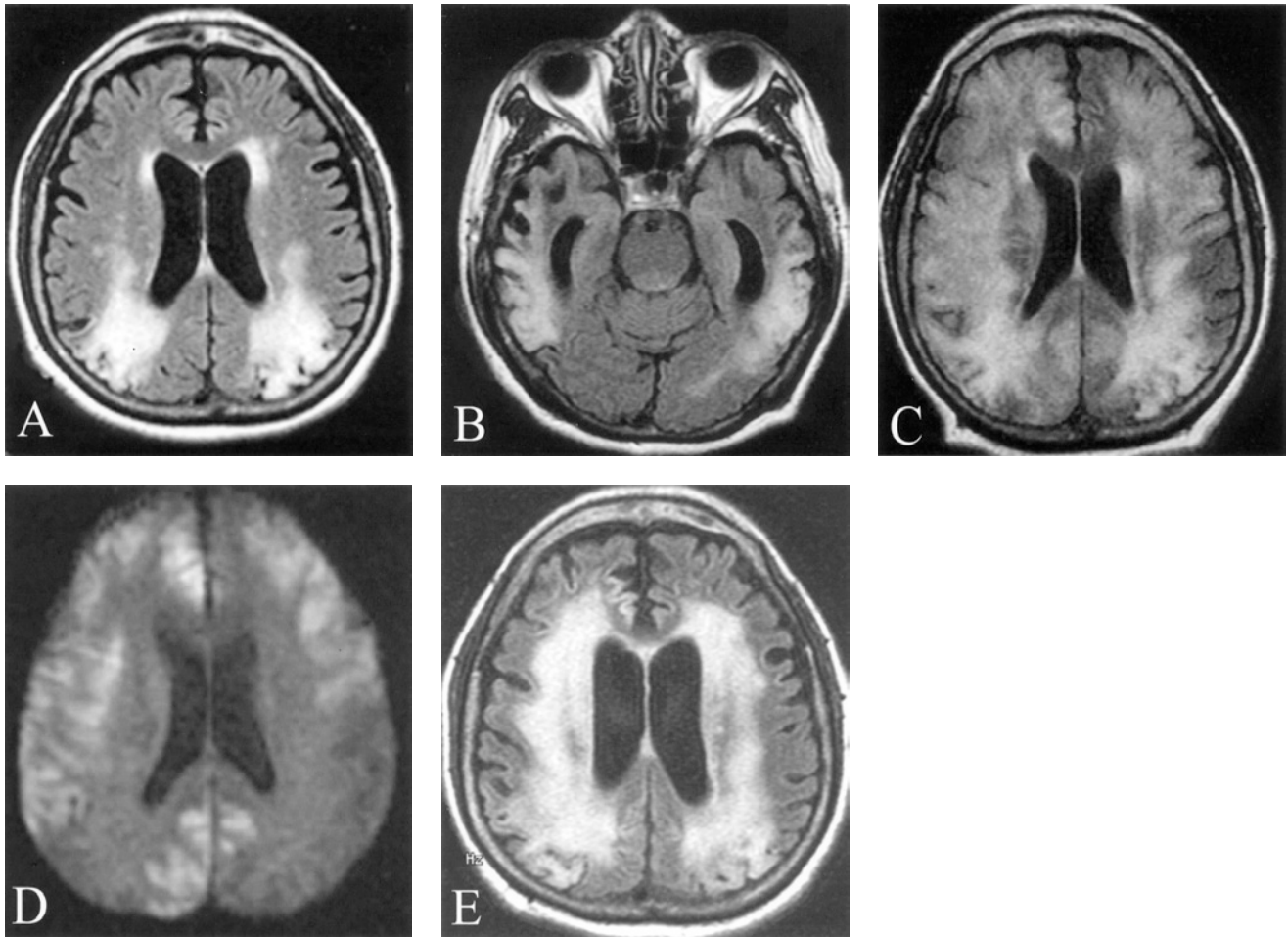


## MRI of Adult-onset Type II Citrullinemia

**Key words:** citrullinemia, CTLN2, MRI, pseudoulegyria



**Figure 1.** Fluid attenuated inversion recovery (FLAIR) images (A, B) on admission showed diffuse atrophy and hyperintense bilateral temporo-occipital lobe. With acute exacerbation, severe edema was found on FLAIR (C) and diffusion-weighted images (D). Two months later, the progression of atrophy with more widespread hyperintensity areas was revealed on FLAIR images (E).

A 56-year-old woman was admitted with repeated episodes of consciousness disturbance and progressive dementia, and bilateral temporo-occipital hyperintensity on MRI (Fig. 1A, B). Thereafter, she acutely developed consciousness disturbance, showing elevation of plasma levels of ammonia, citrulline and arginine, and extensive brain edema (Fig. 1C, D). The liver biopsy revealed a selective decrease of argininosuccinate synthetase, leading to the diagnosis of adult-onset type II citrullinemia (CTLN2). Two months later, the brain showed severe atrophy (Fig. 1E). Neuropathologically, CTLN2 shows the characteristic cortical atrophy, designated as “pseudoulegyria”, and severe edema in exacerbations (1, 2). Our serial MRI demonstrates how the “pseudoulegyric” changes progress.

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