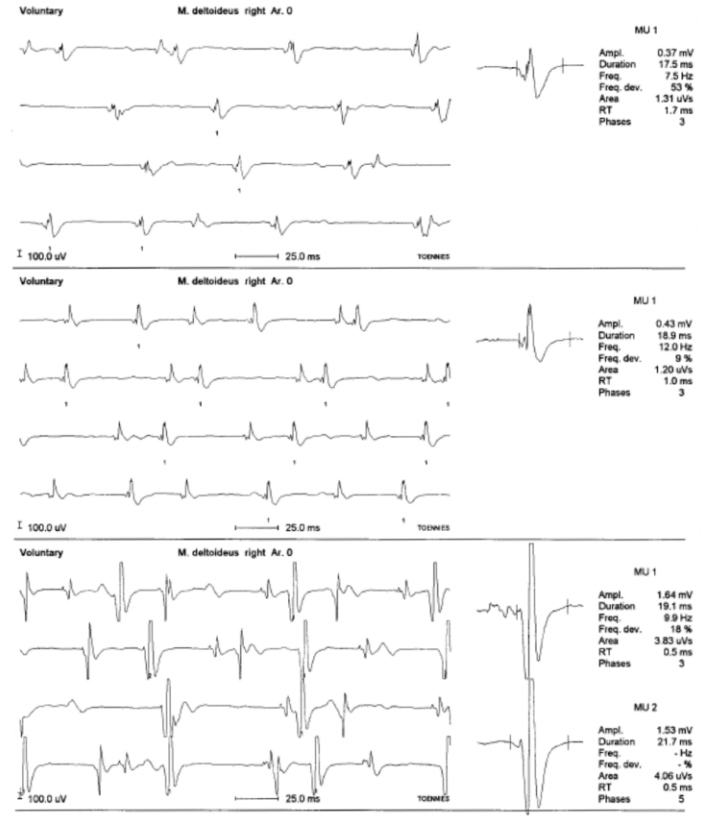
NCV motor:

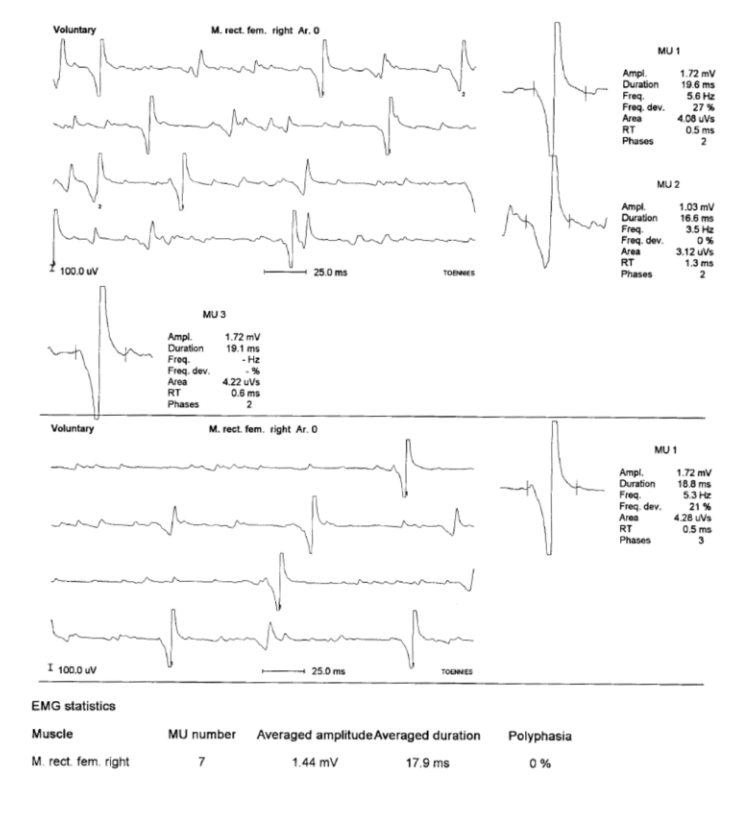
Nr	Nerve	Rec. site	Distance cm	Temp. °C	Lat. ms	Ampl. mV	NCV m/s
A1	N. peronaeus dist R	M. ext. dig. br.	6.5	0.0	3.2	10.1	-
B1	N. peronaeus proximal	M. ext. dig. br.	17.0	0.0	6.3	10.1	54.8



