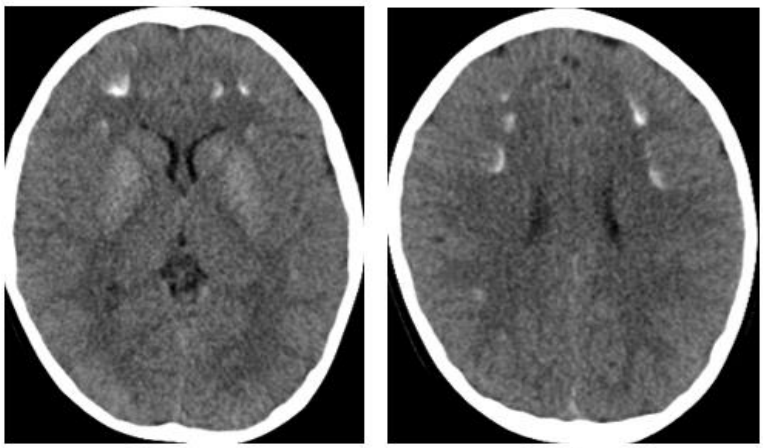
A Case of the 10 days

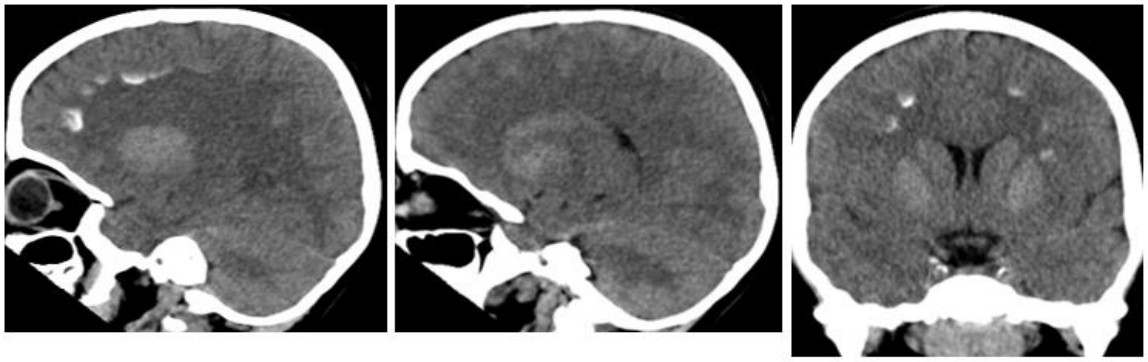
Case 356

This case report is the same as Case 298 which was previously presented as Leigh disease. However, this case was later revised as young-typed MELAS syndrome rather than Leigh disease. Then, we submit this case as a new version.
A four-year-old girl was previously transported to our hospital by ambulance for convulsion and loss of consciousness. At that time, after initiation of lunch in kinder garden, she dropped sliding down from a chair with face redness and tonic convulsion lasting for around 10 minutes. Laboratory test revealed AST 48 U/l, ALT 28 U/l, ALP 542 U/l, and lactic acid 48 mg/dL.
She had experienced convulsion previously two years ago. She had been admitted in our hospital several times not just for convulsion but also for consciousness disorder, hypo-glucosemia, and hyper-lactic acidemia. She took brain CT and MRI for further investigation (Figs. 1-3). There was a calling from university hospital that her gene investigation revealed she had deficit of KSS regarding mitochondria abnormality, indicative of young type MELAS.



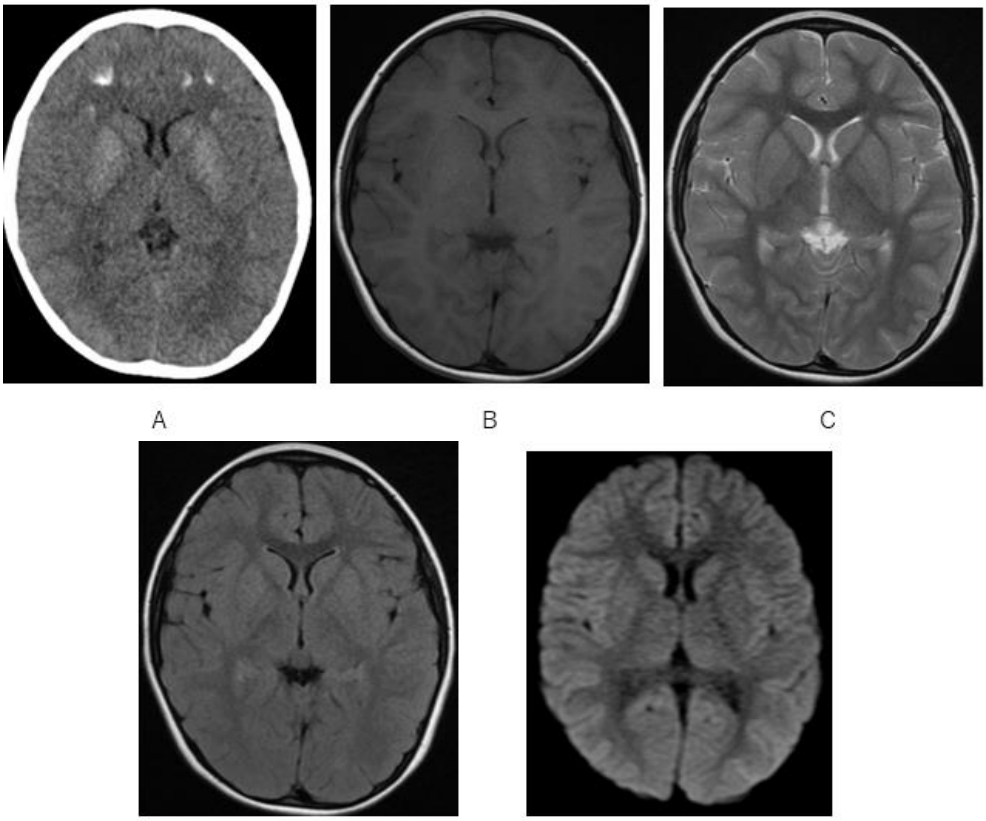
A B

Fig. 1 Axial images of brain CT depict high attenuation areas of cortex adjacent to deep white matter in frontal lobe (A, B) and slight high attenuation of caudate nucleus and basal ganglion (A).



A B C

Fig. 2 Sagittal (A, B) and coronal (C) CT images depict high attenuation areas of cortex adjacent to deep white matter in frontal lobe (A, C) and slight high attenuation of caudate nucleus and basal ganglion (A-C).



A B C D E

Fig. 3 High and slight attenuation areas visualized on CT (A) are not demonstrated on T1WIMRI (B), T2WIMRI (C), FLAIRMRI (D), Diffusion WI (E).

What is the difference of abnormality between MELAS and Leigh disease?

1. **Incidence of abnormal nuclear DNA**
2. **Incidence of abnormal mitochondria DNA**
3. **Incidence of abnormal nuclear mRNA**
4. **Incidence of abnormal nuclear tRNA**

answer

2024.10.4