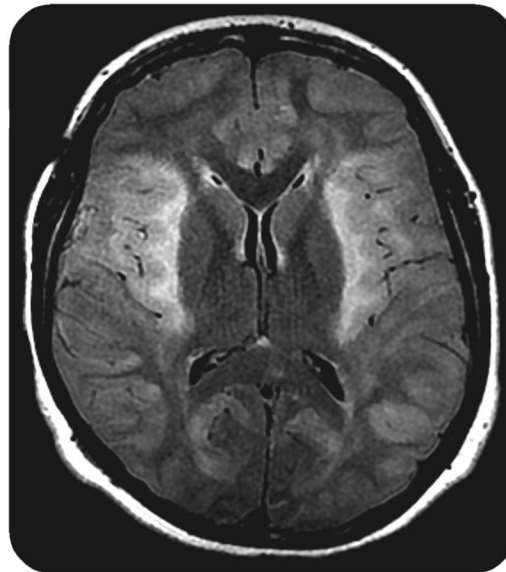


# Teaching NeuroImages: Ornithine transcarbamylase deficiency revealed by a coma in a pregnant woman

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**Figure 1** T2 fluid-attenuated inversion recovery axial MRI



Bilateral temporal, peri-insular, and frontal cortex hypersignal and thickening, with thalamic sparing. Subcortical U fibers were affected.

A 32-year-old pregnant (20 weeks of amenorrhea) woman, after change in dietary intake (due to a trip to Korea), developed over 3 weeks progressive neuropsychological disorders associated with behavior disorders. The patient rapidly worsened, and had impaired consciousness and a coma. Biological tests showed hyperammonemia (173  $\mu\text{mol/L}$ ), glutamine chromatographic peak, and increased urinary orotic acid concentration. Brain MRI (figure 1) and spectroscopy (figure 2) findings were consistent with urea cycle disorders.<sup>1,2</sup> A novel heterozygous mutation p.Ala209Glu (c.626C>A) in *OTC* gene was identified. She was treated with sodium benzoate, sodium phenylacetate, citrulline, hemofiltration, and reduced protein intake. Consciousness improved, and pregnancy was carried to completion, but mild cognitive impairment persisted 3 months later. The baby girl also carried the mutation but had no sequelae at 11 months.

## AUTHOR CONTRIBUTIONS

Pierre Bailly and Jean-Baptiste Noury took care of the patient in the ICU and wrote the article. Serge Timsit's opinion was requested to confirm diagnosis. He read and corrected the article. Douraid Ben Salem performed the MRI. He read and corrected the article.

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No targeted funding reported.

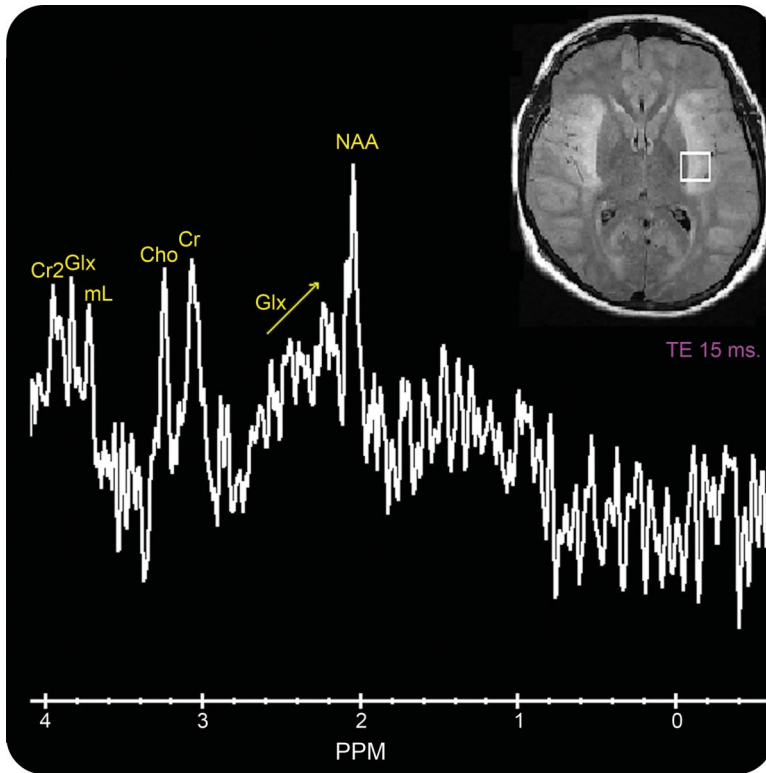
## DISCLOSURE

The authors report no disclosures relevant to the manuscript. Go to [Neurology.org](http://Neurology.org) for full disclosures.

## REFERENCES

1. Bireley WR, Van Hove JL, Gallagher RC, Fenton LZ. Urea cycle disorders: brain MRI and neurological outcome. *Pediatr Radiol* 2012;42:455–462.
2. Gropman A. Brain imaging in urea cycle disorders. *Mol Genet Metab* 2010;100(suppl 1):S20–S30.

Figure 2 Magnetic resonance spectroscopy



Increased levels of glutamine-glutamate (Glx) and slight decrease in levels of myo-inositol (mI). The arrow indicates Glx peaks. Typical MRI and MRS findings in ornithine transcarbamylase deficiency are T1 and T2 hypersignals, located in the cerebral cortex, preferentially in peri-insular regions and the basal ganglia and an increase in Glx. This is due to the excess of ammonia and is not pathognomonic of urea cycle disorders. Cr = creatine; Cho = choline; NAA = N-acetylaspartate; TE = echo time.

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