Expanded View Figures

Figure EV1. Neuroimaging of Subjects 1, 2 and 3 reveals signal abnormalities suggestive of mitochondrial disease involvement.

A Axial T2-weighted imaging of Subject 1 at 21 months showed bilateral hyperintense signals in the pons (arrows).
B Coronal T2-weighted imaging of Subject 1 at 21 months showed bilateral hyperintense signals in the thalami and substantia nigra (arrows).
C Axial T2-weighted imaging of Subject 2 at 10 days of age showed bilateral hyperintense signals in the basal ganglia (arrows).
D Sagittal T2 imaging of Subject 2 at 10 days of age showed Dandy-Walker malformation and thinning of the corpus callosum (arrows).
E, F Axial and coronal (respectively) T2-weighted imaging of Subject 3 at 5 months of age showed bilateral hyperintense signals in the thalami (arrows).

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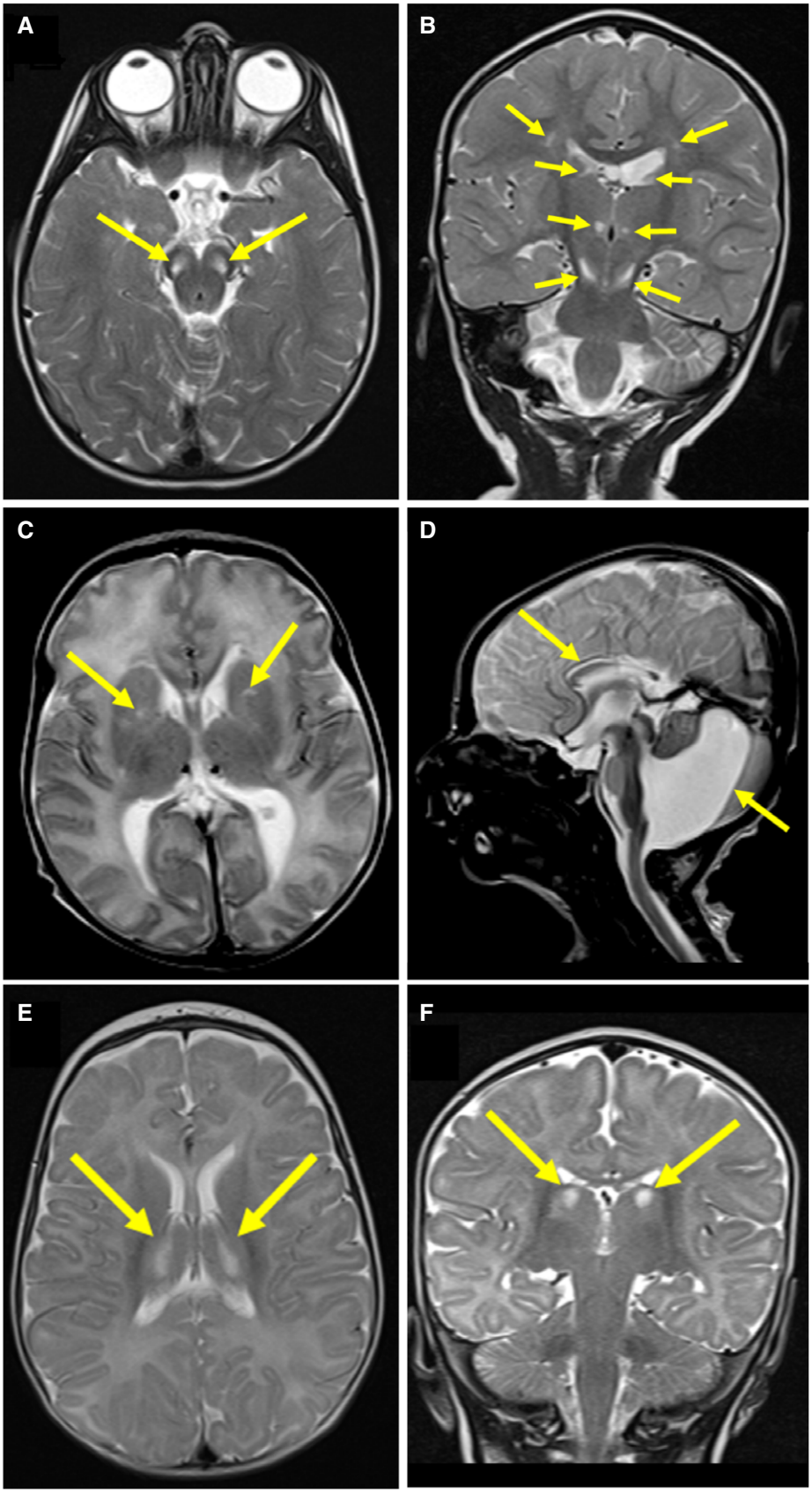

Figure EV1.