NCV sensory orthodromic:

Nr	Nerve	Rec. site	Distance cm	Temp. °C	Lat. ms	Ampl. µV	NCV m/s
A1	N. medianus dist R		13.0	30.0	2.3	11.6	57.0



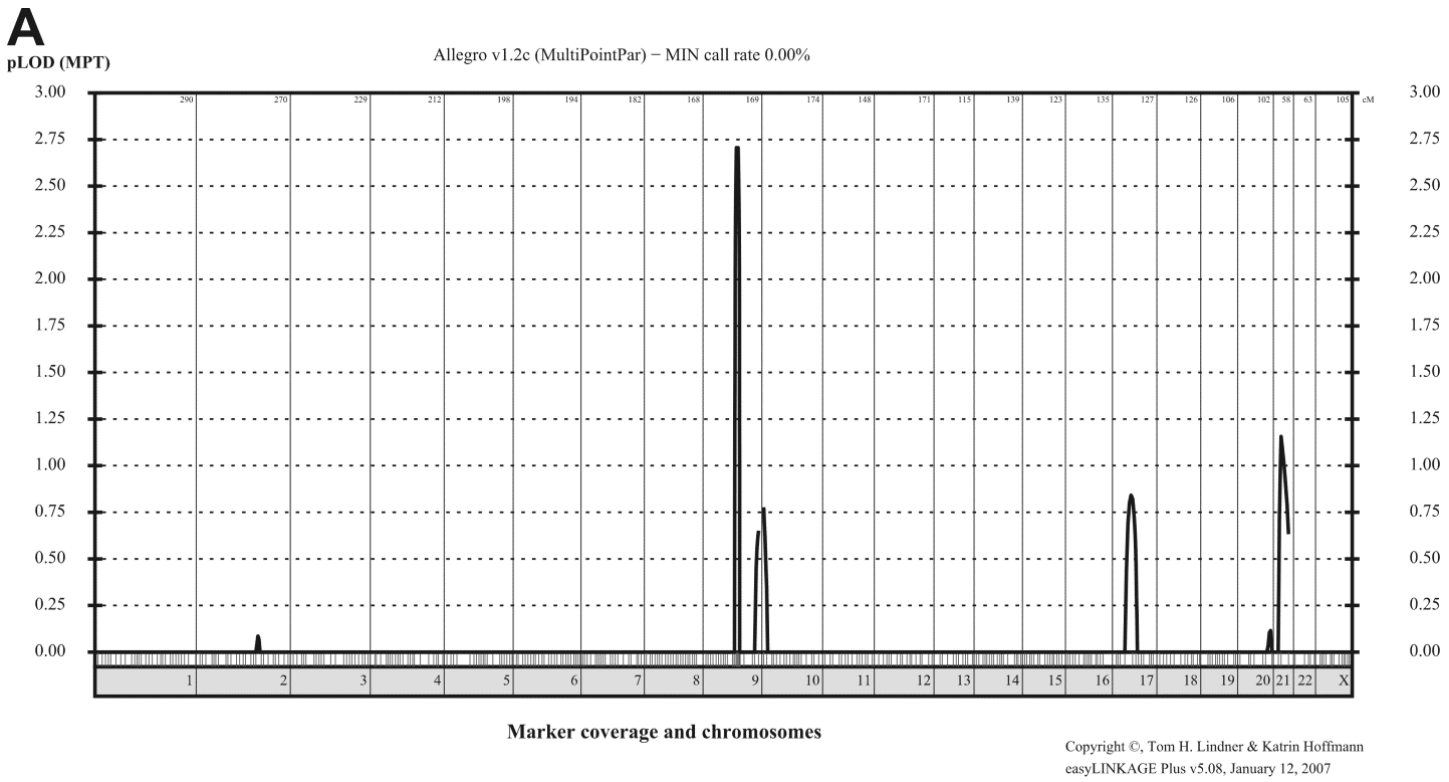


EMG statistics

Muscle	MU number	Averaged amplitude	Averaged duration	Polyphasia
M. rect. fem. right	7	1.44 mV	17.9 ms	0 %

Figure S1. Nerve Conduction and EMG Findings in Individual 177.IV.3

Normal motor (A) and sensory (B) conduction velocities. Anterior horn involvement with giant potentials from EMG of upper (C) and lower (D) limbs.



