Figure E4: Neuropathology of homozygous loss of DIAPH1. A) Brain MRIs of patient F1.P1 at the age of 8 years and 8 months, patient F1.P2 at the age of 1 year, patient F2.P3 at the age of 4 years and 1 month and patient F3.P4 at the age of five days. T1-weighted midsagittal images show microcephaly and thin chiasm (I, arrows) of the patients F1.P1, F1.P2 and F2.P3. T2-weighted axial images of these patients demonstrate abnormal hyperintense occipital subcortical and periventricular white matter and cortex bilaterally (II, arrows). Brain MRI of patient F3.P4 at the age of five days shows normal findings. B) Occipital (upper panel) and lateral (lower panel) view of the brain from neuropathological examination of patient F1.P2. The abnormal area comprised the whole visual cortex or Broadmann areas. C) Hematoxylin-eosin staining of the occipital cortex gyrus (patient F1.P2) showed abnormal neuronal lamination, and NeuN staining of the adjacent gyrus was gliotic and showed total neuronal loss.