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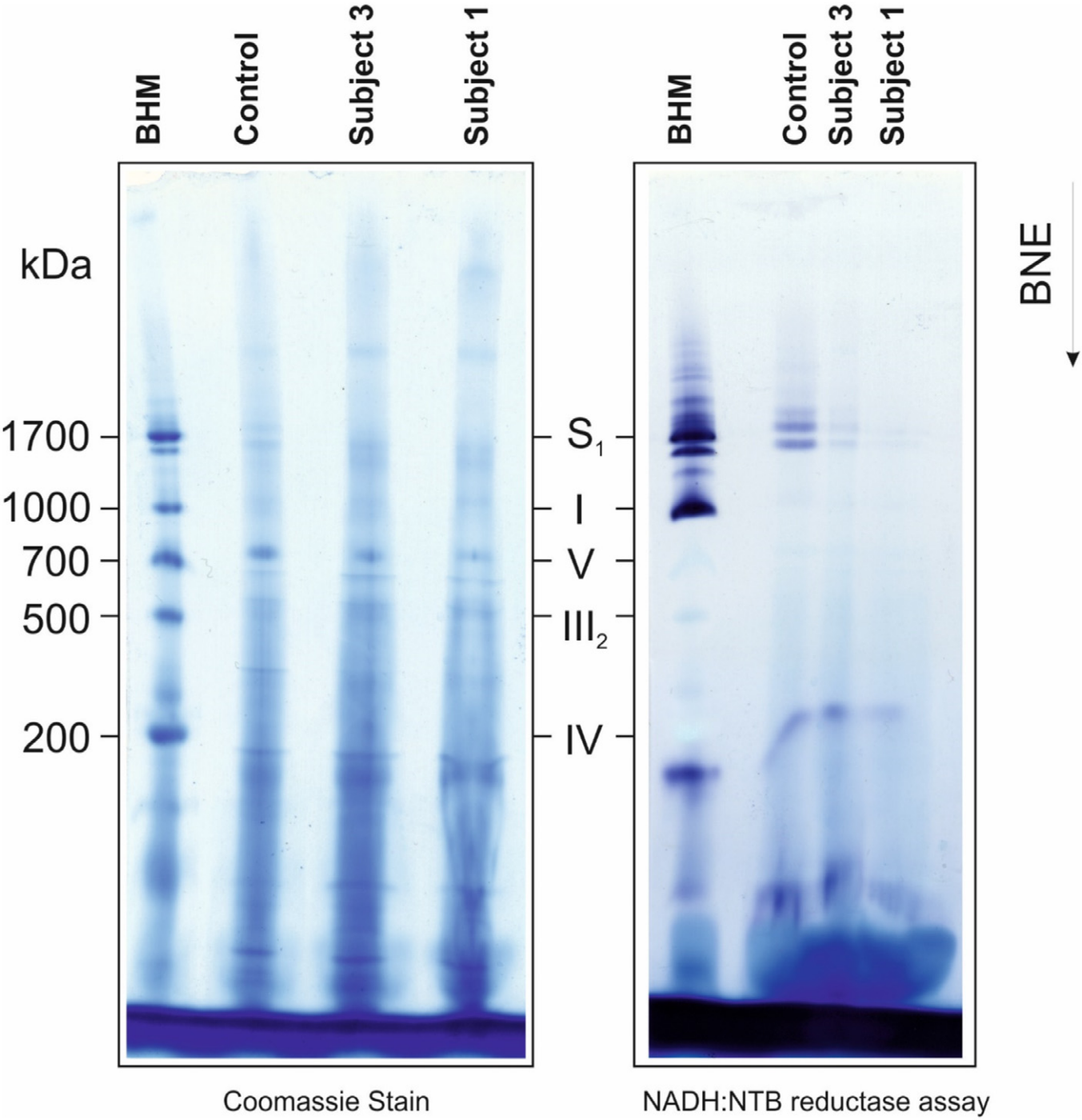

Figure EV2. *NDUFC2* variants cause a severe reduction of complex I-containing supercomplexes.

Coomassie stain (left panel) and an in-gel complex I assay (right panel) in BN-PAGE showed a reduction of complex I-containing supercomplexes in subject mitochondria. Analysis of complex assembly and stability detected a drastic deficiency of complex I-containing supercomplexes.

S_1, supercomplex containing complex I, dimer of complex III and 1 copy of complex IV; V, ATP synthase; I-III-IV, complexes I-III-IV. BHM, bovine heart mitochondria solubilised with digitonin as native mass ladder.

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