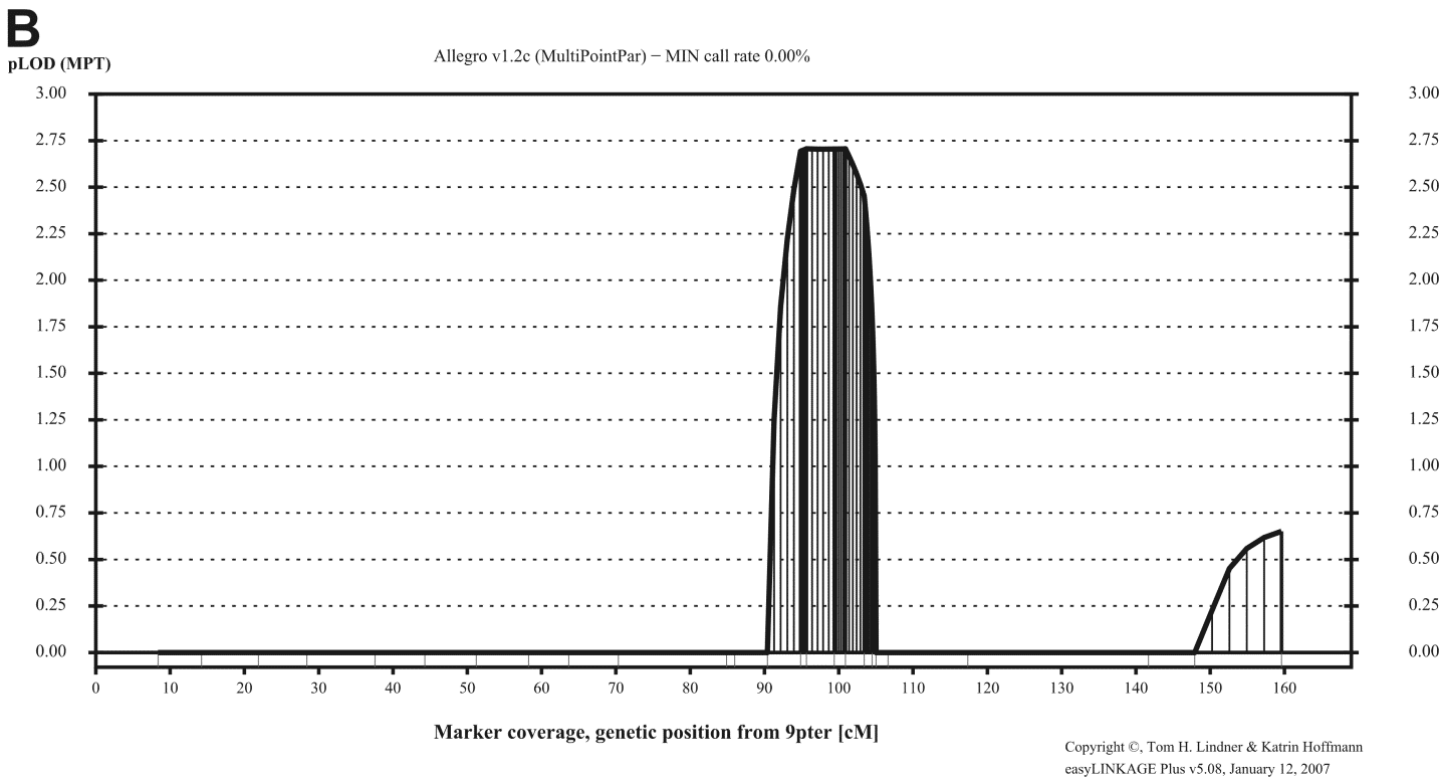


Figure S2. Multipoint Parametric Linkage Analysis in Family 177

(A) Linkage plot of genome-wide analysis with STR markers generated by easyLinkage with Allegro v1.2c and representing the parametric LOD score values on the y-axis in relation to genetic position on the x-axis. Human chromosomes are concatenated from p-ter (left) to q-ter (right) on the x-axis. $pLOD_{max}$ = 2.71 on chr9q. (B) Plot of linkage analysis on chr9 after finemapping with additional polymorphic STR markers. The genetic distance is given in cM.

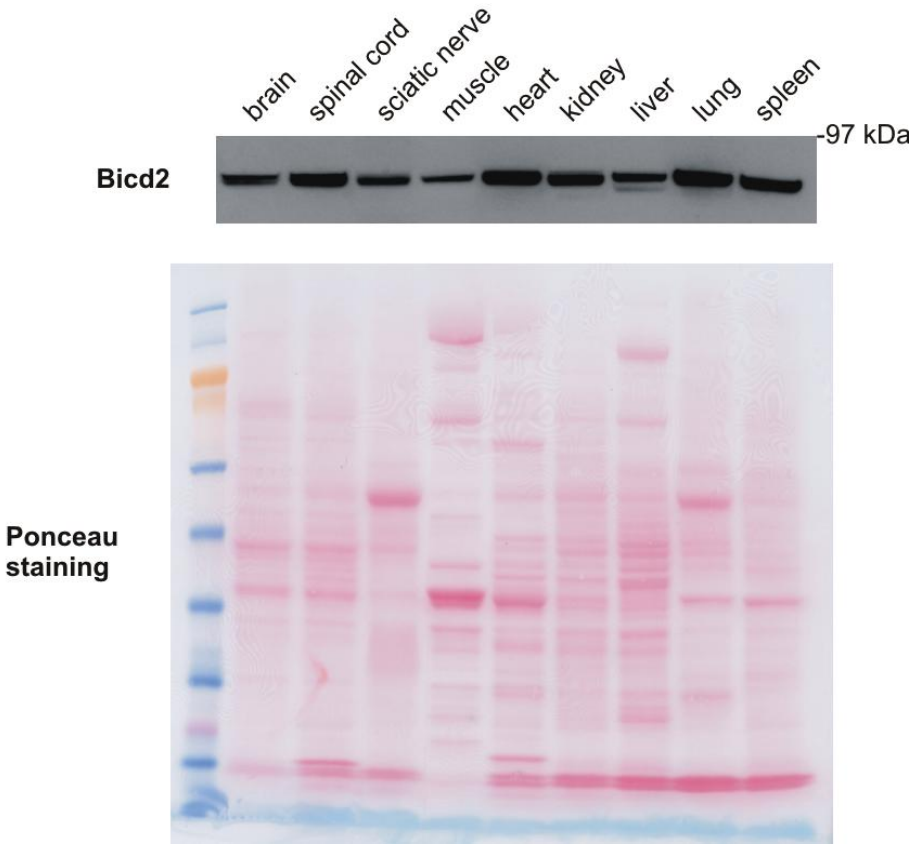


Figure S3. Bicd2 Protein Levels in Mouse Tissues
Immunoblotting analysis showing the Bicd2 protein levels in different mouse tissues. Ponceau staining confirms comparable loading of the samples.